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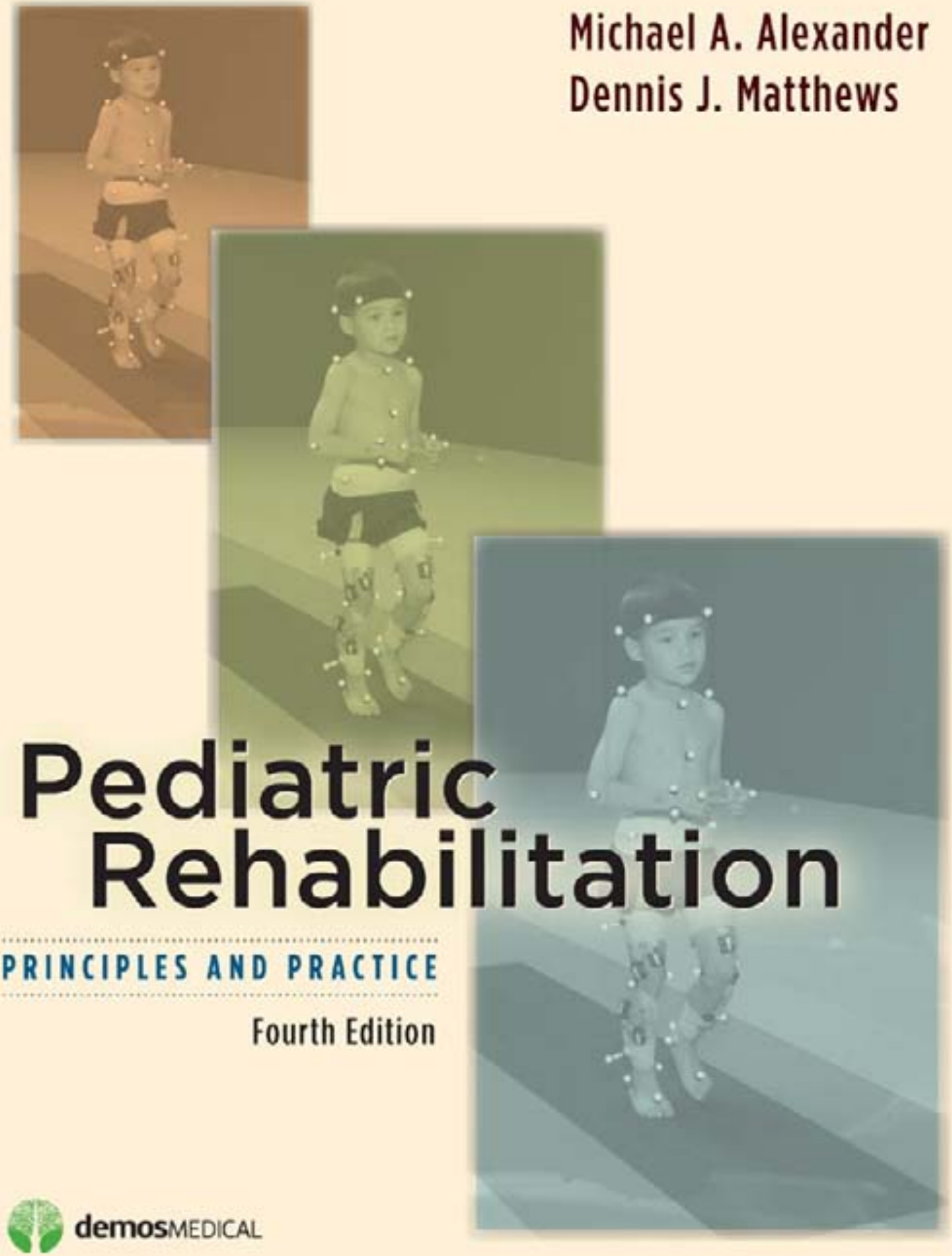

Pediatric Rehabilitation

PRINCIPLES AND PRACTICE

Fourth Edition

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Pediatric Rehabilitation

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FOURTH EDITION

Pediatric Rehabilitation

Principles and Practice

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Dedication

To Dr. Gabriella Molnar, a recognized founder of our field of pediatric rehabilitation medicine. Dr. Molnar created our first textbook, edited the subsequent two editions, and wrote numerous state-of-the-art textbook reviews for the Child with Physical Disability. After escaping from Hungary in 1956 from the Russian occupation and communist regime, Dr. Molnar displayed much foresight and courage throughout her professional career. Her guiding principle has always been that children are not miniature adults, but individuals with changing physical, intellectual, and emotional abilities and needs. At every age, therefore, the principles of rehabilitation medicine have to be adapted to these changing aptitudes. Beginning as a resident at Albert Einstein College of Medicine, Dr. Molnar quickly rose through the ranks from faculty instructor to full tenured professor, while developing and running the Pediatric Rehabilitation Medicine Service. Concluding her career at Children's Hospital and Research Center in Oakland, California, where she created a new Department of Pediatric Rehabilitation Medicine, she finished training her last of over 50 domestic and international fellows. Her speaking career has included invitations from all over the world, including Australia, Europe, Asia, and England. She has served on the editorial boards for the *Archives of Physical Medicine and Rehabilitation* from 1976 to 1994 and *Developmental Medicine and Child Neurology* from 1992 to 1997. She is a recipient of the Krusen Award from the American Academy of Physical Medicine and Rehabilitation (AAPMR), the highest honor obtainable for proven performance in clinical expertise, contributions to the literature, and administration in the field of rehabilitation medicine. Simply stated, Dr. Molnar defines the standard for the rest of us to follow.

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Preface

This is the first edition of *Pediatric Rehabilitation* that has not been under the leadership of Dr. Gabriella Molnar, who is happily retired and living with her "cowboy" in Oklahoma. She has passed the torch to me and Dennis Matthews and we have been stimulated by the challenge. For the most part we have kept the basic structure of past editions while making changes that reflect the advancement of pediatric rehabilitation.

Our field is changing and so we have streamlined some topics and added new chapters on gait labs, as many of us are actively involved in this exciting adjunct to our practice of medicine. We have added a new chapter on aging with pediatric onset disability that will be of great interest to physiatrists.

Medicine and rehabilitation are changing. Many of us see very few arthritis patients. Our rheumatology colleagues are doing a fantastic job with these children and so the need for rehabilitations has decreased. Leg lengthening, limb salvage procedures, limb reattachment, and improved safety of farm machinery have decreased the number of children who need prosthesis. Genetic testing has altered the referral patterns for children who need electromyography. Bladder continence and bowel irrigation surgeries have changed how we manage patients with spinal cord injuries.

More of us are seeing children with concussions as the literature proves the value of screening and following the children serially. Our options for spasticity management are different and more widely used than in the last edition of this book.

We are constantly challenged by children who are surviving cancers to assist in their rehabilitation. Patients with solid organ transplants benefit from our services. Our patients seem sicker, spend less time on our inpatient units, and are now managed in day programs. The chapter authors have diligently incorporated these issues and many more.

A new feature of this edition is "Pearls and Perils" of caring for different types of patients. These pearls and perils are important take-home points some of the authors have for you.

You will notice that some chapter authors have returned and we have asked them to incorporate new pediatric rehabilitation specialists, as it is our hope that these new coauthors will become the senior authors of future chapters and perhaps editors of future editions.

We are happy to present to you this compiled wisdom of the brightest and most enthusiastic clinicians in our tightly knit group of pediatric rehabilitation specialists.

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Pediatric Rehabilitation

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1

History and Examination

Michael A. Alexander and Gabriella E. Molnar

The physiatric history and examination of a child require a blend of medical diagnostic skills to establish or confirm the diagnosis as well as a knowledge of child development and behavior to evaluate functional assets and difficulties for the intervention phase of rehabilitation.

SETTING THE TONE

To ensure the best cooperation, especially in the preschool age, the environment should be child-friendly. Exposure to crying and upset children should be avoided in the waiting room or other areas. If the family brings the child's siblings, someone should take care of them during the appointment so that the parents can focus on the interview without distraction. The examination room should have a small table and chair with an assortment of toys for different ages to make the child comfortable and relaxed. The examiner's attire also influences the child. A good rule is to "lose the white coat." The child is not impressed by it and, in fact, may be intimidated from past medical visits. Pictures of cartoon characters or animals on the wall, small toys, and decals on instruments help to create a playful atmosphere and alleviate the child's fears.

Start the visit by introducing yourself, which should include telling the patient and parents something about yourself, and what will be happening during the visit, and for how long, and then asking the parents to tell in their own words why they came and what specific questions they have for you.

Concerns stated by the referral source should be shared with the parents. Many parents are unsure about what information the visit can provide. This is the opportunity to explain what pediatric rehabilitation is and what it can offer the child and family. The examiner also should explain that it is part of the examination to watch the child so that the parents will not feel offended by the examiner's wandering gaze. Because observation of spontaneous behavior is one of the most informative aspects of evaluating youngsters with a disability, examination begins from the moment the child is in the physician's view. Questions about history and illnesses should be asked in simple terms so that the family can understand them and provide proper information. It is also important to clarify insurance coverage and whether additional tests can be performed on the same day or must await approval.

HISTORY

Prenatal and Perinatal History

The prenatal and perinatal history includes the preconceptual period and the parents' ages and health before and since the birth of the child. Maternal

factors during gestation may lead to fetal malformations. Examples of these associations include febrile illnesses (1) anticonvulsants (2) with spina bifida; maternal diabetes with caudal regression syndrome and sacral agenesis; and rubella, thalidomide, or fetal alcohol syndromes. Feeble or eventually lost fetal movements may be the earliest sign of a motor disability of prenatal origin. Prenatal care, unusual weight gain or loss, hypertension, or any other gestational problems should be explored. Mode and duration of delivery, use of anesthesia, induction, intrapartum complications, and expected and actual date of birth should be noted. History of previous pregnancies, deliveries, and fetal loss is necessary. Prenatal cerebral damage seems to be increased in infants of mothers with previous spontaneous abortions (3). A detailed neonatal history is essential, including birth weight, Apgar scores, onset and success of breastfeeding, and the infant's age at discharge. Weak lip seal and sucking force and inadequate feeding may be preliminary signs of oral motor dysfunction. If the infant needed admission to the neonatal intensive care unit (NICU), what were the problems, medications, and supportive measures? Neonatal seizures may signal pre- or perinatal brain damage. Prematurity, particularly very low birth weight, is a frequent cause of cerebral palsy (2). Large birth weight may lead to intrapartum trauma, brachial plexus palsy, or, on rare occasions, spinal cord injury, particularly with breech or other fetal malposition. When extended hospitalization was required, one should note the infant's age, weight, and condition on discharge, including means of feeding and need for ventilatory or other supportive measures at home, which may predict subsequent, persistent, or recurrent problems.

Developmental History

The developmental history should cover all major aspects of function and behavior. For details of developmental milestones and testing, the reader is referred to Chapter 2. This discussion presents only guidelines for the purpose of diagnostic interpretations. Discrepancies between different areas of functioning provide clues about the nature of medical diagnosis and developmental disability.

Delayed accomplishments, primarily in motor function, suggest a neuromuscular deficit. One of the earliest signs that parents report is a lack of spontaneous movements when the infant is held or placed in the crib. They may add that the baby feels limp or stiff, suggesting hypotonia or spasticity. In all cases of motor dysfunction, it is important to clarify whether the dysfunction was a steady, continuing delay from an early age, suggesting a static disease, or an arrest or regression noted at a particular point. However, the relatively fast pace of early motor development may mask slow deterioration due to progressive neurologic disease for a while.

Developmental history and subsequent assessment must take into consideration the interactive effect of coexistent deficits. A significant cognitive dysfunction by itself may delay gross and fine motor development (4). It also tends to enhance the functional consequences of a neuromuscular disability. Slow development in personal and adaptive tasks that require both motor and cognitive abilities may be related to impairment in either area. A combination of both can create the impression that the motor deficit is more severe than it actually is.

A history of delay in communication development raises several differential diagnostic possibilities: (a) true language dysfunction affecting receptive or expressive domains or both, (b) oral motor dysfunction interfering with speech production, and (c) significant hearing loss. In a child with motor disability, language dysfunction may result from diffuse or focal cerebral lesions, such as head injury or cerebral palsy, particularly when cognitive function is also affected.

The ability to follow simple and, at a later point, complex commands indicates preserved receptive language even in the absence of verbalization. Parents report a variety of responses, such as smiling, cooing, crying, pointing, or vocalization with inflection as a substitute for speech. Oral motor dysfunction is also associated with cerebral palsy, most often with spastic quadriparesis or dyskinetic disorders due to suprabulbar or pseudobulbar palsy. Bulbar palsy in medullary involvement affects speech production, for example, in spinal muscular atrophy or spina bifida with syringobulbia. There is a close association between anatomical structures and neurologic control for speech and oral feeding. Concurrent oral motor dysfunction with feeding difficulties is an additional sign of bulbar or pseudobulbar pathology and confirms the suspicion of speech production deficit. In such cases, history of early feeding is most relevant. For example, was there a good lip seal and strong suction on breastfeeding? When bottlefed, the infant can handle 4 ounces in about 10 minutes, and feedings every three to four hours are generally adequate. The need for longer and more frequent feeding to maintain weight gain, especially during the first few months; coughing; nasal regurgitation of liquids; difficulty with drinking from a cup; and difficulty with introduction of solid food due to chewing problems are early symptoms of oral motor dysfunction and a possible subsequent deficit in speech production. Augmentative communication training should be initiated early in such cases.

Hearing is an essential factor for speech development. Early cooing and babbling are innate characteristics of infants and involve the same vocal components, regardless of the language spoken in their environment. Infants with hearing loss start to fall behind after six to eight months of age when learning of auditory-dependent vocalization begins. Parents may notice a decrease even in spontaneous babbling at that age. All neonates and infants at high risk for developmental disability or recurrent ear infections should have an initial and, if warranted, repeat hearing evaluations. Correction of a hearing deficit should be provided as soon as possible after it is detected (5).

For infants and young children, the history is obtained from parents or caretakers. While gathering information from one person about another, the examiner gains an understanding of both and establishes rapport with parents and child. Early school-aged children can provide some information about themselves and should be encouraged to do so. Preadolescents and particularly adolescents generally prefer to give an account of their problems and achievements. Adolescents often wish to have privacy without the parent present, at least for part of the visit.

General Health History

The examiner should determine whether the patient is an essentially well child with impairment or a sick child who has been hospitalized several times. In the latter case, one should explore in detail the frequency, reasons, tests, and treatments. Even if one has access to records, the parents should be asked to tell the child's history in their own words. Their account provides an insight into their knowledge and participation in the child's care. One should ask how many visits they make to medical centers and therapists and how much time is spent in transit for the child's care.

History of allergies to medications or other substances should be noted. An early history of allergies to different and often inconsistent formulas may indicate that the child in fact had feeding difficulties that were attributed to allergy. Multiple exposures to latex and any signs of allergy should be determined, particularly in spina bifida or after repeated surgeries. Any medications that the child takes regularly, including dietary supplements and homeopathic or alternative medications or aerosols, should be recorded with dosage and schedule.

The risk and incidence of seizures are higher in static and progressive diseases of the central nervous system. Overt or suspicious signs, type and frequency of seizures, anticonvulsants, and their effectiveness and possible side effects should be recorded.

Nutrition, with special consideration for the child's disability, should be reviewed. Feeding difficulties or behavior problems may lead to inadequate consumption of calories and essential nutrients. Dietary intake may be lower than required for the increased energy expenditure on physical activities in children with motor disability. In contrast, caloric intake may be excessive when physical activity level is restricted and lead to obesity, most often in wheelchair users with spina bifida (6) or muscular dystrophy. Dietary information and guidance are fundamental for regulation of neurogenic bowel incontinence. Family eating patterns should be taken into consideration. Injuries, burns, fractures, and spinal cord and head trauma are followed by a catabolic state. Monitoring of weight, nutrition, and fluid intake is essential during inpatient rehabilitation for major injuries. Caloric requirements for children are calculated from age-appropriate standards, which take into consideration growth. In children with motor disability, upward or downward adjustment in height and weight may be needed, depending on their level of physical activity and individual growth trend. Specific recommendations are available for children with spina bifida to avoid obesity (7,8).

History of respiratory complications, past or present, should be explored in certain disabilities. Central ventilatory dysfunction (CVD) is a complication of Arnold-Chiari malformation in spina bifida (9). Syringobulbia may cause similar symptoms. Nightmares, insomnia, and night sweating are complaints associated with hypercapnia, and may be reported in advanced stages of muscular dystrophy or atrophy. Hypercapnia and sleep apnea may occur in diseases of the central nervous system. Intercostal muscle paralysis in high thoracic paraplegia with spinal cord injury or spina bifida, spinal muscular atrophy, or advanced muscle diseases leads to inefficient pulmonary ventilation and handling of secretions. With severe spastic or dyskinetic cerebral palsy, the respiratory musculature may lack coordination. Such children are prone to recurrent bouts of pulmonary infections. Coexistent feeding difficulties with minor aspirations, or restrictive pulmonary disease due to spinal deformities are additional adverse factors.

Restricted mobility of the spine and thoracic cage may be present in ankylosing spondylitis or severe systemic-onset juvenile rheumatoid arthritis. Detailed information about home management and use and frequency of equipment must be included in the history. Exercise dyspnea may be a sign of pulmonary compromise or deconditioning due to the high energy cost of physical activities in children with a motor disability. Cardiac decompensation with right-sided failure, a potential complication of pulmonary

dysfunction, is more likely to occur in older children or young adults with the previously mentioned disabilities. Myopathic conduction defects and arrhythmias are often symptom-free in the absence of heart failure. Consultation with pulmonary and/or cardiology specialists should be arranged when history reveals suspicious symptoms.

Visual and hearing impairments are more frequent in childhood disabilities. Inquiry about these aspects of function should not be overlooked in taking the history. The necessity of regular hearing assessment was mentioned earlier. The same applies to visual function. Prenatal infections, anoxic or infectious encephalopathy, metabolic diseases, meningitis, hydrocephalus, and head injury warrant exploration of visual and auditory function. With the development of new antibiotics, acquired hearing deficit due to antibiotic use is not a significant concern. Like all children, handicapped youngsters are prone to a variety of childhood illnesses. In some cases, however, acute symptoms and febrile illnesses may be directly related to complications of a specific disability. Vomiting, headache, irritability, or lethargy may be prodromal signs of decompensating hydrocephalus in spina bifida, cerebral palsy (2), or an intercurrent unrelated illness. Recurrent headaches are also a manifestation of autonomic dysreflexia in spinal cord injury, along with bowel or bladder distention. Fever may represent central hyperpyrexia in severe head injury or hyperthermia due to pseudomotor paralysis in high thoracic spinal cord injury. However, such conclusions can be reached only after other causes of fever have been excluded. In neurogenic bladder, urinary tract infection should always be investigated as a possible cause of febrile illness. A history of the usual pattern of the amount and frequency of voiding is essential in neurogenic bladder dysfunction. Systematic daily recording is a guide for bladder training. Fluid intake, in accordance with pediatric norms, needs to be monitored at home, and records of both bladder and bowel dysfunction should be available on the medical visit.

Immunization history is part of all pediatric visits. Often, a disabled child in good health has not received the recommended vaccinations because of excessive concern on the part of the family or pediatrician. But it also may mean that the child has always looked ill when scheduled for immunization.

History of Behavior

The examiner should ask about the child's behavior in terms of temperament and personality. The parents may state that the child was always a good baby, but this report may mean that the youngster never cried and slept more than expected for his or her age. In other cases, parents may report excessive crying and restlessness while the child is awake and during sleep. Some children may show excessive mood swings from lethargy to hyperactivity, whereas others are even-tempered and react appropriately. One should ask the parents whether the child is friendly, outgoing, and sociable or shy and withdrawn, particularly in group situations. Parental guidance may be needed to encourage interactive behavior by the child. Compliance or problems with obedience, daily activity level, attention span, sleeping and eating habits, and special interests and dislikes are revealing information. Separation from the parents may be a problem for children with disability. The parents may be uncomfortable to leave the child with relatives or other caretakers. In this context, it is important to point out the need and methods to foster the child's independence.

Educational and Social History

Very young children may be enrolled in an early intervention program, home- or center-based. Frequency, length of sessions, components of training, the child's tolerance and cooperation in the program, and its effectiveness, as perceived by the parents, should be clarified. The same applies when the slightly older child attends a preschool program. In school-aged children, information about the type of class—mainstream, integrated, or special education—is important. Academic expectations are different in each of these educational pathways and should be taken into consideration when report card grades are interpreted. Individualized education program (IEP) meetings and environmental accommodations are other pertinent details. The child may have special interests and strengths that should be further developed or difficulties in certain subjects, which may require additional help and adjustment of the IEP. Review of educational status is a consistent part of follow-up visits, and assistance should be offered when problems arise.

Opportunities to meet and play with other children in addition to school or home contacts, visits and sleepovers with friends, and participation in various recreational activities are formative experiences that prepare all youngsters for social functioning and adulthood. Asking the parents to describe the child's daily schedule, including regular and occasional activities on weekdays and weekends, yields a valuable insight into these aspects of the entire family's lifestyle. Time spent in school, therapy, homework, play, and leisure activities with family members, friends, or alone should be noted. Housing, employment of the parents, siblings and their ages, and social support of the family provide further understanding of the physical and social environment. Some families with a

disabled child experience social isolation. Information about or referral to community resources is helpful in all cases.

Family History

In motor or other developmental disabilities, a detailed family history must be obtained to rule out the possibility of an inheritable genetic disease. Health and function of the parents, siblings, and other family members on the maternal and paternal sides should be explored through several preceding generations. One should ask specifically whether there are other children in the family with developmental delay or adults with known motor disability, limb deficiency, or other malformations. Historical information is at times incomplete until further questioning brings to light additional facts. Family albums and pictures of relatives may be helpful to detect dysmorphic facial or other features. Consanguinity is an increased risk for genetic disease, including diseases with a recessive autosomal inheritance pattern. In some autosomal-dominant conditions, mild variants of a disease may be missed until a thorough investigation of suspected family members is carried out. Congenital myotonic dystrophy and facioscapulohumeral dystrophy are examples. Affected males with familial history on the maternal side are typical of X-linked conditions. Multifactorial inheritance, such as spina bifida, creates a complex situation, with or without known familial history (2,7). Referral for genetic workup is necessary whenever a genetic condition is known or suspected. Pregnant mothers of affected children should be referred for genetic counseling; prenatal diagnostic tests for detection are also available.

EXAMINATION

This chapter provides only general guidelines for the format and structure of the pediatric rehabilitation examination at different ages. Specific details of diagnostic signs and interpretation of findings are discussed in subsequent chapters about different disabilities.

Observation

As emphasized previously, the examination begins as soon as the family and child enter the examination room, before the child is actually touched or asked to perform. Sometimes, it may be the most informative phase of the examination. Specific behaviors to observe and note include reaction to separation from the parents (in young children); apparent visual and auditory awareness; temperament (calm or hyperactive, compliant, or difficult); spontaneous exploration and interest in toys, games, or books in the room; style, concentration, attention span, or distractibility during play; level and manner of motor activities; attempts to engage the parents and the examiner in conversation, vocabulary, complexity of language, and quality of speech; and interaction with parents or examiner (appropriate, shy, or demanding). Observations of the parents' response and their way of handling the child's behavior are also revealing.

Examination by Age

For infants and young children, the examiner must create an atmosphere of trust. Friendly advances during history-taking or while the child is at play allay initial fears and anxiety. At this age, most, if not all, of the examination can be accomplished with the child in the parent's lap if the child remains fearful. Interactive play in this phase of the examination can incorporate developmental testing by offering toys for grasping or raisins to test pincer grasp. Hearing, vision, cranial nerves, and postural abnormalities also can be observed.

As the parent gradually undresses the child, gentle touch and tickling or funny sounds with a smile help to maintain relaxation and to facilitate hands-on examination. Inspection and palpation of body parts and gentle movements to examine tone are performed at this point. The examiner should be prepared to improvise if the child shows increasing anxiety.

The actual hands-on examination, consisting of bodily handling and manipulation, is the last stage; anxiety-provoking or painful tests are deferred to the end. If the examination requires placement of the child on a table, the mother can sit at the end and let the child's head rest in her lap. With anxious children, performance of gross motor activities, such as sitting, crawling, standing, or walking, also can be conducted through the parent. One should note the quality of movements, postures, weakness, incoordination, asymmetry, or reflex abnormalities that reflect a motor deficit. Range of motion, deep tendon reflexes, or primitive reflexes that need physical manipulation should be examined after evaluation of active mobility. Tests that require instrumentation, such as sensation, fundoscopy, otoscopy, and oral function, conclude the examination.

Giving choices involves the preschool child in the examination. For example, the examiner may ask, "Should we look at your arm or leg now?" On the other hand, questions such as "Can I look at your arm?" should be avoided because if the child says "no," confrontation results. Parents can often bring out many capabilities of their children without the examiner touching them.

School-aged and Adolescent Patients

The customary method of systematic medical examination is applicable. Children with cognitive deficit need to be approached according to their mental rather than chronologic age. Children in this age group, particularly adolescents, are usually embarrassed about walking in underwear in front of their parents. Shorts or a bathing suit is more acceptable. Adolescents need to be seen with and without their parents. Their concerns may be different from those of the family and should be addressed with respect for their privacy.

The scope of the examination is expanded to reflect the growing child's increasing functional needs in activities of daily living (ADLs) and other areas of competence. A comprehensive examination includes screening in educational achievements, reading, writing, and arithmetic. Formal psychological or psychoeducational testing follows in case of deficits.

Growth

Parameters of physical growth should be routinely measured on each visit and plotted on the standard growth chart. Height and weight are obtained at all ages, and head circumference is measured in children under three years and thereafter in children with deviations. Serial monitoring is necessary in hydrocephalus, regardless of etiology, and microcephaly, which reflects defective brain growth. In spina bifida and other disabilities that require full-time wheelchair use, arm span measurement is recommended instead of height (7). Extremity length and girth are recorded in children with localized growth disturbance due to neurogenic weakness, epiphyseal fracture, or arthritis. In growth disturbances that involve one side of the body, one must determine whether the condition represents hemihypertrophy or hemiatrophy. Hemihypertrophy unrelated to neurologic causes requires investigation for renal tumor.

Inspection

General appearance and special features may help to establish a diagnostic entity. Dysmorphic facial features, epicanthal folds, increased intercanthal distance, external ear anomalies, and malformations of the toes or fingers suggest a prenatal disorder, possibly teratogenic or genetic, and at times, an identifiable syndrome (l0). Blue sclerae are a sign of osteogenesis imperfecta. Asymmetric facial and palpebral fissures and pupils may indicate facial palsy or Horner's syndrome, whereas craniofacial asymmetry and vertical strabismus develop in torticollis. Dolichocephaly is typical in premature infants and children. A bald spot or area of short, thinning hair over the posterior skull is a sign of weak neck muscles, most likely associated with generalized weakness. Extraocular, facial, and tongue muscle weakness may represent cranial nerve dysfunction, myopathy, or other neurologic disease. Involuntary eye movements and nystagmus are noted in cerebellar or other CNS disorders.

The skin should be inspected for telangiectasias, nevi, or other lesions. Cafe-au-lait spots or pigmented skin areas are seen in neurofibromatosis. In children with ataxia, telangiectasias are usually present over the flexor surface of the knees and elbows. Malar rash suggests a rheumatic disease. Adenomatous rash, seizures, and hemiplegia are present in tuberous sclerosis. Hairy patches, dimples, or other skin lesions over the spine are frequent signs of spina bifida occulta (7). A small sinus, dermal tract, or pylonidal cyst in the gluteal crease also may accompany occult spina bifida. Sudden weakness in such cases may indicate an infection penetrating into the spinal canal or a neurologic complication related to underlying malformation in or around the spinal cord. In children with sensory deficit, the entire area must be routinely examined for skin lesions, pressure abrasions, ulcerations, and infections. Foot deformities, varus or valgus deformity, or claw toes lead to abnormal weight distribution and callus formation consistent with the pathologic posture. Calluses over the dorsum of the feet and knees, the so-called "housemaid's knee," develop in older children whose preferred mode of locomotion is crawling. Multiple scars, bruises, and abrasions in various stages of healing may indicate frequent falls or child abuse.

Asymmetry in the size of skeletal muscles should be noted in terms of location and distribution. Anterior axillary and upper chest muscle atrophy may represent absent pectoralis muscle or wasting due to an old brachial plexus injury. Congenital clubfeet or multiple joint deformities are manifestations of prenatal muscle weakness due to spina bifida, arthrogryposis, or myotonic dystrophy, or may be idiopathic. A hypertrophic, "muscle-bound" appearance is a sign of myotonic dystrophy. Deformed, fusiform, dimpled joints are seen in arthrogryposis. Lower extremity joint positions reflect the distribution of muscle weakness in newborns with spina bifida. Hypertrophy of the calf muscles is an early sign of Duchenne muscular dystrophy. Hypertrophic musculature of the shoulder girdles and upper extremities is a convincing indication of functional crutch walking or effective wheelchair locomotion. An enlarged limb with bruit detectable by palpation or auscultation may signal an arteriovenous shunt and increased blood flow in the extremity.

Flaring of the ribs, or the so-called bell-shaped chest, suggests ineffective intercostal muscle function in children with motor unit disease or high spinal cord dysfunction. In scoliosis, the thoracic cage is asymmetric.

Palpation

In infants and young children, the fontanelles and cranial sutures should be palpated for patency, tension, and size with the child in sitting position and while the child is quiet and not crying. A tense fontanel in a vigorously crying child does not necessarily mean increased intracranial pressure. In case of ventriculoperitoneal shunt, the reservoir should be located and checked for ease of emptying and speed of refill. The skin should be felt for texture, temperature, and absent or excessive perspiration. Pseudomotor paralysis in spinal cord injury eliminates sweating below the level of the lesion, and compensatory excessive perspiration occurs above the level of the lesion with high environmental temperature. Vasomotor dysfunction with coldness to touch and paleness or slight cyanosis of the skin may be present in severe upper motor neuron impairment. It is seen in the lower extremities of some children with cerebral palsy. Subcutaneous abnormalities may be palpable, such as hard calcific deposits in dermatomyositis or neurofibromatous nodules along the course of peripheral nerves. When arthritis is suspected, each joint should be felt for the cardinal signs of inflammation, warmth, discomfort, and swelling due to synovial thickening and effusion.

Much can be learned from palpation of muscles. Tone and bulk are reduced in lower motor neuron paralysis; in longstanding denervation, the muscle tissue feels less resilient and fibrotic. The pseudohypertrophic calf muscles in Duchenne muscular dystrophy have a typical rubbery, doughy, hard consistency. A fibrotic nodule is usually palpable in the sternocleidomastoid muscle in congenital torticollis. In an infant who has an isolated knee extension contracture, a palpable nodule in the quadriceps indicates fibrotic muscle changes at the site of previous repeated intramuscular injections. Localized pain and swelling accompany injuries to soft tissue or bone. Osteoporotic fractures in lower motor neuron lesions with sensory deficit show swelling but are painless. Tenderness in many muscle groups with weakness, fatigue, or skin rash is suspicious for myositis due to collagen disease or parasitic or viral infections.

Organ Systems

Although the primary health care of children with disabilities remains the responsibility of the pediatrician, the pediatric physiatrist should perform a selective general physical examination. The emphasis is placed on organ systems that are at increased risk in certain handicaps and may affect both overall health and successful rehabilitation.

Vital signs, including blood pressure and heart rate, are obtained in all patients. In myopathies and collagen diseases, cardiac auscultation should be performed because of the possibility of associated heart disease. In a child with developmental delay, the presence of a heart murmur may suggest an undiagnosed syndrome. Blood pressure monitoring is particularly important in spinal cord injury, neurogenic bladder, Guillain-Barré syndrome, and residual poliomyelitis, as well as in children receiving stimulant medications.

In disabilities that cause ineffective ventilation and involve the risk of minor aspirations, auscultation of the lungs must be a routine procedure. Myopathies, thoracic spinal cord dysfunction due to injury or malformation, severe spastic quadriparetic cerebral palsy, and any disability with oral motor dysfunction are such indications.

Abdominal and rectal examinations are essential in children with neurogenic bladder and bowel dysfunction to evaluate bladder distention, bowel or rectal impaction, and anal sphincter tone. Stool consistency, intermittent or continuous bladder incontinence, and gross appearance and microscopic examination of the urine should be noted. Umbilical movements in response to eliciting superficial abdominal reflexes help to delineate the spinal cord level in thoracic lesions. Absent abdominal muscles result in loose skin folds resembling a prune; hence, the name prune-belly syndrome.

Neuromuscular System

Examination of neuromuscular function consists of testing reflexes, tone, active motion, strength, and coordination. Limited understanding and cooperation in infants and young children requires adaptation of traditional methods of testing. After four to five years of age, the standard examination is generally applicable.

In infancy, reflex testing includes age-appropriate responses that reflect early immaturity and subsequent maturation of the central nervous system. In newborns and young infants, state of alertness, activity, and comfort influence muscle tone (11–14). If the baby is anxious, upset, restless, or crying, this part of the examination should be postponed. Valid assessment may require several attempts. In the first few months of life, flexor tone predominates. Hypotonia or hypertonicity signals neurologic abnormalities. Increased tone is the symptom of corticospinal or basal ganglion

damage. Myopathy, cerebellar dysfunction, and lower motor neuron lesions due to anterior horn disease, neuropathy, or spina bifida all can result in hypotonia. However, a hypotonic stage usually precedes the appearance of increased tone in perinatal anoxic brain damage (15). This stage of hypotonicity tends to last longer in dyskinetic cerebral palsy than in spastic types. On passive motion of hypotonic muscles or extremities, no resistance is felt. The infant with generalized hypotonia is limp and floppy with handling and, in severe cases, may feel like a "rag doll"—a descriptive term for this finding. In hypotonia related to motor unit disease or lower motor neuron lesion, deep tendon reflexes are diminished or absent. In contrast, they are present or increased in floppy infants during the transient hypotonic phase of central nervous system damage (15).

Spastic hypertonicity and related postures are influenced by position in space and the effect of gravity. The child should be examined in supine, prone, and vertical positions to elicit typical postures. Examples include increased scissoring, extension, and plantar flexion of the legs when a child with spastic cerebral palsy is suddenly lifted into vertical suspension. Resistance to both slow and fast stretching of muscle should be tested to differentiate rigidity from spasticity (16). In infants and young children, one may use a number of developmental reflexes to examine active movements and strength (17). The Moro reflex includes shoulder abduction followed by forward flexion of the arm. Eliciting palmar or plantar grasp reflexes demonstrates finger or toe flexor function. Asymmetric responses in the upper extremities may suggest Erb's or Klumpke's paralysis or hemiplegia. Unilateral or bilateral absence of protective extension response is likewise suggestive of weakness in the respective extremity. A four-month-old infant elevates the head and trunk on extended arms in the prone position. Scapular winging during this activity is a sign of a weak serratus anterior muscle (18). In older children, the wheelbarrow maneuver demonstrates the same finding (18). Lifting up under the axilla elicits spontaneous active shoulder depression. When these muscles are weak, the shoulders slide upward, virtually touching the ears. These signs suggest myopathy with proximal weakness.

Young children often adopt ingenious substitutions or vicarious movements to cope with weakness of particular muscles. With weakness of the deltoid, they may fling the arm forward by momentum or substitute the long head of the biceps for shoulder flexion. In advanced shoulder and elbow weakness, they may "walk up" the arm on the torso, using their fingers to get the hand to the mouth. Combat crawl is a usual way of crawling in lower extremity paralysis. Deformities around a joint reflect an imbalance of strength in muscles acting on the joint. The deformity or deviation is in the direction of over-pull. Such imbalance may be spastic or paralytic.

Visual observation during performance of functional activities to detect muscle weakness should consider the child's age and the achievements expected for the child's developmental stage. Walking on tiptoes, squatting and rising without using the arms for assistance, and straight sitting up from the supine position without rolling to the prone position or to the side are mastered by children around three years of age (19). Thus, inability of younger children to perform these activities in a mature pattern should not be interpreted as weakness of the plantar flexors, hip and knee extensors, or abdominal muscles. Testing for Trendelenburg's sign and grading the triceps surae by having the child rise on the toes of one leg must be deferred until four years of age, when children develop adequate balance.

The standard technique of manual muscle testing can be used after school age, except in children who have serious behavioral problems or mental retardation (20–23). The customary grading system of scores from 0–5 or zero to normal is used. Above fair grade, the wide range of normal variations in growth patterns should be considered in judging good versus normal strength. Because children are adept in using substitution movements, the examiner must pay special attention and adhere to precise technical conduct of testing individual muscles. Side-to-side comparison may detect even mild neurologic weakness, although disuse atrophy or mild bilateral neurologic weakness may escape detection. Quantitative strength determination with comparison of both sides is helpful to demonstrate unilateral disuse atrophy in such strong muscles as the quadriceps. This determination is particularly advisable in teenage athletes after knee injury. Resumption of training for competition before virtually equal bilateral quadriceps strength is regained predisposes to recurrent injuries. Testing of strength in upper motor neuron lesions requires the well-known considerations for position in space and orientation of head and major joints, which may affect recruitment of motor units and produce synergistic movement patterns.

A common sign of central movement disorders is impaired coordination. Proprioceptive sensory loss or parietal lobe syndrome may contribute to incoordination. Movement abnormalities associated with cerebellar dysfunction, basal ganglion disease, dyskinetic disorders, or spastic incoordination present with specific distinguishing signs. Detection of coordination deficit is based mostly on observation of gross and fine motor function in children less than two to three years of age. Concurrent mild delay of motor development is not unusual. After three years of age, the examination

becomes more specific for testing the quality of performance in complex and more advanced developmental skills. Around three years of age, the child can walk along a straight line, unsteadily placing one foot in front of the other. In comparison, facility at tandem walking at five years of age is a good illustration of continuing refinement of motor skills with age. The pediatric physiatrist may be asked to evaluate the appropriateness of coordination in children without an overt physical disability (24). Clumsiness of handwriting and drawing, difficulties in physical education or sports, and other subtle signs may be present. Such children may have a motor incompetence of apraxic nature, sometimes related to visuomotor perceptual deficit (25). It also may be associated with learning and behavioral dysfunction. A number of tests are available for examining motor proficiency and dexterity in children without physical disability (26,27). Tasks to evaluate youngsters with minor neurologic dysfunction include imitation of gestures (28), hopping (29), hand-clapping (30), and pegboard performance (31,32).

Musculoskeletal System

Examination of the musculoskeletal system includes inspection and palpation of bones and soft tissues, measurement of active and passive joint range of motion, and assessment of stance and gait (33–36). It is complementary to neuromuscular assessment. As in previous parts of this chapter, only developmental variations are discussed.

Bone configuration and joint mobility change during the growing years (37,38). Full-term infants may lack as much as 25 degrees of elbow extension because of predominant flexor tone. In contrast, joint hyperextensibility and hypotonia allow increased passive motion in preterm infants. The scarf sign is a good illustration of excessive joint mobility in premature babies. Holding the infant's hand, the examiner draws one arm across the chest, like a scarf, toward the contralateral shoulder. In premature infants, the elbow crosses the midline, indicating hypotonic laxity of the shoulder and elbow joints. Full-term neonates have incomplete hip extension with an average limitation of 30 degrees as a result of early flexor tone predominance (37,38). The limitation decreases to less than 10 degrees by three to six months. At birth and during early infancy, hip external rotation exceeds internal rotation (37,39). With the resolution of early hip flexion attitude, internal rotation gradually increases. Differences between bilateral hip abduction, apparent shortening of one leg, and asymmetric gluteal and upper thigh skin folds are highly suggestive of congenital or acquired hip dysplasia or dislocation (38). Alignment of the femoral neck in neonates is consistent with prenatal coxa valga and increased anteversion. Femoral inclination is 160 degrees, and the angle of anteversion is 60 degrees. Respective adult measurements of 125 and 10 to 20 degrees develop postnatally and are accelerated by weight bearing.

Persistent fetal configuration in nonambulatory children with physical disabilities enhances the effect of neurogenic muscle imbalance on the hip joint and contributes to acquired hip dislocation in spina bifida and cerebral palsy. The popliteal angle is 180 degrees in the hypotonic preterm infant, compared with 90 degrees in full-term neonates. A combination of increased flexor tone and retroversion of the proximal tibia causes this limitation of knee extension in mature newborns. By 10 years, tibial retroversion resolves spontaneously. An early varus configuration of the tibia contributes to the physiologic bowleg appearance in infancy and corrects itself by two to three years of age. A systematic review of skeletal development, with examination of the spine and extremities, is presented in Chapter 14.

Normal variations of stance and gait should not be mistaken for pathology in the growing child (35,40,41). Gait abnormalities evident on clinical observation include asymmetric stride length and stance phase in hemiparesis; toe walking and scissoring with lower extremity spasticity; crouch posture and gait in diplegic cerebral palsy; Trendelenburg's gait in motor unit diseases and hip dislocation; gastrocnemius limp with lack of push-off in L4–L5 weakness due to spina bifida; and various types of gait deviations associated with involuntary movements, such as ataxia, tremor, or dyskinesias, in dysfunction of the central nervous system.

Sensory Examination

A complete examination of all peripheral sensory modalities is possible only in older children(42). Nevertheless, some modalities can be tested in infants and young children, and provide significant information. An infant who cries and squirms to move away from pinprick obviously perceives pain(43). A sleepy infant may be slow to respond and requires repeated stimuli. Withdrawal of the leg from painful stimuli may represent the triple flexion spinal withdrawal reflex in thoracic spinal cord lesion and should not be mistaken for active movement and presence of sensation. Comparing the infant's reaction to pinprick on the arms or face differentiates actual sensory perception in such cases. Older infants respond to touch and vibration by turning toward or moving away from the stimulus. Presence of superficial reflexes signals an intact afferent and efferent reflex arc. The neurosegmental levels are T8–T12 for abdominal reflexes,

L1–L2 for the cremasteric reflex, and S4–S5 for the anocutaneous reflex. In spina bifida, absence of these reflexes generally coincides with sensory deficit in the respective dermatomes. In young children who cannot be tested for proprioceptive function, ataxia and incoordination may suggest absence of this sensation. Testing of position sense is usually reliable by school age.

Cortical sensory function is impaired in parietal lobe damage (42,44). The most frequent childhood example is hemiparetic cerebral palsy. Disproportionately poor spontaneous function, neglect, and visual monitoring during use of the arm and hand are suspicious signs. Objective evaluation is generally feasible after five to six years of age, using the same technique as in adults for stereognosis, two-point discrimination (45). and topognosia with single or double sensory stimulation. Testing for graphesthesia may be attempted by using a circle or square. Around eight years of age, the traditional number identification gives more accurate information. Cutaneous sensation and proprioception must be intact, and adequate cognitive ability is a prerequisite for testing cortical sensory function.

The child's age and ability to cooperate need to be considered in the examination of special senses. Moving a bright light or attractive object across the visual field is used to test vision in infants. At one month, the infant will follow to midline and at three months, from side to side through a 180-degree arc. The Stycar test and the illiterate E chart are used for screening preschool children at risk for visual deficit (46,47). At an early age, unilateral impairment or loss of vision and visual field defects, such as hemianopsia, are more likely to remain undetected than bilateral deficits. A child with strabismus or suspicion of diminished vision should see an ophthalmologist as soon as the problems are discovered. Early treatment with eye patching or corrective lenses is necessary to prevent amblyopia ex anopsia (48,49). Central dysfunction of visual attentiveness, discrimination, and information processing may be misinterpreted as diminished vision and require both ophthalmologic and neuropsychologic investigation.

Screening of auditory function is a routine procedure in the neonatal nursery, pediatric office, and school. The examination of handicapped infants and children also should include simple screening of hearing, eliciting the blink or startle reflex. Responses by hand clapping to speech of conversational loudness or whisper; perception of finger rubbing near the ear; and reaction to tuning fork, bell, or cricket toy are methods of testing. Absent, lost, or delayed speech, articulation deficits, inattentiveness to sound, a history of recurrent otitis media, head injury, or failure to pass the screening test indicates a need for complete evaluation of auditory function (43,48,50,51).

Functional Evaluation

The pediatric rehabilitation examination is meaningless if the physiatrist does not construct from it a coherent picture of the child's functional achievements. This evaluation both complements and integrates the variety of information derived from all phases of the examination.

The developmental diagnostic evaluation is a convenient, functionally oriented assessment tool for infants and preschool children (19,52). Language, fine motor and adaptive skills, gross motor abilities, and personal–social behavior are the four major areas of function in the organizational framework of developmental testing. The same functional domains are considered in the evaluation of older children and adolescents. However, in these age groups, the examination includes a wider range of developmental expectations and abilities to function in school and society. ADLs and gross mobility skills need to be assessed in this context. In addition to speech, testing of language function includes other modes of communication: reading, writing, spelling, and, if indicated, augmentative communication. Drawing, design construction, arithmetic problems, and questions about handling hypothetical situations in daily life offer a brief, preliminary insight into cognitive and learning abilities. A number of specific assessment instruments were designed for various childhood disabilities (53–56). These instruments are useful functional assessment tools for their designated conditions and appropriately complement the customary developmental evaluation.

INFORMING INTERVIEW

Informing the family about the findings of the examination and their implications is an important responsibility of the physician. Factual information must be imparted with a caring attitude. Informing the parents about a newly established diagnosis should be considered as crisis intervention. A diagnostic label is insufficient without explanation of its meaning. The parents need to know the estimated prognosis, including the uncertainties of early prognostication, particularly in central nervous system dysfunction, with the possibility of multiple handicaps. Future needs in care and functional rehabilitation should be outlined. One should emphasize the need to avoid focusing on the physical disability alone and to consider the child's developmental and social needs. Effective counseling and communication skills are essential for establishing a partnership between the physician and family to ensure the successful outcome of a comprehensive rehabilitation program.

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Medical Care of Children with Disabilities

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Children with special health care needs (CSHCN) are those children who have been or are at risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally(1). An estimated 12.8% of children in the United States had a special health care need in 2001(2). Due to the large number of CSHCN, primary care providers (PCP) and pediatric subspecialists care for this complicated group of children on a regular basis. Routine health maintenance visits are frequently omitted in favor of visits for acute illnesses, which can result in a failure to discuss routine health care issues, such as growth and development, immunizations, vision, hearing, and dental care. Evaluation of children during an acute illness can pose unique challenges to care providers relating to many factors, including extensive past medical and surgical histories, lengthy lists of medications, and the lack of typical signs and symptoms normally present during a typical childhood illness. This chapter will focus on a discussion of the provision of medical care to CSHCN using a medical home model, the routine health maintenance issues for children with disabilities, and the common acute medical issues that a PCP may be asked to evaluate in this group of children. Finally, this chapter will discuss the issue of palliative care for children with special health care needs.

MEDICAL HOME

The concept of a medical home has long been endorsed by the American Academy of Pediatrics as the optimal model for the provision of primary care for all children. As defined in 1992, a medical home should provide care that is "accessible, continuous, comprehensive, family-centered, coordinated, and compassionate. It should be delivered or directed by well-trained physicians who are able to manage or facilitate essentially all aspects of patient care. The physician should be known to the child and family, and should be able to develop a relationship of mutual responsibility and trust with them" (3,4). The provision of "culturally effective" care is an additional mandate of a medical home.

Given the multiplicity of the needs of CSHCN, access to a medical home, as defined previously, is of critical importance. Beyond the provision of acute and routine medical care, the medical home can provide both "vertical links" within the medical community and "horizontal links" to the wider community. Within such a network, families should feel that they have a supportive, effective, informed, and caring network to rely on to help them meet the acute, chronic, and often unanticipated problems of a child with special health care needs.

Within the medical community, families rely on the primary care physician to make appropriate referrals to and communicate with the multiple subspecialists

who also provide care to many of these children. It can be of enormous benefit to have a designated individual in the office or clinic who is able to coordinate multiple appointments on the same day, thus lessening the burden of travel for these families. Having translator services available, as well as written materials in the family's primary language, is an additional benefit.

Children with special health care needs often require therapeutic as well as supportive services. Examples of therapeutic services include home nursing; physical, occupational, or speech therapy; and in some cases, mental health services. Supportive services may include the provision of letters of medical necessity, assistance with transportation, acquisition of durable medical equipment, provision of information regarding financial entitlements and respite care, connections to community support groups, and communication with schools. Care coordination can and should be facilitated by knowledgeable individuals within the medical home, typically experienced registered nurses (RNs) and social workers. The provision of such services can play a pivotal role in decreasing the care burden on the family, promoting maximal independence of the child and enabling full participation in community life. The medical home has additionally been described as an effective model for implementing a successful transition to adult medical care as children with special health care needs age.

ROUTINE HEALTH MAINTENANCE

Children with special health care needs see their PCP more frequently than a typical child. (5) Many of the visits are for routine care, such as well-child checks, immunizations, and school and sports physicals. Routine visits, however, can often be redirected quickly toward a disease-specific focused discussion. Although important, the basic well-child visit is no longer the focus. It is important for the PCP to ensure that routine health care needs are being addressed, even if it means that a second appointment is scheduled. The following are examples of health care topics that should be addressed during routine visits.

Growth and Nutrition

Assessment of growth is a basic element of the routine physical examination of children. Routine care in a typical child becomes a challenge when dealing with a child with a disability. Length, weight, and head circumference should be obtained at each health maintenance visit in the very young child, and length and weight should be obtained for the older child. Obtaining a weight on a child who is unable to stand on the office scale is often accomplished by having parents hold the child while stepping on the scale themselves. This is more effective in a smaller child, but is more difficult in a larger child or one with severe spasticity or hypotonia. It is recommended that an office who cares for large numbers of children who are nonambulatory obtain a wheelchair scale, which allows the child to be weighed easily in his or her own wheelchair. Assessment of the length of a child is also problematic when the ability to stand is limited. Use of arm span as a substitution for height may be an acceptable option. Alternatives to standing height also include individual measurements of lower extremity segments when significant joint contractures are present. To obtain segmental measurements, the child is placed in the supine position on the examination table and the assessment is done by adding all of the measurements obtained from the head to pelvis, the pelvis to knees, and knees to feet. Use of knee height has also been used as another means of monitoring a child's growth (6–8).

Plotting a child's anthropometric data on a growth chart will allow the PCP to track a child's nutritional status. A weight-to-length ratio below the fifth percentile may represent failure to thrive. However, growth velocity is the more important piece of information. Many children with disabilities will be below the fifth percentile for their age, but as long as their weight and length increase in parallel to a normal curve, growth may be appropriate. A child's age should be corrected for prematurity until 2 years of age. One must remember that some children with special health care needs have short stature as part of their disease process or syndrome. Special growth charts are available for children with Down's syndrome and Turner syndrome (9,10).

A nutritional assessment should be completed during routine health maintenance visits. When there is concern about a child's growth, a more careful investigation into the food intake is necessary. The amount, variety, and consistency of food eaten may provide the examiner with information regarding the caloric intake of the child. The amount of food eaten is important, but the amount of time it takes a child to complete a meal also is essential. It is not unusual for a child with severe cerebral palsy or an infant with spinal muscular atrophy to eat a meal over a prolonged period. The amount of energy that is expended during this lengthy meal may cost more in energy expenditure than is actually gained in caloric intake. Additional information that is important to obtain includes how safe the child appears while eating. This is best achieved by watching an infant or child eat or drink during their routine visit. Signs and symptoms of feeding problems include coughing or choking while eating, a wet vocal quality during or after the meal, poor sucking, gagging easily, and vomiting after a meal. A referral to

a comprehensive feeding clinic should be considered if there is a concern about the weight of the child or his or her safety while eating. An interdisciplinary clinic may include an occupational and speech therapist, nutritionist, physiatrist, gastroenterologist, and/or developmental or rehabilitation physician.

Immunizations

Routine immunization against childhood diseases should be recommended for all children with disabilities. The most current schedule can be obtained through the Centers for Disease Control and Prevention (CDC) and is approved by the American Academy of Pediatrics and American Academy of Family Physicians. (11) Special consideration must be given to children with special health care needs. Although children with disabilities are not necessarily at higher risk for contracting childhood infections, they may have greater morbidity when ill with one of these infections. One of the more controversial subjects is administration of the diphtheria and tetanus toxoids and acellular pertussis (DTaP) or measles, mumps, rubella (MMR) vaccine to children with a personal or family history of seizures. Administration of these vaccines can increase the risk of seizures in this group of children (12). The seizures are typically short in duration, generalized, self-limited, and associated with a fever. Because the pertussis immunization is given during infancy, the onset of a seizure after the vaccine can be confusing. Frequently, parents implicate the vaccine as the cause of a new-onset seizure disorder, such as infantile spasms, when in fact, the association is coincidental. It is recommended that the DTaP be delayed until a complete neurologic evaluation is completed and the cause of the seizure determined. The MMR, on the other hand, is not recommended to be withheld, even with a recent history of seizure, because it is typically first given after the onset of infantile seizures and the etiology of the seizure is generally already known.

Special attention should be given to children who are immunocompromised. Children with physical disabilities, such as those with rheumatologic diseases and Duchenne muscular dystrophy who are on chronic corticosteroids, are included in this special population. In general, it is not recommended that children who are immunocompromised from corticosteroid use receive live bacterial or viral vaccines. Although definitive guidelines do not exist, the current Red Book recommendation is that children receiving high doses of systemic corticosteroids given daily or on alternative days for more than 14 days not receive live-virus vaccines until 1 month after the discontinuation of the medications. High-dose corticosteroids are defined by receiving >2 mg/kg per day or >20 mg/day if the child weighs more than 10 kg. In the case of Duchenne muscular dystrophy, it is recommended that children receive all of their immunizations prior to the initiation of corticosteroids (13).

Immunization against influenza of CSHCN, families, and medical providers on a yearly basis is critical to decrease the potential devastating morbidity and mortality associated with this virus. Chemoprophylaxis during an influenza outbreak is also recommended to decrease the ongoing spread. Influenza immunization of all high-risk children older than 6 months of age and their close contacts should be strongly encouraged each fall (14). High-risk children being seen in the rehabilitation clinics should include those with recurrent pneumonias or upper respiratory infections and those with neuromuscular diseases such as spinal muscular atrophy (SMA), congenital myopathies, and muscular dystrophies. Children who may have increased risk from complications due to pneumococcal disease should receive the pneumococcal conjugate and/or polysaccharide vaccine (12).

Dental

Tooth decay is one of the most common diseases of childhood (15). Tooth decay and poor dental hygiene in children with disabilities is related to swallowing problems, drooling, and gastroesophageal reflux. The administration of medications with sweeteners to make the taste more palatable or those that cause gingival hyperplasia such as phenytoin also contribute to tooth decay. Routine dental care of a child or adolescent with severe developmental disabilities may be challenging for parents and caregivers due to an oral aversion, a tonic bite reflex, or the inability of the child to follow instructions to open his or her mouth. Other daily care activities, such as administration of multiple medications or respiratory treatments, may make dental hygiene less of a priority. Once a child takes over the care of his or her own teeth, the quality of cleaning may not be optimal because of cognitive and physical limitations.

Dental health of children with cerebral palsy (CP) compared to children with other disabilities is most frequently described in the literature. The incidence of dental caries in children with CP is similar to the general population, although the quality of the caries is different. The size of the carious lesions is greater than what is seen in typical children (16–18). Periodontal disease is more prevalent in children with CP compared to their typical peers, likely due to the presence of gingival hyperplasia from phenytoin administration (19). Malocclusion and developmental enamel defects were also more common in children with CP (20–24). Erosion of primary and permanent teeth has been attributed to chronic gastroesophageal (GE) reflux.

The severity of erosion has been correlated with the duration of the GE reflux disease, frequency of vomiting, pH of the acid, and the quality and quantity of saliva (25–28). Despite the fact that children with CP don't participate in high-risk activities as frequently as their able-bodied peers, dental trauma is more common (29,30). These injuries, most commonly to the maxillary incisors, are related to trauma during transfers or falls.

There is little information about dental problems for children with spina bifida. An important issue that must be addressed at each visit is to ensure that the dental office or operating room provide a latex-free environment (31). Families may need to remind the dentist and hygienist of the child's risk for an allergic reaction to latex. Latex-free gloves must be available to reduce the risk of an allergic reaction. Boys with Duchenne muscular dystrophy (DMD) can have malocclusion with anterior and posterior open bites, which are associated with lip incompetence, mouth breathing, and macroglossia. Deteriorating oral muscle function as the child gets older is associated with increased plaque and calculus formation and gingival inflammation, but not necessarily with the presence of dental caries (32,33). Boys with Duchenne muscular dystrophy have a greater risk of malignant hyperthermia when anesthesia is used for dental care (34,35).

Routine examinations and cleaning to maintain optimal dental hygiene should be performed by a dentist comfortable in the care of children with special needs. Some of the dental care may need to be accomplished under anesthesia in order to obtain the maximum benefit. Combining dental procedures with other necessary procedures, such as a brainstem auditory evoked response (BAER), local intramuscular injections with phenol or botulinum toxin, or certain orthopedic procedures, may limit the exposure to anesthetic agents. The American Academy of Pediatrics Policy Statement on oral heath care states that children with special health care needs be referred to a dentist as early as 6 months of age and no later than 6 months after the eruption of their first tooth, or 12 months of age (whichever comes first) (36). Visits will provide the dentist with the opportunity to provide specific education to the family to allow for optimal dental care.

Vision

Vision screening and eye examination should be a component of all routine health care visits. The American Academy of Pediatrics recommends that the evaluation begin in the newborn period and then at all subsequent visits, with the goal of identifying conditions that might result in visual impairments or represent serious systemic diseases (37). In the child with a disability, this is especially important, given the frequent association of visual disorders with neurologic diseases. The eye evaluation from birth to 3 years should include a vision assessment, which is accomplished by having the infant or young child fix on an object. The examiner assesses the child's ability to maintain the fixation and follow the object into different gaze positions, a skill that by 3 months of age is developmentally appropriate. Further evaluations of the young child should also include external inspection of the eye and lids, pupillary and red reflex examination, and ocular alignment. Assessment of the child older than 3 years should also include age-appropriate visual acuity measurements and an attempt at ophthalmoscopy.

Ophthalmologic disorders frequently seen in children with cerebral palsy require very close follow-up with an ophthalmologist (38). Annual evaluation for cataracts should be completed in children with myotonic dystrophy or those on chronic corticosteroids, such as boys with Duchenne muscular dystrophy or a child with a juvenile rheumatoid arthritis (13,39). Detailed and accurate documentation of the ophthalmologic examination of a child with spina bifida can be helpful when assessing possible ventriculoperitoneal (VP) shunt malfunctions. For example, a malfunctioning VP shunt may cause papilledema or changes in extraocular movements. These are early indications that may manifest prior to more obvious signs, such as headaches or lethargy.

The eye examination of a child with a disability is best performed by a pediatric ophthalmologist due to the child's high risk for ophthalmologic problems. The ophthalmologists have the skill needed to obtain a thorough assessment. A referral to a pediatric ophthalmologist for specialized tests, such as an electroretinogram (ERG), may be useful in assisting with the diagnosis of rare neurologic conditions, such as mitochondrial diseases.

Hearing

Newborn hearing screening is standard of care in the United States. In 1999, the American Academy of Pediatrics endorsed the implementation of a universal newborn hearing screening program (40). Two technologies are used for newborn hearing screening: brainstem auditory evoked response (BAER) and otoacoustic emissions (OAEs). It is important that all newborns be screened and is particularly imperative for children with disabilities. Periodic reassessments of children with disabilities are important, since a hearing impairment can significantly affect their developmental skills.

Primary care providers should pay special attention to children with specific disabilities, as they are at greater risk for developing hearing loss. For example, children with Down's syndrome are at increased risk

of otitis media and concomitant transient conductive hearing loss (41). Children with congenital cytomegalovirus (CMV), both symptomatic and asymptomatic at birth, are at risk for progressive and late-onset hearing loss (42). Children with athetoid cerebral palsy due to kernicterus have a high incidence of hearing loss, as do children who have been treated with ototoxic antibiotics for systemic infections (43,44).

ACUTE ILLNESS IN THE PRIMARY CARE OFFICE

Children with disabilities present to primary care providers with the same childhood illnesses of their typical peers, but the presenting signs and symptoms may be quite different. Medical personnel who care for these children need to be acutely aware of these differences in order to accurately and efficiently diagnosis and treat the acute illness. It is important that the primary care providers understand the disease-specific complications and how they may present. Referring to previous medical records can be helpful in determining the unique issues for a particular child. The American Academy of Pediatrics (AAP) provides specialized forms for families and medical personnel to maintain an up-to-date record of a child's medical history, current medications, past medical complications and how they typically present, and a treatment plan based on presenting signs and symptoms (45). An up-to-date form can provide medical care providers information in a critical situation. The following section reviews specific acute and chronic complications and strategies for the primary care provider when approaching a child with special health care needs in order to facilitate an appropriate diagnosis and treatment plan.

Respiratory Complications

Drooling

Difficulty in managing oral secretions in children with disabilities results from poor oral motor control. Parents may express concern over their child's drooling, frequent cough, or increased upper airway congestion. Management of drooling can be pharmacologic or surgical. Treatment is recommended when drooling causes significant skin irritation, social problems, or the child is having recurrent respiratory infections secondary to poor secretion management (46).

When oral secretions are copious, use of a suction catheter by caretakers can keep the oral cavity and upper airway clear. Families should be instructed in the appropriate technique of oral cavity suctioning and have a portable suction machine that can be used when out of the home setting. Use of medications such as glycopyrolate or the scopolamine patch can decrease the volume of secretions. The use of botulinum toxin injections into the submandibular and parotid glands is being recommended more frequently for children with cerebral palsy (47). Surgical ligation of the glands is typically reserved for cases that are unresponsive to medications.

Drooling may indicate that a child is having difficulty with eating, drinking, or swallowing. In a child with a degenerative neuromuscular disease, such as spinal muscular atrophy, the development of increased drooling or difficulty managing oral secretions should prompt a further investigation into his or her feeding status. A referral to a feeding team should be made for consideration of performing a modified barium swallow. Alternative feeding modalities, such as a nasogastric tube or a gastrostomy tube, may be necessary. Use of medications to dry secretions in a child with muscle weakness may be counterproductive, as thicker secretions may be more difficult to clear.

Respiratory Distress

Children with upper respiratory infections commonly present to their primary care provider with fever, increased work of breathing, and tachypnea. The evaluation and treatment of a child with a disability who presents with these symptoms should be similar to a typical child. However, the deterioration may be accelerated, requiring a rapid diagnosis and initiation of treatment. The assessment should begin with a review of vital signs, including pulse oximetry. The physical examination focuses on assessing the child's level of alertness, his or her work of breathing, and a chest examination. Children with neuromuscular diseases will frequently increase their respiratory rate in order to maintain oxygen saturation. Unfortunately, a child can decompensate quickly in this situation as a result of significant fatigue. Oxygen saturations can be falsely reassuring in the face of hypoventilation.

Diagnostic testing may include pulse oximetry, chest x-ray, venous or arterial blood gas, sputum culture looking for a bacterial etiology, and viral studies for identification of common viruses such as influenza and respiratory syncytial virus (RSV). Viral etiologies are the most common causes of upper respiratory infections in both disabled and typical children.

Use of antiviral medications should be considered in children with disabilities because of their high risk for significant morbidity. Enteral or parenteral antibiotics should be reserved for suspected bacterial etiologies. Coverage for anaerobic bacteria should be initiated when aspiration pneumonia is suspected.

A child with a neuromuscular disease, such as spinal muscular atrophy or Duchenne muscular dystrophy, may need assistance with secretion mobilization and airway clearance. Secretion mobilization can be addressed with chest percussion or a vibratory vest, skills that a family should be comfortable performing. Airway mobilization can be accomplished with the use of a cough-assist machine. The In-Exsufflator, a commercially available device that provides a positive pressure breath followed by a large exhalation, improves peak cough expiratory flow rates (48,49). Children and caregivers should be familiar with the different techniques and initiating their use at the first signs of a respiratory illness. When symptoms increase and evidence of hypoventilation is present, use of noninvasive and invasive respiratory support may be necessary. Noninvasive support may include negative pressure ventilation or positive pressure ventilation with bilevel positive airway pressure (BiPaP) (50). It is important for primary care providers to have knowledge of the various options for respiratory support and to understand the family's wishes on the extent of treatment the family wants in the case of acute decompensation. Acute events are less stressful when families and their primary care providers have discussed their wishes while the child is well and prior to the event.

Neurological Complications

Seizure Activity

Children with cerebral palsy are at increased risk of having seizures (51). The primary care provider is frequently asked to evaluate a child who is having increased seizure activity. Identification of an intercurrent illness, which may lower the seizure threshold, and a review of adherence to the current medication regimen are critical questions that must be asked. Obtaining levels of the antiepileptic medication is useful in determining whether suboptimal levels are the etiology of the increased seizure activity and there is a subsequent need to increase the medication dose if levels are low. A recent increase in weight might trigger the need to adjust the current dose. A referral for an electroencephalogram (EEG) and consultation with a pediatric neurologist may be generated if the pattern of seizures is determined to be changing. Empowering a family to treat seizure activity with fast acting benzodiazepine in the home setting is an important way to decrease the need for emergency room visits.

New-onset seizures in a child with a disability should be evaluated thoroughly. For example, a new seizure in a child with spina bifida and shunted hydrocephalus may represent a shunt malfunction with subsequent worsening hydrocephalus.

Spasticity

The majority of children with cerebral palsy have spasticity as a component of their upper motor neuron disorder. Primary care providers are frequently asked to evaluate a child with increasing tone. The acute onset of increased tone may represent an intercurrent illness, such as an otitis media or a urinary tract infection, causing pain in a child, which is manifested as spasticity. The sole presenting signs of an acute fracture of an extremity in a child who is nonverbal may be increased tone. A careful assessment of all extremities is necessary when the diagnosis is unclear. Treatment of the increased spasticity should focus on treatment of the underlying illness. Use of antispasticity agents such as diazepam and baclofen may be a necessary adjunct when tone is markedly increased. Acute withdrawal from a malfunctioning intrathecal baclofen pump (see chapter on cerebral palsy) may present with increased tone, diaphoresis, tachycardia, hypertension, and irritability. The irritability may be related to the pruritis that is an idiosyncratic reaction not associated with a rash. Primary care providers who care for children who have intrathecal baclofen pumps should be familiar with these common presenting symptoms and management of the withdrawal from baclofen. Immediate administration of oral baclofen or intravenous (IV) diazepam will help decrease the symptoms, but referral to a center that can evaluate and treat the malfunction is needed to resolve the problem (52).

Orthopedic Complications

Fractures

The incidence of fractures associated with minimal trauma is increased in children with cerebral palsy, spina bifida, and Duchenne muscular dystrophy (53). This is related to reduced bone mineral density secondary to immobilization or limited mobility. Children with cerebral palsy or Duchenne muscular dystrophy with an acute fracture typically present with pain and/or irritability. However, children with spina bifida or a spinal cord injury may only present with swelling of the limb due to their lack of sensation. Radiographs should be utilized when swelling of a limb is present, even when no trauma history is elicited.

Treatment of fractures in children with disabilities varies, depending on the diagnosis, type, and location of the fracture. Casting of a limb in a child may depend on his or her degree of mobility. In other words, in a child who is wheelchair-dependent, a bulky splint may be applied. This is especially true for a child who is insensate, since a plaster or fiberglass cast may lead to pressure sores. Prophylactic treatment of reduced bone mineral density in children with special health care

needs is controversial and should be addressed by a specialist in disorders of bone metabolism (54).

PALLIATIVE CARE

Primary care for children with special health care needs may include consideration for palliative care services. Children who should be referred for palliative care are those with potentially life-threatening diseases. This diverse group includes children with diagnoses of advanced or progressive cancer, neuromuscular diseases, severe cerebral palsy, acquired brain injuries, severe central nervous system (CNS) malformations, complex and severe cardiac abnormalities, and chromosomal or metabolic abnormalities. Children with HIV infection, severe immunodeficiency, cystic fibrosis, and severe epidermolysis bullosa also meet the criteria for palliative care. Palliative care for children can and often does include life-prolonging treatments, such as a tracheostomy placement for a boy with Duchenne muscular dystrophy, as well as potentially curative treatments, such as chemotherapy for a child with advanced cancer.

In a policy statement on palliative care, the American Academy of Pediatrics states, "Palliative treatments focus on the relief of symptoms (eg, pain, dyspnea) and conditions (eg, loneliness) that cause distress and detract from the child's enjoyment of life. It also seeks to ensure that bereaved families are able to remain functionally intact."(55) Palliative care focuses on the quality of the life remaining to the child. Palliative care addresses not only the physical symptoms, but also the psychological, social, and spiritual issues of a child who lives with life-threatening or terminal conditions.

CONCLUSION

In summary, children with special health care needs account for a large percent of children in a primary care practice. It is imperative that the primary care provider be familiar with the associated medical conditions of each child and the need to provide routine health care. Furthermore, it is critical for the primary care provider to be familiar with the frequently occurring acute medical illnesses in children with disabilities and the common manner in which they may present.

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3

Psychological Assessment in Pediatric Rehabilitation

Jane A. Crowley and Kayla White-Waters

The goal in pediatric rehabilitation is not an end point as in the adult world. Rather, it is a process toward the continued development of ever-changing abilities and emotional, behavioral, and cognitive structures. The goal of any pediatric rehabilitation process is to foster the continuing work of childhood. This additional distinguishing dimension of pediatric rehabilitation relates to the central imperative of the pediatric population—development. There is a dual goal: rehabilitation to prior levels and habilitation for the remaining development in that child's or teen's life. An important tool for establishing current levels, setting future goals, and tracking progress over time is psychological assessment.

The rehabilitation physician and the team will treat a wide array of medical conditions among their patients. Rehabilitation medicine departments will encounter requests for treatment for those with congenital disability, acquired disability from illness or injury, and chronic medical conditions. A recent estimate of the incidence of severe chronic illness seen in rehabilitation is more than 1 million children in the United States (1). These children, and those who survive catastrophic illness or injury, are a growing population due to medical advances that reduce mortality, covering the full age range from infancy to young adulthood.

In acknowledging that normal development assumes an intact sensory, motor, and overall neurologic system for interaction with the environments of family and the larger world, the children and teens we work with do not have the standard equipment or inter-relationships among skills. For example, a child's motor disability can easily alter the basic emotional developmental tasks. The protraction of physical dependence that is a reality for a child with a congenital disability like spina bifida, at the very least, risks altering the psychological milestones of separation/individuation. Cognitive sequelae of that central nervous system (CNS) disorder can also result in academic and adaptive behavior deficits. In these cases, standard developmental schema often do not apply (2). Not only because of deficits, but because there are unique tasks to be mastered with a disability. Functional use of a wheelchair, doing activities of daily living (ADLs) with one arm, self-catheterization, and visual competence with a field cut are but a few specific "milestones" our patients face. In the case of a traumatic injury, the disruption of a normal life, with typical developmental progress and engagement in the world, is an emotional maelstrom for the patient and his or her family (3).

Potential distortions in many aspects of the nurturing and individuating demands of competent development abound in children with disabling conditions (4). The barrage of medical technology and interventions is vast in variety and effectiveness. Yet, the psychological cost of these necessities can be high. The challenges of hospitalization, a disruption of familiar routine, the

therapy demands of rehabilitation, absence of parents, and intrusive or painful medical procedures are additional tasks against which to bulwark the patient (5). In a broader context, there is prejudice against those with disability, and children must face the extra demands of bridging ignorance and misconceptions.

In line with the centrality of development, the objects of assessment constitute a "moving target." Environmental demands change, as does the child's or teen's abilities to meet them. At school age, the child must now function competently in the ever-increasing demands for independence reflected in the school setting. Furthermore, the medical condition can itself change over a child's development. A disease process can progress (for example, juvenile rheumatoid arthritis), or increasing body size can change the nature of mobility (for example, spina bifida), and prior function can be lost. The task is to have these experiences remain *challenges* to development and not become *barriers*. This argues for continued monitoring throughout a child's development as a vital factor and the importance of psychological assessment as a vital part of that monitoring to be utilized throughout the pediatric course of a patient's life.

The relationship of family functioning to outcome in pediatric disability has been widely demonstrated (6,7). The challenge to a family is to walk an unfamiliar path, as few families have direct experience with childhood disability. The effects may be bidirectional (4), with the deficits from the medical condition interacting with parental features or the child's status resulting in disrupted parenting approaches. Parents often must assume an additional role as case manager and advocate in the medical and educational systems. In addition, they have to "translate" their child's issues to other family members at the nuclear and extended family levels. The family becomes a vital arena of intervention. The family is the first-order site of development and stimulation as well as a filter for the larger world.

ADJUSTMENT VERSUS PSYCHIATRIC DIAGNOSIS

It is important to recognize the distinction between *psychiatric disturbance* and *adjustment problems* as conversant concepts in assessment for a pediatric rehabilitation population. Indeed, psychiatric disturbance is not common in children with chronic conditions, as some studies show that their functioning is better than children in the mental health clinic population. Taken together, however, children and teens seen in rehabilitation medicine settings do have a greater risk for adjustment problems (1). Their medical condition acts as a life stressor not encountered by their healthy peers. To use psychiatric diagnoses in this population belies the reality of behavioral symptomatology that is indeed adaptive to the conditions and situations of a child's medical condition. Though some behaviors may be unusual in the healthy child, they may be adaptive to this population (8). The concept of *adjustment* encompasses the variability that these patients encounter. It can express the unique trajectory that these children's lives will take, and recognizes it as adaptive in that it is age-appropriate for those conditions and oriented ultimately toward healthy adult functioning.

A wide body of literature addresses the adjustment of children with chronic physical conditions. This group was twice as likely to have adjustment problems as healthy children in a meta-analysis of 87 articles by Lavigne and Faier-Routman (9). Though the specific prevalence rates were higher yet among these children, only a minority showed maladjustment. Such children, then, are more vulnerable than those who are healthy. Newer assessment instruments have been developed that utilize the concept of *quality of life* (10) and will be discussed here in the section "Population-Specific Assessments." With this approach, the nature of a child's or teen's adjustment and the reflection of the uncontrollable factors in his or her situation are captured for a wider rubric than the inadequate dichotomy of normal versus abnormal.

However, the use of psychiatric diagnoses can be appropriate in both this population and those with neurodevelopmental disabilities. The latter group showed a rate six times that of the general population for significant emotional and behavioral problems (drawn from an outpatient clinic population) (11). The identified problems run the gamut, encompassing a breadth of disorders, and are more likely to persist into adulthood. There is the primary impairment of the neurologic disorder and a secondary impairment of psychosocial support problems (4). With the primary disturbance in the brain, these findings are not surprising. Other factors exist as well to either exacerbate or ameliorate the brain's abnormality, but the overall picture is one of significant neurologic and psychological morbidity, with the source of disability either congenital or traumatic.

The most common disabling injury of childhood is, in fact, traumatic brain injury (12), which carries a substantial risk for long-term cognitive disability as well as behavioral deficits (13). Expressed differently, incidence figures are such that by the tenth grade, 1 in 30 students would have had a traumatic brain injury (across severity ranges). This population will be a substantial part of a pediatric rehabilitation medicine practice. The causative link between cognitive and behavioral functioning represents the juncture of thinking and adaptive behavior that can be devastating to the ongoing development of a child survivor (14). There is a particular danger in the

misattribution that easily occurs. Behavioral deficits are attributed to more common etiologies, as opposed to the organic brain disorder from the injury. With misattribution comes inappropriate treatment. Cognitive limitations are not accounted for in treatment efforts, or the wrong premise (for example, antecedent versus contingent programming) used, and failure occurs or even exacerbation of the original problems. Awareness and consideration of the brain damage from an injury means assessment must encompass a wide focus. Neuropsychological testing is the centerpiece in these children and teens, representing a subtype of general psychological assessment that will be an important aspect of many rehabilitation cases.

NATURE OF MEASUREMENT

The essence of psychological assessment lies in the construction of the instruments used to explore various concepts of adjustment, personality functioning, behavior, and cognition. This construction always has at its core the notion of standardization through its reference to a norm group, whose performance is characterized by a transformation of the raw score earned by an individual. Even the most skilled observer could not provide the richness of the information gleaned from a psychometrically sound test. Such a test allows for the comparison of that subject to the typical performance of his or her peers in a fair and objective way. The value of standardized assessment depends on some core concepts, elucidated in the following sections.

Norm-Referenced Measurement

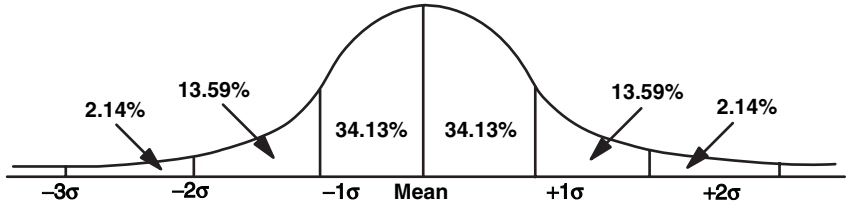
Norm-referenced tests are standardized on a clearly defined group, referred to as the norm group, and scaled so that each individual score reflects a rank within the norm group. The examinee's performance is compared to the group, generally a sample that represents the child population of the United States. The comparison is carried out by converting the raw score into some relative measure. These are derived scores and indicate the standing of a patient relative to the norm group. These scores also allow for comparison of the child's performance on different tests. Stanines, standard scores, age- and grade-equivalent scores, and percentile ranks are the most common tests.

A central concept in the expression of individual performance as compared to a norm group is the normal curve. The normal curve (Fig 3.1) is a bell-shaped curve. It represents the distribution of many psychological traits, with the greatest proportion at the "middle" of the curve, where it is the largest, and the abnormal levels—both below- and above-average—at the two "tails." All derived scores have a distinct placement on the normal curve and are varying expressions of the location of an individual's performance on that curve.

Stanines are expressed as whole numbers from 1 to 9. The mean is 5, with a standard deviation of 2. Substandard performance would be judged with stanines in the range of 1–3 and above average at 7–9. In this transformation, the shape of the original distribution of raw scores is changed into the normal curve.

Standard scores are generally the preferred derived score (15). Their transformation of raw scores yields a mean for the normative group and a standard deviation. This places a given score across the normal curve, and the scores express the distance from the mean of that patient's performance.

T scores, *z* scores, and the well-known *IQ* of the Wechsler scales are all standard scores. Like all standard scores, the *z* score derives a constant mean and standard deviation across all age ranges The *z* score has a mean of 0 and a standard deviation of 1. It expresses below-normal performances with the minus sign and above-average with the plus sign, with scores in a range of −3 to +3. These scores are often transformed into other standard scores to eliminate the positive and negative signs (see Figure 3.1). T scores



Z-scores	–3.0	–2.0	–1.0	0.0	1.0	2.0	3.0
Percentile	0.13	2.5	16	50	84	97.5	99.87
T-score	20	30	40	50	60	70	80
IQ (SD=15)	55	70	85	100	115	130	145
CEEB or SAT score	200	300	400	500	600	700	800
Stanine	1	2	3	4 5 6	7	8	9

Figure 3.1 The Normal Curve.

and the IQ scores are drawn from the z score, with different numerical rubrics that eliminate the plus or minus sign associated with *z* score.

Multiplying by 10 and adding a constant of 50 yields a *T* score ranging from 20 to 80, with an average of 50. Another transformation occurs by multiplying the standard score by 15 and adding 100. This provides a range from 55 to 145, with a mean of 100 and a standard deviation of 15 or 16, depending on the test used. This is the method that produces the *Deviation IQ*, the form of derived score used on the Wechsler intelligence batteries. The alternative to the Deviation IQ is the *Ratio IQ,* which is the ratio of mental age to chronological age multiplied by 100, used in the Stanford–Binet tests. The statistical properties of this are poor, and it is not generally used or well regarded.

What appear more understandable, but are not as psychometrically sound as standard scores, are percentile ranks and age- and grade-equivalent scores. Percentile ranks offer easy interpretation, with the rank reflecting the point in a distribution at or below which the scores of a given percentage of individuals fall. To a lay audience, this is often confused with percentages, which are not referenced to a normative population—only to the number correct compared to the total number of items. For example, function at the 50th percentile is average performance, whereas a grade of 50% on a test would be considered failing.

Even more straightforward appeal exists for age- and grade-equivalent scores. These scores are obtained by discerning the average raw score performance on a test for children of a given age or grade level. The individual patient's score on that test is compared to that value. Grade equivalencies are expressed as tenths of a grade (for example, a grade equivalency of 4.1 represents the beginning of fourth grade). Despite their appeal, there are limitations with these forms of derived scores. First, a grade-equivalency value does not mean that a child is performing at that particular level within his or her own school, as the curricular expectations of the school might be different from the mean score established by the normative sample. Some actual age- or grade-equivalency values might not have been earned by any specific member of a normative sample, but instead are extrapolated or interpolated from other points of data. Furthermore, age or grade equivalencies may not be comparable across different tests. The meaning of a first grader who obtains a raw score similar to a third grader is not that the child is functioning as a third grader in that subject. He or she shares that score, but the assumption that the child in first grade has all the skills of a third grader is inappropriate. Similarly, a 12-year-old patient who achieves an age equivalency score of 8 years, 4 months seldom actually functioned on the test the way a typical 8-year-old child would, and certainly should not be treated like an 8-year-old for most issues in rehabilitation programming.

Finally, as is the case with percentiles, age- and grade-equivalents cannot be used in statistical tests, as there is an unequal distribution of scores. Both require conversion to another scale before they can be used in data analysis.

Reliability

This concept of *reliability* refers to the ability of a test to yield stable (ie, reliable) results. There needs to be a consistency and stability of test scores, and the nonsystematic variation reduced as much as possible. Psychometric theory holds that any score is composed of the measurement of the actual trait that a child possesses as well as an error score, which represents the variation or error of measurement. The reliability coefficient is the conversant statistic to express this property. It can vary from 0.00, indicating no reliability, to 1.00, indicating perfect reliability. High-reliability coefficients are considered particularly important for tests used for individual assessment. In the case of cognitive and special ability tests, a reliability coefficient of 0.80 or higher is required for sufficient stability to be a useful test. Reliability coefficients are calculated for a test across three conditions. One is *test-retest*, meaning the capacity of the test to yield a similar score if given a second time to a child. Another is *alternate-form* reliability, where the child is tested with an alternate form of the test, measuring the same trait and in the same way as the initial testing. A third kind refers to *internal stability* in a test, where in the ideal test, item responses are compared to another item on the test to demonstrate the equivalence of items in measuring the construct in a replicable manner. Active judgments must be made in the choice of tests, with reliability coefficients reviewed in the process of test selection.

Validity

This is another vital consideration in the construction and use of standardized tests. *Validity* is the extent to which a test actually measures what it intends to measure and affects the appropriateness with which inferences can be made based on the test results. Validity of a given test is expressed as the degree of correlation, with external criteria generally accepted as an indication of the trait or characteristic.

Validity is discussed primarily in terms of *content*—whether test items represent the domain being measured as claimed—or *criterion*—the relationship between test scores and a particular criterion or outcome. The criterion may be concurrent, such as comparison of performance on neuropsychological

test measures with neurophysiologic measures (eg, computer tomography, electroencephalography).

Alternatively, the criterion may be *predictive*—the extent to which test measures relate in a predictive fashion to a future criterion (eg, school achievement). In the rehabilitation context, various events and contingencies may affect predictive validity. An appropriate determinant of predictive validity is the likelihood that the individual's test performance reasonably reflects performance for a considerable period of time after the test administration. Acute disruption in physical or emotional functioning could certainly interfere with intellectual efficiency, leading to nonrepresentative test results. In contrast, chronic conditions would be less likely to invalidate the child's performance from a predictive standpoint because significant change in performance as a function of illness or impairment would not be expected over time. With therapeutic interventions, a patient's performance could improve, so test results from prior to that would not be valid. The more time that passes between test administrations, the more likely extraneous factors can intervene and dilute prior predictive validity. Anxiety, motivation, rapport, physical and sensory handicaps, bilingualism, and educational deficiencies can all effect validity (15). For an inpatient population, the effects of acute medical conditions (eg, pain, the stress of hospitalization, medical interventions themselves, fatigue) can also affect validity. Wendlend and colleagues (16) noted that in a study of cognitive status post-polymyelitis, the deficit seen could well have been due to the effect of hospitalization as opposed to the disease.

Construct validity refers to the extent to which the test relates to relevant factors. Another important component of validity is *ecological validity*, which refers to the extent to which test scores predict actual functionality in real-world settings. Test scores are typically obtained under highly structured clinical testing situations, which include quiet conditions, few distractions, one-on-one guidance, explicit instructions, praise, redirection, and so on. These conditions do not represent typical everyday tasks or settings (17). This disconnect between the test setting and real life is especially relevant in children with brain-related illness or injury. These children, who have high rates of disordered executive functioning (eg, distraction control, organization, planning, self-monitoring, etc.) benefit disproportionately from the highly directive nature of clinical testing, and test scores may overestimate true functional capacity for everyday tasks (18).

A test's reliability affects validity in that a test must yield reproducible results to be valid. However, as detailed previously, validity requires additional elements.

In the rehabilitation population, all of these issues have particular import. Most tests are developed on a physically healthy population. Motor and sensory handicaps and neurologic impairment are not within the normative samples. Issues of validity predominate here, though with transitory factors as noted previously, reliability can be affected as well. Standardized procedures may have to be modified to ensure that a patient is engaged in the testing in a meaningful way.

USES OF ASSESSMENT

Psychological assessment has a wide variety of purposes in pediatric rehabilitation. These purposes encompass issues directly related to the medical setting, but often have equal utility in educational planning. Unique to the field of pediatric rehabilitation is this necessity for interaction between what are arguably the two biggest public systems for children: medicine and education. Both have their productive and counterproductive forces and hold a vital place in the individual child's or teen's life. Furthermore, both can act to hinder or potentiate the salutary effect of the other. The needs and parameters of engagement with both is at the crux of the navigation of development for our patients, and psychological assessment contributes significantly to this process.

Psychological testing is often associated solely with IQ testing. The intelligence quotient (IQ) concept of intellectual development is too narrow for many of the applications in a pediatric rehabilitation setting. Instead, the evaluation of the broader aspect of *cognition* is the more important activity. Cognitive assessment covers testing the wide array of known components of the brain's thinking skills. Assessing these intake, processing, and output modalities of thinking, their individual elements or the combination of these skills are vital factors in school or in medical rehabilitation. School is children's work, and the interface with this system is critical, as it is the arena where many key adjustment and developmental issues are played out. Psychological adjustment—indeed, overall functioning—is intimately tied to cognitive status. Coping with frustration, functioning within a group, and inhibiting for long-term goals, are examples of processes vital to school that have cognitive capacity at their center.

Within the schools, the psychological assessment performed has typically included only intellectual and achievement testing as prime components. Though that is changing in some settings, it is not yet common that *cognitive* processes are assessed. For the populations common to a rehabilitation medicine practice, many conditions have brain involvement (eg, traumatic brain injury). Their needs are clearly beyond the limitations of typical school testings. Eligibility for services within the special education system under the qualifying

conditions of traumatic brain injury (mandated by the federal government in 1998) cannot be done without consideration beyond IQ and achievement testing. Indeed, traumatic brain injury (TBI) as its own inclusion category was done to reflect the serious misunderstanding of the disorder when only evaluated by IQ and achievement testing alone. The intellectual assessment of children with spina bifida needs explication beyond IQ testing as well. Often, the component parts of the Full-Scale IQ score are so divergent in children with spina bifida and other brain conditions that it does not represent a true summary score. To understand a child's condition fully, further assessment of cognitive processes needs to be done. Pertinent abilities are attention, concentration, memory, and executive functions. In the wide array of conditions known to affect brain functioning there are primary and secondary effects. Primary effects are seen from brain tumors, seizure disorders, or cancer processes. Secondary effects on cognitive processes are seen in the process of infectious disease or cancer treatment. It is necessary to evaluate a broader array of abilities rather than relying solely on IQ to understand the full spectrum of required cognitive skills for competent development.

In order to promote the fuller understanding of medical conditions and their effects on cognitive functioning, the rehabilitation practitioner will often be consulted for more specialized assessment to capture the full nature of functioning within his or her patients. Input into the Individualized Educational Plan (IEP), which is the centerpiece of planning in the special education system, is essential in brain-based disorders to ensure full consideration of the medical condition, its own process, and its unique effect on brain functioning. The dynamic nature of recovery is notably absent from most students receiving special education services, but is often a primary part of the course in traumatic brain injury, brain infectious processes, cancer, or strokes. The need for frequent reassessment, specific remediation-focused services, or specialized support in re-entry to school are several of the unique concepts that are vital to sound educational planning in our population but are largely unknown to the traditional process of special education. This is the most critical juncture of school and medical factors in a pediatric rehabilitation process.

As per Section 504 of the Rehabilitation Act, *accommodations* are often sought on either a long-term or transitory basis in rehabilitation medicine patient groups. These are efforts to "level the playing field" within the school setting in acknowledgement of disability that skews a student's ability to benefit from the standard educational setting. These students do not require the breadth or type of actual intervention or service gained through special education classification, but instead need modifications in the system in order to demonstrate their capacities or adequately access the learning environment. Results of psychological/neuropsychological evaluations can be useful in demonstrating such need related to cognitive issues. For example, deficits in information processing speed can have a global effect on functioning within the group instructional environment of school. Accommodations such as reduction in homework, extended time for tests, or lecture notes, among others, can all be sought with the documentation provided by evaluation results. The issue of how long the accommodations are required can be answered by repeated testing. An example is in the case of a brain injury where recovery occurs and accommodations may no longer be needed.

It is important for the clinician to recognize the role he or she can play in securing vital, but not typical, medical treatment for a patient. This includes speech and language or occupational therapy, cognitive remediation, or adjustment-focused cognitive behavioral work. The documentation of that need, based on the medical diagnosis or history, can be obtained much quicker and with the proper focus through the medical system in terms of both insurance coverage and proper treatment frequency and formulation. Obtaining assessment from a public school system can be a lengthy process. For rehabilitation patients, this can waste valuable time and, therefore, cannot meet the time frame needed for an acute recovery. A typical school psychological assessment could miss acute issues and be even less likely to detect weaknesses that could hamper development or skill acquisition distant from the injury or illness. Such evaluation needs the medical framework of rehabilitation psychology to be timely and pertinent. Furthermore, with a rehabilitation psychology perspective and knowledge, appropriate documentation emerges to secure services covered by medical insurance or from legal settlement funds, if such exists. Keeping the intervention within the medical perspective can make it more integrated with disease or injury sequelae and, therefore, more targeted and appropriate in terms of goals and treatment techniques.

It can be seen that the assessment of a child's or teen's learning process is essential to both the school and medical setting. Memory processes, language abilities, planning, or capacity to inhibit are essential functional elements in either system. The preference of one modality over another, or the explication of memory functioning, can be of great use in school issues and in rehabilitation. The need to master specialized tasks, such as wheelchair skills or self-catheterization, can be enhanced when general learning styles of an individual patient can be discerned.

This understanding of a patient's cognition can inform educating the patient about his or her medical disorder, or the rationale about a medical procedure. The feelings of victimization that can evolve around a painful surgery and the subsequent effect on adjustment or even personality formation are secondary

sources of potential morbidity in a child's development. The child's or teen's sense that he or she was regarded enough in the consideration of procedures to be included in the decision and planning process. The experience of this and the skill to be a meaningful participant are vital long-term skills and are promulgated by knowing the proper way to present material in a way to ensure understanding. Decisions about a child's ability to benefit from a specific treatment such as biofeedback, relaxation training, or the varieties of behavioral programming available are part of diagnostics that guide treatment.

Change as the result of intervention can be quantified by assessment. However, change without overt intervention, but to chronicle the long-term outplay of a medical condition, is arguably the most common use of assessment in rehabilitation. The risk for long-term sequelae in traumatic brain injury or from cancer processes and treatment is well known (13,19). The serial assessment of a patient, particularly through known critical developmental periods or illness interventions, is at the core of sound pediatric rehabilitation practice. A developmental lag becomes the object of treatment, whether to spur development or to teach compensatory strategies. As the physical process of a disease is monitored through traditional outpatient clinic visits, so the status cognitive/behavioral of functioning in relation to the demand of one's medical condition or to changing developmental expectations is equally important to monitor.

Baseline assessment is the initiation of such a process. It most often has been understood as measuring function at the outset of illness or injury against which to calibrate future change. Now this concept has been expanded to include the characterization of a healthy child or teen prior to exposure to risk. Specifically, this paradigm defines the process of baseline assessment in sports as regards the risk of *concussion*. Participation in all sports has exploded in recent years in children of all ages (20). With the use of baseline cognitive testing, the determination of a child's or teen's unique cognitive profile prior to a concussion are quantified (21). In a much shorter time frame than the one implied previously for more serious illness or injury processes, the degree of concussion and recovery are discerned by repeated testing post-concussion within weeks or months. Cognitive assessment is generally regarded as essential in the diagnosis and monitoring of concussion, as delineated by the International Conference on Concussion in Sport held in Zurich in 2008. The increased vulnerability of the adolescent athlete relative to adults is well recognized as to duration of symptoms and differential recovery pattern. Furthermore, the effect of repeat concussion, treatment options, school demands, restriction of exposure to risk (continued sports participation)—both during recovery and subsequently—and the potential effect on a developing brain (22) are all factors that argue for the role of neuropsychological assessment in the care of such patients. In the next section, the nature of this type of baseline testing will be explored.

Understanding the individual experience of a child or teen in relation to his or her body experience is another use of assessment. Understanding the experience, whether through a questionnaire about pain, assessment of specific mood states like depression or anxiety, or a general personality assessment of that patient, can be quite useful. Differential diagnosis can be important, as in the case of post-traumatic stress disorder, where cognitive symptoms of that disorder can be mistaken for the effects of a mild brain injury or concussion. In that circumstance, the deficits are due to the effects of the stress and not to the mechanical disruption of trauma.

TYPES OF ASSESSMENTS

The purpose of psychological assessment is to discern the status of an individual in relation to an appropriate peer group. Jerome Sattler discerns four pillars of child assessment as norm-referenced tests, interviews, observations, and informal assessment (15). This is a broader list than many referral sources would recognize, as typically "tests" are all that might be considered as psychological assessment or evaluation. However, a central tenet in psychology is that test scores or results cannot be interpreted in isolation. Information from naturalistic settings must be sought through the methods of interview, observations, and informal assessments, as enumerated by Sattler.

In a discussion of cognitive testing, the issues of single tests versus batteries is an important consideration. Single tests are designed to tap a specific dimension of cognition, like verbal learning or visual–motor abilities. As useful as they are for more in-depth examination of a single construct, this strength is a source of limitation as well. Seldom is the question at hand to be answered by examining a single ability. Abilities are not the unitary concepts that evolve from theoretic models. The influence of other overarching cognitive abilities, such as attention or processing speed, is not addressed directly and is discernible only through observation. Normative samples for single tests can be restricted and not large enough or representative enough to draw firm conclusions as to standing within one's peer group.

Therefore, the use of a *test battery* is preferred. The best-known example of a test battery is the Wechsler batteries for intelligence assessment, comprised of a number of subtests. These collections cover an array of abilities. In neuropsychologic assessment, the concept of a fixed battery versus a flexible battery exists. A *fixed battery* is a group of subtests developed to tap a spectrum of either a specific function—for instance, memory

or attention—or a comprehensive view of cognition. The Wechsler memory scales and the NEPSY described in the next section are examples of a battery for a specific function, in the case of the memory scales, and a comprehensive assessment, in the case of the NEPSY.

Fixed batteries provide for the strongest basis for comparison of a patient's performance across the subtests, as the norms are based on this arrangement of tests, given in the established order to the normative group. Because all subjects receive the same subtests, there can be an expression of both strengths (what a patient can do) and deficits. This is particularly useful in the construction of rehabilitation plans. A flexible battery is composed of a number of single tests, assembled with the patient's referral question or known medical condition in mind, with an eye to tapping tests most likely to explicate suspected deficits.

Lezak and colleagues (23) noted a survey of neuropsychologists where 70% responded that they use a flexible battery approach. They note the position that fixed batteries involve more testing than some patients need and can't accommodate the practice of adding tests either newly developed or needed to explicate a deficit seen but in need of further examination.

Automated or computer use in testing has increased substantially since the 1980s. Prior to that, automated and later computerized administration and scoring of tests was quite limited. Initially, computerized testing of attention was developed (Gordon Diagnostic System, Connors Continuous Performance Test). More recently, computerized tests have been developed for concussion diagnosis and monitoring (as noted in the prior section), but also for research purposes. Such techniques offer repeatability, sensitivity to subtle cognitive changes, and ease of administration. Reliability, validity, and other considerations pertinent to general issues in more traditional so-called pen-and-paper tests are pertinent to this type of assessment as well.

Evaluation, then, is a robust and multifactorial process, not to be confined to a set of test scores or descriptions of test performance, but also to include natural setting data. The norm-referenced placement of a patient has a role, but the assessment setting in and of itself imposes a high degree of structure. While this one-to-one administration is not replicated in real life, it is necessary for the standardization of administration and the reference to a normative sample, as described previously. Therefore, the addition of perspectives from natural settings of the home, school, and community are necessary, as is the consideration of the aspects of the medical condition.

Maureen Dennis (24) captured the interaction of these factors in the following, which she calls an "outcome algorithm." Though Dennis is specifically referring to disorders that affect the central nervous system, the same factors apply in understanding other medical disorders as well. She explains it as

> . . . biological risk associated with the medical condition, moderated by the child's development; by the time since onset of the condition; and by the reserve available within the child, family, school, and community.

The interpretation of standardized tests must take these factors into account: issues about the course of a disease or injury recovery, the unique interface that the course of an illness or recovery has on the timetable of childhood development, and the actual length of the struggle with the medical condition. Her inclusion of the word "reserve" with which to respond and cope dovetails with the requirement of assessment that examines these factors as well.

Some of these elements are captured in a good history taking and/or record review. Reserve factors concerning coping and response are also gathered in history but can additionally be tapped by standardized questionnaires, whose responses are sought from a variety of sources. These encompass figures from the major settings in a child's life (ie, parents and teachers). The value of such instruments is that they can reference responses to those of a normative population such that the degree of divergence from standard development can be expressed. Some include consistency scales that add information about the nature of the responses given.

Culture-Sensitive Assessment

Psychological assessments with culturally diverse children are challenging under any circumstances. Most measures have a culture bias in terms of content and validity, and normative data are seldom adequately representative of diverse groups. Not all examiners are sufficiently sensitive to the impact of cultural issues on test performance, and when interpreted without caution, results can be misleading. The assessment of English language learners, children who have reduced mastery of the English language because their parents' primary language is not English, is particularly challenging. Use of interpreters or test translations carries limitations, such as lack of equivalent concepts in the two languages, minimal provision for dialectical variations, and possible changes in the level of difficulty or meaning of translated words (15).

Several "culture-fair" tests have been developed to reduce culture bias by limiting the amount of verbal exchange, using more abstract content that is less grounded in culture and language, and using more diverse groups during the norming process. This represents an important step in culturally sensitive assessments, and some of these tests are discussed in

following sections. However, there is no way to truly eliminate cultural bias from tests, and demographic data on normative groups must be carefully examined before assuming that it is any more representative of the specific patient than traditional tests. For example, many of the "culture-fair" tests are normed only on children in the United States. Their use for students with different backgrounds, such as children from refugee camps in Africa with little to no formal schooling, is clearly limited.

Culture-sensitive assessments in pediatric populations are made even more complicated by the frequency of mild to severe motor impairment. Examiners assessing individuals with motoric impairment rely heavily on tests of verbal cognitive skills and try to reduce the number of tasks that require speeded or complex motor responses. Examiners assessing individuals from linguistically or culturally diverse backgrounds rely heavily on tests of nonverbal cognitive skills and try to reduce the verbal component. Examiners assessing individuals from linguistically diverse backgrounds with motor impairments are limited indeed in terms of valid options. Even in pediatric groups that do not have motor impairment, the higher frequency of *discrepancies* in functioning (significant strengths and weaknesses in a single individual, such as may be caused by damage to right versus left hemisphere or cortical versus subcortical areas) makes the traditional practice of assessing nonverbal skills and considering the results representative of *general* functioning highly questionable. School and community-based clinicians may not be aware of the complexity of issues involved and may provide scores without adequate caution regarding limitations.

SPECIFIC INSTRUMENTS

Neuropsychological Evaluation

Originally, the neuropsychological assessment was directed at diagnosing the presence, nature, and site of brain dysfunction. The focus has shifted from diagnosis to assessment of a child's function to identify and implement effective management, rehabilitation, or remediation services.

Neuropsychological Batteries

As mentioned earlier, neuropsychological batteries have been developed to provide a comprehensive evaluation of cognitive abilities. The two most common in practice today are the downward extensions of the Halstead Reitan Neuropsychological Battery and the NEPSY-II, developed specifically for children.

The Halstead Reitan Battery has been refined and redefined over the years since Ward Halstead's original conceptualization in the 1940s to a larger series of tests to diagnose so-called brain damage for ages 14 and above (25), and subsequently the downward extension for ages 9–14, called the Halstead Neuropsychological Test Battery (HRNB) for Older Children. It takes approximately four to six hours to administer and uses subtests from the adult Halstead Reitan Battery, with some modifications. The battery for children ages 5–8 is called the Reitan Neuropsychological Test Battery and requires a similar time interval for administration. These batteries, in wide usage earlier, are criticized for a number of pivotal problems. The first is on conceptual grounds, in that the battery was not developed for children, but for adults, and is perhaps reflected in the minimal assessment of memory, academics, and language, with no direct measure of attention. The psychometric properties are widely acknowledged to be quite poor, such that reliance on those alone for interpretation is inappropriate. Considerable clinical acumen is required to interpret findings. Dean concludes a review of the batteries saying, "The HRNB cannot be recommended for general clinical use without considerable training and familiarity with research on the battery (26)." Considering norms published in the interim, Lezak et al. (23) is more favorable to the HRNB in saying that what statistics it yields are misappropriated by "naïve clinicians," implying the same point as Dean.

The only neuropsychological battery ever developed specifically for children is the NEPSY-Developmental Neuropsychological Assessment (27), with the newest version, the NEPSY-II (28), published in 2007. Both batteries are based on the diagnostic principles of the Russian neuropsychologist Alexandr Luria. The original NEPSY had two forms and covered ages 3–4 and 5–12, with a core battery of 11 to 14 subtests represented to tap five functional domains: attention and executive functions, sensorimotor functions, language, visuospatial processing, and memory and learning functions. This original version was criticized for its content and psychometric properties (29). It is well standardized, and though some instability is noted in some subtests, this may indeed reflect the reality of the developmental status of the brain.

The most recent version has not been tested enough to generate a literature on its strong points or weaknesses. It does expand the age range to 16 years, extending one ostensible benefit of a battery that covers the childhood range, allowing for the ideal serial assessment. The content has also changed, with targeted groupings of subtests for various diagnoses, nonverbal elements, and new measures of executive functioning, memory and learning, which reportedly solves some of its statistical problems. A functional domain in social perception has been added as well.

Attention, Concentration, and Information Processing

The processes of attention, concentration, and information processing are often central concerns for any patient with a medical condition involving the brain (30). In many ways, they form the basis on which the other component processes occur. Overall cognitive productivity suffers from losses or failures to develop these skills.

Attention has been conceptualized in a number of ways, generally relating to an organism's receptivity to incoming stimuli. Most do regard the issues of automatic attention processes versus deliberate/voluntary as central dimensions. Other characteristics include sustained, purposeful focus—often referred to as concentration—and the ability to shift attention as required by a stimulus. Being able to ward off distractions is usually seen as part of concentration (31). Vigilance is conceptualized as maintaining attention on an activity for a period of time. There are the needs to respond to more than one aspect of a stimulus or competing stimulus—the capacity to divide attention—alternating with shifts in focus.

The multitude of processes subsumed in the concept of attention are necessary because of the overall effect. Most notable is the developmental nature of attention in childhood and adolescence. Increasing demands in school participation are seen in the shifting requirements throughout the academic process. In the early grades, a child is more directly engaged by the teacher, but as the years progress, the capacity for independent (ie, voluntary/deliberate) processes grows. Attentional processes are a central aspect of the changing capacity of normal development. Attention's vulnerability to normal variation, as with fatigue or anxiety, is a part of typical functioning. Attentional processes require a certain "tone" to the brain's functioning, attention and its concomitants are often affected in brain disorders. Furthermore, with acquired deficits in the disordered brain, the demands are higher, as an individual struggles with recognizing the need to attend along with implementing a specific compensatory task.

Lezak and colleagues (23) note that underlying many attention problems is *slowed processing*. This can be misinterpreted as a memory disorder (32), as competing stimuli in normal activity interrupt the processing of the immediately preceding stimuli and something is "forgotten," in common parlance. The discernment of this specific problem is important, as strategies alleviating the effects of slowed processing would be different from those for memory per se.

All of these aspects warrant examination, notably in those with a brain disorder, due to the overall effect on functioning and the demand for acquisition of academic and adaptive behaviors throughout childhood. The effects of anxiety about an illness process, its treatment, and demands for coping can all affect attention, and in a competent diagnosis are differentiated from primary brain disruption.

Because of the issue of time in competent attention processes, computerized testing has real utility to control for calibration of presentation and response. Absent a fully computerized administration, the use of taped auditory stimulus in attention testing allows for standardized presentation increments. Typically, the computerized tasks involve visual stimulus and the taped presentations involve auditory ones. This differentiation between verbal and nonverbal, or auditory versus visual, is necessary to capture these two central aspects of stimulus processing.

Recent development of a battery of attention tasks for children, the Test of Everyday Attention-Children will be described next. It attempts to cover a number of aspects of attention processes and for the comparison of subtest scores to allow for relative differentiation of components.

Inattention, slowness, and poor concentration have a wide-ranging effect on competent cognitive and adaptive functioning. Other processes may be quite competent, but attention and its aspects can be a primary "rate limiting" factor. These should be addressed in even a screening of functioning, whether at bedside or in the clinic, both as an overall indicator of current cognitive activity, but also as a harbinger for developmental problems to come, signaling the need for more stringent monitoring. Commonly used tests are described in Table 3.1.

Problem-Solving and Executive Functioning Tests

As with attention processes, deficits in these realms can have a devastating effect on overall functioning. Cognitive process can be intact, but with executive functioning impairments, the output can be substantially derailed. The basic tasks of life can suffer, along with the ever-present demand in childhood to acquire new skills. These deficits can be more obscured in children than in adults, as there is a natural support of activity by parents or other family members. Return to school can be the point at which executive functioning problems can clearly be seen for the first time since an acquired illness or injury. Traumatic brain injury presents a particular vulnerability to deficit in these skills. Executive functions are associated with the frontal and prefrontal areas of the brain, where, due to the mechanisms of closed head injury and the shape of the brain and skull convexities, damage can be focused across the full range of severity. Rehabilitation efforts suffer, both in commitment to the process and in learning strategies to compensate for deficits (39).

3.1 Tests of Attention and Speed of Processing

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Test of Everyday Attention Test of Everyday Attention-Children (TEA-Ch) (33)	Batteries of 8 or 9 tasks for ages 17 and above; TEA-CH ages 6–16	Taps visual/auditory attention including dual tasks; selective, sustained and executive control
Gordon Diagnostic System (GDS) (34)	Normed for ages 6–16 years; includes 3 tasks: delay, vigilance, distractibility; has preschool version for ages 4–5 yrs	Freestanding and portable electronic devices used for administration and calculation of summary data; parallel version for vigilance task; relies solely on visual task
Paced Auditory Serial Addition Test (PASAT) Children's Paced Auditory Serial Addition Test (CHIPASAT) (35)	Adding pairs of digits presented at 4 rates of speed, controlled by the audiotape presentations; adult and child forms; ages 8 and above	Highly sensitive to deficits in processing speed; sensitive to mild disruption, but can be stressful test to take, as many items can be missed at normal ranges
Continuous Performance Tests (36)	Covers a category of tests; visual or auditory stimulus where must respond to a target stimulus in the presence of distractors; various versions for ages 4 and up	Many versions exist; sustained, vigilance and inhibition tapped; Connors Continuous Performance Test II and Test of Attention are well known.
Symbol Digit Modalities (SDMT) (37)	Oral or written; requires visual scanning and tracking to match preset symbol and number pairs	Taps information processing; Spanish version with norms; seen as selectively useful.
Trail-Making Test (TMT) (38)	Subject draws lines to connect consecutively numbered (Part A) and alternating numbers and letters in order (Part B). Ages 9 and up	Part of Halstead-Reitan battery; test of speed, visual search, attention, mental flexibility, and fine motor; needs interpretation with other tests; Part B is most sensitive

The competent measurement of these skills requires a multidimensional approach and is quite complex (40). Testing of these functions imposes a degree of structure required by standardization such that vital elements can be obscured. Attempts at quantification in real-life situations becomes particularly important. Questionnaires for parents and teachers elicit descriptions of behavior that can be compared to normative expectations. Particularly for parents, this can be useful in understanding the need for treatment. Teachers have a normal sample of age-appropriate peers in the classroom and can be more aware of such problems. In that circumstance, the questionnaire process can illuminate the component elements to be addressed, as a deficit in classroom performance can be composed of many factors, with differing contributions to the overall presentation. The Behavior Rating Inventory of Executive Functions (BRIEF), described in the section on "Psychosocial Evaluation," is an instrument focused on these behaviors. It covers the preschool period through adolescence, as well as a self-report questionnaire for older children, with basic forms for teachers and parents to complete.

Lezak (23) differentiates these brain skills as

> "Questions about executive functions [are generally phrased \as *how* or *whether* a person goes about doing something (eg, Will you do it and, if so, how and when?); questions about cognitive functions are generally phrased in terms of *what* or *how much* (eg, How much do you know? What can you do?)."

There are many models, as in attention, as to what comprises these skills and how to measure the components, since it is far from a unitary concept. Again, as in attention, the developmental progress of these skills is a central aspect of childhood and adolescence. In the teenage patient, assessment of these skills is vital, as adult-like capabilities for work, driving, and independence can be severely affected and, in the particular case of driving, have disastrous results. The enactment of graduated driver license requirements for teen driving in some states implies the centrality of these skills and their necessity for that activity. Stepwise exposure and supervision of driving for teens allows for a graduated experience before full driving privileges are granted. Specialized assessment through

rehabilitation-based driving evaluations using computer simulation should be considered by a rehabilitation team in any teen with a history of brain disorder.

Definitions of executive skills include the capacity for planning and flexible use of strategies, and the ability to generate, maintain, and shift cognitive sets; to use organized search strategies; and to use self-monitoring and self-correction, as well as the capacity to utilize working memory. It is distinct from general intelligence, though it does correlate at lower levels of intelligence. Again, as in attention skills, these skills are vulnerable and easily disrupted in many circumstances, as they are largely acquired throughout childhood as an essential central process of competent development. Therefore, deficits acquired can be "silent" until they are called on for future development. The range of tasks is wide, from inhibiting behavior in the absence of visible authority to planning how to accomplish several assignments due at the same time.

Though the cognitive aspects are difficult to quantify, the literature on these is substantial. However, the emotional and behavioral aspects of executive skills is less studied (41). Executive skills act to regulate behavior (42), inhibit and manage emotions, tolerate frustration, and provide persistence. Notable is the result of limited empathy (ie, taking the position of the other). They are observed collaterally in any sound testing process, but are captured better, to the extent possible, in the questionnaire approach discussed previously. The effect of impairment in these skills can be widespread and debilitating, especially as expectations for empathy and self-awareness increase in adolescence.

One of the questionnaires does differentiate these two factors. In the BRIEF (131), questions about such skills yield feedback for the behavioral regulation composite, as differentiated from another composite reflecting the cognitive aspect, metacognition. A list of tests that cover this wide-reaching domain is listed in Table 3.2.

Nonverbal/Visual–Perceptual Function Tests

This type of cognitive task is seen as one of the two major classes of cognitive input/output. At its base is the perceptual capacity of vision. From earliest infancy, humans already have sufficient visual perception to mimic another's facial expression (29). Related to the developmental aspects of childhood, by age 9, visual processes are integrated with tactile and proprioceptive functions. Tests of visuoperceptual, visuospatial, and visuomotor function are all within this domain and include the ability to discriminate between objects, distinguish between left and right, judge spatial orientation and the relationship among objects in space, copy a model, understand symbolic representations of maps and routes, and solve nonverbal problems.

3.2 Tests of Problem Solving and Executive Function

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Halstead Category (HCT) (43)	Versions exist for ages 5–9 and 9–14, as well as through adulthood; part of Halstead-Reitan battery	Machine and booklet forms; measures conceptualization and abstraction abilities
Wisconsin Sorting Test (WCST) (44)	Revised manual offers norms for ages 6.5 and above.	Requires inference of correct sorting strategies and flexible use
Tower of Hanoi (TOH) (45)	Computer and standard administration; ages 4 and up	Taps working memory, planning, rule use and behavioral inhibition
Tower of London (TOL) (46)	Arrange balls on pegs to match picture; norms for 7 and up	Taps inhibition, working memory, anticipatory planning
Stroop Color-Word Test (47)	Well-known test, quick administration in paper form; several versions	Test of inhibition, selective attention, and switching sets
Matching Familiar Figures (MFFT) (48)	Must find identical match for stimulus picture; ages 6 and up	Measures impulsivity
Fluency Tasks Verbal and Design	Speeded tasks of response generating to verbal and nonverbal stimuli	Taps self-monitoring, initiating, and shifting; included in many batteries
Delis-Kaplan Executive Function System (D-KEFS) (49)	Battery of 6 subtests; age 8 and above	Battery aids comparison of subtest scores

Direct functional outputs include being able to navigate the environment and depth perception. Testing of these functions can illuminate visual field cuts, visual neglect, and apraxia.

However, these tasks often involve other aspects of cognition, such as attention, memory, speed of thinking, and motor impairment. At the base is the requisite of normal visual acuity, screened for in the pediatric clinic setting or in school admission testing. However, after a brain injury or illness, the intactness of the basic perceptual components of nonverbal functioning should not be assumed. Issues like cortical visual defects or loss of binocular vision require examination by vision specialists; in more severe TBIs, such injuries are not uncommon. Consideration of the myriad factors involved in this domain's assessment requires examining other test performances to discern patterns, as well as factors that may affect visual function output but not be a deficit in these processes per se. The importance of these functions as a basic component of input and output of cognitive function is seen in their presence in all intelligence and neuropsychological batteries. Table 3.3 provides a representative listing of these tests.

Language Functioning Tests

To examine this function is to assess what is felt to be a central aspect of brain functioning and the foundation of human thought. Language development is well delineated, and proceeds from spoken to written competency in normal childhood. Receptive and expressive abilities require separate assessment, as do the modalities of written language. Speech as the mechanical aspect of oral communication is assessed as distinct from language per se. One element can be intact, while the other has significant impairment, and evaluation distinguishes between the two.

An acute injury most notably, but even an illness, may produce a frank aphasia, but this often resolves from its most dramatic state. Deficits remain that are more subtle, but important to address. Common deficits after injury differ between an adult and a child. In childhood and adolescence, deficits in word finding, dyscalculia, and problems with formulating written language are common (29). Functional problems, like difficulty with instructions or following commands, require analysis to determine the degree of linguistic difficulty versus other factors, such as attention or memory. As is always the case, the determination of concurrent difficulty in other tasks provides the diagnostic information to hone in on the core problem(s).

Components like phonologic processing, naming, language comprehension (oral and written), and understanding the syntactic structure of language, as well as the productive aspects of language, should be covered. The profound impact of language deficits on academics makes it a particularly important aspect for assessment. Problems with reading comprehension are often seen, and because reading is one of the primary tools for learning once a child enters elementary school, the impact can be widespread on competent school functioning.

There are many well-accepted tests to measure all these aspects. For receptive language, the Peabody Picture Vocabulary Test-III (54), Token Test (55), or the Bracken Basic Concept Scale-Revised (56) are widely used. These vary from single-word comprehension to grammatical/syntactical structure to linguistic

3.3 Tests of Nonverbal/Visual–Perceptual Function

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Rey-Osterreith Complex Figure Test (ROCF) (50)	Copying of complex figure and a delayed recall condition; alternate forms exist for repeat administration; ages 6 and up	Taps planning, visual organization, and memory for complex visual information
Beery Developmental Test of Visual-Motor Integration. 4th ed. (VMI) (51)	Design-copying test of 24 forms of progressive difficulty; supplemental tests of visual perception and motor coordination; Ages 3+; procedures for younger children	Long-standing test with new additions; visuoperceptual, visuomotor integration is assessed
Facial Recognition Tests (52)	Requires direct matching and side profiles of photos of human faces; norms for ages 6+	Present in standalone tests, but also in batteries, including memory aspect; implications of right hemisphere function
Wide Range Assessment Battery of Visual Motor Ability (WRAVMA) (53)	Battery of 3 tests: drawing, matching, and pegboard. Can be administered individually or as a battery yielding a composite; ages 3–16.	Sound psychometrics yield standard scores and percentile for each subtest

concepts. Similar tests exist to investigate expressive oral language. Batteries like the Clinical Evaluation of Language Functions (57) or the Test of Written Language-2 (58) offer the advantages of batteries, while covering various aspects of language so that differential levels can be discerned.

A long tradition in neuropsychological evaluation is the evaluation of aphasia, the disturbance in the basic language capacity of the brain. This capacity begins at the level of auditory discrimination and phonologic awareness, proceeding to words, then meaningful word combinations. The Boston Diagnostic Aphasia Examination (59), though its full utility with children has been questioned has long been in use. Issues such as fluency skills, where the ability to generate words within a parameter, such as beginning sound, or rapid naming are basic language skills that can be lacking due to developmental or acquired problems. Though they are not everyday language skills, they represent an automaticity of language that can affect more complex skills, such as reading.

Memory and Learning Tests

Memory involves cognitive mechanisms used to register, retain, and retrieve previous events, experience, or information (23). All aspects of this activity need to be assessed to provide sound diagnostic information in addition to developing remediation or compensatory strategies. Questions of ecological validity are particularly cogent in memory evaluation, as necessary types of memory cannot be assessed in the testing situation. Adaptive behavior questionnaires and devices that attempt to incorporate real-life situations, like the Rivermead Behavioral Memory Tests, are useful to round out more traditional assessment tools, which are largely based on theoretical laboratory models. The nature of material to be remembered in everyday life is different, but so is a naturalistic setting, and the attendant natural distractions are part of many instances where memory is needed.

A full examination of memory covers a number of distinctions, including declarative/explicit versus implicit/procedural memory, recognition versus recall, encoding versus retrieval issues, prospective and remote memory, short-term/working memory versus long-term memory. A prime distinction is verbal versus nonverbal memory, and it should be included as a referral question in most situations.

Findings need to be viewed in the context of the recognized developmental changes in memory functioning through childhood (60). Developmental changes that mark the progression toward mnemonic competence are attributable to the child's growing proficiency in the use of strategies to aid encoding and retrieval of information.

In situations of traumatic brain injury, there is a specific role for monitoring the time span where brain functioning was insufficient to record ongoing environmental input, referred to as post-traumatic amnesia (PTA). This is done through tracking orientation and return of continuous recall. The latter refers to the brain's resumption of the capacity to register everyday occurrences on an automatic basis. For pediatric rehabilitation, the Children's Orientation and Amnesia Test (COAT) was developed for this purpose by Ewing-Cobbs and colleagues (61), based on the Galveston Orientation and Amnesia Test for adults. The duration of PTA has been shown to more reliably predict recovery than the Glasgow Coma Score (GCS), the rubric used in general medicine to judge severity and, by implication, prognosis. Retrograde amnesia should also be assessed, representing the time span for which formation of long-term memory was disrupted, so that minutes, hours, and sometimes days prior to the injury are not recalled. This also requires serial monitoring, as restoration of retrieval processes results in more information being recalled as the brain recovers. For retrograde amnesia, the monitoring is essentially just patient responses to questioning of events leading up to the injury (Table 3.4).

Sensory–Perceptual and Motor Tests

Tests of these functions can be illuminative for laterality issues as well in determining the extent of impairment in the corresponding cerebral hemisphere. Peripheral disorders must be ruled out as the cause of discrepancies or abnormal scores. There are well-established norms from age 3 and up pertinent to motor sequencing, various hand movements, and reciprocal coordination. This area includes tests of tactile discrimination and fine motor or hand–arm movements. Rates of competence between the sides in simple items and in items with gradually increasing complexity are done for both tactile and fine motor functioning. The techniques of A.R. Luria (67) are often used for fine motor examination, with elements of executive function abilities intrinsic to completion of the more complex movements. Specific tests would include the Grooved Pegboard (68) for skill motor movements—a timed task involving peg placement in holes at various orientations to the shape of the pegs. Though interpretation must be done in the context of other data, such tests can provide information about the course of a disorder. An example is in chronic hydrocephalus, where monitoring with tactile proprioception as in finger recognition and number-writing perception can signal progression of the cerebral pathology.

Brief Smell Identification (69) allows for standardized, forced-choice odor identification, with 12 microencapsulated odorants as a screening test for olfactory

3.4 Tests of Memory and Learning

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Rivermead Behavioral Memory Test, 2nd ed. Children's Version (62)	Tasks are analogues of everyday memory; has immediate and delayed tasks; two versions: adult (age 11+) and children's (ages 5–10); Four parallel forms.	Novel approach with everyday tasks increases utility in case planning and remediation. May miss moderate to mild deficits; alternative forms very useful, though enough statistics aren't given for full utility; shows general disruption.
Wide Range Assessment of Memory and Learning 2 (WRAML 2) (63)	Traditional memory battery covers nonverbal and verbal, immediate, recognition and delayed; ages 5–90	Excellent psychometrics; widely used; Has a screening form
Test of Memory and Learning -2 (TOMAL-2) (64)	Ages 5–60; traditional battery; covers nonverbal, verbal, immediate, delayed, and cued recall	Good psychometrics; Easy to administer
Child Memory Scale (CMS) (65)	Ages 5–16; battery; parallel structure of adult Wechsler Memory Scale	Widely used; enables comparison with IQ and achievement as part of Wechsler series
California Verbal Learning Test -C (CVLT-C) (66)	Ages 5–16; verbal memory assessed; short and long delay (20 min) procedures	Limited (only tests verbal abilities); hard to score by hand; good psychometrics

function. Many studies have documented a high incidence of olfactory dysfunction post-brain injury in adults, correlated with higher-order cognitive skills that can be elusive to discern in direct fashion. The role in the developing brain is less delineated. Norms have been developed from age 5 and up.

Computerized Assessment

Within this area, a number of devices have already been listed under other sections, notably in the attention/processing speed section. The discussion here will be of the relatively recent use of computerized testing of cognitive functions specific to abilities disrupted by concussion. These abilities include speed of processing and reaction time, and are done with varying stimuli. An inherent limitation is the lack of auditory presentation in these instruments, where all stimuli are visual in presentation, even though language stimuli are used in conjunction with nonverbal stimuli (spatial location, line drawings) in one test listed. The repeatability and ease of administration is an advantage of these tests and so can be used for the serial monitoring recommended for complex concussion recovery. Scores on these devices serve as guidelines of functional capacity that determine return to activities, whether that is around cognitive demand (school) or physical demand (gym class, sports, bike riding, etc.). Balance assessment can also be used as a specific monitor representing a high-level dynamic function of the brain's motor control and an ability required for competent physical activity participation. There is ongoing debate about the sensitivity of cognitivie versus balance deficits as the most sensitive indicator of concussion sensitivity.

There are only two computerized batteries that have norms within the pediatric population. Both include a symptom report. The HeadMinder Concussion Resolution Index (CRI) (70) has norms for ages 18–22 and "under 18." The latter refers to a normative sample down to age 13, with analysis yielding no difference in the scoring of adolescents from ages 13–18 (71). The CRI is an Internet-based platform with six subtests, taking 25 minutes to administer. It yields three scores: processing speed index, simple reaction time index, and complex reaction time index. Verbal (written) stimuli were specifically avoided, with all stimuli in a visual icon format to minimize error due to language disability or English-as-second-language issues.

Immediate Post-Concussion Assessment and Cognitive Testing (ImPACT) (72) is available in Windows and Macintosh applications as well as through an online version. An on-field Palm-based version is also available and includes a brief on-field mental status evaluation. It does use verbal stimuli, and there is reading involved in testing instructions, with a sixth grade required reading level (73). It has eight subtests in its current version and registers demographic/history data, current concussion details (including information about anterograde and retrograde amnesia), as well as somatic and cognitive symptoms. There are four scores from ImPACT: verbal memory, visual memory, reaction time, and visuomotor speed. ImPACT has

norms for ages 11 and above. Adolescent norms on this battery are extensive, and there is an extant literature on its use. Though developed primarily for sports concussion management, it has recently been used to characterize concussions presenting to an emergency room (74). A version is being developed for children ages 5–10.

Cognitive and Intellectual Measures

A central component of all psychological assessment has been a measurement of intellectual or cognitive ability. As this pertains to children, the purpose is typically to predict and plan for academic capacity and appropriate educational programming. Tests of this nature have also allowed clinicians and educators to detect students who may be at risk for learning problems and benefit from special services.

Of the major general cognitive tests, each is based on different theoretical models, but all share a fundamental similarity: separate assessment of verbal and nonverbal skills, with scores combined to yield a general composite. In the rehabilitation population, children whose illness or disability differentially affects verbal or visual–spatial skills require a more sophisticated selection and analysis of tests. These children are more likely than the typical population to show significant differences on different types of skill sets, and composite scores may not provide much useful information. For example, a child who scores in the average range on visual–spatial tasks and in the impaired range on verbal tasks may be given an overall composite score in the low-average range—which does little to describe the child's actual abilities and even less in terms of guiding programming.

In cases of significant physical or sensory impairment, such as hemiparesis, clinicians are simply not able to fully and adequately assess the full range of intellectual functioning. Tests that require rapid bilateral fine motor skills have to be modified, thus negating valid interpretation, and replaced with less involved tests that require pointing. These tests cannot be assumed to measure precisely the same skills—and may even be skipped altogether in favor of using scores on verbal-response tests as the primary index and then assuming that the score reflects general capacity across domains. This practice is ill-advised even in normal populations, much less in children where there is evidence of neurologic impact that may differentially affect various skill sets. In general, with children like these, scores on cognitive tests should be carefully interpreted, with cognizance of limitations, and used *as part* of a larger body of neuropsychological assessment that uses more sophisticated and specified measures to best assess the full span of skills that are commonly affected by illness or disability.

The Wechsler scales include the Wechsler Intelligence Scale for Children, 4th Edition (WISC-IV) (75), the Wechsler Adult Intelligence Scale, 3rd Edition (WAIS-III) (76), and the Wechsler Preschool and Primary Scale of Intelligence, 3rd Edition (WPPSI-III) (77). The factor structure of the WISC-IV was significantly changed from the previous edition. The WISC-IV includes a full-scale score made up of four separate composites, each of which is made up of several different subtests. The four composites are verbal comprehension, perceptual reasoning, working memory, and processing speed. The core working memory subtests are primarily verbal in nature, and the core processing speed subtests are primarily nonverbal in nature. The WISC-IV is designed for use with children ages 6–16 years. The WAIS-III is used with individuals ages 16–89 years. It yields a full-scale score comprised of verbal and performance (nonverbal) scaled scores. The verbal scale includes two separate indexes: verbal comprehension and working memory. The performance scale includes the perceptual organization and processing speed indexes. Each index is made up of several different subtests. The WPPSI-III has two different score structures, depending on age level. For children age 2½ to 4 years, there is a full-scale score comprised of verbal, performance, and general language composites. For children ages 4 to 7 years, 3 months, there is one additional composite score: processing speed. Important considerations in the assessment of preschool-age children are addressed in the following section, "Instruments for Use With Young Children."

The Stanford-Binet Intelligence Scales, 5th Edition (78) is designed for use with individuals age 2–89 and up. The full-scale score is made up of five factor indexes: fluid reasoning, knowledge, quantitative reasoning, visual–spatial processing, and working memory. Each factor index includes separate assessments of nonverbal and verbal skills. It should be noted that some of the "nonverbal" tasks require significant receptive language skills, which may complicate interpretation in a child with a basic discrepancy in verbal and nonverbal skills.

The Kaufman Assessment Battery for Children, 2nd Edition (K-ABC-II) (79) was designed for use with children ages 3–18. It is unusual in that guidelines are provided for interpreting results within two different theoretical models: the Luria neuropsychological model and the Cattell-Horn-Carroll psychometric model. Using the Luria model can provide some coherence within a broad neuropsychological assessment. Under this model, there are five scales (sequential processing, simultaneous processing, planning ability, knowledge, and learning ability), each comprised of multiple subtests. There is also a distinct nonverbal index that can be administered entirely through

nonverbal gestures and responses, which can be useful for children with certain disabilities.

Instruments for Use With Young Children

Tests of infant ability have been developed in an attempt to measure developmental status of infants and young children. Such tests are primarily useful in describing current developmental status, with minimal relationship of these early childhood competencies to skills considered crucial during later developmental phases (80). Predictive validity is considered viable only with infants who are significantly developmentally delayed in the first year of life (81,82). Furthermore, tests of infant abilities heavily emphasize assessment of motor skills and cooperative behavior, which are areas compromised in a child with chronic or acquired disability, causing additional complications for achieving test validity in this population.

Research generally indicates that the younger the child, the less predictive intelligence tests are of later test scores and academic performance as the child ages (83,84). The assessment of young children typically requires adaptation and expansion of existing tests to obtain reluctant and valid information. Factors to be considered are that the young child cannot be expected to perform on request and exceptional efforts may be necessary to elicit the degree of responsiveness and cooperation necessary to obtain sufficient and meaningful information. According to Stevenson and Lamb (85), an infant's response to a strange adult-influenced test performance and "sociably friendly" infants scored higher on measures of cognitive competence. Ulrey and Schnell (80) noted that preschool children have had minimal experience with test situations, show minimal concern for responding correctly, and have limited experience with the feedback process that is contingent on being right. Usually, the process of merely asking young children to complete a task may not yield an accurate indication of their capabilities. It is, therefore, incumbent on the examiner to make a judgment about the extent to which the child's performance represents optimal functioning. The likelihood of obtaining ecologically valid information can be enhanced by incorporating observations and analyses of infants' or young children's interactions with the environment (eg, parents, siblings, or caregivers) during spontaneous play.

The Bayley Scales of Infant and Toddler Development, 3rd Edition (Bayley-III) (86) can be used to measure cognitive and motor ability in children age 1–42 months. The cognitive scale measures memory, visual preference, visual acuity, problem solving, number concepts, language, and social development. The language scale measures social communication, semantics, morphology and syntax, prelanguage vocalizations, and comprehension. (Separate receptive and expressive language subtests are included.) The motor scale measures functional grasp and hand skills, object manipulation, visual–motor integration, head control, trunk control and locomotion, motor planning, and quality of movement. (Separate fine and gross motor subtests are included.) There is also a social-emotional scale (covered in the section on psychosocial assessment) and an adaptive behavior scale that is the same as the early childhood version of the Adaptive Behavior Assessment System-II (87), which is covered in the section on adaptive behavior. The Bayley-III is considered the best available instrument for infant assessment (88).

The Brazelton Neonatal Assessment Scale (BNAS) (89) is administered to infants between 3 days and 4 weeks of age to generate an index of a newborn's competence. This scale includes 27 behavioral items and 20 elicited responses to assess. Test scores may be most useful when the test is repeated over the first several weeks of life, so that *changes in scores* can be examined to assess the infant's ability to respond to parenting and recover from the stress of birth. It is this recovery pattern that predicts later functioning in childhood more than a single score (90). Scores have also been used to teach parents how to provide sensitive and confident care to their infants, with small to moderate effects (91).

Alternative Tests of Cognitive Function

Alternative tests of cognitive ability are of particular utility with rehabilitation populations, where patients often have specific impairments (eg, motor impairments, sensory impairments) that preclude the valid use of more common measures. Some of the alternative measures rely less on verbal responding, or reduce requirements for motor output or speed of responding. In a pediatric rehabilitation population, it is often necessary to use alternative assessment measures to accommodate a range of conditions that may interfere with the child's ability to meet requirements of standardized test administration on traditional measures.

Given that many of these alternative measures were designed for particular populations, scores generated are not interchangeable with scores of the major intelligence scales. Furthermore, the special formatting of these tests limits the applicability of results to "real-world" environments, where such intensive accommodations are not always made, and scores may not be as predictive of actual functioning in major settings such as school, home, or community. These instruments may be most useful as screening or supplemental tools in the assessment or interpretation processes.

The Universal Nonverbal Intelligence Test (UNIT) (92) is a test of intelligence that is designed to be completely nonverbal. It can be used with children ages 5–17 years. Administration is done through eight specified pantomime gestures. Responses are also entirely nonverbal, and consist of pointing, paper–pencil, and manipulating items. Multiple standardized teaching items are provided to help ensure that the examinee understands the purpose of gestures. The UNIT is most useful for children who have significant hearing or oromotor limitations, or who do not speak English. Relatively normal fine motor functioning is required for valid use of the test. There are four overlapping scales (memory, reasoning, symbolic, and nonsymbolic), and a full-scale score. The nonsymbolic scale is designed to measure abstract symbolic functioning, which is typically measured through *verbal* scales on cognitive tests. Some children who *can* hear seem to find the examiner's complete reliance on nonverbal pantomime to be somewhat off-putting at first.

The Leiter International Performance Scale-Revised (Leiter-R) (93) is a nonverbal test of intelligence for use with individuals ages 2–20 years. There are two batteries: visualization and reasoning, and attention and memory. The test is administered through nonverbal pantomime. Respondents manipulate items. Motor responses are relatively simple, and thus the test can be used with people with some degree of motor impairment. However, some of the items are scored for speed of response, in which case, even mild motor impairments could yield misleading results. This test is useful with individuals with hearing or oromotor limitations, or who do not speak English.

The Comprehensive Test of Nonverbal Intelligence (C-TONI) (94) is designed to assess intelligence in individuals ages 6–89 years. It includes an overall composite and two subscales: pictorial and geometric. The test can be administered orally or in pantomime. The option of oral administration is for use with children who are not hearing impaired, as these children can be confused when a test is administered completely nonverbally. The C-TONI has the additional advantage of requiring no more complex motor response than pointing to the correct answer. Tests requiring only pointing are sometimes further modified by clinicians to accommodate severely impaired children for whom even pointing is too difficult (eg, the examiner points to each option and the examinee provides indication through predetermined head or trunk movements when the correct choice is reached).

Raven's Progressive Matrices include three separate forms: Coloured Progressive Matrices (95) designed for children ages 5–11, Standard Progressive Matrices (96) for children ages 6–17, and Advanced Progressive Matrices (97) for older adolescents and adults, including individuals suspected of above-average intellectual ability. The tests are brief measures made up of abstract visual arrangements, with the examinee required to select one of multiple choices to complete the arrangement. Instructions can be administered orally or through pantomime. These tests can be used with children with oromotor or hearing impairments, or who do not speak English. The examinee responds by pointing, so it is useful for children with motoric impairment. They are limited as a measure of general cognitive functioning because they assess only one specific type of skill, which may be particularly problematic in a neurologic population where highly specific strengths and weaknesses are often seen.

The Peabody Picture Vocabulary Test-III (PPVT-III) (98) is a receptive vocabulary test, where the respondent is given a vocabulary word and points to the best match from a series of pictures. It is sometimes used as a screening device to estimate verbal cognitive abilities for students with expressive speech and/or motor difficulties, though, of course, great caution is warranted, as the PPVT-III assesses only a single skill set. Visual–perception and native English skills are required. The PPVT-III can be used with children ages 2.6–90+ years.

As noted previously, the K-ABC-II (79) includes a distinct nonverbal index that can be administered entirely through nonverbal gestures and responses, which can be useful for children with certain disabilities. This test requires relatively complex and rapid motor responding, and would not be appropriate for use with individuals with even mild motoric impairment. Table 3.5 provides a complete listing.

Achievement Tests

The assessment of academic achievement represents an integral component of the evaluation of children and adolescents, as school is the "work" of childhood. An important task of assessment is separating academic knowledge from rate of production (referred to as academic fluency) in children with response speed deficits due to motoric impairment or brain injury. Many tests of achievement include a speeded component. Overall scores may be less helpful than specific scores that separate out fluency and basic skills. In addition, academic testing in youth with recent-onset illness or injury may overestimate long-term academic capacity. Academic testing generally measures previously learned knowledge, which may be intact in children whose illness or disability has not yet affected schooling. Whether a child can continue to make progress is a critical question. This is particularly true in brain-injured youth whose deficits in attention, executive functions, and anterograde memory have a strong impact on mastery of new academic skills, and applies to other types of recent-onset

3.5 Alternate Tests of Cognitive Ability

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Universal Nonverbal Intelligence Test (UNIT) (92)	Nonverbal test that measures both symbolic and nonsymbolic cognitive skills in the nonverbal domain. Age range: 5–17.	Requires some fine-motor functioning; designed to reduce cultural bias; easy to administer; useful with individuals with auditory or oromotor limitations, or who do not speak English.
Leiter International Performance Scale-Revised (Leiter-R) (93)	Nonverbal test developed for use with hearing- or language-impaired subjects; measures visual–spatial reasoning and nonverbal attention and memory. Age range: 2–20.	Motor responses are relatively simple, but some items are scored for speed, so motor impairments may affect results. Useful with individuals with auditory or oromotor limitations, or who do not speak English.
Comprehensive Test of Nonverbal Intelligence (C-TONI) (94)	Nonverbal test with pictorial and geometric subscales to measure concrete and abstract nonverbal skills. Only motor skill required is pointing, and this can be further adapted for severely motor-impaired individuals. No time limits.	Nonverbal test with option for oral administration in English-hearing individuals. Useful for individuals with combined limited motor functioning and auditory or oromotor limitations or who do not speak English.
Raven's Progressive Matrices Tests (95,96)	Measures nonverbal reasoning; three different forms for different age ranges; limited motor skills required; advanced version is useful for individuals considered to have above-average intelligence; no time limits	Limited in that it uses a single type of task; useful for individuals with auditory, oromotor, or physical disabilities, or who do not speak English.
Peabody Picture Vocabulary Test-III (PPVT-III) (98)	Multiple-choice test of receptive vocabulary; for individuals aged 2.6–90+; pointing is the only response required, and further adaptations can be made for severely motor-impaired; no time limits.	Useful as a screening device for measuring verbal functioning in children with significant expressive verbal or motor impairments; sometimes used to estimate general cognitive functioning in individuals who cannot participate in other types of assessment, but should be interpreted with great caution.
Kaufman Assessment Battery for Children-II (KABC-II) (79)	General intelligence battery that includes a nonverbal index that can be administered entirely without spoken language. Relatively complex and rapid motor responses are required.	Suitable for individuals with auditory or oromotor impairments, or non-English speakers; not for use with individuals with even mild motor impairment

conditions that place higher coping demands on the child, leaving fewer resources available for basic academic learning.

Some of the more frequently used, individually administered, norm-referenced, and wide-range screening instruments for measuring academic achievement spanning kindergarten through twelfth grade include the Kaufman Test of Educational Achievement, 2nd Edition (K-TEA-II) (99), and the Wechsler Individual Achievement Test, 2nd Edition (WIAT-II) (100), and the Woodcock Johnson Psychoeducational Battery, Third Edition (WJ-III) (101). The Wide Range Achievement Test, 4th Edition (WRAT-IV) (102), is frequently used, but is a brief measure that yields limited information. The Peabody Individual Achievement Test-Revised (103) addresses generally similar content areas as the other major assessment tools, but minimizes the verbal response requirement by using a recognition format (eg, point to correct response based on four choices). Although this format may allow assessment of children presenting with certain impairments, language or motor, the results may not provide the best indication of expectations for student performance in the classroom, where recall and more integrated answers are required.

New assessment guidelines under the Individuals with Disabilities Education Act (IDEA, 2004) for diagnosing learning disabilities in public education settings include options for using response to intervention (RTI), which is a process of assessing progress in skill acquisition in response to scientifically supported interventions, using frequent brief assessments rather

than a single cluster of standardized testing. While RTI is not specified for use in qualifying children under other special education diagnostic categories, such as health impairment, orthopedic impairment, sensory/physical impairment, or brain injury, the RTI model provides a potential structure for assessing *progress* in the school setting.

The use of frequent brief assessments can be useful in the aforementioned situation of recent-onset conditions, where it is important to identify children who are not making sufficient *progress*, despite showing intact pre-injury/illness skills. This method of frequent assessment can also be useful in identification of children who, due to neurologic condition or medication side effects, show significant fluctuations in cognitive functioning. The AIMSweb assessment system (104) provides multiple alternate forms of brief assessments that can be administered weekly. Scores are compared against normative data, and patterns of progress are compared against typical rates of improvement among same-grade students. Various measures are offered in the areas of early numeracy and literacy, math calculation, reading fluency and comprehension, and written expression. There are Spanish versions of some measures. Psychometric data is strongest for the reading fluency measures. Not all school districts use the AIMSweb system.

The Dynamic Indicators of Basic Early Literacy Skills (DIBELS) (105) include literacy measures for grades K–sixth. They can be downloaded at no charge. Guidelines are provided for score interpretation, and *patterns of progress* over time are measured. Physicians should be aware that RTI is provided as an *option* for identification of learning disabilities under federal law. Not all school systems will have a structure in place for using it, but for those that do, *inclusion of the patient in the RTI process may yield valuable information*. Table 3.6 provides a listing of achievement measures.

Adaptive Behavior

Adaptive behavior includes behaviors and skills required for an individual to function effectively in everyday life at an age-appropriate level of independence. The American Association on Mental Retardation (AAMR) distinguishes three major categories of

3.6 Measures of Achievement

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Kaufman Test of Educational Achievement-II (KTEA-II) (99)	Reading (decoding and comprehension), math (computation and applications), and written language composites (spelling and composition), as well as additional subtests measuring reading-related skills and oral language. Ages 4.6–25.	Age- and grade-based norms provided; norms broken down by fall, winter, spring; reading-related subtests help identify specific deficits in phonological awareness or rapid naming.
Wechsler Individual Achievement Test-II (WIAT-II) (100)	Subtests measure pseudoword decoding, word reading, comprehension, numerical operations, math reasoning, written expression, spelling, oral language, and listening comprehension. Ages 4–85.	Age- and grade-based norms provided; norms broken down by fall, winter, spring; co-normed with the Wechsler Intelligence Scale for Children-IV to promote statistically sound comparisons between IQ and achievement scores.
Woodcock Johnson III Tests of Achievement (WJ-III) (101)	Scales assess reading, oral language, mathematics, written language, and knowledge. Separate scales assess basic skills, applications, and fluency for reading, math, and written language. Multiple additional scales of highly specified skills are included. Ages 2–90+.	Age- and grade-based norms provided; scoring provided through use of computer software only; lack of hand-scoring option limits clinician in interpretation in some cases; specific fluency scores useful in populations with processing speed deficits; written expression subtest relatively simplistic.
Wide Range Achievement Test-IV (WRAT-IV) (102)	Subtests include sentence comprehension, word reading, spelling, and math computation Ages 5–94.	Brief measure that does not assess some critical aspects of academic functioning.
Peabody Individual Achievement Test-Revised (PIAT-R) (103)	Includes subtests for general information, reading recognition, reading comprehension, mathematics, spelling, and written expression. Ages 5–18.	Uses a recognition format that accommodates individuals with language and motor impairments; measures relatively limited set of skills compared to other tests

adaptive functioning. Conceptual skills include language, functional academics, and self-direction. *Social* skills include establishing friendships, social interaction, and social comprehension. *Practical* skills include basic self-care skills and navigation of home, school, and community tasks and environments. In later adolescence, vocational functioning is also assessed as part of the practical domain.

Deficits in adaptive behavior are one of the core criteria in determining a diagnosis of mental retardation, along with significantly impaired intellectual functioning. Adaptive functioning is assessed primarily through structured interviews and rating scales completed by persons familiar with the child in natural settings, such as parents and teachers. These scales are open to the response bias inherent in this type of assessment, but are also directly linked to programming assistance. There is great utility in using responses to adaptive skills to identify target skills for rehabilitation. Several issues are especially noteworthy in using these assessments with rehabilitation populations. First, adaptive scores may be disparate with intellectual testing scores in a traumatic brain injury population, because they represent more procedural learning and are often less affected directly after the injury. The failure to gain subsequent abilities can be a source of substantial disability as time goes on, due to impairments in sensory or cognitive abilities. Second, in contrast to individuals with developmental mental retardation, who may be expected to show a general pattern of mastery of easier skills and nonmastery of more difficult skills on each scale, the rehabilitation population is more likely to show uneven peaks and valleys across skills even within the same domain. For example, a person with motoric impairment may struggle with some "easier" self-care skills, but have the cognitive and adaptive ability to handle more "difficult" skills in the same domain. In these individuals, standardized scores may not provide a meaningful picture, but analysis of specific items can provide direction for rehabilitation programming.

The Vineland Adaptive Behavior Scales-II (106) is a widely used set of scales that has four forms: Survey Interview, Parent/Caregiver Rating, Expanded Interview, and Teacher Rating. Each assesses four broad domains. The communication domain assesses expressive, receptive, and written communication. The daily living skills domain assesses personal, community, and domestic skills. The socialization domain assesses interpersonal relationships, play and leisure time, and coping skills, The motor skills domain assesses fine and gross motor skills for young children. The domain scores are combined to yield a composite index. A maladaptive behavior domain surveys inappropriate social or behavioral displays. The survey interview and rating scales take 20–60 minutes to complete, while the expanded interview is lengthier. The second edition includes updated content and increased coverage of early childhood adaptive behavior for use down to early infancy.

The Adaptive Behavior Assessment System-2 (ABAS-2) (87) includes five forms, each taking 15–20 minutes to complete: Parent/Primary Caregiver form for birth to 5 years, Teacher/Daycare Provider Form for children ages 2–5 years, the Teacher Form for ages 5–21 years, the Parent form for ages 5–21 years, and the Adult form for ages 16–89. In the second edition of the system, the domains are closely aligned with the AAMR definition of adaptive behavior. The conceptual domain assesses communication, functional academics (or pre-academics), and self-direction. The social domain assesses leisure and social skills. The practical domain assesses self-care, home/school living, community use, health and safety, and, for older adolescents and adults, work skills. The scales are well validated. Table 3.7 provides a complete listing of these tests.

Psychosocial Evaluation

The assessment of psychosocial status has different conceptual bases, depending largely on the age of the child. A multimethod, multisource assessment is critical, as different sources are sensitive to different areas of functioning (107). Structured interview, observational methods, performance evaluation, and careful analysis of both medical data and psychosocial variables should be combined, and, where possible,

3.7 Measures of Adaptive Functioning

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Vineland Adaptive Behavior Scales-II (106)	Age: Birth to 90 years. Measures four domains: communication, daily living skills, socialization, and motor. Also includes a maladaptive behavior scale.	Assessment of adaptive motor skills relevant for a rehabilitation population. Rating scale and interview formats available.
Adaptive Behavior Assessment System-II (ABAS-II) (87)	Age: Multiple scales covering birth to 89 years. Measures three domains: conceptual, social, and practical.	Composite areas specifically match AAMR guidelines.

Abbreviation: AAMR, American Association on Mental Retardation.

multiple sources of information should be included, such as parents, teachers, and child self-report.

Caveats

One of the trickiest issues in psychosocial assessment in rehabilitation populations is the need to account for the biologic factors on assessment results. Most psychosocial assessment tools are not specifically designed for use with children with disabilities or chronic illness. It must be appreciated that a wide range of adjustment levels exists. While children with chronic physical conditions appear to be at increased risk for psychological adjustment problems, the majority of children in this population do not show evidence of maladjustment (107). Furthermore, assumptions based on group membership by disability or medical condition can be inaccurate. For example, intuitive reasoning would indicate that individuals with disfigurements, such as amputations or burns, would be particularly affected. Such is not the case, however, as demonstrated in research of these groups (108).

It is important to be aware that some items on psychosocial assessment scales can elicit medical as opposed to psychological distress. Particularly in children, "somatization"—or the tendency to express high levels of physical symptoms—is often assessed in scales measuring emotional functioning. A high level of somatization is considered indicative of internalizing problems such as depression and anxiety in general child populations, and high somatization scores can lead to high scores on composite scales meant to measure general internalizing problems. Obviously, in youth with chronic illness, the extreme physical symptoms relating to the medical condition may, *even in the absence of other areas of significant symptomology,* yield a score on the somatization subscale that is high enough to lead to elevated "total" emotional symptoms scores. It is incumbent on the professional to analyze the general profile and individual items in these cases. If there are low rates of *other* indicators of emotional distress besides those symptoms specific to the medical condition, it is important not to overinterpret the elevated scores. At the same time, high total scores should not be disregarded just because they are in part due to medical symptoms, as this population does frequently show elevated symptoms of distress, even when somatic items are not included in scoring (109). An intimate familiarity with the items making up the measure and the specific variables associated with the individual child's medical condition is required for psychosocial assessment in this population. Physicians should be wary of scores provided by school and community clinicians who are not specifically familiar with the challenges in assessment for this population. *Referral to clinicians who specialize in pediatric rehabilitation should be strongly considered when psychosocial concerns are an issue.*

Unique to the arena of personality of psychosocial functioning is the empirically based or criterion-group strategy of assessment. This approach grew in response to the serious liabilities presented by self-report tests, which used items that had face validity. For example, an item that asks about arguing with others was a direct question, just as could be asked in a live interview. There are great liabilities to that approach; it assumes that subjects can evaluate their own behavior objectively, that they understand the item in the way it was intended, and that they chose to respond candidly. In a radical departure, the developers of what came to be know as the Minnesota Multiphasic Personality Inventory (MMPI) formulated the test with the main premise that nothing can be assumed about the meaning of a subject's response to a test item—the meaning can be discerned only through empirical research. Items are presented to criterion groups, such as depressed, schizophrenic, or passive-aggressive personality disorders, and control groups. By their answers as a diagnostic group, the items *become* indicative of a given disorder or personality outplay, regardless of what the content of the items was or an intuitive judgment of what it *should* indicate. This approach also allows for the determination of respondent's bias—whether an adolescent self-reporting, as in the case of the Minnesota Multiphasic Personality Inventory-Adolescent (MMPI-A), or parents filling out a behavioral checklist such as the Personality Inventory for Children-2.

In young children, temperament is a more cogent concept than that of personality. The dynamics of psychological functioning are the effect of innate temperament in interaction with parents and other caregivers within the basic sensorimotor exploratory nature of infancy and early childhood. If school is children's work, play is the work of this youngest group. What an interview or a self-report measure yields in older children, the observation of play provides in the preschooler. To quote Knoff (110), "This information reflects the preschooler's unique perceptions of his or her world, perceptions that are important in any comprehensive assessment of a referred child's problems." Projective techniques such as the Rorschach are not recommended in this population because of the need to interpret ambiguous visual stimuli. The active developmental maturation of visual–perceptual systems and the attendant normative variability mitigate against the appropriateness in preschoolers.

Individual Assessment Tools

Functional behavior assessment (FBA) is highly appropriate when young children, as well as older youth, with disability or illness are displaying significant

behavior problems (111). When the ability to effectively communicate or independently access one's wants and needs is inhibited by cognitive or physical disability, rates of inappropriate behaviors can increase as the child learns (sometimes subconsciously) that these behaviors can effectively serve a function. FBA is a structured assessment method for determining the underlying function (ie, purpose) of inappropriate behavior. This assessment method has the advantage of being directly linked to intervention strategies—when a function is identified, environmental interventions can be developed to teach the child to use more appropriate behaviors to meet his or her purpose. There is an adaptive emphasis for children who cannot use developmentally appropriate language or mobility, and children with even severe impairments in cognitive, language, sensory, or motor functioning can be assessed through this method. Functional Behavior Assessment includes structured interviews examining the antecedents and consequences of behavior, structured observations of behavior in naturalistic settings to identify environmental mediators, and experimental manipulation of environmental conditions (functional analysis) to determine whether behaviors serve to meet children's need for attention, tangible items or activities, to escape from nonpreferred situations, or to meet internal needs, such as the release of endorphins through self-injury.

Transdisciplinary play-based assessment (TPBA) (112) is a standardized observation of play. It provides an exhaustive listing of developmentally cogent play behaviors under four domains: cognitive, language and communications, sensorimotor, and social–emotional development. It allows the child to engage in the most natural of activities, but is limited in that there may not be an expression of a specific behavior of interest but rather a global picture of the child in interaction with the environment. Because of the limitations of individually administered tests in the young child, this acts as cross-validation of parental report and is less influenced by the demanding characteristics of traditional testing. The advantage of hearing spontaneous language production is particularly useful, for this is often the primary *shutdown* of younger children in an evaluation setting (113). There are other systems for play observation. Some are designed for the more evocative structure of play designed to tap certain themes (eg, abuse) used in children. In the rehabilitation population, nonpathologic issues such as adjustment and developmental integrity predominate, so the TPBA offers an excellent choice.

The Bayley Scales of Infant and Toddler Development, 3rd Edition (86) provides a normative framework for this domain by providing scaled scores for the popular Greenspan Social-Emotional Growth Chart (114), which is a parent-report instrument to assess early indicators of social–emotional functioning in children ages 0–42 months.

The Minnesota Multiphasic Personality Inventory MMPI-A (115) is based on the criterion group strategy described in the introductory comments to this section. It is the first revision of the original MMPI specifically for use with adolescents. For the original test (MMPI), adolescent norms were developed in the 1970s, but it was only a downward extension at best. Now, new items tap specific adolescent developmental or psychopathologic issues. There are new supplemental scales that give feedback relative to alcohol and drug problems and immaturity. There are 15 new content scales in addition to the original 10 clinical scales. Development of the validity and response bias of the subject was expanded by devising response-inconsistency scales.

The original MMPI interpreted with adolescent norms had been used extensively with adolescent medical populations, including those with physical disability (116). For the development of the MMPI-A, extensive rewriting and some revision of test items were done. A national representative adolescent sample was used for normative data (not the case in the original MMPI). The new length is 478 test items presented in a booklet form, with true/false response. Reading level required is best considered to be seventh grade, although it had been designed with the goal of fifth-grade comprehension. In actuality, the range is from fifth to eighth grade. The test is available in an audiotape format as well, which takes about 90 minutes. Each item is read twice. This aspect was designed for access by the visually impaired, but doubles for individuals who have reading comprehension problems. Language comprehension level required for the audiotape format is fifth grade. A computer-administered form is also available that presents items singly and with a response entered on the keyboard.

The effective use of the MMPI-A with pediatric rehabilitation patients is contingent upon cautious interpretation. For example, elevated scores on scales such as "hypochondriasis" or "lassisitude-malaise" will be interpreted differently in a patient with chronic illness than in general populations. A correction factor is recommended for use with spinal cord injury to obviate responses to items that reflected the reality of the medical condition, as opposed to the criterion value assigned to the item (117). Recommended uses for the MMPI-2, which would also appear appropriate for the MMPI-A, in medical assessments include assessment of response bias, as the validity scales allow for assessment of the accuracy of the patient's self-report, identification of emotional distress factors relating to the medical condition that may influence recovery, and comorbid psychiatric conditions that would be expected to affect recovery and participation

in rehabilitation. Attempts to use the MMPI-2 (and likely the MMPI-A) to differentiate between organic and functional conditions are discouraged, as research suggests that elevated scores on scales suggestive of somatic preoccupation can reflect the *effect*s of the medical condition (118).

The Personality Inventory for Children, 2nd Edition (119) is a behavior rating scale for children ages 5–19. It is comprised of 275 items to be completed by a parent. There is a brief form that takes about 15 minutes to complete. Composite scales include cognitive impairment, impulsivity and distractibility, delinquency, family dysfunction, reality distortion, somatic concern, psychological discomfort, social withdrawal, and social skill deficits. Three validity scales are designed to assess response biases, including inconsistency, dissimulation, and defensiveness, that may invalidate responses. Sattler (120) finds that additional research is needed on the reliability and validity of this new version of the scale, and there have been some concerns noted about the use of previous versions with specific rehabilitation populations—notably those with brain injury.

The Achenbach System of Empirically Based Assessment (121,122), including the Child Behavior Checklist for Ages 6–18 (CBCL/6–18), the Child Behavior Checklist for ages 1.5–5 (CBCL/1.5–5), the Youth Self-Report (YSR), and Caregiver-Teacher Report Forms (TRF), are commonly used measures of psychosocial adjustment. They were each developed through factor analysis (or the *statistical* grouping of items into clusters/scales, as opposed to using clinical judgment to group items), but also include DSM-oriented scales developed through clinical judgment. Broad domains include internalizing symptoms and externalizing symptoms. The CBCL/6–18, TRF, and YSR each include 112 items in eight scales. The CBCL and TRF are designed for completion by parents or teachers, respectively, of children ages 6–18 years. The YSR is designed for self-report of adolescents ages 11–18, and requires a fifth-grade reading level. The CBCL/1½–5 and Caregiver-Teacher Form, for use with younger children, each consist of 100 items, separated into seven and six scales, respectively. The scales are commonly used in children with chronic physical conditions (107). Limitations of its use with children in this population include limited sensitivity to milder adjustment problems, a possible confound by medical symptoms, incomplete assessment of social functioning, and methodological concerns (123).

The Behavior Assessment System for Children-2 (BASC-2) (124) includes three parent rating scales (Preschool, ages 2–5 years; Child, ages 6–11; and Adolescent, ages 12–21); three teacher rating scales, following the same age ranges; and three self-report of personality scales (Child, ages 8–11 years; Adolescent, ages 12–21 years; and Young Adult, ages 18–25 years, attending a post-secondary school). Each scale takes 20–30 minutes to complete and requires a third grade reading level. Parent rating scales include composite scores for adaptive skills, behavioral symptoms, externalizing problems, and internalizing problems. Teacher rating scales measure these four areas and add a school problems scale. The self-report scales include composite measures of emotional symptoms, inattention/hyperactivity, internalizing problems, personal adjustment, and school problems. The BASC-2 scales also include several indexes to measure response sets that would indicate invalid scores, such as high rates of negative answers, high rates of positive statements, endorsement of nonsensical or implausible items, or inconsistent responses. The BASC-2 system is well validated and provides an integrated multisource system of assessment (120).

The Rorschach Inkblot Technique (125) remains a widely used test in children and adolescents. It is the classic technique of 10 inkblots presented with the instruction to say what it looks like to the examinee. An alteration in administration with younger people is to follow up each card with the inquiry, asking why it looked like whatever the response was, whereas with adults, this is done only after all blots are viewed. Normative data on this technique for children and adolescents began appearing in the 1970s; however, these are not representative of the general population, being overrepresentative of children with above-average intelligence, with incomplete attention to race and socioeconomic status (125). Despite the fact that some norms exist down to age 2 years, most authors agree that the Rorschach should not be used with children below the age of 5 years. There is little experience with this type of test in assessing the type of adjustment issues common to the rehabilitation population. Therefore, it should be used guardedly.

Children's Apperception Test (CAT) and Thematic Apperception Test (TAT) (126) represent another type of projective test, but this time, the stimuli are ambiguous pictures and the subject is asked to make up a story concerning what is happening, what led up to the scene in the picture, and what will happen next. It requires considerable skill on the part of the examiner, and should be given only by the professional, as is the case with all projective techniques. There is usually follow-up questioning about the story given, and the recording is verbatim. There are no real normative data on the CAT, but some authors believe that it remains a powerful technique in discerning children's personalities (127). Some believe it taps themes of confusion and conflict, with the child's resolution being a central focus of interpretation. It is based on the author's personality theory as opposed to a pathologic model. The entire set contains 20 cards, although a standard administration

uses only selected pictures. Over the years, individual cards have been identified as being particularly useful with certain age groups. There are concerns regarding lack of adequate reliability and validity data (120).

In these days of cost-efficiency considerations, more specific measures are of great utility. The choice of a specific construct is often suggested by the results of other examinations or by knowledge of the presenting problem. Anxiety is a common correlate of chronic physical conditions (128). The Revised Children's Manifest Anxiety Scale for Children (RCMAS) (129) is a single-construct measure of anxiety. The RCMAS has 37 short statements to which the child responds yes or no. There is a total anxiety score, as well as a lie subscale that examines the candidness and honesty of the response set. The brevity of the instrument results in the three anxiety subscales that can be generated but are of limited use. The standardization sample was large and representative of socioeconomic status, demographics, race, and gender. Validity and reliability are extensively reported in the manual and are helpful in informed interpretation. Reading level is third grade, so a wide variety of children and adolescents can use this device. Because of its brevity and specificity, it should be only one part of a battery.

The Children's Depression Inventory (CDI) (130) is a well-recognized self-report measure of depressive symptoms in children ages 7–17 years. There are five subscales: negative mood, interpersonal problems, ineffectiveness, anhedonia, and negative self-esteem. Reliability for the total score is stronger than for subscales. Though a popular measure, questions have been raised about the psychometric properties (120).

The Behavior Rating Inventory of Executive Function (BRIEF)system (131) includes a preschool version of Parent and Teacher Rating Scales (ages 5–18) that can be completed by parents or teachers/daycare providers (ages 2–5) and a Self-Report (ages 11–18). The behavioral rating of executive functioning is an important addition to the assessment of psychological functioning in any child with neurologic impairment. Soliciting the observation of executive functioning in natural environments is especially important in light of previously mentioned concerns regarding ecological validity of clinical tests of executive functioning due to the highly structured, directive nature of clinical assessment. The preschool version of the BRIEF includes three broad indexes—inhibitory self-control, flexibility, and emergent metacognition—and a global composite, as well as two validity scales to identify excessive negativity or inconsistency in responding. The other versions have two broad indexes—metacognition and behavioral regulation—and a global composite, as well as the two validity scales. Table 3.8 provides a complete listing.

Family Environment

The instruments noted here are part of the ever-growing recognition of the pivotal importance of family functioning in the face of a child's disability and adjustment. The most dramatic impetus has been the requirement of a family service plan in all early-intervention services for children up to 3 years of age. Beyond the case to be made in the youngest age group, many studies show a strong relationship between family functioning and a child's psychological adjustment across a number of different medical conditions (132). The importance of such considerations is clear. The following are synopses of two widely used instruments for populations often within the scope of a rehabilitation practice.

The Home Observation for Measurement of the Environment Sale (HOME) (133) is a checklist designed to assess the quality of a child's home environment. It is an involved process including observation of the home setting and interview with parents. Six areas are assessed: responsiveness of parent, parental acceptance of child, organization of physical environment, provision of appropriate play materials, parental involvement with child, and opportunities for variety in stimulation. In young children, the home setting is a strong predictor of later functioning.

The Family Environment Scale (FES) (134) rates parental perception of the social climate of the family, and is rooted in family systems theory. It contains 90 true–false items that break down into 10 subscales: cohesion, expressiveness, conflict, independence, achievement orientation, intellectual–cultural orientation, active-recreational orientation, moral–religious orientation, family organization, and family rules. Scores are plotted on a profile, with two forms available—the actual state of the family as perceived by individual members and the ideal state. Profiles derived from each parent can be compared, from which the family incongruence score is calculated.

There has been controversy about the psychometric properties of the FES relative to the stability of its factor structure. It was suggested that the factor structure varies, depending on which family member's perceptions were used. There is some caution expressed about its use as a clinical diagnostic tool in a rehabilitation setting with adults (135). Others have used it successfully in studies of children with chronic medical conditions. In one such study by Wallander and colleagues (136), family cohesion made a significant contribution to social functioning in children with spina bifida. A measure of family functioning specific to children with disabilities

3.8 Measures of General Psychosocial Functioning

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Functional Behavior Assessment (FBA)	A style of observation-based behavioral assessment geared toward identifying the underlying purpose of problem behavior.	Results are directly linked to interventions for behavior change. Can be successfully used with individuals with severe disabilities in any domain.
Transdisciplinary Play-Based Assessment (TPBA) (112)	Normed for 6 months to 6 years. Administered in home or clinic. Structured play observation.	Designed with intervention development as primary goal. Taps a naturalistic activity; more engaging for young children.
The Bayley Scales of Infant and Toddler Development-III (86)	Ages 0–42 months. Provides normative framework for major social–emotional milestones.	Co-normed with the cognitive measures on the Bayley Scales.
Minnesota Multiphasic Personality Inventory-Adolescent (MMPI-A) (115)	Objective self-report for adolescents ages 14–18. Revision of most widely used personality test for this age. Detailed assessment of response bias.	Excellent standardization and psychometric properties. Audiotape administration available. Likelihood of continued widespread uses facilitates comparison across different groups. Length can be problematic in terms of engagement by subjects. Some subscales specifically measuring physical complaints must be interpreted carefully.
Personality Inventory for Children-2 (PIC-2) (119)	Two versions cover ages 3–16 years. Parent report rating scale. Separate norms for mother and father as respondents. Assesses response bias.	Well normed for clinical population, but less research in rehabilitation population. Some concerns noted in use with brain injury.
Achenbach System of Empirically Based Assessment (121)	Includes parent report (CBCL), and teacher report (TRF), scales ranging from ages 1.5–18 years, and a self-report scale (YSR) for ages 11–18. Empirically driven and *DSM*-oriented scales provided.	Parent and teacher forms are widely used instruments in rehabilitation and nonrehabilitation populations. Does not assess response bias. Subscales measuring physical complaints must be interpreted carefully in a rehabilitation population.
Behavior Assessment System for Children-2 (BASC-2) (124)	Age: Parent and teacher scales range from 2–21 years. Self-report scales range from 8–25 years. Several scales measuring response bias.	Computer-scoring program provides easy comparison of information from multiple sources. Subscales measuring physical complaints must be interpreted carefully in a rehabilitation population.
Rorschach Inkblot Technique (125)	Projective personality test using inkblots as ambiguous stimuli. Standardized scoring norms provided for ages 5–16.	Psychometrically unsound. Concerns regarding impact of visual–perceptual impairments in rehabilitation population.
Children's Apperception Test (CAT) and Thematic Apperception Test (TAT) (126)	Projective personality test using ambiguous pictures. Some structured scoring.	Assesses themes of confusion and conflict, but requires careful interpretation. Absence of psychometric/normative data.

DSM, Diagnostic and Statistical Manual of Mental Disorders.

(PCDI) is presented in the following section on population-specific assessments. Table 3.9 provides a full listing of these tests.

Population-Specific Assessments

While most of the measures listed previously are designed for general use in the assessment of psychosocial functioning in children and adolescents, an increasing number of measures are being developed specifically for use with pediatric rehabilitation populations. Population-specific measures are more sensitive to the unique adjustment challenges that these youth face.

The Parents of Children with Disabilities Inventory (PCDI) (137) was designed to assess not only the frequency of disability-related stressors, but also parent perceptions of the stressors, which are an important factor in family adjustment. Four areas of concern are measured: medical and legal, concerns for the child,

3.9 Measures of Single Dimensions

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Revised Children's Manifest Anxiety Scale for Children (RCMAS) (129)	Self-report of anxiety. Includes a lie scale to assess response bias.	Items assessing physiological symptoms must be interpreted with caution in rehabilitation population.
Children's Depression Inventory (CDI) (130)	Self-report measure of depression. Five subscales: negative mood, interpersonal problems, ineffectiveness, anhedonia, and negative self-esteem.	Well-recognized scale. Some questions have been raised about the psychometric properties.
Behavior Rating Inventory of Executive Function System (BRIEF) (131)	Parent, teacher, and self-report rating scales. Measures behavior regulation and metacognition. Two response-bias scales included.	Allows for assessment of executive skills in naturalistic environment, which is important, as this can be hard to validly assess in clinical settings.

concerns for the family, and concerns for the self. Limited psychometric data is available, though initial estimates of reliability and concurrent and construct validity appear adequate. Further validation and normative studies are needed.

The Pediatric Inventory of Neurobehavioral Symptoms (PINS) (138) has the advantage of having been specifically designed for the assessment of personality, emotional, and behavioral issues associated with traumatic brain injury. It has the disadvantage of having less research support, though there is some evidence of construct validity. It is comprised of 54 items, and can be completed by parent or teacher. Five general scales are obtained: mental inertia, social inappropriateness, dissociation of affect and behavior, episodic symptoms, and biologic symptoms.

The Pediatric Pain Questionnaire (PPQ) (139) is a structured interview completed with patients and parents. It measures both pain intensity and location, using body outline and visual analogue, as well as the emotional and perceptual experience. There are separate forms for children, adolescents, and parents. The adolescent form also covers the social and environmental influences on the experience.

History taking is an integral part of the process, including extensive history of treatments, child and family pain history, and environmental aspects. The analogue scale provides no numbers or markings, but instead elicits present and worst pain intensity of the past week. Different semantic anchors are used for children (*not hurting* versus *hurting a lot*), along with happy and sad faces. The adolescent and parent versions are anchored by *no pain* and *severe pain* and pain descriptors of hurting and discomfort. The body outlines are age-appropriate on the children and adolescent forms. The child can indicate four levels of pain intensity by coloring in the body outline with a choice of eight crayons. The child chooses colors to demonstrate the intensity gauged by four categories of pain descriptors. In this way, the child can show multiple sites and register the appropriate range of intensity in each. A separate list of pain descriptors is provided that assesses the evaluative, emotional, and sensory quality of the child's own experience. Words are provided for younger children or anyone who may have trouble generating labels.

The multidimensional aspect of the PPQ is appealing for anyone who has struggled to understand the experience of pain in children. It allows for engaging visual representations as well as standard language expression. Expecting parent reports to match the child's is erroneous. As in the adult literature, the subjectivity of the pain experience mitigates against this being the case. Comparison of child and parent reports is useful more as a gauge of convergence in the relationship between parent and child, not as a validating measure. Despite the unusual structure of some of its components, reliability and validity have been shown for the PPQ, and it holds considerable promise.

Measurement of health-related quality of life (HRQOL) represents an important component in the assessment of psychosocial functioning in pediatric populations. The PedsQL (140) is designed to measure HRQOL through brief child and/or parent ratings, with separate scales designed for different age groups within the 2-to-18-year range. Physical, emotional, social, and school functioning scales are included in the generic core scale, and supplemental condition-specific modules are available for asthma, rheumatology, diabetes, cancer, and cardiac conditions. Additional disease-specific measures of HRQOL are available for use with other populations such as epilepsy (141) and cystic fibrosis (142). A listing of population-specific measures is shown in Table 3.10.

The assessment of disease-related knowledge should not be overlooked. Most children with chronic

3.10 Population-Specific Measures

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Parents of Children with Disabilities Inventory (PCDI) (137)	Assesses frequency and perceptions of family stressors in the areas of medical/legal, concerns for child, concerns for family, concerns for self.	Limited psychometric data available. Assessment of perceptions of stressors is important, as this construct is related to adjustment.
Pediatric Inventory of Neurobehavioral Symptoms (PINS) (138)	Designed to assess sequelae associated with traumatic brain injury. Five domains assessed: mental inertia, social inappropriateness, dissociation of affect and behavior, episodic symptoms, and biologic symptoms.	Limited research on scale, though some construct-validity data is available.
Pediatric Pain Questionnaire (PPQ) (139)	Assesses pain intensity and location, as well as emotional and perceptual experience. Different scales for children, adolescents and parents.	In-depth assessment of highly subjective experience.
PedsQL (140)	Measures health-related quality of life through child and parent ratings. Generic core scale measures physical, emotional, social, and school functioning. Condition-specific modules available for asthma, rheumatology, diabetes, cancer, and cardiac conditions.	Measures important aspect of functioning in pediatric populations. Disease-specific measures tap unique issues within separate illnesses.

illness or disability face the dual challenge of needing to cope with higher demands (as compared to normal populations) in terms of medical treatment regimens, using lower general coping resources due to primary symptoms and secondary deficits. Treatment adherence is of critical concern. Assessment of general developmental maturity and psychosocial adjustment is a key indicator for addressing this issue. There is also evidence that knowledge of the disease and treatment is important in children and especially adolescents (143). Informal assessment of patient understanding may help identify barriers to treatment adherence.

CONCLUSION

This chapter seeks to be a reference primarily to the physician, but also all potential rehabilitation team members. It details the uses of psychological assessment and hopefully acts as a primer of sort on how to be a "good consumer" of such services. Since the first edition of this book an important development in the CPT codes has occurred. In 2002, the addition of the health and behavior assessment and intervention codes established diagnostic interview and interventions for psychosocial adjustment and psychoeducational purposes as a legitimate and billable activity. Prior to this, treatment or even a referral for evaluation would require assigning a psychiatric diagnosis for CPT coding, which was wholly inappropriate. With this change, there is now a complete framework for the supportive and intervention role of mental health staff to assist patients and families in the significant coping challenges in the medical setting. This recognition removes all barriers to the inclusion of valuable psychosocial interventions to enable our patients and their families to have the services vital to the optimum outcome of the rehabilitation process.

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4

Language Development in Disorders of Communication and Oral Motor Function

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Communication, as defined by the National Joint Committee for the Communicative Needs of Persons with Severe Disabilities (1), refers to "any act by which one person gives to or receives from another person information about that person's needs, desires, perceptions, knowledge, or affective states. Communication may be intentional or unintentional, may involve conventional or unconventional signals, may take linguistic or nonlinguistic forms, and may occur through spoken or other modes." Communication is clearly a dynamic process used to exchange ideas, relate experiences, and share desires.

It takes a variety of forms, including speaking, writing, gesturing, and sign language. As we know, interference with the physical ability to perform any of these acts has a significant impact on communication. Oral motor and neurologic impairments that affect communication may also significantly affect swallowing.

The purpose of this chapter is to provide a basic understanding of the acts of communication and swallowing, as well as an understanding of the primary disorders resulting from abnormal development or acquired injury of structures or systems related to these acts. The chapter is divided into two parts: The first part describes speech and language development and disorders. First, the primary components of speech and language are defined and described, and brief examples of deficits that result from disruption in these components are provided. Acquisition of speech and language skills is then outlined, including primary milestones for each. Some of the most common speech and language disorders, both developmental and acquired, as well as common associated disorders, are then outlined. Finally, speech and language assessment and intervention are briefly described.

The second part describes feeding and swallowing processes and disorders. Development of feeding skills, including expected milestones, is described. Anatomy and physiology of the swallowing mechanism is illustrated and described. Common disorders of deglutition, both congenital and acquired, are described. Finally, feeding and swallowing assessment and intervention are addressed.

SPEECH AND LANGUAGE DEVELOPMENT AND DISORDERS

Within the field of communication sciences and disorders, we think of communication as broadly

comprised of speech and language. Speech generally refers to aspects of communication that involve motor output for production of speech sounds. Production of speech sounds requires functional input from respiratory, phonatory, and articulatory systems (Table 4.1). Language generally refers to the process by which we both encode and process meaning within messages, and is divided into three primary components: form, content, and use. These components can be further subdivided based on five key aspects of language—specifically, phonology, morphology, syntax, semantics, and pragmatics.

Speech Components

As noted previously, production of speech requires input from *respiratory*, *phonatory*, and *articulatory* systems. An airstream is generated by the lungs, passes through the vocal cords, and is then shaped by the articulators to form speech sounds. Impairments in any of these systems most likely will have a significant impact on speech production.

Respiration

The *respiratory* system is composed of the upper and the lower airways. The upper airway consists of the nose, mouth, pharynx, and larynx, and the lower airway consists of the tracheobronchial tree and the lungs (2).

Upper Airway. The upper airway has many functions. The mucous membranes covering much of the upper airway structures are softer, looser, and more fragile in infants and young children than in older children and adults, and more susceptible to edema and injury from trauma.

4.1 Components of Speech

RESPIRATORY	PHONATORY	ARTICULATORY
Upper Airway	*Larynx*	Lips
Nose	Vocal Cords	Tongue
Mouth	Cartilage	Palate
Pharynx	Muscle	
Nasopharynx	Mucous	
Oropharynx	Membrane	
Hypopharynx	Ligaments	
Lower Airway		
Trachea		
Lungs		

Nose. All children are obligate nasal breathers during the first six months of life, during which time the soft palate is in close anatomic approximation with the epiglottis. This factor, combined with the relatively large size of the tongue relative to the oral cavity at this age, renders nasal patency essential for maintaining an airway. Those children with nasal obstructions such as choanal atresia are at risk for respiratory compromise (cyanosis) during feeding.

Mouth. The lips, mandible, maxilla, cheeks, teeth, tongue, and palate are the most important components of the oral cavity with regard to manipulation of airflow for respiration and speech production. The infant tongue takes up a larger area in the mouth and rests more anteriorly in the oral cavity than that of the adult. There are numerous congenital craniofacial anomalies, often associated with syndromes, that have an adverse impact on airflow. Some anomalies, such as cleft palate, prevent sufficient valving of the airstream, resulting in inaccurate production of speech sounds. Other anomalies, such as glossoptosis (oropharyngeal or hypopharyngeal obstruction during feeding caused by tongue retraction, and common in Pierre Robin Sequence), can result in blockage of the airstream and subsequent respiratory distress.

Pharynx. The pharynx, a muscular tube shared by the respiratory and digestive tracts, is sometimes referred to as the aerodigestive tract, and serves vital functions for both respiration and swallowing. It is divided into three portions: the nasopharynx, oropharynx, and the hypopharynx. The pharynx in an infant is gently curved, and as the child grows and develops, the angle increases to approximately 90 degrees.

The *nasopharynx* is the portion of the pharynx directly behind the nasal cavity, extending from the roof of the nasal cavity to the roof of the mouth. In addition to conducting air, the nasopharynx acts as a resonator for voice. The Eustachian tubes from the middle ear open into the nasopharynx.

The *oropharynx* is that portion of the pharynx directly behind the oral cavity, extending from the roof of the mouth (pharyngeal aspect of the soft palate) down to the base of the tongue, at the level of the tip of the epiglottis. Movement of the pharyngeal walls in this portion, together with elevation of the soft palate and the posterior portion of the tongue, are crucial for velopharyngeal closure. Inadequate closure, or velopharyngeal incompetence, can result in disordered speech production.

The *hypopharynx* extends from the base of the tongue at the level of the hyoid bone and tip of epiglottis down to the entrance of the larynx and esophagus.

Lower Airway

The lower airway consists of the tracheobronchial tree and the lungs. The tracheobronchial tree consists of a system of connecting tubes that conduct airflow in and out of the lungs and allow for gas exchange.

Trachea. The trachea is situated anterior to the esophagus, beginning at the cricoid cartilage and extending inferiorly to the carina, where it bifurcates into the right and left main-stem bronchi. It is composed of C-shaped cartilage rings joined by connective tissue. These cartilage rings assist in keeping the trachea open during breathing. As noted previously, the mucous membranes of the trachea are softer, looser, and more fragile than those of the adult and more susceptible to damage, increasing the risk of obstruction from edema or inflammation.

Lungs. The lungs are situated in the thoracic cavity, enclosed by the rib cage and diaphragm, the major muscle of ventilation, which separates the thoracic cavity from the abdominal cavity. The diaphragm in an infant is flatter than that of an adult, resulting in less efficient functioning for respiration. The air passages in infants and small children are much smaller, increasing their susceptibility to obstruction. The respiratory bronchioles, alveolar ducts, and alveoli grow in number until about 8 years of age, after which they continue to grow in size. Impairments in lung function can occur as a result of birth-related conditions such as bronchopulmonary dysplasia and diaphragmatic hernia, or due to acquired disorders such as spinal cord injury. These impairments often require tracheostomy and/or mechanical ventilation, which in turn have an impact on speech production.

Contribution of Respiratory Dysfunction to Speech Disorders

Speech disorders related to respiratory dysfunction are often secondary to the presence of tracheostomy and/or ventilator dependence. The primary diagnoses of children requiring chronic tracheostomy and/or ventilator dependence include conditions due to trauma such as brain injury, spinal cord injury and direct injury to the trachea; congenital conditions; progressive neurologic disorders; and acquired nontraumatic conditions such as Guillain-Barré syndrome and anoxic encephalopathy (3). It is important to note that the causes of respiratory failure and subsequent need for mechanical ventilation are not always respiratory disease or disorder. The lungs themselves may be healthy, but access to them or the systems that contribute to their function may be impaired.

A primary means of airway management in the presence of chronic respiratory insufficiency is a tracheostomy. A tracheostomy is an artificial opening created between the outer surface of the neck and the trachea between the second and third tracheal rings. The opening itself is referred to as the stoma, and the tracheostomy tube inserted into the trachea through the stoma serves to maintain the opening, as well as provide means for connecting mechanical ventilatory devices. Tracheostomy provides a secure airway, long-term airway access, and a means for interface with mechanical ventilatory devices, and as such, is the most frequently used method of airway management. Placement of the tracheostomy tube diverts airflow away from the trachea through the tube and out the neck, bypassing the upper airway, including the vocal cords. Depending on the size and type of tracheostomy tube, a portion of the airflow will still pass around the tube and through the vocal cords; this may or may not be sufficient to produce sound. In the event that it is not sufficient, options to facilitate sound include downsizing of the tracheostomy tube to a smaller diameter and use of a unidirectional flow valve such as the Passy-Muir valve (4), which directs greater air flow through the upper airway and out the nose and mouth. Table 4.2 reviews factors for

4.2 Tracheostomy Tube Decision Flow Chart

TTS—CUFF ↓ → ⇓	LW PRESS AIRCUFF↓ → ⇓	LW PRESS AIRCUFF↑ → ⇓	TALKING TRACH →	AAC
CUFFLESS	CUFF ↓	PRTCUFF ↓ ADJ VENT → ⇓		
SAME SZ → ⇓	ADJ VENT ⇓			
PHON W/LEAK → ⇓	PHON W/LEAK → ⇓	PHON W/LEAK → ⇓ CUFF↓DAY	ENT-VF EXAM	
UFV IN LINE	UFV IN LINE	CUFF↑NT ⇓ UFV IN LINE		

Note: A double-arrow pointing down indicates progression if successful with that step; arrow pointing to the right indicates progression if that step was not successful. Single arrow pointing down indicates deflate/decrease pressure. Arrow pointing up indicates inflate/increase pressure.

TTS, tight-to-shaft cuff; Same SZ, same size; AAC, augmentative and alternative communication; LW PRESS, low pressure; ADJ, adjust; PRT cuff, partial cuff; ENT, otolaryngologist; VF, vocal folds; Phon, phonation; UFV, unidirectional flow valve.

Source: From Ref. 5.

consideration when determining the most efficient tracheostomy tube to use (5).

Phonation

The *phonatory* system is comprised of the larynx, and provides the sound source for speech. When this sound source is disrupted, it may result in alterations in voice quality, thus affecting communication.

The larynx is made up of cartilage, ligaments, muscles, and mucous membrane. It protects the entrance to the lower airway and houses the vocal cords (Fig. 4.1) (6).

Sound is *generated* in the larynx, and that is where pitch and volume are manipulated. The strength of expiration of air from the lungs also contributes to loudness, and is necessary for the vocal folds to produce speech (Fig. 4.2) (6).

Most of the muscles of the larynx receive their innervation via the recurrent laryngeal branch of the vagus nerve. This branch descends downward and wraps around the aorta, and for this reason, children who undergo cardiac surgery can sometimes experience voice disorders. If the recurrent laryngeal nerve is stretched or damaged during surgery, innervation to the vocal cords can be disrupted, and vocal hoarseness can occur.

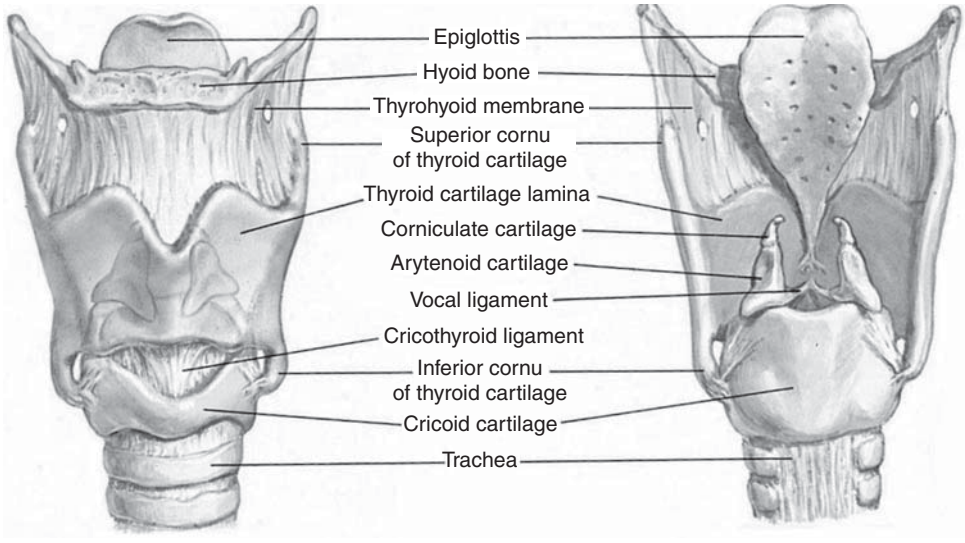


Figure 4.1 The larynx.

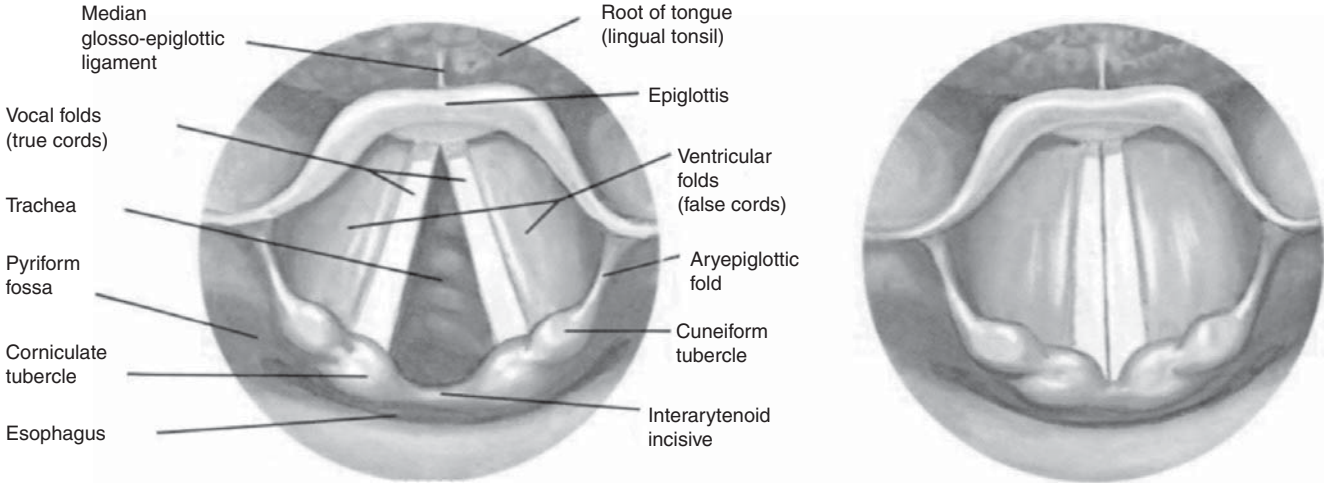


Figure 4.2 The vocal cords.

Contribution of Phonatory Dysfunction to Speech Disorders

Speech disorders related to *phonatory* dysfunction are generally classified as voice disorders, and include dysphonia (abnormal voice quality) and aphonia (loss of voice). Dysphonia is an impairment of voice secondary to cranial nerve involvement, laryngeal pathology or tracheostomy, and is characterized by varying degrees of breathiness, harshness, and vocal strain. Dysphonia may be a prominent feature of dysarthria related to cranial nerve involvement. Laryngeal pathologies resulting in dysphonia may include polyps, granulomas, nodules, or other lesions affecting the vocal fold mucosa. A common vocal fold trauma resulting in dysphonia is traumatic intubation following serious injury requiring assisted ventilation.

Articulation/Resonance

The *articulatory/resonatory* system is composed of the structures of the oral and nasal cavities, which modulate the airstream into the acoustic waveforms perceived as speech. Articulators responsible for production of speech sounds include the lips, tongue, and palate.

In addition to the placement of articulators, successful production of accurate speech sounds requires adequate functioning of the oral and nasal cavities as resonating chambers (resonance). Modulation of the airstream by these structures is a complex process that relies on intact structures as well as precise neuromuscular coordination. Fig. 4.3 illustrates where various articulators are located, as well as places of articulation for various speech sounds. Impairment in one or more of these components is likely to result in a disorder of articulation/resonance.

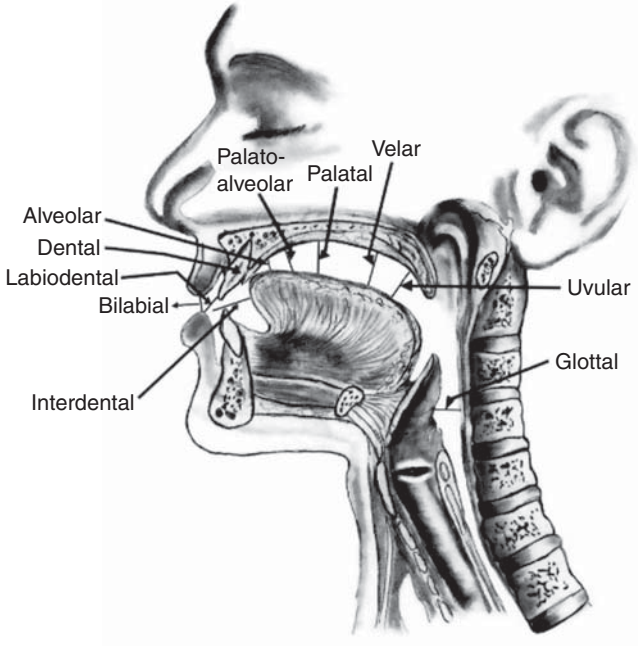


Figure 4.3 Places of articulation.

Contribution of Articulatory/Resonatory Dysfunction to Speech Disorders

Speech disorders related to *articulatory/resonatory* dysfunction include disorders that result from impairment in any component of the *articulatory/resonatory* system, and as such are quite comprehensive. They include all motor speech disorders, including dysarthria and apraxia (Table 4.4), as well as disorders resulting from congenital conditions such as cleft palate.

Although the three components of speech described previously are considered separately as individual components, they function as a single coordinated and interactive unit for production of speech, and as such, are subsystems of a complex motor act requiring precise coordination of muscle groups. It is easy to understand how impairments in any of these components can have an impact on communication, as the extent and complexity of the speech system make it susceptible to the influence of a myriad of factors.

Motor Speech Disorders

Motor speech disorders are a collection of communication disorders involving retrieval and activation of motor plans for speech, or the execution of movements for speech production (7). Subcategories include dysarthria and apraxia of speech. Motor speech disorders occur in both children and adults. They may be acquired or developmental in nature (Table 4.3).

Acquired: adverse event (usually neurologic) occurs that impedes continuation of previously normal speech acquisition

Developmental: no specific identifiable etiology to explain delays in speech acquisition

Dysarthria refers to a group of related motor speech disorders resulting from impaired muscular

4.3 Motor Speech Disorders

DEVELOPMENTAL	ACQUIRED
Phonological disorder	Dysarthria
Verbal apraxia	Verbal apraxia
Articulation disorder	Articulation disorder

control of the speech mechanism, and manifested as disrupted or distorted oral communication due to paralysis, weakness, abnormal tone, or incoordination of the muscles used in speech (Table 4.4) (8). It affects the following:

Respiration: respiratory support for speech, breathing/speaking synchrony, sustained phonation

Phonation/Voice: loudness, quality

Articulation: precision of consonants and vowels

Resonance: degree of airflow through nasal cavity

Prosody: melody of speech, use of stress and inflection

Movements may be impaired in force, timing, endurance, direction, and range of motion. Sites of lesion include bilateral cortices, cranial nerves, spinal nerves, basal ganglia and cerebellum.

Associated characteristics of dysarthrias include slurred speech; imprecise articulatory contacts; weak respiratory support and low volume; incoordination of the respiratory stream; hypernasality; harsh or strained/strangled vocal quality; weak, hypophonic, breathy vocal quality; involuntary movements of the oral facial muscles; spasticity or flaccidity of the oral facial muscles; and hypokinetic speech.

Some common etiologies for dysarthria in children include stroke, brain tumor, aneurysm, traumatic brain injury, encephalopathy, seizure disorder, cerebral palsy, and high-level spinal cord injury.

Oral apraxia refers to an impairment of the voluntary ability to produce movements of the facial, labial, mandibular, lingual, palatal, pharyngeal, or laryngeal musculature in the absence of muscle weakness.

Verbal apraxia (also called apraxia of speech, or AOS) refers to an impairment of *motor speech* characterized by a diminished ability to program the positioning and sequencing of movements of the speech musculature for volitional production of speech sounds. Apraxia is not the result of muscle paralysis or weakness, but may lead to perceptual disturbances of breathing/speaking synchrony, articulation, and prosody. Site of lesion is generally the left precentral motor or insular areas.

Developmental verbal apraxia (also called developmental apraxia of speech, or DAOS) refers to a speech disorder resulting from delays or deviances in those processes involved in planning and programming movement sequences for speech in the absence of muscle weakness or paralysis. Associated characteristics of DAOS include receptive-better-than-expressive language, presence of oral apraxia (may or may not exist with DAOS), phonemic errors (often sound omissions), difficulty achieving initial articulatory configuration, increase in errors with increase in word length and/or phonetic complexity, connected speech poorer than word production, inconsistent error patterns, groping and/or trial-and-error behavior, and presence of vowel errors.

4.4 Types of Dysarthria

	SPASTIC	HYPOKINETIC	HYPERKINETIC	ATAXIC	FLACCID	MIXED
Site of Lesion	Bilateral upper motor neuron	Extra-pyramidal system	Extra-pyramidal system	Cerebellum	Unilateral or bilateral lower motor neuron	Multiple sites of lesion
Associated characteristics	Spasticity of orofacial muscles Imprecise articulatory contacts Strained/strangled voice quality Monopitch Reduced stress Reduced rate	Rigidity of orofacial muscles Imprecise articulatory contacts Hypophonia Monopitch Reduced stress and inflection Transient increased rate/rapid rate	Involuntary movements of orofacial muscles Imprecise articulatory contacts Harsh voice quality Incoordination of the respiratory stream Transient increased rate	Irregular articulatory breakdown Harsh vocal quality Incoordination of the respiratory stream Excess and equal stress pattern Reduced rate	Flaccidity of the orofacial muscles Imprecise articulatory contacts Breathy voice quality Low vocal volume Reduced stress and inflection Hypernasality	Characteristics dependent on site of lesion
Example of disorder	Cerebral palsy	Parkinson's disease	Dystonia	Friedreich's ataxia	Bulbar palsy	Amyotrophic lateral sclerosis

Source: From Ref. 8.

Children with motor speech disorders may demonstrate impaired phonological systems because their ability to acquire the sound system of their language is believed to be undermined by difficulties in managing the intense motor demands of connected speech (9).

Language Components

With regard to models of language, the prevailing school of thought follows Bloom and Lahey's philosophy, which proposes three main components of language: form, content, and use (Fig. 4.4). According to Bloom and Lahey, language can be defined as "a knowledge of a code for representing ideas about the world through a conventional system of arbitrary signals for communication (10)."

These three components can be subdivided further into *phonology, morphology, syntax, semantics,* and *pragmatics*, as described in the following sections.

Form

Form with reference to language refers to the rule-based structure humans employ to formulate language, ranging from phonemes to sentences, and comprises *phonology, morphology,* and *syntax*.

Phonology refers to the rule-governed system by which sounds, or phonemes, are combined to create meaningful units, or words. The English language contains 44 recognized phonemes, which are classified as consonants or vowels. This distinction involves presence or absence of interruption of the air stream. Vowels are formed through modulation (without interruption) of the air stream via variation in position of the lips and tongue (Fig. 4.5) (11).

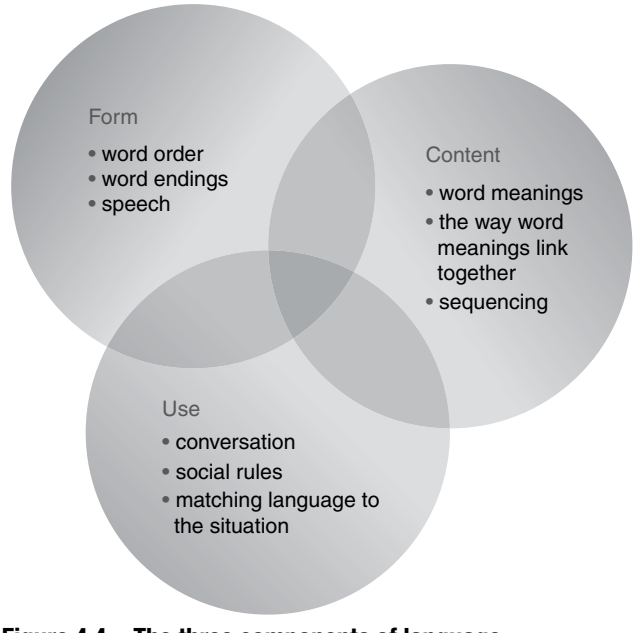


Figure 4.4 The three components of language.

Variations in tongue position for production of different vowels are systematically characterized as high, mid, or low, as well as front, central, or back, and can further be described as tense or lax (Fig. 4.6) (12). For example, the vowel /i/, pronounced "ee," is considered a high, front, tense vowel, as the front of the tongue is

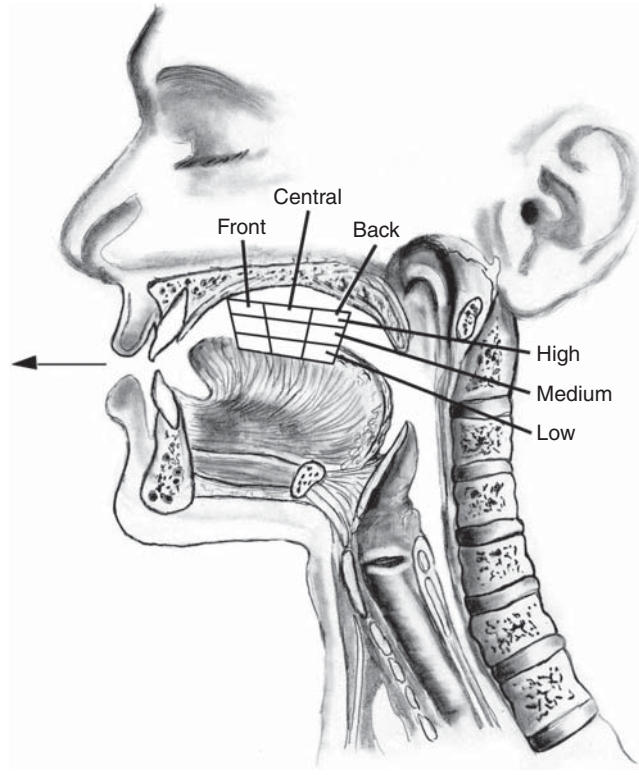


Figure 4.5 Vowel areas.

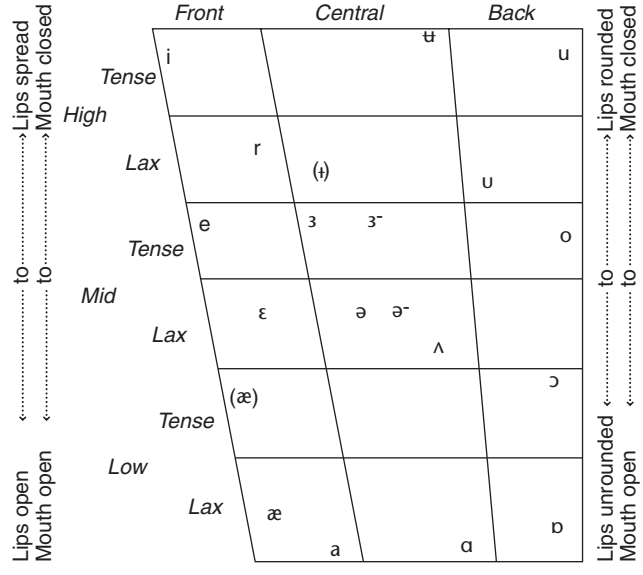


Figure 4.6 Tongue positions for vowel production. (Reprinted with permission from Bronstein AJ. The pronunciation of American English. New York: Appleton-Century-Crofts, Inc., 1960.)

high and the tongue is tensed. Diphthongs are combinations of vowels, and require movement of the tongue from one position to another during production.

Consonants are formed through a combination of varying degrees of interruption of the airstream and variations in tongue and lip posture (see Fig. 4.3). Phonemic acquisition in children follows a systematic sequence, and it is believed that children acquire phonemes not in isolation, but rather in the context of their relationship to other sounds in a word (Table 4.5) (13).

Table 4.6 provides a graphic representation of the typical age ranges during which most children acquire consonant sounds (14,15). This is useful in determining at what age a child is considered outside of the norm for acquisition of a specific sound and when intervention might be indicated.

With regard to how well one can expect to understand a child's speech over the course of phonemic acquisition, Lynch et al provide an estimate of speech intelligibility at different ages, summarized in Table 4.7 (16).

Phonological disorders are a subset of sound production disorders in which linguistic and cognitive factors, rather than motor planning or execution, are thought to be central to observed difficulties (common etiologic variables include otitis media with effusion, genetics, and psychosocial involvement) (17). Developmental phonological disorders result when children fail to progress in their acquisition of specific phonemes. Currently accepted theory regarding phonology in children proposes the existence of phonological processes that are present in the phonological systems of all children as they develop language, and are systematically eliminated at predictable ages in a standard developmental progression. Failure to eliminate, or resolve, these processes, results in a phonological processing disorder. An example of a developmental phonological process is "stopping of fricatives," in which a child systematically substitutes a stop sound (a sound that stops airflow, such as /p, t, k/) for a fricative sound (a sound that produces friction through partial interruption of airflow, such as [th, s, z, f, v]), producing words such as "dum" for "thumb," "tun" for "sun," or "dip" for "zip." These sound substitutions are systematic and applied by the child in the same context each time that sound occurs. Nondevelopmental phonological processes are indicative of disordered versus delayed phonological development, and are rarely seen in normal development. An example of a nondevelopmental phonological process is initial consonant deletion, in which a child deletes the initial sound in a word, such as "ee"/"key" or "ake"/"make."

Table 4.8 illustrates the typical developmental sequence for resolving phonological processes (18).

Morphology refers to the rule-based system by which words are constructed and altered, often through addition of prefixes and suffixes, to reflect concepts such as number, possession, and verb tenses. For example, addition of the phoneme "-s" to the end of a word makes it plural. The "-s" in this instance is considered a morphological marker signifying the notion of "plural."

Disorders affecting morphology are most typically developmental and result when children have difficulty mastering the acquisition of rules for applying morphological markers. Difficulty with use of morphological markers can also be seen following certain types of focal brain injury, such as damage to Broca's area, when expressive language becomes telegraphic in nature, losing the nuances provided by morphological markers.

Syntax refers to the system of rules by which words are combined to create phrases, clauses, and sentences. The various parts of speech in English (eg, nouns, pronouns, verbs, adverbs, adjectives, etc.) serve different functions within these constructions, such as description, action, and attribute, and as such have specific rules for combination with each other. For example, the basic word order in English is subject-verb-object.

As with morphology, disorders affecting syntax are typically developmental and are the result of difficulty mastering the acquisition of rules for creating grammatically correct sentences.

4.5 Phonemic Acquisition: Age at Which 75% of Children Tested Correctly Articulated Consonant Sounds

AGE (YEARS)	SOUNDS
2	m, n, h, p, ŋ
2.4	f, j, k, d
2.8	w, b, t
3	g, s
3.4	r, l
3.8	š (she), tš (chin)
4	ð (father), Z (measure)
4+	dž (jar), Θ (thin), v, z

Source: From Ref 13.

Content

Content with reference to language refers to the *semantics*, or meaning, of words, as they relate to, or represent, objects, actions, and relationships. *Semantics*, or meaning, is conveyed through the use of words or other symbols within a given context. Development of semantics in children reflects growing and changing concepts related to experiences, culture, and cognitive

4.6 Acquisition of Consonant Sounds

2	3	4	5	6	7	8
_____	p_____					
_____	m_____					
_____	h_____					
_____	n_____					
_____	w_____					
_____	b_____	_____				
	_____k	_____				
	_____g	_____				
	_____d	_____				
	_____t	_____	_____	_____		
	_____ng	_____	_____	_____		
	_____	f_____				
	_____	y_____				
		_____r	_____	_____		
		_____l	_____	_____		
		_____s	_____	_____	_____	_____
		____	ch_____	_____	_____	
		____	sh_____	_____	_____	
		____	z_____	_____	_____	_____
			_____j	_____	_____	
			_____v	_____	_____	_____
			____	th ____ (voiceless_as in "**th**ink")	_____	
				_____TH (voiced as in "**th**at")	_____ zh (as in "trea**s**ure")	_____ _____

Source: From Refs. 14 and 15.

4.7 Speech Intelligibility in Children

By 18 months, a child's speech is normally 25% intelligible.

By 24 months, a child's speech is normally 50% to 75% intelligible.

By 36 months, a child's speech is normally 75% to 100% intelligible.

Source: From Ref. 16.

level. An example of a changing semantic notion is that of overgeneralization. Children first learn the meaning of a word based on one representation of that word and initially overgeneralize it to apply to all similar representations. Hence, "dog" may at some point be applied to denote all four-legged creatures.

Child language disorders affecting semantics may be developmental and related to general cognitive development, or they may be acquired. Examples of disorders that involve semantics include specific

4.8 Resolution of Phonological Processes: Ages by Which Phonological Processes Are Eliminated

PHONOLOGICAL PROCESS	EXAMPLE	GONE BY APPROXIMATELY (YEARS;MONTHS)
Context sensitive voicing	pig = big	3;0
Word-final de-voicing	pig = pick	3;0
Final consonant deletion	comb = coe	3;3
Fronting	car = tar ship = sip	3;6
Consonant harmony	mine = mime kittycat = tittytat	3;9
Weak syllable deletion	elephant = efant potato = tato television =tevision banana = nana	4;0
Cluster reduction	spoon = poon train = chain clean = keen	4;0
Gliding of liquids	run = one leg = weg leg = yeg	5;0
Stopping /f/	fish = tish	3;0
Stopping /s/	soap = dope	3;0
Stopping /v/	very = berry	3;6
Stopping /z/	zoo = doo	3;6
Stopping 'sh'	shop = dop	4;6
Stopping 'j'	jump = dump	4;6
Stopping 'ch'	chair = tare	4;6
Stopping voiceless 'th'	thing = ting	5;0
Stopping voiced 'th'	them = dem	5;0

Source: From Ref. 18.

language impairment (SLI), semantic–pragmatic language disorder, and Landau-Kleffner syndrome (19). In all these cases, children exhibit some degree of difficulty understanding the meaning of words and sentences. For children with semantic processing difficulties, the more abstract a concept is, the more difficult it is to understand. This holds true for things that require interpretation beyond the literal meaning, such as might be required in an idiom or slang expressions. Deficits related to semantics can also result in difficulty identifying the key points in a sentence or story, which in turn may lead to problems with topic maintenance.

Use

Use with reference to language describes the function language serves within a social context, and is governed by pragmatics. Pragmatics refers to how we use the language we have acquired to communicate in social situations. Within a social interaction, language may be used in many different ways, such as to make comments, to ask questions, to acknowledge comments, and to answer questions. In 1976, Elizabeth Bates described three critical components of pragmatics: the ability to use speech acts to express intentionality in order to accomplish a given purpose

(function), the ability to use social understanding and perspective-taking ability to make presuppositional judgments, and the ability to apply rules of discourse (eg, quantity, quality, relevance, clarity) in order to engage in cooperative conversational exchanges (20).

Child language disorders affecting pragmatics are most typically those associated with disorders on the autism spectrum. Acquired injuries that may have an impact on pragmatics include traumatic brain injury affecting the frontal lobes. Frontal lobe injury often impairs executive functioning and increases impulsivity, resulting in impaired judgment. This, in turn, may impair one's ability to understand perspective and to apply rules of discourse appropriately.

To summarize, language competence requires the successful intersection of form, content, and use. As simple as it may seem, having a successful conversation is a complex act requiring integration of many aspects of language and involving a blending of linguistic features with sociocultural understandings. "Conversation is not a chain of utterances, but rather a matrix of utterances and actions bound together by a web of understandings and reactions"(21).

Speech and Language Acquisition

Acquisition of speech and language skills follows a fairly systematic progression, with easily identifiable milestones associated with specific ages in each area, as briefly outlined here (22,23).

- Birth–3 Months
  - □ Makes pleasure sounds such as cooing
  - □ Develops differential cries for different needs
  - □ Develops social smile
- 3–6 Months
  - □ Increase in variety of vocalizations
  - □ Babbling sounds more speechlike, with increased consonant productions
  - □ Uses sounds and gestures to indicate wants
- 6–12 Months
  - □ Reduplicative babbling occurs (eg, dada, bibi, etc.)
  - □ Uses speech sounds to get attention
  - □ First words emerge (~10–12 months)
  - □ Responds to simple requests
  - □ Imitates speech sounds
- *18–24 months*
  - □ Uses words more frequently than jargon
  - □ Has expressive vocabulary of 50–100 words
  - □ Has receptive vocabulary of 300+ words
- *2–3 Years*
  - □ Uses two- to three-word sentences
  - □ Points to pictures in books
  - □ Speech is understood by familiar listeners most of the time
- *3–4 Years*
  - □ Uses simple sentences with negatives, imperatives, and questions
  - □ Talks about activities at school and home
  - □ Understands simple "wh-" question words
- *4–5 Years*
  - □ Mean length of utterance (MLU) = 4.6–5.7 words
  - □ Uses grammatically correct sentences
  - □ Relays a long story accurately
- *5–6 Years*
  - □ MLU = 6.6 words
  - □ Uses all pronouns consistently
  - □ Comprehends 13,000 words
- *6–7 Years*
  - □ MLU = 7.3 words
  - □ Comprehends 20,000–26,000 words
  - □ Refines syntax

Speech and Language Disorders

Speech and language disorders in children can be conceptualized as falling into two categories: developmental and acquired. Within the category of developmental, we can also distinguish between developmental delay and developmental disorder. Developmental language delay refers to delay in the acquisition and development of age-appropriate language skills, typically across all domains. This can be due to medical or psychosocial factors. A developmental language disorder is characterized by atypical development of language skills in one or more domains, often with aberrant or interrupted development. As noted previously, there are specific milestones associated with each age as a child acquires speech and language skills. It is important to monitor development and watch for any signs that might indicate delay or disorder. The following is a list of danger signals of communication problems by age (24):

- *By 6 months*
  - □ Does not respond to the sound of others talking
  - □ Does not turn toward speaker out of view
  - □ Makes only crying sounds
  - □ Does not maintain eye contact with caregiver
- *By 12 months*
  - □ Does not babble
  - □ Does not discontinue activity when told "no"
  - □ Does not follow gestural commands, such as "want up" or "give me"
- *By 24 months*
  - □ Does not say a meaningful word
  - □ Does not refer to self by name
  - □ Does not follow simple directions
  - □ Does not talk at all at 2 years
  - □ Vocabulary does not seem to increase
  - □ Does not have any consonant sounds
  - □ Does not answer simple yes/no questions

■ *By 36 months*
- □ Does not say whole name
- □ Does not seem to understand "what" and "where" questions
- □ Uses jargon a great deal
- □ Answers your question by repeating the question
- □ Continues to echo statements made by others
- □ Does not use two- to three-word utterances
- □ Points to desired objects rather than naming them
- □ Does not name any objects in pictures
- □ Leaves off the beginning consonants of words
- □ Cannot be understood even by parents
- □ Does not respond when you call name

An acquired language disorder is characterized by language deficits in one or more domains secondary to neurologic insult. This can and often does result in aberrant development due to interruption in the normal course of language acquisition. When considering a speech and language disorder resulting from a congenital disorder such as cleft palate or Pierre Robin Sequence, classification becomes more difficult. The disorder does not fit the definition of a developmental delay, in that the development is atypical secondary to structural deficits. The disorder is also not considered acquired, as the structural deficit leading to the disorder occurred at birth, before the child began to develop language.

Some common causes of loss or deterioration of language in childhood include head injury, unilateral cerebrovascular lesions, cerebral infections, brain tumors, seizure disorders, and cerebral anoxia. These disorders can result in acquired childhood aphasia (25). *Acquired childhood aphasia* is defined as a language disorder secondary to cerebral dysfunction in childhood appearing or occurring after a period of normal language development. The cerebral dysfunction may be the result of a focal lesion of one of the cerebral hemispheres, a diffuse lesion of the central nervous system (CNS) above the level of the brainstem (TBI, cerebral infection), a diffuse lesion related to convulsive activity, or unknown etiology Landau-Kleffner syndrome (LKS). In general, pediatric-acquired aphasia tends to be characterized by nonfluency, with primary deficits in verbal expression, with parallel deficits in written expression and auditory comprehension relatively intact.

Pediatric traumatic brain injury (TBI) can result in more generalized dysfunction secondary to diffuse axonal injury caused by acceleration forces. Although such damage can have a significant impact on a variety of brain functions, the damage, sustained at the axonal or cellular level, is often not detected by brain scans. The definition of TBI, written by the federal Division of Special Education as part of Public Law 101–476 (Individuals with Disabilities Act, or IDEA), was published in 1992 as the guideline for state departments of education to use in determining how to provide educational services to these children. It reads as follows (26):

> "Traumatic Brain Injury" means an acquired injury to the brain caused by an external force, resulting in total or partial functional disability or psychosocial impairment, or both, that adversely affects a child's educational performance. The term applies to open or closed head injuries resulting in impairments in one or more areas, such as cognition; language; memory; attention; reasoning; abstract thinking; judgment; problem-solving; sensory, perceptual, and motor abilities; psychosocial behavior; physical functions; information processing; and speech. The term does not apply to brain injuries that are congenital or degenerative, or brain injuries induced by birth trauma (Federal Register, Vol. 57, no. 189).

Other common acquired disorders that can affect speech and language development include high-level spinal cord injury (SCI) and hearing loss. High-level SCI often affects some of the cranial nerves that are responsible for movement of the articulators necessary for speech production (Table 4.9) (27).

With regard to hearing loss, if children acquire hearing loss during the period of speech and language acquisition, they are at significantly increased risk for communication disorders.

There are many congenital disorders that can have an impact on speech and language development. Some of the most common include cerebral palsy, cleft palate/craniofacial anomalies, hearing loss, and autism.

Cerebral palsy (CP) is defined as a group of disorders of development of movement and posture, causing

4.9 Cranial Nerves Involved in Speech and Swallowing

Trigeminal (V)	**Vagus (X)**
Face (sensory)	Larynx (sensory and motor)
Head (sensory	Hypopharynx
	Soft palate
Facial (VII)	Cricopharynx
Taste (anterior 2/3)	
Ear (sensory)	**Spinal Accessory (XI)**
Facial expression (motor)	Soft palate (motor)
	Tongue (motor)
Glossopharyngeal (IX)	Pharynx (motor)
Pharynx (motor)	
Oropharynx (sensory)	**Hypoglossal (XII)**
Posterior tongue (sensory, taste)	Tongue (motor)
	Hyoid (motor)
	Extrinsic larynx

Source: From Ref. 17.

activity limitation, that are attributed to nonprogressive disturbances that occurred in the developing fetal or infant brain. The motor disorders of cerebral palsy are often accompanied by disturbances of sensation, cognition, communication, perception, and/or behavior, and/or by a seizure disorder (28). CP may significantly affect tone, which in turn affects ability to use those muscles appropriately to perform the necessary movements for speech production. As noted previously, speech production is a complex motor act requiring precise coordination of muscle groups, including respiratory, phonatory, and articulatory systems. When abnormal tone is present, either hyper- or hypotonicity, this interferes with coordination both within and across these systems, resulting in motor speech dysfunction, specifically dysarthria. The most common types of dysarthria associated with cerebral palsy include spastic, ataxic, and hyperkinetic (see Table 4.4).

Children with spastic cerebral palsy are more likely to exhibit imprecise articulatory contacts, strained/strangled voice quality, and reduced rate. Children with ataxic cerebral palsy typically exhibit irregular articulatory breakdown, harsh vocal quality, incoordination of the respiratory stream, and reduced rate. Children with athetoid cerebral palsy exhibit imprecise articulatory contacts, harsh vocal quality, incoordination of the respiratory stream, and transient increased rate.

Treatments for hypertonicity, such as intrathecal baclofen, selective dorsal rhizotomy, and various oral medications, may have an influence on speech and communication. These treatments frequently result in improvements, but in some cases may worsen impairment (29,30). Authors of this chapter report clinical observations of improved breath support for voice production and improved articulation with intrathecal baclofen therapy.

The presence of combined motor and cognitive impairments makes assessment of communication difficult. There is great need for a standard classification system along the lines of the Gross Motor Classification System (GMFCS) (31) and Manual Ability Classification System (MACS) (32). Van Tubbergen and Albright developed a five-level ordinal scale to classify levels of expressive language: the ExPRS (Expressive Production Rating Scale) (33). Like the GMFCS and MACS, the ExPRS provides a descriptive classification system for expressive communication, including the use of alternative or augmentative communication (Table 4.10). Further investigation on the reliability and validity of the ExPRS is needed to enhance its potential in transdisciplinary settings.

Cleft palate and other craniofacial anomalies involving the oral cavity most typically affect a child's articulation as well as resonance. A cleft palate prevents ability to valve the airstream at the level of the palate, making it impossible to close off the nasal passage during speech. This results in hypernasal speech. A number of other syndromes, such as velocardiofacial syndrome (also known as DiGeorge syndrome), affect the ability of the soft palate to function properly, resulting in velopharyngeal incompetence, in turn resulting in impaired resonance (hypernasality).

Congenital hearing loss can have a significant impact on the development of speech and language, depending on the severity of the loss. Speech and language disorders resulting from hearing loss may affect multiple areas of communication, including language comprehension, syntax, vocabulary, and articulation. The nature and extent of communication disorders in children with hearing impairment are influenced by type and degree of hearing loss, causative factors, age at onset, cognitive status, and environment. Early identification and intervention are critical to maximize potential for developing communication skills in children with hearing loss. Intervention can include provision of hearing aids, environmental modifications (eg, FM or frequency modulation systems in the classroom), aural habilitation/rehabilitation, sign language, total communication (combination of auditory–vocal language, signs, gesture, and speech reading), or surgical implant (cochlear implant).

Autism is one of the fastest-growing childhood disorders in our nation today. The current estimate is that 1 out of every 150 children is diagnosed with autism. The spectrum of autism disorders is broad, including pervasive developmental delay—not otherwise specified, autism, Asperger's syndrome, Rett's disorder, and childhood disintegrative disorder. Within the DSM IV criteria (34), the current classification system used to diagnose children with an autism spectrum disorder,

4.10 Expressive Production Rating Scale (ExPRS)

Child's communication: Mark the item that best describes your child's typical abilities:

___ Speaks in a generally age-appropriate way; minor limitations, if any.
___ Speaks with some difficulty; speech may be slow or somewhat difficult to understand by a new listener.
___ Speaks with significant difficulty; speech is slow or quite difficult to understand by a new listener.
___ Communicates independently with limitations; individual uses adapted techniques such as signing or an augmentative communication device.
___ Communication is severely limited even with the use of augmentative technology.

Source: From Ref. 33.

deficits in some aspect of communication are present in all the disorders; in fact, 10 of the 15 characteristics listed to characterize autism are directly related to communication.

Organized according to presence/absence of communication, they are as follows:

- Five criteria relating to language:
  - delay in, or total lack of, the **development of spoken language** (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
  - in individuals with adequate speech, marked impairment in the ability to **initiate or sustain a conversation** with others
  - **stereotyped and repetitive use of language** or idiosyncratic language
  - lack of varied spontaneous **make-believe play or social imitative play** appropriate to developmental level
  - delays or abnormal functioning in language as used in social communication, with **onset prior to age 3 years**
- Five criteria relating to social interaction:
  - marked impairment in the use of multiple **nonverbal behaviors**, such as eye-to-eye gaze, facial expression, body postures, and gestures, to regulate social interaction
  - failure to develop **peer relationships** appropriate to developmental level
  - a lack of **spontaneous seeking to share enjoyment**, interests, or achievements with other people (eg, by a lack of showing, bringing, or pointing out objects of interest)
  - lack of **social or emotional reciprocity**
  - delays or abnormal functioning in social interaction, with **onset prior to age 3 years**
- Five criteria relating to patterns of behavior, interests, and activities:
  - encompassing preoccupation with one or more **stereotyped and restricted pattern of interest** that is abnormal either in intensity or focus
  - apparently inflexible adherence to specific, **nonfunctional routines or rituals**
  - stereotyped and **repetitive motor mannerisms** (eg, hand or finger flapping or twisting, or complex whole body movements)
  - persistent **preoccupation with parts of objects**
  - delays or abnormal functioning in **symbolic or imaginative play**, with onset prior to age 3 years

In addition to delayed development of receptive and expressive language, the hallmark characteristic for children with autism is a deficit in the pragmatics, or use, of language. These children typically have impaired reciprocal social interaction and, in more severe cases, lack intent to communicate.

Assessment and Treatment of Speech/Language Disorders

Speech-language pathologists provide diagnostic, treatment, and educational services to children who are experiencing impairments of speech, language, voice, fluency, communicative–cognitive, memory, and swallowing skills. The primary disorders are outlined in Table 4.11 (35), divided into developmental versus acquired.

Assessment

In assessing language disorders in children, it is crucial to understand the normal developmental level associated with the chronological age of the child to determine premorbid developmental levels and to assess the impact of the neurologic event or other interruption in typical developmental maturation on that development.

It is equally important to identify children at risk, as we know that speech and language delays/disorders in infancy and toddlerhood can result in difficulties in academic learning, social interaction, and development of appropriate peer relationships throughout childhood (36,37).

Areas of assessment in pediatric communication disorders include pragmatics, cognition, orientation, attachment/interaction, prelinguistic behaviors, phonological development/intelligibility, oral motor function, language comprehension (auditory and reading), language production (verbal and written), fluency, voice, hearing, and feeding and swallowing. These areas are assessed formally through test batteries, objective procedures, and parent interview questionnaires, as well as informally through direct observation of and interaction with children in naturalistic contexts. Detailed description of specific assessment materials and procedures in each of these areas is beyond the scope of this chapter. It should be noted that assessment is often done as part of a multidisciplinary evaluation, and input from other disciplines is often vital in providing the most comprehensive diagnosis and treatment plan. One area of common need for multidisciplinary input is augmentative and alternative communication. For children who are nonverbal or who have significant motor impairment, a reliable means of access to augmentative communication devices and to computers must be identified, and this process may require input from speech pathology, occupational therapy, rehabilitation engineering, and sometimes physical therapy. Once a child has undergone a thorough evaluation, results are carefully

4.11 Primary Disorders of Speech, Language, and Swallowing

	DEVELOPMENTAL	ACQUIRED
Motor speech disorders	Phonologic disorder Verbal apraxia Articulation disorder	Dysarthria Verbal araxia Articulation dsorder
Language disorders	Language delay Language disorder	Aphasia
Voice disorders	Aphonia Dysphonia	Aphonia Dysphonia
Fluency disorders	Nonfluency Dysfluency/stuttering	Dysfluency/stuttering
Communicative–cognitive disorders	Learning dsabilities Autism	Traumatic Brain Injury Aphasia
Memory disorders		Short-term memory deficit Long-term memory deficit Verbal learning deficit
Swallowing disorders	Oral aversion Discoordination of suck–swallow–breathe	Oral dysphagia Pharyngeal dysphagia Oropharyngeal dysphagia

Source: From Ref. 35.

reviewed, a diagnosis is made, and treatment recommendations are formulated. A child's parents or caregivers are included as much as possible in the assessment process, as well as in the development of the treatment program.

With regard to the diagnosis, it is important to have a clear understanding of a child's medical history and any contribution that medical status may have made to the child's communication disorder. This will determine whether the deficit is considered developmental or acquired, and the diagnosis will then drive the treatment recommendations, including specific goals and objectives, treatment timeframe, and projected outcome (prognosis). A clear understanding of a child's cognitive level is also crucial in making appropriate diagnoses as well as treatment recommendations. If a child's cognitive level is commensurate with level of language ability, expectations for improvement and prognosis are different than for a child exhibiting a significant discrepancy between language and cognition.

Assessment tools and strategies that are accessible and appropriate for individuals with speech and other impairments are critical. Typical standardized tests specify the modality in which information is presented to the child and the modality in which the child must respond. Most procedures require clear speech for full participation. For example, most tests of phonological awareness require the participant to verbally present words or sounds to demonstrate skills. For an individual with significant apraxia, it is difficult to determine whether errors are due to underlying deficits in phonological awareness, effects of apraxia, or other reasons. For individuals who use alternative or augmentative communication, most communication requires the individual to make selections from preprogrammed arrays. This presents a further confound in that the ability to make choices of preference may be more developed than the ability to answer a factual question on demand if there are impairments in pragmatics (38).

Given the dearth of accessible speech, language, and cognitive assessment tools for individuals with communication impairments, especially if there are concurrent motor impairments, efforts to develop such instruments is a priority to optimize educational and medical interventions, as well as to provide accurate and meaningful diagnoses.

In addition to developing treatment recommendations, it is important to make any other referrals as appropriate. For example, if a child's history includes language regression, a referral to pediatric neurology may be indicated. If a child with documented speech and language delay has not had a formal

hearing assessment, a referral to audiology is warranted. Finally, if a child is exhibiting characteristics consistent with a disorder on the autism spectrum, a referral to pediatric psychology may be necessary to obtain a formal diagnosis.

Treatment

Once a child has been evaluated, recommendations for treatment are made. These include specific goals and objectives in the identified deficit areas. Treatment for children with developmental speech and language delay or disorder differs in a number of important aspects from treatment for children with an acquired speech and language disorder. First, we distinguish between developmental *delay* and *disorder* in that *delay* implies typical but slowed or late development of communication skills. *Disorder* implies aberrant development of communication skills. For example, most typically developing children overgeneralize certain semantic concepts in the course of acquiring expressive vocabulary. At some point, they may use the word *dog* to refer to all four-legged animals, or *juice* to refer to all drinks. For children with developmental delay, they would be expected to persist in these overgeneralizations beyond predicted ages. In contrast, children with developmental disorders may exhibit atypical language patterns, such as reversing word order or leaving out certain parts of speech (eg, verbs) completely in their development of expressive language. These errors are not part of the typical pattern of language acquisition, and thus would be considered a disorder.

Treatment for children with developmental speech and language delay will typically focus on general language stimulation within the specific areas of delay. For example, for a child with delay in expressive language, a general goal might be for a child to use language successfully to get daily needs and wants met. Objectives within that goal might be to increase expressive vocabulary, increase utterance length, ask and answer questions, or improve speech intelligibility. Treatment for children with developmental disorders will need to be more tailored to the specific errors exhibited, which will not necessarily fall within the typical acquisition of speech/language milestones. Children with the diagnosis of autism would fall under the category of developmental disorder, in that their language development does not follow the typical developmental progression. There are a number of treatment programs for children with autism, ranging from applied behavioral analysis (ABA) (39,40) to the "floor-time" (DIR) approach (41). The decision regarding which treatment approach to use in part is determined by the severity of the communication disorder; children with more severe disorders are often referred to ABA programs due to the increased amount of structure. Children with milder disorders may benefit more from a play-based approach such as DIR.

Treatment for children with acquired communication disorders can be somewhat more complex, as it requires a detailed understanding of the specific deficits as well as how they related to the child's development of communication as a whole. In addition, it requires the ability to distinguish between gains due to spontaneous recovery from injury, gains due to typical expected development, and gains due to treatment. One of the most common areas of treatment in acquired communication disorders is traumatic brain injury.

Janet Lees proposes three stages of recovery in pediatric brain injury: acute period, lasting from emergency admission to reestablishment of stable conscious state; consistent recovery, lasting from reestablishment of stable conscious state to the point where progress begins to slow, or plateau; and the slowed recovery, or plateau stage (42). The period during which a child makes the greatest progress is the second stage, in which intensive therapy and educational input can maximize recovery. The period where long-term residual deficits become apparent occurs during the third stage. The length of each stage varies, depending on the severity of the head injury. When treating children with acquired traumatic brain injury, it is important to keep in mind the unique characteristics and needs specific to pediatric brain injury. For example, pediatric brain injury occurs on a moving baseline of normal development upon which further development is expected. For this reason, assessment tools need to be appropriate for the developmental age of the child; in young children, this means some functions will not be accessible. Plasticity in the developing nervous system may allow the preservation of certain functions, particularly those related to language. In addition, plasticity could theoretically involve relocation of function to the opposite hemisphere or elsewhere in the same hemisphere. Normal recovery may occur, can be a most dramatic and unexplained phenomenon, and should not be confused with plasticity. Finally, critical periods for the development of a particular function may exist, which, at most, cannot be retrieved. This may, for example, apply to the development of social communication in young children at relatively high risk of the development of autistic features (43).

When it is not possible to promote or maintain verbal communication in children, regardless of whether they have a developmental or acquired disorder, it may be necessary to provide augmentative or alternative options for communication. Numerous options are available for nonverbal children, ranging from sign language to high-tech augmentative communication devices. Common low-tech solutions include signing,

pictures (eg, Picture Exchange Communication System, or PECS) (44), and recordable devices with finite selections, such as the Cheap Talk Device. (see article by Elizabeth Libby Rush at http://enablingdevices.com/ask-Steve/assistive_technology_devices_used_in_education_1). Children in need of augmentative or alternative communication typically are evaluated by speech pathology first, and if a more comprehensive assessment is indicated, a second evaluation may be done as part of a multidisciplinary assessment, including occupational therapy and rehabilitation engineering. Children who have significant motoric impairments often need input from occupational therapy regarding access solutions. Children who have complex needs requiring more custom solutions often benefit from input from rehabilitation engineering.

FEEDING AND SWALLOWING PROCESSES AND DISORDERS

During the first 12 months, infants have a number of unique anatomic and physiologic characteristics that gradually diminish with growth and maturation (Fig. 4.7) (45). For example, the larynx in infants is positioned higher in the neck than in older children and adults, with close approximation of the epiglottis and soft palate, resulting in added airway protection, as well as obligate nasal breathing (Fig. 4.7A, 4.7B) (46,47). This is important in promoting the suck–swallow–breathe sequence, the most complex sensorimotor process undertaken by the newborn infant. Structural or functional abnormalities in the upper airway of infants put them at greater risk for feeding difficulties. Other unique features of infants include sucking pads in the cheeks to provide additional stability during sucking and a significantly larger tongue with respect to the oral cavity, which restricts tongue movement to the anterior–posterior direction characteristic of suckling.

Infants also exhibit a number of unique physiological aspects that are important for successful feeding and swallowing. These include reflexes that assist with development of feeding, such as the suck–swallow reflex, the rooting reflex, and the phasic bite reflex. As cortical development advances, these automatic reflexes

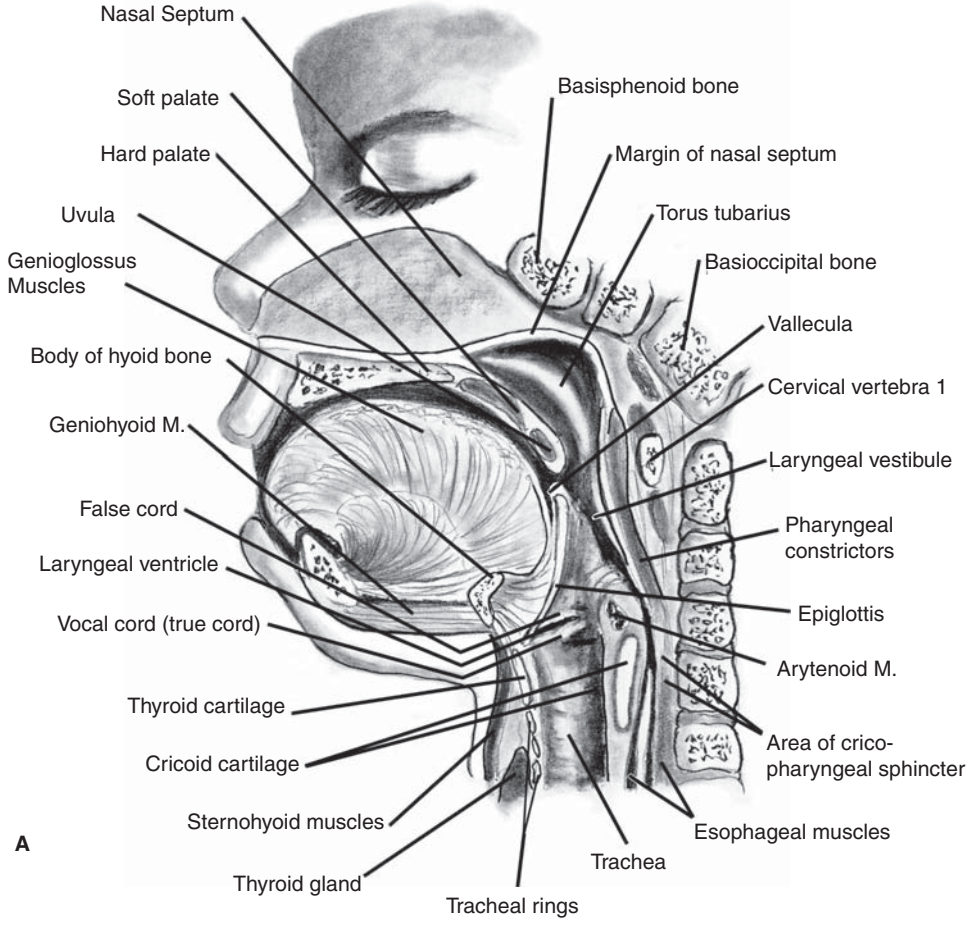


Figure 4.7A The pharynx: infant.

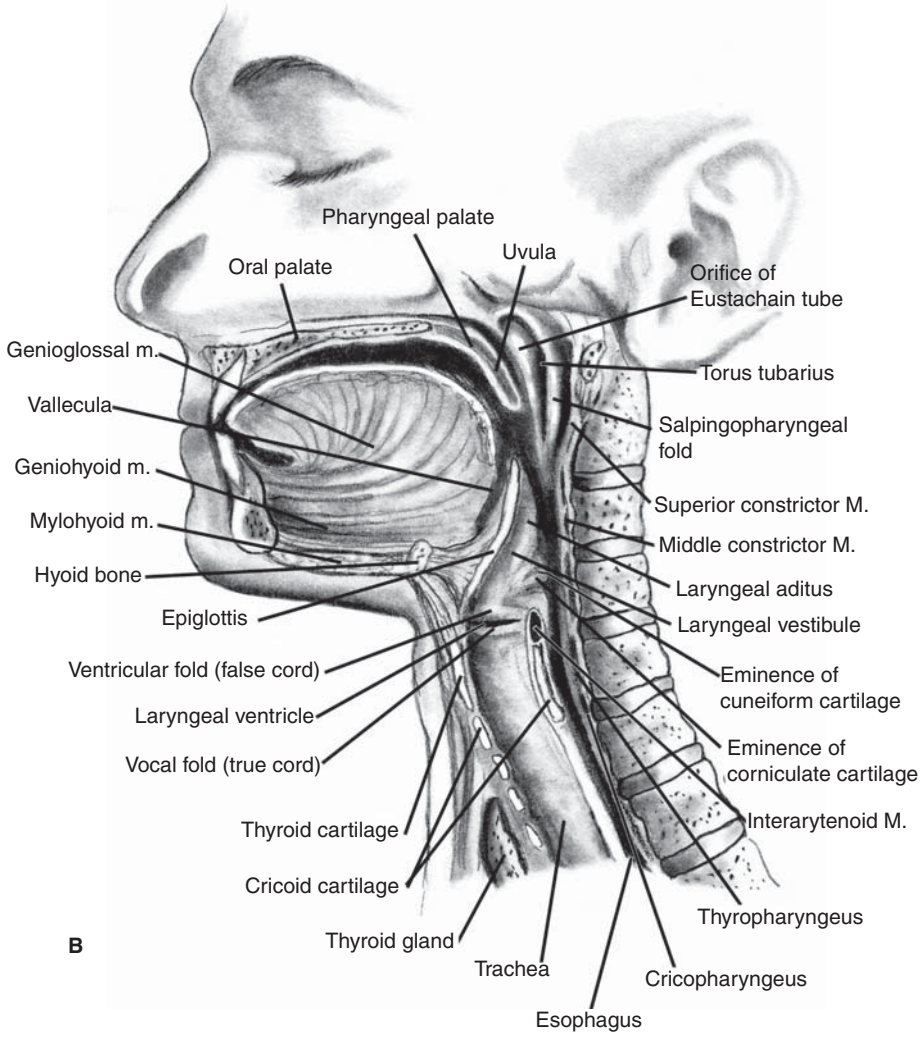


Figure 4.7B The pharynx: adult.

gradually evolve into more volitional actions, beginning during the period from 4 to 6 months of age. For example, at about 6 months of age, the transition from suckling to sucking begins to occur, with anatomic and neurologic maturation resulting in gradual lowering of the jaw, allowing more space for tongue movement, and gradual increase in volitional control permitting increased refinement and control of movements. The development of motor milestones in infants and toddlers is accompanied by attainment of feeding and swallowing skills, as outlined in Table 4.12 (45,48).

Critical periods are believed to exist in the development of normal feeding behavior. This can sometimes become problematic when caregivers are not sensitive to these critical stages. For example, caregivers may choose to maintain children on pureed foods due to apprehension regarding readiness to handle solid, chewable foods. However, research shows that delaying introduction of solid foods can result in food refusal and sometimes the development of food aversions (49). By the time children reach the age of 3 years, their ability to chew and swallow has matured and, with the exception of laryngeal position, their anatomy and physiology closely approximate those of the adult.

Infants with anatomical or physiologic abnormalities are at even greater risk for developing significant difficulty with establishing and maintaining oral feeding due to inability to initiate oral feedings within age-appropriate time frames. It is crucial for clinicians to have a thorough understanding of normal anatomical and physiologic development for feeding and swallowing in order to understand the implications of disorders.

Feeding and swallowing abilities involve multiple, interrelated anatomical and physiologic components within the body (eg, oral motor, pharyngeal, esophageal, respiratory, gastrointestinal). For this reason, effective management of children with feeding and swallowing disorders typically requires input from many specialists. These specialists may work separately or ideally may work within an interdisciplinary feeding

4.12 Attainment of Feeding and Swallowing Milestones

AGE (MONTHS)	DEVELOPMENT/POSTURE	FEEDING/ORAL SENSORIMOTOR
Birth to 4–6	Neck and trunk with balanced flexor and extensor tone Visual fixation and tracking Learning to control body against gravity Sitting with support near 6 months Rolling over Brings hands to mouth	Nipple feeding, breast, or bottle Hand on bottle during feeding (2–4 months) Maintains semiflexed posture during feeding Promotion of infant–parent interaction
6–9 (transition feeding)	Sitting independently for short time Self-oral stimulation (mouthing hands and toys) Extended reach with pincer grasp Visual interest in small objects Object permanence Stranger anxiety Crawling on belly, creeping on all fours	Feeding more upright position Spoon feeding for thin, smooth puree Suckle pattern initially suckle→suck Both hands to hold bottle Finger feeding introduced Vertical munching of easily dissolvable solids Preference for parents to feed
9–12	Pulling to stand Cruising along furniture First steps by 12 months Assisting with spoon; some become independent Refining pincer grasp	Cup drinking Eats lumpy, mashed food Finger feeding for easily dissolvable solids Chewing includes rotary jaw action
12–18	Refining all gross and fine motor skills Walking independently Climbing stairs Running Grasping and releasing with precision	Self-feeding: grasps spoon with whole hand Holding cup with 2 hands Drinking with 4–5 consecutive swallows Holding and tipping bottle
18–24	Improving equilibrium with refinement of upper extremity coordination Increasing attention and persistence in play activities Parallel or imitative play Independence from parents Using tools	Swallowing with lip closure Self-feeding predominates Chewing broad range of food Up–down tongue movements precise
24–36	Refining skills Jumping in place Pedaling tricycle Using scissors	Circulatory jaw rotations Chewing with lips closed One-handed cup holding and open cup drinking with no spilling Using fingers to fill spoon Eating wide range of solid food Total self-feeding, using fork

Source: From Refs. 45,48.

and swallowing team, providing the added benefit of coordinated care. An interdisciplinary approach is recommended at institutions where professionals evaluate and treat children with complex feeding and swallowing problems. Table 4.13 describes the members and functions of a comprehensive feeding and swallowing team.

Primary components of clinical assessment of pediatric feeding and swallowing skills include a thorough history, a prefeeding evaluation, and a feeding observation or trial feeding. If aspiration is suspected or risk of aspiration is a factor, instrumental assessments of swallowing, such as videofluoroscopic swallowing assessment (VFSS) or fiber-optic endoscopic evaluation of swallowing (FEES) may also be necessary following the clinical evaluation.

Feeding and swallowing difficulties can occur within a broad range of disorders, including anatomical or structural defects, neurologic deficits, systemic conditions, or complex medical conditions. Congenital

4.13 Feeding and Swallowing Team Members

TEAM MEMBER	FUNCTION
Parents	Primary caregivers and decision makers for child
Physician	Medical leader
(Pediatric physiatrist, gastroenterologist, developmental pediatrician)	Team co-leader Pediatric health and neurodevelopmental diagnosis Medical and health monitoring within specialty area
Speech-language pathologist	Team co-leader (active in feeding clinic and coordinates programmatic activities) Clinic and inpatient feeding and swallowing evaluation VFSS with radiologist FEES (with otolaryngologist)
Occupational therapist	Evaluates and treats children with problems related to posture, tone, and sensory issues such as oral defensiveness Oral sensorimotor intervention program
Dietitian	Assesses past and current diets Determines nutrition needs Monitors nutrition status
Psychologist	Identifies and treats psychological and behavioral feeding problems Guides parents for behavior modification strategies Directs inpatient behavioral feeding program
Nurse	Organizes preclinic planning Reviews records and parent information Coordinates patient follow-up Changes gastrostomy tubes
Social worker	Assists families for community resources Advocacy for the child
Additional specialists	
Otolaryngologist	Physical examination of upper aerodigestive tract Detailed airway assessment FEES with speech-language pathologist Medical and surgical treatment of airway problems
Pulmonologist	Lower airway disease—evaluation and management
Radiologist	VFSS with speech-language pathologist CT scan of chest Other radiographic diagnostic studies
Pediatric surgeon	Surgical management of gastrointestinal disease
Cardiovascular surgeon	Surgical management of cardiac disease
Neurologist/neurosurgeon	Medical and surgical management of neurologic problems
Physical therapist and rehab engineer	Seating evaluations and modifications to seating systems

Abbreviations: CT, computed tomography; FEES, fiberoptic endoscopic evaluation of swallowing; VFSS, videofluoroscopic swallow study.

anatomical or structural defects commonly affecting swallowing include tracheo-esophageal fistula (TEF), choanal atresia, and cleft palate. Acquired anatomical defects include laryngeal trauma. Neurologic deficits commonly affecting feeding and swallowing include cerebral palsy, traumatic brain injury, genetic syndromes, hypoxic/ischemic encephalopathy, meningitis, and Arnold-Chiari malformation. Systemic conditions typically associated with feeding and swallowing disorders include respiratory disease such as bronchopulmonary dysplasia (BPD) and Reactive Airway Disease (RAD), and gastrointestinal disorders such as gastroesophageal reflux (GER). Complex medical conditions resulting in swallowing disorders include prematurity and cardiac abnormalities. Given the interrelated nature of systems contributing to swallowing function, abnormalities (congenital or acquired) in any one of these systems can result in a feeding or swallowing disorder. For example, premature infants or infants with cardiac abnormalities often have abnormally high respiratory rates. If respiratory rates are above 60 breaths per minute, successful feeding is often not possible because energy expended for breathing leaves no energy for feeding, resulting in breakdown in coordination and increased risk for aspiration (50). Infants and children with reflux are at increased risk for feeding difficulties, as reflux contributes to negative experiences associated with feeding (gastroesophageal pain/discomfort, aspiration), and subsequent feeding aversion may develop. Structural defects such as vocal fold paralysis, laryngeal cleft, tracheoesophageal fistula, glossoptosis, or choanal atresia can result in difficulty protecting the airway, resulting in aspiration. Thus, obtaining a thorough medical history is crucial to understanding the etiology of a child's swallowing disorder.

In addition to medical history, a feeding history is important to obtain, as this will determine how to approach feeding assessment. If a child has been eating but his or her diet has been restricted to specific consistencies secondary to swallowing difficulties, this will be important to know. If a child has never been an oral eater, this is also critical information in subsequent clinical assessment decisions. Also, if a child has specific feeding utensils that he or she is accustomed to using, these should be used during the clinical assessment.

In addition to indirect assessment through parent interview and thorough review of medical records, direct observation of the child prior to introducing food should address alertness, ability to tolerate oral stimulation, and presence of a non-nutritive suck or ability to manipulate a bolus. Oxygen saturation and respiratory rate during these activities may need to be monitored. Positioning restrictions secondary to physical limitations or medical interventions should also be identified, as these may have an impact on the child's ability to feed. A complete oral motor examination should also be completed to determine the presence of any structural or functional abnormalities of the oral musculature. Presence/absence of swallow response, laryngeal elevation, and vocal fold function should all be screened prior to introduction of food.

With regard to level of alertness, children with TBI and associated cognitive impairment are at increased risk for aspiration related to decreases in cognitive level. A retrospective study completed by the authors found a significant correlation between Rancho Los Amigos Level of Cognitive Functioning and swallowing ability (51).

Regarding oral presentation of materials, there are a number of aspects to consider. Until recently, the Evan's Blue Dye Test or modified Evan's Blue Dye Test (MEBD) was commonly used to detect aspiration at the bedside. Its use has recently become somewhat more controversial. A recent report in the literature of a retrospective study comparing results from the use of MEBD, FEES, and VFSS documents low sensitivity of this measure to aspiration and cautions the clinician regarding false negative results (52). Another study, reported by Tippett and Siemens in 1996, notes 90% sensitivity of the MEBD in detecting aspiration of dyed foods for a group of 34 consecutive patients with tracheostomies (53). Thus, although the validity of the study for determining aspiration remains controversial and requires further objective study, it remains a useful component of the bedside swallowing assessment for some children in determining safety for oral intake.

When using foods during the bedside assessment, a number of variables can be manipulated, including the presenter, the consistency, the mode of presentation, and the bolus size (54). Food can be presented by the clinician, the parent, or the child, depending on the readiness and medical stability of the child and the availability and willingness of the parent. The child's age, current oral motor status, and premorbid feeding abilities will all affect decisions regarding consistency, mode of presentation, and bolus size.

If aspiration is suspected during the bedside assessment (coughing/choking, drop in oxygen saturation, wet vocal quality), further instrumental assessment such as a VFSS is generally indicated. Instrumental studies will assist in providing more detailed information, such as when the aspiration occurs (eg, before, during, after the swallow), what factors caused the aspiration (eg, premature spillage, unprotected airway, cricopharyngeal dysfunction), and what compensations, if any (eg, food consistency, positioning, presentation), may improve the swallow. The VFSS assesses three phases of swallowing: oral, pharyngeal, and esophageal (Fig. 4.8) (55). Figure 4.9 illustrates the position of the bolus during each of the three phases.

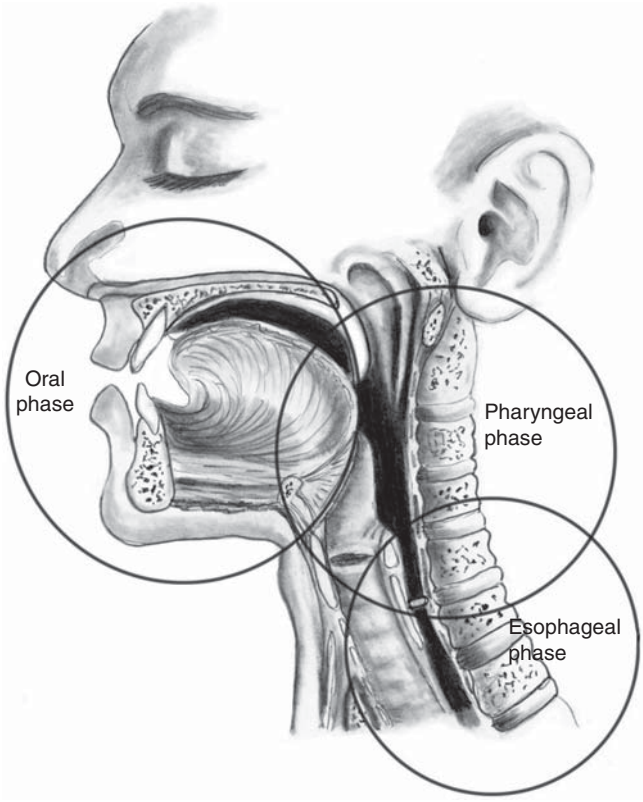
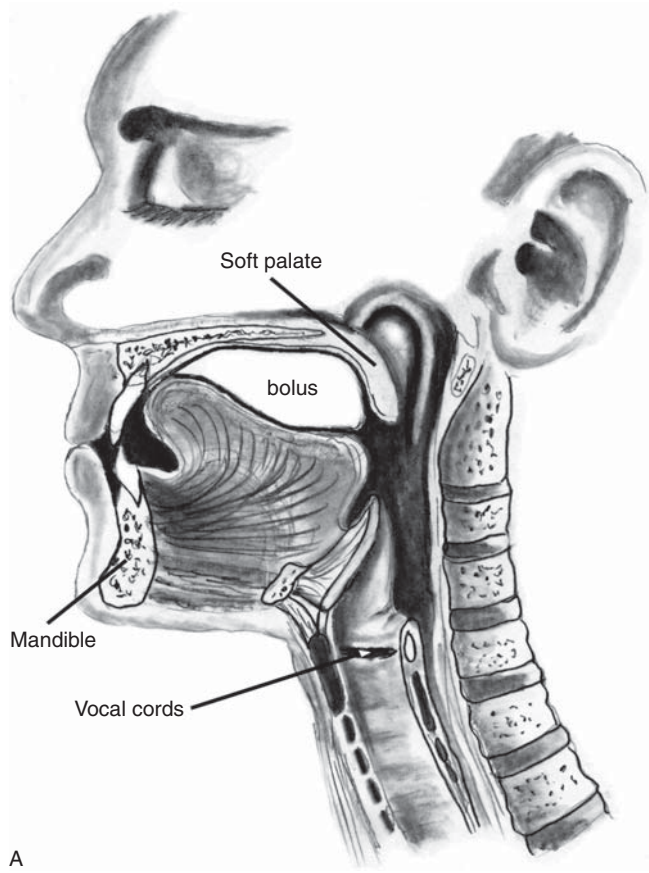
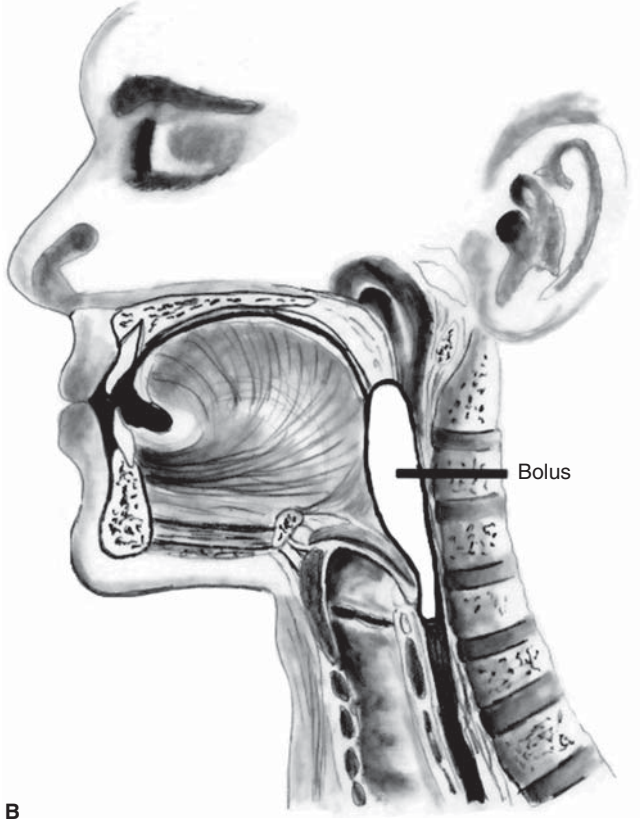


Figure 4.8 Phases of swallowing.



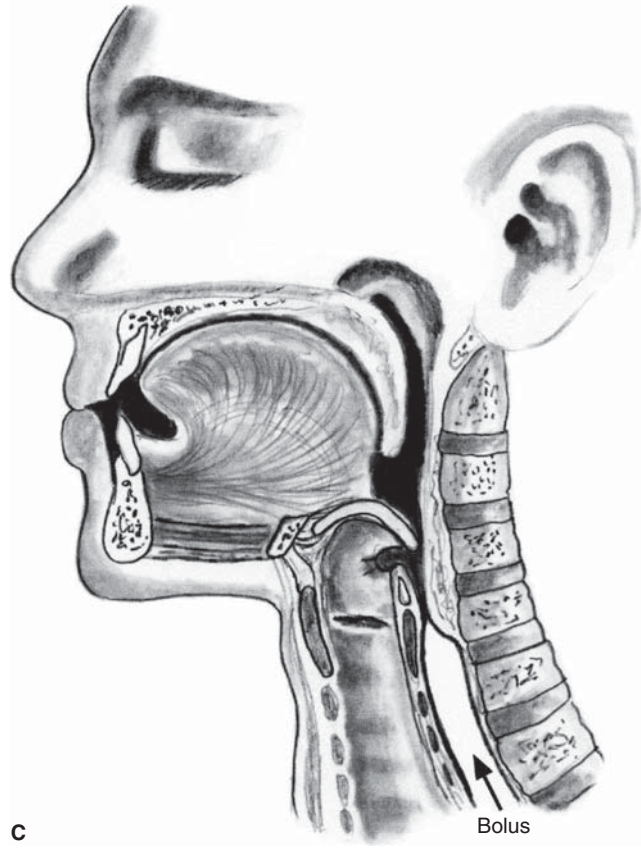
A

Figure 4.9 (A–C) Position of the bolus during phases of swallowing.



B

Figure 4.9 *Continued*



C

If there is no evidence of aspiration during the bedside assessment, recommendations are made for oral feeding based on the results of the trial feeding, the child's level of ability to feed orally, and the child's nutritional needs.

An alternative procedure, fiber-optic endoscopic evaluation of swallowing (FEES), is sometimes recommended instead of VFSS (56). It involves passage of a flexible fiber-optic endoscope transnasally to the area of the nasopharynx superior to the epiglottis, allowing observation of the swallowing mechanism from the base of the tongue downward. Use of FEES in the pediatric population has been established in the literature as a "practical and effective means of evaluating swallowing in children of all ages" (57,58). An advantage of VFSS is the ability to observe the actual aspiration event and to visualize the aspirated material in the airway. An advantage of FEES is the ability to observe amount and location of secretions and residue.

Instrumental examinations can be helpful in delineating pharyngeal and esophageal physiology as it pertains to swallowing. Decisions regarding when to perform an instrumental examination are guided by a number of factors, including risk for aspiration by history and clinical observation, documented incoordination of suck–swallow–breathe sequence during infant feeding, clinical evidence of pharyngeal or upper esophageal phase-swallowing deficits, prior aspiration pneumonia or similar pulmonary problems that could be related to aspiration, or etiology suspicious for pharyngeal or laryngeal problem, such as neurologic involvement commonly associated with feeding and swallowing problems.

Factors determining which type of instrumental exam to use are outlined in Table 4.14.

Management decisions with regard to feeding may be complex, and a number of factors must be considered, including medical, nutritional, oral sensorimotor, behavioral, and psychosocial. Treatment may include direct and indirect strategies, depending on the swallowing deficit. Examples of direct strategies include use of positioning maneuvers such as chin tuck or supraglottic swallow. Examples of indirect treatment strategies include diet modifications (eg, thickening liquids), changes in feeding routine (eg, small amounts frequently throughout the day), or changes in presentation of food (eg, Sippy cup versus bottle).

Diet texture modification is a common practice in management of dysphagia. Given the wide variation across clinicians and facilities, the American Dietetic Association attempted to establish some standard terminology and practice of texture modification through creation of The National Dysphagia Diet (NDD), published in 2002. The NDD was developed through consensus by a panel that included speech pathologists, dietitians, and food scientists. It proposes a hierarchy of four diet levels of semi-solids and solids, as well as two levels for liquids (see Table 4.15) (59).

One treatment option for children that is somewhat controversial involves oral sensorimotor intervention. This treatment method is typically performed by either speech pathology or occupational therapy, and involves techniques that are directed toward improving a child's ability to accept, manipulate, and swallow foods successfully. These techniques may include work with the jaw, lips, cheeks, tongue, and palate, both with regard to desensitizing and improving function. The benefits of such treatment approaches are still inconclusive, with little evidence to date documenting efficacy, efficiency, and outcomes. Some children appear to improve oral function with variations in texture, tastes, and temperature of foods. Other children benefit from posture and positioning changes. To be

4.14 Instrumental Swallowing Assessment

FINDINGS BETTER VIEWED ENDOSCOPICALLY (FEES)	FINDINGS BETTER VIEWED FLUOROSCOPICALLY (VFSS)
Airway closure	Tongue control and manipulation of bolus
Amount and location of secretions	Tongue contact to posterior pharyngeal wall
Frequency of spontaneous swallowing	Hyoid and laryngeal elevation
Pharyngeal/laryngeal sensitivity	Cricopharyngeal opening
Residue build-up	Airway closure at level of arytenoid to epiglottal contact
Aspiration before the swallow	Epiglottic retroversion
Aspiration after the swallow	Esophageal clearing
Coordination of the bolus and airway protection	Aspiration during the swallow
Coordination of breathing and swallowing	Amount of material aspirated
Ability to adduct TVFs for supraglottic swallow maneuver	
Fatigue over a meal	
Altered anatomy contributing to dysphagia	
Effectiveness of postural change on anatomy	

FEES, fiberoptic endoscopic evaluation of swallowing; VFSS, videofluoroscopic swallow study.

4.15 Dysphagia Diet Levels

DYSPHAGIA DIET CONSISTENCIES	EXAMPLES	INDICATIONS FOR USE
Thin liquids	Water, juice, soda	Adequate strength and coordination of lip and tongue musculature
Thick liquids	Nectars, milkshakes, cream soups. honey	Premature spillage of thin liquids with increased risk for aspiration
Mashed solids/purees	Yogurt, pudding, pureed meats and vegetables, cream of wheat	Mastication not required. Child may have weak tongue/mandibular musculature or reduced mastication.
Semi-solid	Minced meats/fish, cottage cheese, scrambled eggs, soft mashed fruits or vegetables	Some mastication possible. Fair oral motor control, although with some degree of oral weakness.
Soft chunk solid	Poached or hard-boiled eggs, bananas, canned fruit, mashable vegetables, bread, cold cereal, pancakes, pasta, rice, noodles, cake, pie	Mastication necessary. Appropriate for patients with adequate oral motor control but decreased endurance.

Source: From Ref. 59.

most effective, treatment of swallowing disorders in children should ensure safety while promoting a pleasurable experience. Treatment should also include the primary caregiver in every session, as well as provide home programs and suggestions for how to work with children at home on a daily basis (60–62).

In conclusion, communication and swallowing are both complex acts that require coordination of multiple systems, and disruption in a single component in any one of those systems can and most often does result in some degree of communication or swallowing impairment. Assessment and treatment of these impairments requires thorough knowledge of development and disorders of relevant pediatric anatomy and physiology, as well as an understanding of how to apply that knowledge in evaluation and treatment to ensure the best possible outcome. As our field advances, and as we advocate for the most appropriate treatment for the children we serve, reliance on evidence-based practice has become, and will continue to be, a crucial component for success.

PEARLS OR PERILS

- Children with tracheostomies and those on ventilators are capable of oral communication and oral eating.
- Speech and language delay refers to typical development at a slower pace, while speech and language disorder refers to atypical development when compared with peers.
- The majority of characteristics (10 out of 15) as per the DSM IV criteria used to formally diagnose an autism spectrum disorder involve communication deficits.
- Use of augmentative communication systems (devices, sign language, PECS) does not impede development of oral communication, and may, in fact, promote it.
- Liquids are the least safe alternative when initiating feeding following traumatic brain injury due to delayed reaction times associated with cognitive level of recovery.

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5

Adaptive Sports and Recreation

Ellen S. Kaitz and Michelle Miller

Adapted sports for the disabled (DA) were born in the mid-twentieth century as a tool for the rehabilitation of injured war veterans. They have blossomed to encompass all ages, abilities, and nearly all sport and recreational activities, from backyards to school grounds to national and Paralympic competitions. The trend in recent years has been away from the medical and rehabilitation roots to school- and community-based programs focused on wellness and fitness, rather than on illness and impairment. However, rehabilitation professionals remain connected in a number of important ways. Sports and recreation remain vital parts of a rehabilitation program for individuals with new-onset disability. Furthermore, rehabilitation professionals may be resources for information and referral to community programs. They may be involved in the provision of medical care for participants or act as advisors for classification. As always, research to provide scientific inquiry in biomechanics, physiology, psychology, sociology, technology, sports medicine, and many related issues is a necessary component.

HISTORY

Sports and exercise have been practiced for millennia. Organized activities for adults with disabilities have more recent roots, going back to the 1888 founding of the first Sport Club for the Deaf in Berlin, Germany. The International Silent Games, held in 1924, was the first international competition for DA athletes. Deaf sports were soon followed by the establishment of the British Society of One-Armed Golfers in 1932. Wheelchair sports are younger still, having parallel births in Britain and the United States in the mid-1940s. Sir Ludwig Guttman at the Stoke Mandeville Hospital in Aylesbury, England, invented polo as the first organized wheelchair team sport. "It was the consideration of the over-all training effect of sport on the neuro-muscular system and because it seemed the most natural form of recreation to prevent boredom in hospital . . ." (1). Within a year, basketball replaced polo as the principle wheelchair team sport. In 1948, the first Stoke Mandeville Games for the Paralyzed was held, with 16 athletes competing in wheelchair basketball, archery, and table tennis. This landmark event represented the birth of international sports competition for athletes with a variety of disabilities. The games have grown steadily, now comprising more than two dozen different wheelchair sports. The competitions are held annually in non-Olympic years, under the oversight of the International Stoke Mandeville Wheelchair Sport Federation (ISMWSF).

While Guttman was organizing wheelchair sports in Britain, war veterans in California played basketball in the earliest recorded U.S. wheelchair athletic event. The popularity flourished, and, a decade later, the first national wheelchair games were held. These games also included individual and relay track events. With the success of these games, the National Wheelchair

Athletic Association (NWAA) was formed. Its role was to foster the guidance and growth of wheelchair sports. It continues in this role today under its new name, Wheelchair Sports USA.

The U.S. teams made their international debut in 1960 at the first Paralympics in Rome. The term "Paralympic" actually means "next to" or "parallel" to the Olympics. In the 40 years since, the number and scope of sport and recreational opportunities has blossomed. The National Handicapped Sports and Recreation Association (NHSRA) was formed in 1967 to address the needs of winter athletes. It has more recently been reorganized as Disabled Sports USA (DS/USA). The 1970s saw the development of the United States Cerebral Palsy Athletic Association (USCPAA) and United States Association for Blind Athletes (USABA). In 1978, Public Law 95–606, the Amateur Sports Act, was passed. It recognized athletes with disabilities as part of the Olympic movement and paved the way for elite athletic achievement and recognition.

In the 1980s, a virtual population explosion of sport and recreation organizations occurred. Examples of these organizations include the United States Amputee Athletic Association (USAAA), Dwarf Athletic Association of America (DAAA), and the United States Les Autres Sports Association (USLASA; an association for those with impairments not grouped with any other sports organizations), the American Wheelchair Bowling Association (AWBA), National Amputee Golf Association, United States Quad Rugby Association (USQRA), and the Handicapped Scuba Association.

While the history of sports for the DA can be traced back a century, the development of junior-level activities and competition can be measured only in a few short decades. The NWAA created a junior division in the early 1980s that encompassed children and adolescents from 6 to 18 years of age. It has since established the annual Junior Wheelchair Nationals. Junior-level participation and programming have been adopted by many other organizations, including the National Wheelchair Basketball Association (NWBA), DS/USA, and American Athletic Association of the Deaf (AAAD). Sports for youth with disabilities are increasingly available in many communities through Adapted Physical Education (APE) programs in the schools, inclusion programs in Scouting, Little League baseball, and others.

EXERCISE IN PEDIATRICS: PHYSIOLOGIC IMPACT

It is widely accepted that exercise and physical activity (PA) have many physical and psychological benefits. Much research has been done to support this in adults. Only recently has data been presented to describe the benefits of exercise in both healthy children and those with chronic disease.

Exercise programs in healthy children have resulted in quantifiable improvements in aerobic endurance, static strength, flexibility, and equilibrium (2). Regular physical activity in adolescence is associated with lower mean adult diastolic blood pressures (3). However, a survey of middle school children showed that the majority are not involved in regular physical activity or physical education (PE) classes in school (4). Despite this, school days are associated with a greater level of PA in children at all grade levels than free days (5). Requiring PE classes in school improves the level of PA in children, but does not lower the risk for development of overweight or obesity (6) without dietary education and modification (7). Children attending after-school programs participate in greater amounts of moderate and vigorous physical activity than their peers (8).

Obesity is increasing in epidemic proportions among children in developed countries. It has been linked to development of the metabolic syndrome (defined as having three or more of the following conditions: waist circumference ≥ 90th percentile for age/sex, hyperglycemia, elevated triglycerides, low high-density lipoprotein [HDL] cholesterol, and hypertension) (9); both obesity and metabolic syndrome are more common in adolescents with lower levels of physical activity (10). Insulin resistance is reduced in youth who are physically active, reducing the risk of developing type 2 diabetes (11). Exercise in obese children can improve oxygen consumption and may improve cardiopulmonary decrements, including resting heart rate (12). An eight-week cycling program has been shown to improve HDL levels and endothelial function (13), though in the absence of weight loss, had little effect on adipokine levels (14).

Exercise has positive effects on bone mineralization and formation. Jumping programs in healthy prepubescent children can increase bone area in the tibia (15) and femoral neck, and bone mineralization in the lumbar spine (16). The effects of exercise and weight bearing may be further enhanced by calcium supplementation (17). The effects on postpubertal teens are less clear.

In children with chronic physical disease and disability, the beneficial effects of exercise are beginning to be studied more systematically. Historically, it was believed that children with cerebral palsy (CP) could be negatively impacted by strengthening exercises, which would exacerbate weakness and spasticity. Recent studies show this to be untrue. Ambulatory children with CP who participate in circuit training show improved aerobic and anaerobic capacity, muscle strength, and health-related quality-of-life scores (18).

In ambulatory adolescents with CP, circuit training can reduce the degree of crouched gait and improve perception of body image (19). Performing loaded sit-to-stand exercises results in improved leg strength and walking efficiency (20,21).

Percentage body fat is greater, and aerobic capacity (VO_2/kg) is lower in adolescents with spinal cord dysfunction than healthy peers. Their levels mirror those in overweight peers. They also reach physical exhaustion at lower workloads than unaffected controls (22). Participation in programs such as BENEfit, a 16-week program consisting of behavioral intervention, exercise, and nutrition education, can produce improvements in lean body mass, strength, maximum power output, and resting oxygen uptake (23).

Supervised physical training can safely improve aerobic capacity and muscle force in children with osteogenesis imperfecta (24). Patients with cystic fibrosis who participate in stationary cycling for aerobic conditioning dislike the tedium of the exercise, but improve their muscle strength, oxygen consumption, and perceived appearance and self-worth (25). Pediatric severe-burn survivors have lower lean body mass and muscle strength compared with nonburned peers; however, both are significantly improved following exercise training (26).

Children with polyarticular juvenile idiopathic arthritis have safely participated in aerobic conditioning programs, with improvements noted in strength and conditioning. Those with hip pain may be negatively impacted, having increased pain and disability (27). The exercise prescription in children experiencing hip pain should be modified to reduce joint forces and torques.

Joint hypermobility and hypomobility syndromes commonly result in pain. These patients demonstrate lower levels of physical fitness and higher body mass indexes, likely secondary to deconditioning (28). These and other children with pain syndromes benefit from increased exercise and physical activity.

EXERCISE IN PEDIATRICS: PSYCHOSOCIAL IMPACT

Regular physical activity in early childhood through adolescence fosters not only improvements in physical health, but also psychosocial health and development (29,30). The amount and quality of physical activity has significantly declined over the past several decades and even able-bodied (AB) children are no longer meeting the recommended guideline of one hour or more of moderate-intensity physical activity on five or more days a week (31). In disabled children, the amount of physical activity is even more restricted due to a variety of factors, including the underlying disability, physical barriers, and availability of resources (32). Sit et al. noted that the amount of time spent by children in moderate physical activity at school during PE and recess was lowest for children with a physical disability, at 8.9%, and highest for children with a hearing impairment, at 16.6% of recommended weekly minutes (33). Studies involving AB children have demonstrated that providing game equipment and encouragement from teachers can significantly increase moderate activity levels during recess time (34). Deviterne et al reported that providing participant-specific written and illustrated instruction concerning sporting activities such as archery to adolescents with motor handicaps improves their skill performance to a level similar to an AB adolescent at the end of the learning session that can foster increased self-esteem (35).

Many studies have demonstrated increased social isolation with fewer friendships among disabled children and adolescents. The Ontario Child Health Study revealed that children with a chronic disability had 5.4 times greater risk of being socially isolated and 3.4 times greater risk of psychiatric problems (36). Mainstreaming seems to have a positive impact, although concerns regarding AB peer rejection are still pervasive (37). Children in integrated PE programs were more likely to view their disabled peers as "fun" and "interesting" compared to children who were not integrated (38). One study of teacher expectations in mainstreamed PE classes revealed significantly lower expectations for the disabled student's social relations with peers (39). The attitude toward mainstreamed PE among high school students was significantly more positive in the AB group as opposed to the disabled population (40). Disabled children often view their lack of physical competence and secondly the status among their peers as the major barriers in social competence (41).

In addition to regular physical activity, play is a major component of childhood and important in psychosocial development of children. In preschool children with developmental delay or mental retardation, they were more likely to play on their own or not participate in play compared to the typically developing peers. Placing them in an integrated playgroup increased peer interactions compared to a nonintegrated playgroup, but did not correct the discrepancy in sociometric measures (42). There have also been discrepancies noted in the type of play for children with developmental delays. These children are less likely to participate in imaginative or constructive play (ie, creating something using the play materials) and more likely to participate in functional (ie, simple repetitive tasks) and exploratory play (43). It has been suggested that play should be taught, and one study by DiCarlo demonstrated that a program that taught pretend play

increased independent pretend toy play in 2-year-old children with disabilities (44).

Play for children with physical disabilities is also impaired. Children rely on technical aids such as bracing, walkers, wheelchairs, or adult assistants to access play areas and play equipment. Studies have shown that they are seldom invited to spontaneous playgroups and rarely take part in sporting activities unless the activity is geared toward children with disabilities (45). In a study by Tamm and Prellwitz, preschool and schoolchildren in Sweden were surveyed about how they viewed children in a wheelchair. They were willing to include disabled children in their games, but saw barriers to participation in outdoor activities due to the inaccessibility of playgrounds and the effect of weather. They did not feel disabled children would be able to participate in activities like ice hockey, but could play dice games. They felt sedentary and indoor activities were more accessible. The children also felt that disabled children would have high self-esteem, although most literature has documented that disabled children have low self-esteem (46).

In another study, children with motor disabilities were surveyed regarding how they perceived their technical aids in play situations. Younger children viewed their braces, crutches, walkers, or wheelchairs as an extension of themselves and helpful in play situations. Older children also saw the equipment as helpful, but a hindrance in their social life, as it made them different from their peers. Both older and younger children saw the environment as a significant barrier to play. Playgrounds often had fencing surrounding the area, sand, and equipment such as swings or slides that were not accessible without the assistance of an adult. The weather impacted accessibility due to difficulty maneuvering on ice or through snow. Children often took on an observational role on the playground or stayed inside. It was noted that the lack of accessibility sent the message that the DA children were not welcome and further isolated the DA group. As far as adult assistance, the younger children often incorporated the adult as a playmate. As children became older, they viewed their adult assistants as intrusive and a hindrance in social situations. Older children often chose to stay at home and be alone rather than going somewhere with an adult (45).

The research has highlighted many areas for improvement in accessibility for play and social interaction. Several articles detail ways to create accessible playgrounds, and these playgrounds are now becoming more prevalent in the community (Fig. 5.1). Playground surfaces can be covered with rubber, and ramps can be incorporated throughout the play structure to allow access by wheelchairs, walkers, and other assistive devices. Playground equipment can include wheelchair swings and seesaws that allow a wheelchair placement (47).

Figure 5.1 Playground equipment can be adapted to include children of all abilities, including pathways for wheelchair and walker access.

ADAPTED SPORTS AND RECREATION PROFESSIONALS

A variety of fields provide training and expertise in adapted sports, recreation, and leisure. They include adapted physical education teachers, child life specialists, and therapeutic recreation specialists. Physical and occupational therapists often incorporate sports and recreation into their treatment plans as well. However, their involvement remains primarily within a medical framework, and will not be discussed here.

Adapted Physical Education (APE) developed in response to the Individuals with Disabilities Education Act, which states that children with disabling conditions have the right to free, appropriate public education in the least restrictive environment. Included in the law is "instruction in physical education," which must be adapted and provided in accordance with the Individualized Education Program (IEP). APE teachers receive training in identification of children with special needs, assessment of needs, curriculum theory

and development, instructional design, and planning, as well as direct teaching (48,49). The APE National Standards (50) were developed to outline and certify minimum competency for the field. The standards have been adopted by only 14 states thus far. APE teachers provide some of the earliest exposure to sports and recreation for children with special needs, and introduce the skills and equipment needed for future participation.

Therapeutic recreation (TR) has its roots in recreation and leisure. It provides recreation services to people with illness or disabling conditions. Stated in the American Therapeutic Recreation Association Code of Ethics, the primary purposes of treatment services are "to improve functioning and independence as well as reduce or eliminate the effects of illness or disability" (51). Clinical interventions used by TR specialists run the gamut, from art, music, dance, and aquatic therapies to animal, poetry, humor, and play therapy. They may include yoga, tai chi chuan, aerobic activity, and adventure training in their interventions. While some training in pediatrics is standard in a TR training program, those who have minored in child life or who have done internships in pediatric settings are best suited for community program development. TR specialists are often involved in community-based sports for the DA, serving as referral sources, consultants, and support staff.

Child life is quite different from TR. Its roots are in child development and in the study of the impact of hospitalization on children. Its focus remains primarily within the medical/hospital model, utilizing health care play and teaching in the management of pain and anxiety and in support. Leisure and recreation activities are some of the tools utilized by child life specialists. Unlike TR specialists, child life workers focus exclusively on the needs and interventions of children and adolescents. There is often overlap in the training programs of child life and TR specialists. The role of the child life specialist does not typically extend to community sports and recreation programs.

PARTICIPATION IN PHYSICAL ACTIVITY

A number of scales have been developed to measure participation in activities. One example is the World Health Organization Health Behavior in Schoolchildren (WHO HBSC) survey. It is a self-reported measure of participation in vigorous activity that correlates well with aerobic fitness and has been shown to be reliable and valid (52). The Previous Day Physical Activity Recall (PDPAR) survey has been shown to correlate well with footsteps and heart rate monitoring, and may be useful in assessing moderate-to-vigorous activity of a short time span (53).

The Physical Activity Scale for Individuals with Physical Disabilities (PASIPD) records the number of days a week and hours daily of participation in recreational, household, and occupational activities over the past seven days. Total scores can be calculated as the average hours daily times a metabolic equivalent value and summed over items (54).

The Craig Hospital Inventory of Environmental Factors (CHIEF) is a 25-item survey that identifies presence, severity, and frequency of barriers to participation, and is applicable to respondents of all ages and abilities. A 12-item short form, CHIEF-SF is also available. When applied to a population with diverse disabilities, the CHIEF measure revealed the most commonly identified barriers to participation are weather and family support (55).

Pediatric measures include CAPE, which stands for Children's Assessment of Participation and Enjoyment. This tool has been validated in AB and DA children aged 6–21 years. It is used in combination with the PAC, the Preferences for Activities of Children. Together, they measure six dimensions of participation (ie, diversity, intensity, where, with whom, enjoyment and preference) in formal and informal activities and five types of activities (recreational, active physical, social, skill-based, and self-improvement) without regard to level of assistance needed. The scales can be used to identify areas of interest and help develop collaborative goal setting between children and caregivers. Identification of interests and barriers can facilitate problem solving and substitution of activities fulfilling a similar need (56). The European Child Environment Questionnaire (ECEQ), has been used to show that intrinsic and extrinsic barriers are equally important in limiting PA among DA youth (57).

Using these and other measures, one finds that participation in physical activity varies widely, even among nondisabled populations. The Third National Health and Nutrition Examination survey found that the prevalence of little to no leisure-time physical activity in adults was between 24% and 30%. The groups with higher levels of inactivity included women, older persons, Mexican Americans, and non-Hispanic blacks (58). A number of factors have been positively associated with participation in healthy adults, including availability and accessibility of facilities, availability of culture-specific programs, cost factors, and education regarding the importance of physical activity (59). Likewise, in healthy adolescents, physical activity is less prevalent among certain minorities, especially Mexican Americans and non-Hispanic blacks. Participation in school-based PE or community recreation centers are positively correlated with physical activity, as are parental education level and family income. Paternal physical activity, time spent outdoors, and attendance at nonvocational schools are

more common among children with higher levels of physical activity (60). Access to parks increases participation, especially in boys. Lower levels of moderate or vigorous physical activity are seen in those who reside in high-crime areas (61).

When followed over time, adolescents tend to decrease their participation in physical activity from elementary to high school. Boys who are active have a tendency to pursue more team sports, whereas girls are more likely to participate in individual pursuits (62). Coaching problems, lack of time, lack of interest, and limited awareness have been cited as other barriers to physical activity (63). Overall, however, informal activities account for more participation in children and teens than formalized activities (64).

Ready access to technology is associated with a decline in healthy children's participation in physical activity. Television watching is inversely related to activity levels and positively correlates with obesity, particularly in girls (65). Increased computer time is also related to obesity in teenage girls (66). Interestingly, playing digital games has not been linked with obesity, and *active* video games have, in fact, increased levels of physical activity among children and adolescents (67,68,69).

It is not surprising to learn that many of the barriers to physical activity identified by AB are the same as those experienced by DA children. The most commonly cited are lack of local facilities, limited physical access, transportation problems, attitudinal barriers by public and staff, and financial concerns. Lack of sufficiently trained personnel and of appropriate equipment have also been identified (32,70,71). Among those children with severe motor impairments, the presence of single-parent household, lower family income, and lower parent education are significant barriers (64). Pain is more frequently reported in children with CP and interferes with participation in both activities of daily living (ADLs) and PA (72). The presence of seizures, intellectual impairment, impaired walking ability, and communication difficulties predict lower levels of physical activity among children with CP (73). Many children are involved in formal physical and occupational therapy.

Therapists as a whole have been limited in their promotion of recreation and leisure pursuits for their pediatric clientele (74). Therapy sessions and school-based programs provide excellent opportunities for increasing awareness of the need and resources available for physical activity. Policy and law changes related to the Americans with Disabilities Act are resulting in improved access to public facilities and transportation. Many localities are providing adapted programs and facilities that are funded through local taxation (Fig. 5.2). Impairment-specific sports have grown from grassroots efforts, often with the assistance or guidance of rehabilitation professionals. Organizations such as BlazeSports (www.blazesports.org) have developed programs throughout the United States. The bedrock of BlazeSports America is made up of the community-based, year-round programs delivered through local recreation providers. It is open to youth with all types of physical disabilities. Winners on Wheels "empowers kids in wheelchairs by encouraging personal achievement through creative learning and expanded life experiences that lead to independent living skills." Chapters exist in many cities across the United States and incorporate physical activity into many of the activities they sponsor.

Figure 5.2 Many public facilities have wheelchairs available for rent or use that are designed for use on the beach.

The American Association of Adapted Sports Programs (AAASP) employs athletics through a system called the adaptedSPORTS Model. "This award-winning model is an interscholastic structure of multiple sports seasons that parallels the traditional interscholastic athletic system and supports the concept that school-based sports are a vital part of the education process and the educational goals of

students" (www.adaptedsports.org). The sports featured in the adaptedSPORTS model have their origin in Paralympic and adult disability sports, and are cross-disability in nature. The program provides standardized rules for competition, facilitating widespread implementation. Application in the primary and high school levels can help students develop skills that can lead to collegiate-, community-, and elite-level competition.

In some communities, AB teams or athletes have partnered with groups to develop activity-specific opportunities. Fore Hope is a nationally recognized, nonprofit organization that uses golf as an instrument to help in the rehabilitation of persons with disabilities or an inactive lifestyle. The program is facilitated by certified recreational therapists and golf professionals (www.forehope.org). A similar program known as KidSwing is available to DA children in Europe and South Africa (www.kidswing-international.com). Several National Football League (NFL) football players have sponsored programs targeting disabled and disadvantaged youth. European soccer team players have paired with local organizations to promote the sport to DA children.

Financial resources are also becoming more available. The Challenged Athletes Foundation (CAF) supports athletic endeavors by providing grants for training, competition, and equipment needs for people with physical challenges. Athletes Helping Athletes (www.athleteshelpingathletes.org) is a nonprofit group that provides handcycles to children with disabilities at no cost. The Golden Opportunities fund (www.dsusa.org) provides support and encouragement to DA youth in skiing. More resources can be found at the Disaboom Web site (www.disaboom.com).

INJURY IN THE DISABLED ATHLETE

With more DA athletes come more sports injuries. The field of sports medicine for the disabled athlete is growing to meet pace with the increase in participation. Among elite athletes in the 2002 Winter Paralympics, 9% sustained sports-related injuries. Sprains and fractures accounted for more than half of the injuries, with strains and lacerations making up another 28% (74). Summer Paralympians sustained sprains, strains, contusions, and abrasions rather than fractures or dislocations (75). Retrospective studies have shown 32% incidence of sports injuries limiting participation for at least a day. Special Olympics participants encounter far fewer medical problems than their elite counterparts. Of those seeking medical attention during competition, overall incidence is under 5%, with nearly half related to illness rather than injury. Knee injuries are the most frequently reported musculoskeletal injury. Concerns regarding atlanto-occipital instability and cardiac defects must be addressed in the participant with Down's syndrome.

Among elite wheelchair athletes, upper limb injuries and overuse syndromes are common; ambulatory athletes report substantially more lower limb injuries. Spine and thorax injuries are seen in both groups (76). Wheelchair racers, in particular, report a high incidence of arm and shoulder injuries. The injuries do not appear to be related to distance, amount of speed training, number of weight-training sessions, or duration of participation in racing (77). Survey of pediatric wheelchair athletes reveals that nearly all children participating in track events report injuries of varying degrees. Blisters and wheel burns are most frequent, followed by overheating, abrasions, and bruising. Shoulder injuries account for the majority of joint and soft tissue complaints. Injuries among field competitors are less frequent, with blisters and shoulder and wrist problems reported most often. Swimmers report foot scrapes and abrasions from transfers, suggesting opportunity for improved education regarding skin protection (78).

An important factor in injury prevention for the wheelchair athlete is analysis of and instruction in ergonomic wheelchair propulsion (79). Proper stroke mechanics positively affect pushing efficiency. Push frequency also affects energy consumption and can be adjusted to improve athletic performance (80). Motion analysis laboratories and Smartwheel technology can be utilized to objectively analyze and help improve pushing technique, thus reducing injury (81).

While some injuries are sport-specific, others may be more common among participants with similar diagnoses. Spinal cord–injured individuals are at risk for dermal pressure ulcer development, thermal instability, and autonomic dysreflexia. In fact, some paralyzed athletes will induce episodes of dysreflexia, known as "boosting," in order to increase catecholamine release and enhance performance (82). Education regarding the risks of boosting is essential, as are proper equipment and positioning to protect insensate skin.

Athletes with limb deficiencies may develop painful residual limbs or proximal joints from repetitive movements or ill-fitting prostheses. The sound limb may also be prone to injury through overuse and asymmetric forces (83). Participants with vision impairments sustain more lower limb injuries than upper limb, while those with CP may sustain either. Spasticity and foot and ankle deformities in children with CP may further predispose to lower limb injury. As with all athletes, loss of range of motion, inflexibility, and asymmetric strength further predispose the DA participant to injury. Instruction in stretching, strengthening, and cross training may reduce the incidence and severity of injury.

"Evening the Odds": Classification Systems

Sport classification systems have been developed in an attempt to remove bias based on innate level of function. In theory, this would allow fair competition among individuals with a variety of disabilities. Early classifications were based on medical diagnostic groupings: one for athletes with spinal cord lesion, spina bifida, and polio (ISMWSF); one for ambulatory amputee athletes and a separate one for amputee athletes using wheelchairs; one for athletes with CP; one for Les Autres (International Sports Organized for the Disabled [ISOD]), and so forth. (Table 5.1). These early attempts reflected the birth of sports as a rehabilitative tool. This form of classification continues to be used in some disability-specific sports, such as goal ball for blind athletes and sit volleyball for amputee athletes. Other older systems took into account degree of function. This system unfairly penalized athletes who were more physically fit, younger, more motivated, and so forth.

With the growth of elite competitive sports came the need for more impairment-based classification systems, which shifted the focus from disability to achievement. Impairment-based classifications have the added advantage of reducing the number of classes for a given sport. This results in greater competition within classes and reduces the number of classes only having one or two competitors. Impairment classifications are further utilized in sport-specific definitions, such as in basketball, quad rugby, and skiing (Table 5.2).

The issue of inclusion in elite sports has been quite controversial. Debate exists not only within

5.1 Comparison of medical and functional classifications of les autres athletes

MEDICAL CLASSIFICATION

LEVEL	ATHLETES WITH . . .	EXAMPLES
L1	Severe involvement of all four limbs	Severe multiple sclerosis Muscular dystrophy Juvenile rheumatoid arthritis with contractures
L2	Severe involvement of three or all four limbs but less severe than L1	Severe hemiplegia Paralysis of one limb with deformation of two other limbs
L3	Limited functioning of at least two limbs	Hemiparesis Hip and knee stiffness with deformation of one arm
L4	Limited functioning in at least two limbs; limitations less than in L3	Contracture/ankylosis in joints of one limb with limited functioning in another
L5	Limited functioning in at least one limb or comparable disability	Contracture/ankylosis of hip or knee Paresis of one arm Kyphoscoliosis
L6	Slight limitations	Arthritis and osteoporosis Ankylosis of the knee

FUNCTIONAL CLASSIFICATION

LEVEL	DESCRIPTION
L1	Uses a wheelchair; reduced function of muscle strength and/or spasticity in throwing arm; poor sitting balance
L2	Uses a wheelchair; good function in throwing arm and poor to moderate sitting balance or reduced function in throwing arm with good sitting balance
L3	Uses a wheelchair; good arm function and sitting balance
L4	Ambulatory with or without crutches and braces or problems with balance together with reduced function in throwing arm
L5	Ambulatory with good arm function; reduced function in lower extremities or difficulty in balancing
L6	Ambulatory with good upper extremity function in throwing arm and minimal trunk or lower extremity impairment

Source: United States Olympic Committee, 1998.

5.2 Classification for Alpine Skiers

VISUALLY IMPAIRED	
B1	Totally blind
B2	Partially sighted with little remaining sight
B3	Partially sighted with more remaining sight
STANDING	
LW1	Double above-knee amputees
LW2	Outrigger skiers
LW3	Double below-knee amputees (CP5, CP6)
LW4	Skiers with prosthesis
LW5/7	Skiers without poles
LW6/8	Skiers with one pole
LW9	Disability of arm and leg (amputation, cerebral palsy, hemiplegic)
SITTING	
LW10	Mono skiers (high degree of paraplegia)
LW11	Mono skiers (lower degree of paraplegia)
LW12/1	Mono skiers (lower degree of paraplegia, double above-knee amputees)

Source: International Paralympic Committee, 2008.

sports for the disabled, but also in the inclusion of DA athletes in sports with AB competitors. A few sports such as archery have fully integrated AB and DA competitors. However, in sports such as marathon racing, the AB athlete is at a distinct disadvantage, being unable to achieve the speeds or times of the wheelchair racer. Having classification systems and segregation in DA sports allows for achievement based on ability rather than disability. Yet, there continues to be a discrepancy between the recognition and reward for AB and for DA athletes. The issues of integration and classification continue to be refined and debated. Inclusion at the educational and recreational levels remains much more feasible through Adapted Physical Education and community-based programs.

Adapting Recreation Opportunities

Camping

Camping, mountaineering, and hiking are among the many outdoor adventure activities available to children with disabilities. The National Park Service maintains information on park accessibility and amenities across the United States. The America the Beautiful—National Parks and Federal Recreation Lands Pass is available to any blind or permanently disabled U.S. citizen/permanent resident, and allows free lifetime admission to all national parks for the individual and up to three accompanying adults. Accompanying children under the age of 16 are free. It is obtained at any federal fee area or online at http://store.usgs.gov/pass and allows a 50% reduction in fees for recreation sites, facilities, equipment, or services at any federal outdoor recreation area.

Boy and Girl Scouts of America each run inclusion programs for children with disabilities. Opportunities also exist in dozens of adventure and specialty camps across the United States. Some are geared to the disabled and their families, allowing parallel or integrated camping experiences for disabled children. Participation requires few adaptations, and the Americans with Disability Act has been instrumental in improving awareness in barrier-free design for trails, campsites, and restrooms. Parents should evaluate the camps in regard to the ages of the participants, medical support, and cost. Often, camps are free or offer scholarships and may provide transportation. Some camps will have diagnosis-specific weeks, such as CP, spina bifida, muscular dystrophy, and so on. A nice summer camp resource is www.mysummercamps.com.

There are accessible recreational vehicles (RVs) available for rent as well as purchase. Many manufacturers will customize their RVs during the production process. A number of travel clubs exist across the United States and have Web sites giving information on accessible campsites with an RV in mind. In addition, many have annual gatherings of their members at a chosen campsite. One good Web site is www.handicappedtravelclub.com.

Fishing

Fishing can be enjoyed by virtually anyone, regardless of ability. One-handed reels, electric reels, and even sip-and-puff controls allow independent participation. A variety of options exist for grasping and holding rods as well. These range from simple gloves that wrap the fingers and secure with Velcro or buckles to clamps that attach directly to the rod, allowing a hand or wrist to be slipped in. Harnesses can attach the rod to the body or to a wheelchair, assisting those with upper limb impairments. There are devices that assist with casting as well for individuals with limited upper body strength or control. Depending on the level of expertise and participation of the fisher, simple or highly sophisticated tackle can also be had (84,85).

Both land and sea fishing opportunities are accessible to the disabled. Piers are usually ramped and may have lowered or removable rails for shorter or seated individuals. Boats with barrier-free designs offer fishing and sightseeing tours at many larger docks. These offer variable access to one or all decks, toilet facilities, and shade (84).

Hunting

Adaptations to crossbows and rifles have made hunting accessible for many. The crossbow handle and trigger can be modified for those with poor hand function. Stands for rifles and crossbows are also available for support. Many hunting ranges have incorporated wheelchair-accessible blinds.

Dance

Dancing has become more popular in the able-bodied and disabled populations over the past 10 years. The wheelchair is considered an artistic extension of the body, and many dances have been adapted for the movement of the wheels to follow the foot patterns of classical ballroom dancing. Wheelchair dancing was first begun in 1972 and pairs DA and AB individuals in a variety of dances. Recreational opportunities and competition are available in many states, with classes including duo-dance featuring two wheelchair dancers together, group dancing of AB and wheelchair competitors in a synchronized routine, and solo performances. Wheelchair dance sport has been a recognized sport within the Paralympics since 1998, although it is not currently included in the program. International competition in wheelchair dance has been around since 1977. In addition, ballet, jazz, and modern dance companies offer inclusion for children with disabilities.

Martial Arts

Martial arts classes will include children with a variety of disabilities. The classes can be modified to allow skills at the wheelchair level in forms, fighting, weapons, and breaking. Children are taught self-respect, control, and can advance through the belt system. They are also taught basic self-defense in some settings. There are many different styles of martial arts, and parents should check within their communities for available resources. Equipment adaptations are not needed for this activity.

Scuba and Snorkeling

Freedom from gravity makes underwater adventure appealing to individuals with mobility impairments. Little adaptation to equipment is needed to allow older children and adolescents with disabilities to experience the underwater world. Lower limb–deficient children may dive with specially designed prostheses or with adapted fins, or may choose to wear nothing on the residual limb. Similar to those with lower limb weakness or paralysis, they may use paddles or mitts on the hands to enhance efficiency of the arm stroke. Of particular importance is the maintenance of body temperature, especially in individuals with neurologic disability, such as spinal cord injury or CP. Wet or dry suits provide insulation for cool or cold water immersion. They also provide protection for insensate skin, which can be easily injured on nonslip pool surfaces, coral, and water entry surfaces.

It is crucial that individuals receive proper instruction by certified dive instructors. Most reputable dive shops can provide information and referral. The Handicapped Scuba Association is an excellent reference as well. Disabled divers are categorized based on level of ability. They may be allowed to dive with a single buddy (as with AB divers), two buddies, or two buddies of which one is trained in emergence rescue techniques. Although there is no particular exclusion from diving based solely on disability, a number of medical considerations may preclude scuba diving, including certain cardiac and pulmonary conditions, poorly controlled seizures, and use of some medications. Discussion with the primary care physician and with dive instructors should precede enrollment or financial investment. Scuba diving has also been used as adjunctive therapy in acute rehabilitation programs (86).

Music

Music has been used both as a therapeutic tool and as a means of artistic expression. Attentive behavior was increased in children with visual impairments who participated in a music program (87). There are many options for children who want to play music. Adaptations may be as simple as a universal cuff with a holder for drumsticks or as sophisticated as a computer program to put sounds together to form a musical piece. Two such computer programs are Fractunes and Switch Ensemble. Drumsticks can have built-up rubberized grips. Straps or a clamp may be used to hold a smaller drum onto a wheelchair for a marching band. Woodwind and brass instruments can be fitted with stands and finger pieces adapted for one-handed playing. Mouthpieces may have different angulations to allow easier access for those who have trouble holding the instrument. Some musical instrument makers, including Flutelab (www.flutelab.com), have become quite creative in how they can adapt their instruments. Other individuals have learned to play instruments such as the guitar with their feet (Fig. 5.3).

Figure 5.3 Musical instruments and their video game likenesses may be adapted for use by those with limited strength.

Hippotherapy and Horseback Riding Therapy

Therapeutic horseback riding, or hippotherapy, has been popular in Europe since the 1950s and spread to the United States in the late 1960s. It uses the rhythmic motions and warmth of the horse to work on the rider's tone, range of motion, strength, coordination, and balance. The movement of the horse produces a pattern of movements in the rider that is similar to human ambulation (88). The rider may sit or be placed in various positions on the horse's back or, alternatively, may perform active exercises while on horseback.

There are two recognized treatment options: instructor-directed, recreational horseback riding therapy (HBRT) and licensed therapist–directed hippotherapy. HBRT is directed by nontherapist riding instructors and assistants, and follows the North American Riding for the Handicapped Association's (NARHA) curriculum for riding therapy. It encourages the development of sensorimotor and perceptual motor skills, utilizing the developmental riding therapy methods described by Spink (89). Children are challenged to maintain balance and posture in all body positions as the horse walks and the instructor encourages them to reach and use their upper limbs in a variety of exercises (90). Hippotherapy is directed by a licensed health professional and focuses treatment based on the impairment and functional limitations of children with neuromuscular dysfunction. The horse is considered a therapeutic tool to improve language or gross motor function, including walking, posture, balance, and mobility (91).

Children with any of a variety of disorders that affect muscle tone, strength, or motor skills may benefit from this form of therapy. These disorders include but are not limited to CP, myelodysplasia, cerebral vascular accident, traumatic brain injury, spinal cord injury, amputations, neuromuscular disorders, and Down's syndrome. A careful screening of individuals with spinal pathology should be performed to rule out instability prior to participation. This screening includes the Down's syndrome population, in whom 15% to 20% has atlantoaxial instability (92). In addition, children with a poorly controlled seizure disorder may be excluded. Cognitive or behavioral impairments should not be so severe that they place the rider or others at risk.

Many potential physical, cognitive, and emotional benefits of hippotherapy have been reported. These include improvements in tone, posture, balance, strength, gait, hygiene, attention, concentration, language skills, self-confidence, and peer relations (88,93) Most studies have evaluated the effect on the CP population and children with developmental disabilities. Benda et al noted improvements in back and hip muscle symmetry using remote surface electromyography in children with CP following an eight-minute training session on the horse, as compared to children who sat for eight minutes on a barrel. Unfortunately, the study did not evaluate if these improvements persisted once therapy was completed (94). Sterba studied the effect of an 18-week training session of riding three times a week on children with different types of CP. Significant improvements in the Gross Motor Function

Measure (GMFM) were reported. Progress was noted in all dimensions of the GMFM: lying and rolling; sitting; crawling and kneeling; standing; and walking, running, and jumping during therapy. At six weeks following completion of the program, only dimension E (walking, running, and jumping) had continued improvement, with the other domains returning to baseline (95). In a separate area of study, boys with attention-deficit hyperactivity disorder (ADHD) and/or learning difficulties demonstrated decreased frustration, physical aggression, and difficulties with authority relations after participating in HBRT (96).

Resources

North American Riding for the Handicapped Association (NARHA)—www.narha.org

Aquatic Therapy

Water has been an important therapeutic medium for centuries. In pool therapy, the water's intrinsic buoyancy nearly eliminates the effects of gravity. Therefore, less effort is required for movement and the weight borne on the limbs is minimized. As recovery progresses, activity in the water can be graded to provide varying amounts of resistance. The water temperature can also be therapeutic, with warmer water producing muscle relaxation. Finally, children often view the pool as fun rather than therapy and are often encouraged by the ability to perform movements in the water that they are unable to do on land (97).

The most common indication for pool therapy is muscle weakness, although gains are also noted in range of motion, coordination, endurance, and normalization of tone. It has been recommended for children with CP, neuromuscular disorders, spinal cord injuries, myelodysplasia, arthritis, brain injury, stroke, burns, fractures, and even asthma (97). Children as young as neonates may benefit (98). Aquatic therapy, however, is not indicated for everyone. Caution should be used in children with hypertension or hypotension, open wounds, infective skin lesions, fever, or temperature instability (97). It is contraindicated for children with uncontrolled seizures or excessive fear of the water, or whose cognitive status poses a safety risk for themselves or others.

There are a variety of approaches in aquatic therapy, including Bad Ragaz, Watsu, Halliwick method, Sequential Swim Techniques (SST), and task-specific approaches (99,100). Bad Ragaz is based on proprioceptive neuromuscular facilitation using active and passive techniques (101). The Watsu approach is an energy-release technique in which a body segment is moved while the rest of the body is allowed to drag through the water, thus providing stretch (102). The Halliwick method and SST work on distinct movement patterns with a specific goal, such as swimming. The task-specific approach includes activities such as ambulation (103).

A review of the literature supporting aquatic therapy in children contains little Class 1 evidence. Most studies are small in sample size and fall within level 4 and 5 evidence (104). One study with Class 2 evidence demonstrated improved vital capacity and water orientation skills (standing in the water, floating, and swim positions) in kindergarteners with CP who participated in a six-month aquatic program compared to controls in a land-based program (105). In a recent study by McManus et al, children between the ages of 6 and 30 months with delayed functional mobility completed an aquatic therapy program as part of early intervention (EI). There was a significant improvement in motor skills compared to the control group, who received traditional EI therapy services based on the Gross Motor Subsection of the Mullen Scales of Early Learning. The study was limited by the sample size, variety of diagnoses, and the lack of more accepted testing as accomplished by the GMFM or Peabody (106). The adult literature has more evidence-based support of aquatic therapy, and the same types of studies will need to be replicated in the pediatric population.

Aquatic therapy programs are now offered through many hospital programs as well as local facilities such as the Young Men's Christian Association (YMCA) and Young Women's Christian Association (YWCA).

Yoga/Tai Chi Chuan

Yoga is a mind–body movement therapy with the following components: body mechanics, including breathing skills (pranayama) and simple postures (yogasanas); fitness (sithilikarana, vyayama, and suryanamaskar); and meditation. It has been demonstrated that physiologic changes in the body can be achieved through breathing manipulation, postures, and cognitive control (107,108). There are many different types of Hatha yoga currently being practiced in the United States, each with a different emphasis on the various components.

Studies in the pediatric population have focused primarily on typically developing children, although some have evaluated the effect on those with mental retardation, ADHD, visual impairment, physical impairment, and asthma. The current research has been classified at the 2B level or lower. Primary drawbacks in the studies have been the lack of randomized controlled studies, absent or poor reporting of adverse events, and the wide variety of Hatha yoga protocols used for treatment (109). The existing literature suggests that there can be improvements in mental ability, such as attention, motor coordination, emotional control, and social skills, in children with ADHD or mental

retardation (107,110). There was a positive impact in typically developing children on spatial memory, reaction time, motor planning, motor speed, heart rate, and focused attention (109). Children with visual impairments demonstrated less anxiety and children with physical impairments regained some functional ability, with improved flexibility and balance (111). Children with asthma improved their forced expired volume (FEV), peak flow rate, and distance walked in a 12-minute time period, as well as reported decreased symptoms and medication use (112,113).

Tai chi chuan, or tai chi, has been practiced in China for centuries and has recently gained popularity in the United States. It is a low-intensity exercise with flowing, controlled movement patterns emphasizing semi-squatting postures, balance, relaxation, flexibility, and regulated breathing. Like yoga, it works to balance the mind and body. There are various styles, including Chen, Yang, Wu, and Sun (114).

Most studies of tai chi have been completed in the elderly population and suggest some benefit for overall balance and prevention of falls, strength, flexibility, reduction of blood pressure, memory, and emotional well-being, with decreases in depression and anxiety (114,115). Studies in the treatment of rheumatoid arthritis have been limited by poor methodological quality, and do not definitively support the use of tai chi as a treatment (116). In their review of the literature, Lee et al discussed the possible adverse effects of increased pain in the knee, shoulder, and back, yet acknowledge possible improvements in disability index, quality of life, depression, and mood in the rheumatoid population. There are few studies in the pediatric population. One study presented by Yu-Feng Chang et al. noted improvements in asthmatic children in their forced vital capacity (FVC), FEV1, and peak expiratory flow at rest and post-exercise after completing a 12-week tai chi program. There was no significant change in their reported symptoms when compared to the control group (117). Further studies are needed to delineate the benefit of this therapy in the pediatric population.

Sports for Fun and Competition

Archery

With the exception of the adaptive equipment, archery is essentially unmodified. It is a popular recreational and competitive activity in which individuals with virtually any disability can participate (Fig. 5.4).

Equipment

Trigger release or release cuff: Designed for individuals with a poor grasp or weakness, it assists in the smooth draw and release of the bowstring. Its use is permitted in sanctioned competition only by those with tetraplegia from cerebral palsy or a spinal cord injury.

Wrist and elbow supports: Provide support and stability for the bow arm.

Standing supports: Give the wheelchair user a choice between sitting and standing while shooting.

Bow supports: Provide support and stability of the bow for individuals with weakness or a poor grasp. Its limited use is permitted only in USCPAA competition.

Crossbows and compound bows: For recreational use primarily, although compound bows are allowed in USCPAA competition.

Mouth pieces: Allow archers with upper extremity impairments to draw the bow string with the mouth (36).

Resources

Physically Challenged Bowhunters of America, Inc.: http://pcba-inc.org

Grand National Archery Society (UK): www.gnas.org

U.S. Disabled Archery Team: www.da-usa.org

Baseball

Miracle League is a program facilitating participation of disabled children in a baseball-like activity. In Miracle League play, every player bats once per inning, all base runners are safe, each player scores a run before the inning is over, and the last batter up gets a home run. AB peers and community volunteers assist DA players. Each team and each player wins every game. Another form of the sport is Push N Power Baseball, which utilizes hockey sticks and balls in combination with traditional baseball rules. When unable to catch, pass, or pick up the ball, verbal responses are substituted. Little League baseball also has a division called Challenger, which encourages participation by cognitively and physically challenged children. Teams may have up to 20 players, and may be played as Tee Ball, coach-pitched, or player-pitched.

Equipment

Sports wheelchair, baseball, glove

Super Sport: Upper extremity prosthesis designed for ball sports (37)

Unihoc hockey sticks and balls

Resources

Miracle League: www.miracleleague.com

Push N Power Baseball rules: http://www.geocities.com/CollegePark/Lab/5515/BASEBALL.html

Figure 5.4 Minor adaptations allow participation in bow sports.

Little League: http://www.littleleague.org/Learn_More/About_Our_Organization/divisions/challenger.htm

Basketball

Basketball may be played either as an ambulatory or a wheelchair sport. Teams of five play on a regulation basketball court following National Collegiate Athletic Association (NCAA) rules, with only slight modifications to accommodate the wheelchairs. The National Wheelchair Basketball Association (NWBA) uses a classification point system during competition. A junior program was developed by NWBA with four divisions, each having different age requirements, ball sizes, court measurements, time restrictions, and basket heights. It is a popular sport spanning all disabilities. Adapted versions with no contact, no running, no dribbling, and/or lower baskets are useful for developing skills (36).

Equipment

Sports wheelchair, basketball

Super Sport: Upper extremity prosthesis designed for ball sports

Resources

National Wheelchair Basketball Association: www.nwba.org

International Wheelchair Basketball Federation: http://iwbf.org

Bowling

Recreational bowling may include the use of standard lanes with gutter guards (bumpers) and the use of lighter-weight balls. Rules for competitive bowling may be divided into three divisions: AWBA, Special Olympics, and USCPAA. Lane measurements, rules, and bowling balls are the same as in the AB population under the AWBA. However, assistive devices, such as a handle ball, bowling stick, and bowling prosthesis, are allowed. Under the Special Olympics, target bowl and frame bowl are also allowed. Target bowl uses regulation pins, a two-pound bowling ball, and a carpeted lane that is half the regulation length. Frame bowl uses plastic pins and ball and a shortened lane. Under the USCPAA, there are four divisions with a ramp or chute allowed. Other rules follow the AWBA recommendations.

Equipment

Handle ball: A bowling ball with a spring-loaded retractable handle for individuals with poor finger control.

Bowling stick: A two-pronged stick similar in appearance to a shuffleboard stick.

Bowling ramp/chute: A wooden or metal ramp from which bowlers can push the ball down using their hands, feet, or a head stick.

Bowling prosthesis: Attaches to a standard prosthetic wrist and fits into one of the holes of the bowling ball.

It has a release mechanism activated by stretch on the expansion sleeve.

Resources

American Wheelchair Bowling Association: http://awba.org

Cycling

Cycling is immensely popular as both a recreational and competitive activity. A variety of adaptations are possible to make cycling accessible to a whole range of abilities. Children's tricycles may have blocks, straps, or shoe holders attached to pedals. Backrests and harnesses can be added to the seat to aid in positioning and stability. Adult-sized tricycles can be similarly adapted (Fig. 5.5). Specialized terminal devices for upper limb prostheses make grasping handlebars easier, and both brakes can be controlled by one hand for safety. Recumbent cycles afford maximum trunk support for recreational use by those with poor balance as well as by AB riders. Arm-driven units, which attach to the front of a wheelchair frame, are available with as anywhere from 3 to 48 speeds. Finally, a variety of tandem cycles or tandem conversion kits are on the market. These range from simple tandems to hybrid hand and leg cycles that allow DA and AB to ride together.

Handcycles are arm-driven cycles with rowing or push–pull drives that assist individuals with lower limb impairment or absence. While used for recreation as well, competitive cycling is a rapidly growing sport. Handcycle races may be held in isolation or in combination with bicycling races. In 2004, handcycling was introduced as a Paralympic sport; triathlons that combine swimming, wheelchair racing, and handcycling are increasingly including junior competitors.

Resources

United States Handcycling Federation: http://www.ushandcycling.org

Adaptive Adventures: www.adaptiveadventures.org

World Handcycling: www.worldhandcycling.com

Football (American)

Rules for wheelchair football vary from league to league. There is one national competition, the Blister Bowl, which is held in California. There are six players per team, one of whom must be female or tetraplegic. The asphalt field measures 60 by 25 yards and is divided into 15-yard segments. Play follows NCAA rules and is similar to touch football, with players advancing the ball by running or passing. All players

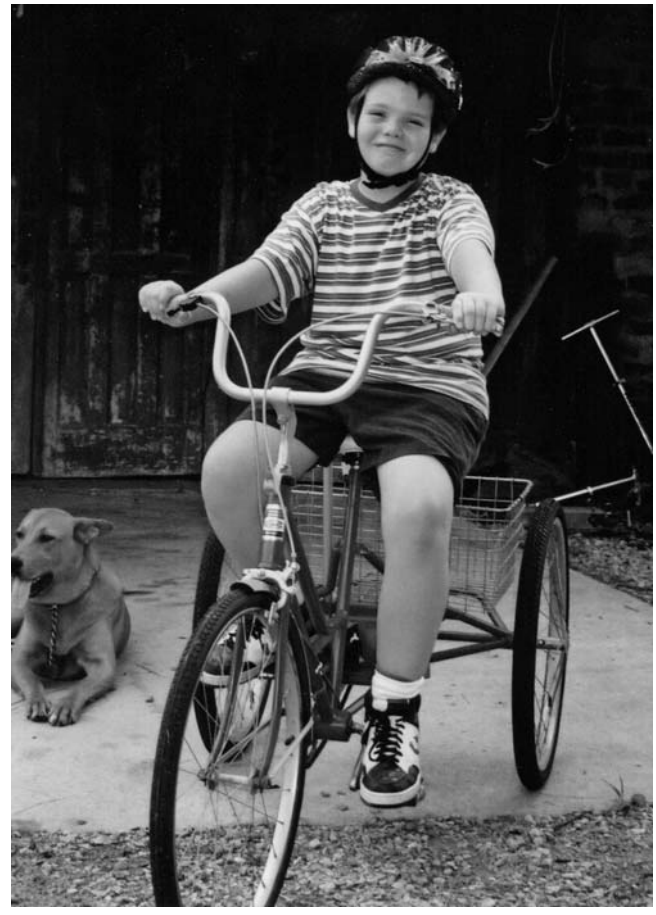

Figure 5.5 An adult-sized tricycle allows a disabled child to join on family rides.

are eligible receivers. Four 15-minute quarters are played. Participants primarily include individuals with amputations, CP, spinal cord injury, and les autres. Wheelchair football is not yet recognized as an "official" sport. The game also may be played on a basketball court indoors.

Equipment

Sports wheelchair, regulation football

Resources

Universal Wheelchair Football Association: http://www.rwc.uc.edu/kraimer/PAGE1.HTM

Hockey

Floor hockey is, in some respects, similar to ice hockey. It is played in a gymnasium with a minimum playing area of 12 × 24 meters and a goal at each end. Teams are composed of six players, who play three nine-minute periods. The puck is a felt disc, and hockey sticks are wood or fiberglass rods. Games may be

either ambulatory or played from wheelchairs. A similar sport, poly hockey, uses a hard plastic puck, a smaller plastic version of the conventional ice hockey stick, and a playing area measuring 12 × 24 meters at a maximum. Canada has further developed a version for power wheelchair users using a three-inch plastic ball rather than a puck and following National Hockey League (NHL) rules. Sledge hockey (sled hockey in the United States) is played on a regulation-sized ice rink using a standard puck or small ball and short sticks called pics. Players are seated on a sledge, which is an oval-shaped frame with two skatelike blades and a runner. Pics are used to propel as well as to advance the puck or ball (Fig. 5.6).

Equipment

Hockey sticks/pics, puck/ball, goals, helmet, knee pads, elbow pads, shin guards, sled

Resources

United States Sled Hockey Association: www.usahockey.com/ussha

Figure 5.6 Sled hockey is as fast-paced and thrilling as its ablebodied counterpart.

Quad Rugby

Quad rugby combines aspects of basketball, hockey, and soccer into an exciting sport developed for tetraplegic individuals. It is played with a volleyball on a regulation-size basketball court with goals at both ends measuring 8 × 1.75 meters. Teams consist of four players in manual wheelchairs, who play four eight-minute quarters. Players are classified from 0.5 to 3.5 in 0.5 increments, based on increasing arm function and trunk control. The combined point value of players on the floor may not exceed 8.0 at any time. The ball must be advanced over midcourt within 15 seconds of possession, and the ball must be bounced or passed within 10 seconds. A goal is scored when two of the player's wheels cross the goal line with the volleyball under control. Penalties may result in loss of possession or a trip to the penalty box, depending on the infraction.

Equipment

Volleyball, gloves, straps (trunk, legs, feet)

Quad rugby wheelchair: Must have antitippers

Resources

International Wheelchair Rugby Federation: www.iwrf.com

Canadian Wheelchair Sports Association: www.cwsa.ca

United States Quad Rugby Association: www.quadrugby.com

Racquetball

Racquetball may be either an ambulatory or a wheelchair sport. It is played on a regulation-size racquetball court and follows the rules of the American Amateur Racquetball Association. There are novice, intermediate, open, junior, two-bounce, and multiple-bounce divisions. It is recommended that players using wheelchairs equip their chairs with roller bars or wheels under the footrest and with nonmarking tires. Racquetball is another of the sports in which DA and AB players can play side-by-side.

Equipment

Standard racquet: A built-up grip or wrapping the handle to the player's hand may be required for those with grip difficulties.

Standard balls, lightweight sports wheelchair

Resources

USA Racquetball: www.usra.org

Road Racing

As running has increased in popularity as a recreational and competitive sport, DA athletes have formed their own running clubs and begun to participate in a variety of road races. Training is usually done on the road or a track. For the wheelchair road racer, rollers are also available. The racing chair is placed on the rollers allowing for free-wheeling and training indoors. The rules for road racing are no different between the AB and DA populations: Whoever crosses the finish line first, wins. DA athletes are placed in functional classes to make the competition more equitable. Power wheelchairs are not permitted in competition. Distances range from the one-mile fun runs to full marathons. Many of the well-known AB marathons now include one or more wheelchair divisions. The longest wheelchair race to date is the Midnite Sun Wheelchair Marathon, which covers 367 miles from Fairbanks to Anchorage, Alaska.

Equipment

Sports wheelchair: Customized racing wheelchairs are available for serious athletes; three-wheelers are most popular.

Gloves

Resources

DS/USA: www.dsusa.org

Cerebral Palsy International Sports and Recreation Association: www.cpisra.org

BlazeSports www.blazesports.org

Wheelchair Sports USA: www.wsusa.org

Adaptive Adventures—www.adaptiveadventures.org

Skiing: Alpine

In the past 30 years, adaptive skiing has grown immensely in popularity. With the advances in adaptive equipment, all disability groups can participate in this sport. Skiing techniques include three-track, four-track, and sit skiing. Three-trackers use one ski and two outriggers, thus creating three tracks in the snow. Outriggers are essentially modified Lofstrand crutches with short skis attached with a hinge. They provide additional balance and steering maneuverability. Single-leg amputees and individuals with hemiplegia are often three-trackers. Four-trackers use two skis and two outriggers. In those with spasticity or poor leg control, a ski bra can be attached to the ski tips. This will prevent the ski tips from crossing. Individuals with muscular dystrophy, spina bifida, paraplegia, and CP typically use four-track skiing. Sit skiing utilizes a mono-ski or bi-ski and two outriggers. All disability groups can sit ski. A tether, which allows the instructor to slow the skier down, is required until the sit ski is mastered. Tethers can also be beneficial during instruction in the ambulatory population. Competitive racing includes slalom and downhill courses.

Equipment

Outriggers, skis, ski bra, ski boots

Ski hand/All-Terrain Ski Terminal Device: specialized terminal device for upper limb amputees

Ski leg: A variety of ski-specific lower extremity prostheses are available.

Resources

United States Ski and Snowboard Association: www.ussa.org

U.S. Ski Team: www.usskiteam.com

Ski Central: http://skicentral.com

Sitski: www.sitski.com

Skiing: Nordic

Standing skiers can often participate in Nordic (cross country) skiing with standard equipment, sometimes modified to accommodate prostheses or braces. Sit skis are also available as in alpine skiing, although the ability of the participant to self-propel is often limited by the weight of the equipment. Tethers may be used to assist in forward movement. Biathlon is a sport consisting of cross-country skiing and target shooting.

Equipment

Outriggers, skis, ski boots, sit ski

Ski hand/All-Terrain Ski Terminal Device: specialized terminal device for upper limb amputees

Ski leg: A variety of ski-specific lower extremity prostheses are available.

Resources

United States Ski and Snowboard Association: http://www.ussa.org

U.S. Ski Team: www.usskiteam.com

Soccer

There are very few modifications to the actual game, and the rules of the United States Soccer Federation

are followed. The modifications include seven players on a team, a smaller field measuring 80 × 60 meters, and occasionally, a smaller goal. These modifications result from fewer participants in a given area. A smaller goal is indicated in the CP population in whom mobility impairments make a larger goal more difficult to defend. Crutches have been allowed for some competitors with lower extremity amputations who do not use a prosthesis (Fig. 5.7).

Equipment

Regulation-size soccer ball

Super Sport: Upper extremity prosthesis designed specifically for ball handling

Resources

American Amputee Soccer Association: www.ampsoccer.org

Top Soccer: www.usyouthsoccer.org/programs/TOPSoccer.asp

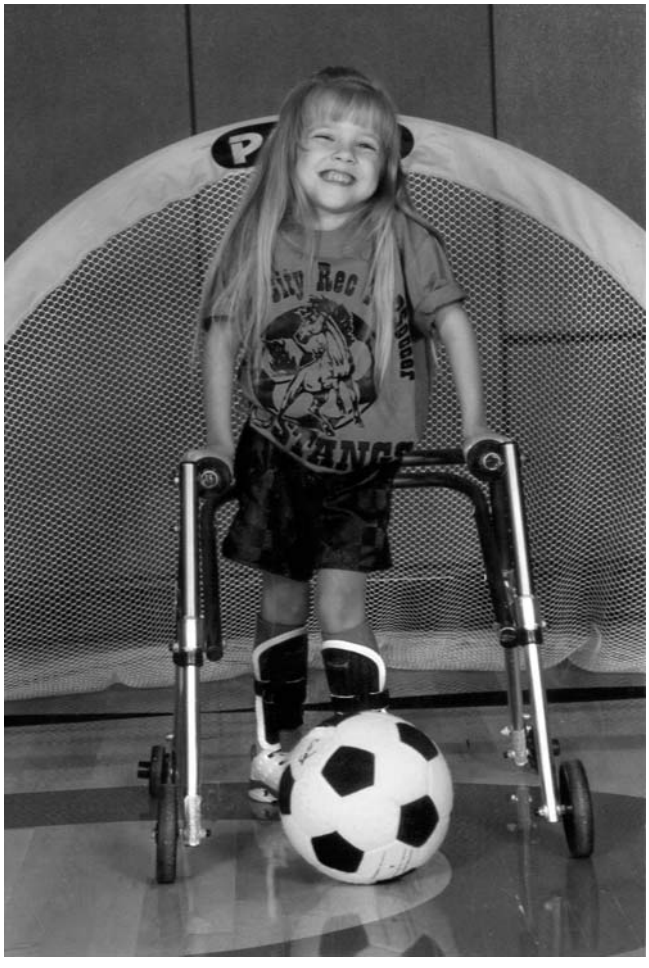

Figure 5.7 Soccer can be played by ambulatory children with gait aids, or by power-wheelchair users utilizing larger balls at indoor facilities.

Softball

Dwarf softball is played according to the rules of the Amateur Softball Association without any modifications. The Special Olympics offers a variety of competitive events, including slow-pitch softball and tee-ball. Wheelchair softball is also available primarily for individuals with spinal cord injuries, amputations, CP, or les autres conditions. It is played on a hard surface with the pitching strip 28 feet from home base and other bases 50 feet apart. Players must use a wheelchair with a foot platform and are not allowed to get out of their chairs. Ten players make up a team, and one of the players must be tetraplegic. The WS/USA point classification is used, and total team points on the field may not exceed 22. A larger ball is used, eliminating the need for a mitt, which would interfere with propelling.

Equipment

Softball, mitt

Prostheses: Upper extremity terminal devices that fit into a mitt or substitute for a mitt are available. A set of interlocking rings can also be attached to the bottom of a bat, allowing an adequate grip by a prosthetic hand.

Resources

National Wheelchair Softball Association: www.wheelchairsoftball.org

Swimming

Swimming is a universal sport in which all disability groups may participate. Numerous competitive events are offered across the United States. These include races of a variety of distances in freestyle, breast stroke, backstroke, butterfly, individual medley, freestyle relay, and medley relay. Classification systems have been developed by each DA sports organization to divide participants into classes based on impairment. In addition, swimmers are grouped according to gender and age. Flotation devices are often recommended, although only allowed in competition in two USCPAA classes. Flotation devices include tire tubes, inflatable collars, waist belts, life vests, head rings, water wings, and personal flotation devices. The use and choice of device is dependent on swimming ability, swimming style, and experience (Fig. 5.8).

Equipment

Flotation device, lift, or ramp

Prosthetics: Includes swim fins attaching to lower extremity prosthetic sockets and swimming hand prostheses. These are generally not allowed in sanctioned competition.

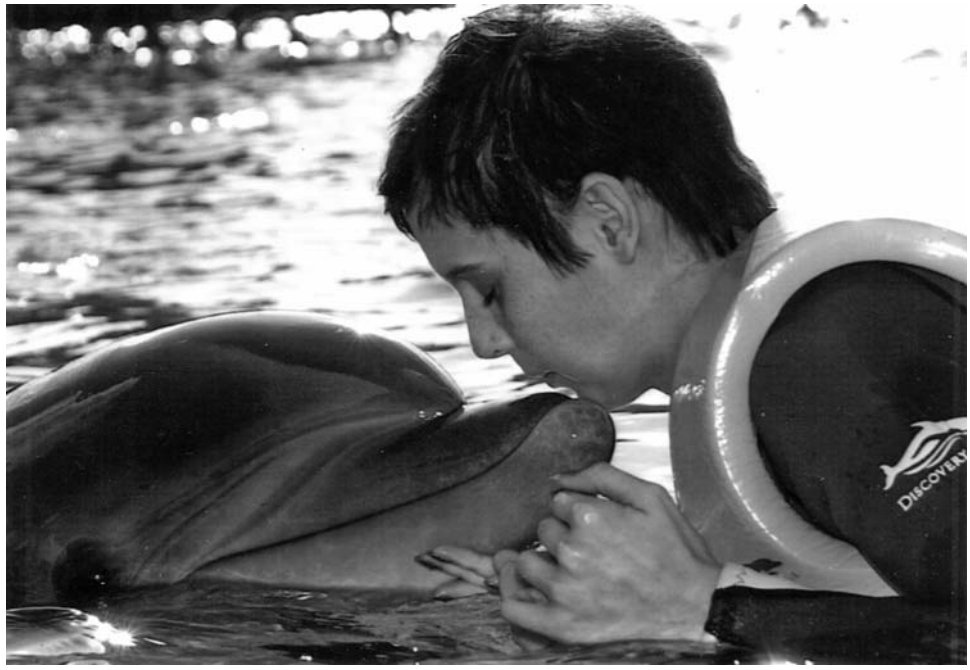

Figure 5.8 Water sports are made easier with flotation devices supporting weak limbs.

Resources

USA Swimming's Disability Swimming Committee: http://www.usaswimming.org

Table Tennis

Only slight modifications involving the delivery of the serve differentiate this sport from AB competition, which follows United States Table Tennis Association rules. The only equipment modifications allowed are to the paddle and, in the case of dwarf competition, floor raisers to make up for height differences. In recreational play, side guards may be added to the table to keep the ball in play longer.

Equipment

Velcro strap or cuffs: Allow correct placement of the paddle in the player's hand.

Regulation-size table, paddles, ball

Resources

U.S. Disabled Athletes Table Tennis Committee: http://www.midy.com/~usatt/parapong/

Tennis

Wheelchair tennis is played on a regulation-size tennis court as either a singles or doubles game. Players are allowed a maximum of two bounces before the ball must be returned. Scoring and other rules follow the United States Tennis Association guidelines. Players are broadly divided into two groups: paraplegic and tetraplegic. Within these divisions, players compete in subdivisions based on their skill. This sport is open to all disability groups. When a wheelchair user plays against an AB opponent, the rules of each one's sport applies to their respective side of the court.

Equipment

Sport wheelchair, tennis racquet, straps (trunk, legs, feet)

Racquet holder: Ace wrap or taping may provide additional support of grip strength if needed. Alternatively, a racquet holder orthosis may be beneficial.

Resources

United States Tennis Association: www.usta.com

International Tennis Federation: www.itftennis.com/wheelchair/

Track and Field

Track and field events are some of the most popular of the adapted sports competitions and involve individuals from all disability groups. Track events may be ambulatory or at the wheelchair level. Ambulatory and wheelchair events range in distance from 10 meters to a full marathon, and take place on

a typical track. Running, walking, and hurdles are all included in the ambulatory division. Power and manual wheelchair slalom races are available in the Special Olympics.

Field events typically include shot put, discus, javelin, long jump, and high jump. The USCPAA has also developed seven events for those athletes who are more physically impaired. These include the distance throw, soft discus, precision event, high toss, thrust kick, distance kick, and club throw. In the distance throw, athletes throw a soft shot as far as possible. The soft discus is similar to the conventional discus, except that the discus is made of a cloth material. For the precision event, six soft shots are thrown at a target, with points awarded for accuracy. The high toss involves throwing a soft shot over a progressively higher bar. Athletes have three attempts to clear the height. In the thrust kick, athletes kick a 6-pound medicine ball away from them, with their foot in constant contact with the ball. The distance kick is similar; however, it uses a 13-inch rubber ball and allows the athlete to initiate a back swing with the foot prior to striking the ball. For the club throw, an Indian club is thrown as far as possible.

Equipment

Racing gloves

Sport wheelchair: Custom-designed racing chairs are available for the serious athlete.

Throwing chair: Provides a stable platform from which athletes may throw

Resources

BlazeSports: www.blazesports.com

Wheelchair Sports USA: www.wsusa.org

Special Olympics: www.specialolympics.org

PEARLS OR PERILS

- Major barriers to participation for children and adolescents with disabilities include lack of transportation, financial constraints, and physical and attitudinal barriers. The presence of an adult assistant further distances disabled children from their able-bodied peers.
- Strengthening exercises in children with spasticity are not contraindicated, and often result in improved strength, aerobic capacity, and quality of life.
- While active weight-bearing exercises such as jumping result in increased bone density, the osteogenic benefits of passive weight-bearing are less clear.
- Access to technology such as video games and computers has resulted in a trend of lowered physical activity and increased obesity among AB and DA youth. However, use of active video games is resulting in increased levels of physical activity. This technology is also being implemented in habilitative and rehabilitative therapy programs.
- Sport and disability specific injury patterns are being recognized among disabled youth, leading to a new field of sports medicine for the disabled. Prescription of appropriate training and equipment are among the tools necessary for the pediatric rehabilitation professional.

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6

Orthotic and Assistive Devices

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Knowledge of orthotic and assistive devices is an important component of rehabilitation practice. Having an understanding of normal upper and lower body movement is fundamental for appropriate recommendation and fabrication of an orthosis. Likewise, clinicians' understanding of normal communication behaviors and language abilities is a prerequisite to the recommendation of an augmentative and alternative communication device.

An orthosis may be defined as any device, applied to the external surface of an extremity, that provides better positioning, immobilizes, prevents deformities, maintains correction, relieves pain, mobilizes joints, exercises parts, or assists or supports weakened or paralyzed parts (19). Orthotic devices may be classified as static or dynamic, depending on the functional need and ability of the extremity. A static orthosis is rigid and supports the affected area in a particular position, whereas a dynamic orthosis allows for some movement. They can be used to substitute for absent motor power, allow optimal function, assist motion, provide for an attachment of devices, and supply corrective forces to increase directional control (18). Several variations of upper and lower extremity orthoses are available that have been proven to increase function for the user.

Assistive technology includes "... products, devices or equipment, whether acquired commercially, modified or customized, that are used to maintain, increase or improve the functional capabilities of individuals with disabilities...," according to the definition proposed in the Assistive Technology Act of 1998. These may include specialize augmentative and alternative communication equipment, such as speech-generating devices, off-the-shelf computer mouse alternatives (such as a trackball), or software that provides special features. Some features that were first used primarily by people with motor and keyboarding disabilities are now included in standard computer operating software (such as speech recognition software).

The key to identifying the most appropriate orthosis or augmentative communication device is being creative and having a proper understanding of the anatomical, biomechanical, and communication needs of the patient and being sensitive to the patient's (or the parents') preferences and desires.

The pediatric population adds a further challenge. Early development is heavily based on fine and gross motor skills. Infants and children use these skills to explore and manipulate their environment. Studies have indicated that the inability to master the environment independently may lead to decreased socialization, learned helplessness, and a delay in normal development (1,5). Therefore, an orthosis should allow for and assist in the growth of the child.

Several team members are involved in prescribing, fabricating, and fitting the orthosis, augmentative communication system, or computer-access option. The physician, often with input from the therapist,

provides patient assessment and a prescription of the orthotic device (21). The therapist and/or orthotist are instrumental in its fabrication and fitting. A team including a speech-language pathologist, an occupational therapist, a special educator, and rehab engineering is often beneficial for augmentative and alternative communication device recommendations. Lastly, the patient and family play an important role in its acceptance and usage. If the device is cumbersome and difficult to manage, it will be rejected and find a home on the top shelf in the closet (3).

UPPER AND LOWER LIMB ORTHOSES

When choosing an orthosis, there are a few key principles to keep in mind. The orthosis should enhance normal movement while decreasing the presence of abnormal postures and tone. It should be simple, lightweight, durable, and strong. It should be easy for the child to use and maintain. Lastly, it should augment functional independence. An orthotic device is not successful unless it assists in improving a child's quality of life. A relatively new medical device in the rehabilitation field is manufactured by the Bioness Corporation. Their devices are neuromodulation products that are designed to service populations with multiple sclerosis, traumatic brain injury, cerebral vascular accident, spinal cord injury and cerebral palsy, and aid in their recovery. Their product can assist both the upper and lower extremity using stimulation to aid in regaining mobility and functional skills so that they can achieve optimal self-care independence, play, and/or work productivity. The upper limb orthosis addresses neurological impairments, while the lower extremity orthosis focuses on regaining the associated foot drop commonly seen in those clients with central nervous system disorders. Both upper and lower extremity orthoses use mild functional electrical stimulation (FES) to improve loss of function from injury associated with a central nervous system disability. The orthoses can be used in the clinic setting or at home. The overall goal of its use with the involved extremity is to reduce the spasticity, minimize the pain and discomfort during use, increase local blood circulation, prevent muscle atrophy, improve or maintain range of motion in the limb, and reeducate muscle use to enhance functional movement. (See the following Web site for additional information: http://www.bioness.com/Bioness_for_Hand_Rehab.php.)

Tables 6.1, 6.2, and 6.3 list some of the more common upper and lower extremity orthoses. Special considerations and limitations are also listed.

Shoe Inserts

Many orthopedic and neurologic pediatric disorders have sequelae that require orthotic management. Shoe inserts may be a viable option in many circumstances. There are many commercially available products to control differing levels of impairment in the hindfoot, midfoot, and forefoot. Heel cups help with shock absorption for joints, heel spurs, bursitis, and tendonitis. In the midfoot, orthoses assist to maintain the arch of the foot in varying degrees of firmness. Numerous products are also available to control disorders of the forefoot and toes. To relieve metatarsalgia, metatarsal bars are available to unload pressure from the metatarsal arch. Pads are available to help to realign hammer and claw toes, cushion bunions and calluses, and to protect toes from friction and irritation. A limitation to these commercially available products is that many times they do not come in pediatric sizes and must be modified to fit.

Orthoses for Positioning, Range of Motion, and Healing

Due to immobility, spasticity, and/or abnormal postures, many children are at risk for joint contractures, musculoskeletal deformity, and skin breakdown. Traditionally, caregivers have used pillows and towel rolls to maintain more appropriate postures. Bony areas such as the occiput, scapular spine, coccyx, femoral head, fibular head, and calcaneus are at greatest risk for skin breakdown from prolonged bedrest or maintenance of one position. Gel pads may be used to distribute weight over a larger area. The child may benefit from positioning pieces to maintain neutral positions and decrease pressure on parts of the body. Foam wedges in various lengths and sizes are commonly used for back support to position a child in side-lying. An abduction pillow may be used to decrease scissoring and increase hip abduction. Foam arm and leg elevators help to reduce edema, and foot splints/boots are available to maintain the foot in a dorsiflexed position with relief for the calcaneus to prevent pressure sores.

The Versa Form pillow is a semipermanent positioning support. These styrene bead bags are available in a variety of sizes and allow for molding to a child in any position. A vacuum pump is required to remove air from the pillow to make it firm. The bead bags need to be reformed after several weeks of use. This new technology gives the practitioner flexibility to change a child's positioning frequently.

Mobility Aids

Transfer Aids

There are a number of commercially available patient care lifts to assist caregivers and/or health care professionals

6.1 Upper Extremity Orthoses

UPPER EXTREMITY ORTHOSIS	COMMON NAME	FUNCTION (REF.)	SPECIAL CONSIDERATIONS (REF.)
STATIC			
Finger	Neoprene thumb abductor	Places thumb in abduction to promote functional use of the hand	Will not overcome severe cortical thumb position
	Static metal orthosis	Places thumb in abduction	Not recommended for fluctuating edema in the joint areas
Hand	Short opponens	Places thumb in abduction and rotated under the second metacarpal. Wrist and fingers are freely mobile.	Allows for full wrist flexion and extension. Should be worn at all times, removing only for hygiene and exercise.
Wrist-hand	Thumb spica	Immobilizes and protects the thumb, positioning it in opposition. Provides a stable post against which the index finger can pinch.	Need to allow for full MCP flexion of the fingers, especially the index finger, and full IP flexion of the thumb.
	Resting hand	Preserves a balance between extrinsic and intrinsic musculature and provides joint support when the hand is put at rest. Prevents deformity.	Should preserve the MCP joint descent and palmar arch following the contour of the distal palmar crease. Pressure at the MCP joint or proximal phalanx should be avoided, as this could cause injury to the MCP joint.
	Wrist cock-up	Supports, immobilizes, or stabilizes the wrist in extension. Increases mechanical advantage for grasp	Must maintain full MCP flexion and CMC motion of the thumb. Monitor the area over the styloid process for pressure changes if a dorsal splint is used.
	Anti-spasticity ball	Positions the wrist, abducts the fingers and thumb, and maintains the palmar arch in a reflex-inhibiting position	Should not to be used for minimal spasticity. (16)
Elbow	Elbow extension	Increases extensor range of motion and prevents flexion	Not recommended for severe flexor contracture or fluctuating tone in either flexor or extensor patterns
Elbow-wrist-hand	Full elbow/hand	Promotes supination at the forearm and provides a long stretch of the limb near end range to decrease tone	Not recommended for flexor tightness
Shoulder	Humeral orthosis	Stabilizes the shaft of the humerus circumferentially	May shift position if not appropriately anchored by straps
	Gunslinger	Supports the shoulder girdle and prevents shoulder subluxation	Make sure the edges around the base of the splint do not cut into the hip area. Check the fitting both in standing and supine positions to accommodate the shift of the splint.
Clavicle	Harness strap	Proximally stabilizes shoulder girdle movement and limits shoulder flexion and abduction movement beyond 90 degrees	Must mark settings for appropriate fit due to increased adjustability. Keep a check on skin integrity around the underarm area.

Continued

6.1 *Continued*

UPPER EXTREMITY ORTHOSIS	COMMON NAME	FUNCTION (REF.)	SPECIAL CONSIDERATIONS (REF.)
DYNAMIC			
Hand	MCP flexion assist splint	Gradually lengthens or gently stretches soft tissue structures that limit joint flexion	Ensure that the traction applied is gentle to guard against soft tissue hemorrhages around the joints, which can cause edema, pain, and increased scarring
	MCP extension assist splint	Passively pulls the proximal phalanx into extension while allowing active flexion	Do not position the proximal phalanx in either radial or ulnar deviation when using dynamic traction
	LMB Finger Spring—PIP extension assist	Gives dynamic traction of the PIP joint without limiting motion at the MCP joint. Assists in reducing tightness or contractures of the PIP joint.	Not recommended for severe spasticity
Elbow	Dynasplint	Brace adjusts to lock out undesired flexion and extension. Settings are adjusted in increments of 10 degrees.	Not recommended for severe spasticity
Power	Smart-WHO (wrist-hand orthosis)	Flexor-hinge hand orthosis that immobilizes the thumb in opposition and semiflexes the IP joints of the index and middle fingers to allow the index and middle fingers to move simultaneously toward the thumb. Variations include using an external power battery pack, SMA actuators, ratchet hand position, and shoulder driven cables (15).	Although design is lightweight and simple, a disadvantage can be the actuator's bulkiness as well as the unsightliness of the orthosis

CMC, carpometacarpal; IP, interphalangeal; MCP, metacarpophalangeal; PIP, proximal interphalangeal; SMO, supra-malleolar orthosis.

6.2 Lower Limb Orthoses

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Solid ankle foot orthosis	AFO, MAFO	Reduces tone, prevents joint contracture, and provides knee and ankle stability. Most appropriate for a child with severe tone, ankle joint hypermobility, and rigid deformities.	Does not allow any ankle movement and therefore limits smooth progression from heel strike to push off
Hinged or articulated ankle foot orthosis	HAFO	A hinged AFO with a plantarflexion stop and free motion into dorsiflexion allows the tibia to translate over the foot in stance. This orthosis allows the foot to dorsiflex for balance reactions and improves ambulation on uneven surfaces and stairs. Posteriorly, a dorsiflexion stop strap can be added to limit the amount of dorsiflexion. A plantarflexion stop in 2–5 degrees of dorsiflexion may assist to control genu recurvatum at the knee.	Does not control "crouched" posture allowing increased dorsiflexion and knee flexion. Children with strong extensor posturing may break the ankle joint. May allow hindfoot to slip, causing midfoot break if insufficient hindfoot dorsiflexion is present.

Continued

6.2 *Continued*

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Anterior floor reaction or ground reaction ankle foot orthosis	GRAFO	Limits a "crouch" posture (stance posture with hip flexion, knee flexion, and ankle dorsiflexion). At heel strike, it encourages a force up through the anterior cuff of this orthosis, giving the knee an extension torque. Knee extension is maintained throughout stance.	A child with significant hamstring or hindfoot tightness or tone will not benefit from this orthosis
Rear-entry hinged floor-reaction AFO		Dorsiflexion stop limits a "crouch" posture while allowing for plantarflexion during the loading phase of stance and at push-off	Active dorsiflexion is required to restrict foot drag during swing
Posterior leaf spring	PLS	The trimlines of this solid AFO are posterior to the malleoli. The slender posterior portion of this AFO gives it flexibility to allow for some dorsiflexion in stance and plantarflexion at push-off.	Does not allow full motion into dorsiflexion or plantarflexion. For medial-lateral ankle stability and arch control, another orthosis may be more appropriate. Does not control foot deformity or extensor tone. Excessive torque on spring may cause skin problems.
Dynamic ankle foot orthosis	DAFO	A supramalleolar orthosis that uses a footboard to support the arches of the foot. Provides medial-lateral ankle stability with control for pronation/supination. Allows some ankle dorsiflexion/plantarflexion.	Difficult to fit into shoes. Difficult for self-donning. Child may quickly outgrow this splint, since it is finely contoured to the foot.
Knee hyperextension splint		Maintains neutral knee and limits knee hyperextension. Uses three points of pressure: superior-anterior surface of the knee, inferior-anterior surface of the knee, and posterior to the knee joint (6,11).	Controls only the knee. Does not control extensor posturing well. It is bulky under clothes and difficult to sit with.
Swedish knee cage	KO	Controls genu recurvatum with the same three points of pressure a knee hyperextension splint and works the same. Uses metal uprights and straps instead of plastic material (7).	Controls only the knee. It is difficult to fit to smaller children, and it is difficult to maintain correct positioning.
Knee ankle foot orthosis	KAFO	Molded plastic upper and lower leg components, usually with a locked or unlocked hinged knee joint. Four most common knee locks are free, drop lock, bail lock, and dial lock. Free knee allows full motion at the knee axis. Knee axis may be straight or offset. Offset axis has an increased extensor moment at the knee joint. The drop lock is a metal collar that slides into place to maintain the knee in extension. The bail lock is a spring-loaded lock that has a trip mechanism to unlock the knee. The dial lock is a lock that may be set in varying degrees of flexion, used to accommodate or decrease a knee flexion contracture (13).	It is bulky and difficult to don/doff. Free knee at times allows too much motion. Drop lock requires fine motor control to lock and unlock. The child must be able to get the knee fully extended to engage the drop lock. Bail locks at times become easily disengaged. Dial locks do not allow free movement through the available range.

Continued

6.2 *Continued*

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Hip knee ankle foot orthosis	HKAFO	Hip belt and joint. Hip and knee joints may be locked or unlocked. Able to progress child to an increasing number of free joints at a time.	Bulky, difficult to don/doff. Difficult to manage clothing for toileting.
Reciprocating gait orthosis	RGO	HKAFOs that are connected by a cable system that links hip flexion on one side with hip extension on the other. This device assists children with active hip flexion and no hip extension to advance legs with a more normalized gait. Allows the child to ambulate with a reciprocal or swing-through gait (13).	Bulky, expensive. Difficult to don/doff. Not appropriate for a child with hip and/or knee flexion contractures. Difficult to manage clothing for toileting.
Hip spica/hip abduction splint		An orthosis made of thermoplastic material and Velcro to position a newborn's legs in abduction and flexion. This splint is used to maintain the femoral head in the acetabulum to mimic normal hip formation. Use of this splint helps to avoid hip subluxation and dislocation. Used from birth up to a year.	Requires frequent repositioning. May need frequent adjustments for growth. Difficult for caregivers to maintain appropriate fit.
Pavlick harness		A soft splint used for children with the diagnosis of congenital hip dislocation. This splint is generally used in the first 9 months of age. Bilateral lower limbs are positioned with hips abducted and flexed to 90 degrees in an attempt to maintain the hips in a reduced position.	Careful positioning required. Caregivers must be vigilant in checking splint positioning.
Parapodium or Variety Village Stander		This device allows the child to stand without upper extremity support, freeing bilateral arms to do activities. Walking with this device and crutches can be quicker than with the Parapodium with a swivel device (13).	More energy expenditure than with the swivel device. Children are unable to independently don/doff or to independently transfer supine to stand and stand to supine. Device is heavy.
Parapodium with ORLAU swivel modification		This orthosis allows the child to walk without use of a gait aid and to use the arms for other activities. Less energy expenditure than with a Parapodium and crutches (14,16).	Same as above. Slower than walking with Parapodium and crutches.
Twister cables		Cables are attached to a pelvic band and traverse the lower limbs to attach on shoes or AFOs. These cables provide control for increased internal rotation. Work well with children with normal to floppy tone to control internal rotation (13).	Do not work well with children with extensor spasticity. They may need to be frequently readjusted as the child grows.

6.3 Trunk Orthoses

TRUNK ORTHOSIS	COMMON NAME	FUNCTION	SPECIAL CONSIDERATIONS
STATIC			
Trunk	Thoracic-lumbosacral orthoses	This device is used to stabilize the spine after surgery, fractured vertebrae, or used therapeutically to provide the trunk with upright support during static or dynamic activities	Can affect respiratory function Brace will not correct spinal deformity, but may alter the progression of the curve Can cause pressure along axilliary line, breast bone, or ASIS area Monitor wear time secondary to heat intolerance
	Theratogs/Benik	This orthotic undergarment and strapping system gives users with sensorimotor impairment tactile positioning cues for improving postural alignment, postural and joint stability and movement, skill and precision	

ASIS, anterior superior illiac spine.

with performing safe transfers for children. The Trans-Aid and Hoyer lift are two examples of patient care lifts. They are designed to transfer children from bed to wheelchair, off the floor, onto a toilet, into a car, and through an 18-inch doorway. Slings are available with heavy-duty support options to further minimize the effort of the caregiver while maximizing safety during the transfer. There are also institutional lifters available, which offer a 400-pound and 600-pound weight capacity, as well as portable home-care lifts, which are lightweight, portable, and designed for home doorways and narrow halls.

Powered overhead transfer lift systems provide families with a unique transfer method. This system enables users to transfer from bed to wheelchair, toilet, or bath using a motorized lift and either manual or motorized lateral movement along a permanent ceiling-mounted track or a free-standing semipermanent rack. However, this transfer system is expensive and usually not covered by insurance.

In addition to patient lifts, there are other smaller devices that can assist children with ease of transfers. One option is a transfer board, while another is an overhead trapeze bar attached to an over-bed frame. The most commonly used transfer board is constructed of maple wood measuring approximately 8 inches wide by 24 inches long. It is ideal for all types of transfers (bed, car, bath bench, commode, etc.). Trapeze bars may be attached overhead to bed frames to assist the child with bed mobility skills and positioning changes. The position is individually set and can be altered as needed. Typically, trapeze bars assist with supine to sitting transfers and initiating rolling side to side. They are often appropriate for use initially, but are soon removed after the child's strength and bed mobility skills improve.

Standers

Numerous passive standing devices are available. These devices offer many potential benefits for the child, including the provision of a sustained muscular stretch, maintenance of trunk and lower limb passive range of motion, facilitation of co-contraction of muscles, decreasing tone, and improvement in trunk and head control. Standers should be used a couple of times a day for up to one hour total. The child should progressively work to increase tolerance in the standing position. However, passive standing should not take the place of the child exploring his or her environment and body.

Three types of standers will be discussed here: supine, prone, and upright. Supine standers go from a horizontal position to approximately 90 degrees upright, depending on the model chosen. Laterals, kneepads, adduction/abduction supports, and head supports all assist to maintain the child's posture while in this stander. Bilateral upper extremity strengthening can be performed in this position, with or without a tray.

However, it does not provide for any upper extremity weight bearing. A further limitation is that it will not work to improve head and trunk control. This stander is recommended for a child with significant extensor tone and posturing and/or a child with poor or absent head control. It is also preferred over the prone stander for the larger child due to the increased ease in positioning.

Prone standers support the child anteriorly. Postural support is supplied through trunk laterals, hip guides, abductor blocks, knee blocks, and shoe holders. These standing devices do come with a chin support to aid children who have limited head control or fatigue easily. However, the child should not be permitted to "hang" on this support; a supine stander is more appropriate if the child lacks fair head control. The stander can be used to improve antigravity head control and promote bilateral upper extremity weight bearing. Its tray may serve as a functional surface for stimulation. This stander may not be appropriate for some children with increased extensor tone. In these cases, gravity increases the work required for neck and trunk extension as well as shoulder retraction, thus feeding into primitive posturing.

Upright standers, such as the Evolv by EasyStand (Fig. 6.1) maintain the child in an erect position through supports at the hips, knees, and trunk. Certain standers are available with a hydraulic or manual lift, making positioning of the larger child easier. This stander mimics a normal standing position and permits the child to work on head control and upper extremity strengthening. The seat swings to the side for ease of transfer.

Figure 6.1 EasyStand Evolv.

Gait Aids

Gait aids are assistive devices designed to improve functional independence and/or expand exercise options through standing and walking. In pediatrics, gait aids assist children to explore and interact with their environment. Improved balance, decreased energy expenditure, decreased impact on joints, improved posture, and decreased pain are all potential benefits of gait aid usage. The most common gait aids are canes, crutches, and walkers.

Canes are available in different sizes with a variety of handles and supports (ie, straight cane versus quad cane). A quad cane provides a better base of support, but a normal gait cycle is more easily mimicked using a straight cane. A hemicane is a combination of a cane and a walker. It has a four-point base and the largest base of support of all the canes. It gives the greatest amount of stability among the canes, but also encourages the child to lean laterally when ambulating.

Crutches generally fall into two categories: axillary and Lofstrand. Axillary crutches are usually constructed of wood or aluminum and have limited adaptability. Some crutches may be modified to offer a forearm support to decrease weight bearing through wrists and hands. The child and family should be cautioned about possible nerve impingement from sustained axillary pressure with improper use. A "Kenney" crutch, not often used in the rehabilitation setting, is an axillary crutch without an underarm support. In place of the underarm support is a leather armband that fits around a child's arm.

Lofstrand crutches are much more flexible. They have a variety of forearm cuff styles, including circumferential or half cuff. Functional independence is increased with the use of Lofstrand crutches because the child is able to reach with his or her hands and the circumferential cuff will stay on the forearm. Half cuffs require less reliance on the cuff for balance, but they will not stay on the forearm if the handgrip is released. Handles may be wide and flat, pistol, or rounded. Rounded handles are the most commonly prescribed. The flat, wide handles may be helpful with tonal issues as well as with carpal tunnel inflammation. Pistol grips provide grooves for finger placement. Newer varieties of Lofstrand crutches are lightweight for children who have limited strength or need shock absorption for their joints.

Another adjustable option for all crutches is the crutch tip. Crutch tips may be constructed with materials of various flexibilities and in different widths to make the crutches more stable. Tips may include a gel, providing some shock-absorbent qualities. In addition, studded cups, which cover crutch tips, are available to make ambulation in rain and snow easier.

Three varieties of walkers are appropriate for the pediatric population: forward, reverse, and gait trainers. Forward walkers are the traditional type of walker. They can be purchased with or without wheels. Children can grip flat handles or use platforms on one or both sides to weight bear through the elbows and forearms. It should be remembered that forward walkers promote trunk flexion in many children.

Reverse walkers, also called posture control walkers, promote an erect posture. The child has increased extension at the trunk and hips when his or her hands are positioned to the sides or slightly in front. A pelvic support can be added to assist with lateral pelvic control and to facilitate trunk extension. Platforms can also be attached to allow forearm weight bearing. These walkers are widely used in the pediatric population. However, due to increased width, adult-sized children may have difficulty with accessibility. Other accessories available with some walkers are swivel wheels, forearm attachments, hip guides, hand brakes, baskets, and seats.

Gait trainers make ambulation a viable option for children who are unable to ambulate with other aids. Intensive body weight–supported treadmill training may be an effective intervention for some children with cerebral palsy who are ambulatory (14). A gait trainer is an assistive device that provides significant trunk and pelvic support (Fig. 6.2). It consists of a metal frame with adjustable-height metal uprights that support the trunk and arms. Adjustable-height seats, which are either slings or a bicycle-type seat, are attached. The seat is not used to support the entire body weight, but rather to keep the child erect. This gait device has been used to teach a more normal reciprocal gait pattern. It may function as a stepping stone to walking with a walker or crutches. Some limitations of gait trainers include decreased transportability, difficulty with positioning, and decreased accessibility. They are wider and longer than traditional walkers are. Gait trainers do have a place in therapeutic rehabilitation—to provide a child with independent means of ambulation when no other assistive device is appropriate and as a therapeutic tool toward ambulation with a more accessible assistive device. Accessories available with gait trainers are trays, wheel locks, harnesses, forearm supports, and differing lower extremity supports.

Figure 6.2 Rifton gait trainer.

For facility use, weight bearing and ambulation aids are available. The Lite Gait is a partial weight bearing gait therapy device. It allows the therapist to control the amount of weight bearing by supporting the patient in a harness system over a floor treadmill. With other therapeutic modalities, this has been shown to improve ambulation and endurance levels (18).

The EVA Walker is a heavy duty walker that has a manual or hydraulic lift that is easily adjustable for a variety of patients. It allows for significant upper extremity weight bearing to assist and improve ambulation for more moderately dependent patients.

Wheelchairs and Seating

The degree of limitation in mobility varies across a broad range for people with physical disabilities (4). Over the years, technology related to wheelchair seating and mobility has enhanced the opportunities for people with disabilities. Many more options exist to match technology with the user than ever before.

In order to begin the process of matching the child's needs to a particular wheelchair, it is recommended that a thorough evaluation be made. Many factors contribute to deciding on a particular seating and mobility system for the pediatric population. These include growth, specific disability, medical interventions, and prognosis of future functional and cognitive abilities. Assess the particular needs of the child, collect medical and surgical history, and perform a physical assessment. A multidisciplinary team approach usually works best. Once the assessment is performed, educate the family on various wheelchairs relative to the child's goals. If possible, simulate the child in as close to the recommended equipment as possible. Finally, determine the particular seating objectives for the child as well as the type of mobility base (22).

Every child has a unique set of challenges that will dictate how his or her rehabilitation needs will be met. Proper seating provides stability and support,

decreases the likelihood of postural deformities, and enhances upper extremity control. Within a wheelchair seating system, maintaining proper body alignment is achieved by using various seating and positioning components (23). Seating systems, including both the seat and the back, can be linear, contoured, or molded. Of the three, linear seating systems provide adjustability that allows the seating system to grow as the child grows. Linear seating systems are the least conforming to a person's body, but they are the easiest to fabricate and most adaptable as the person's orthopedic needs change. The basic materials consist of plywood for the base, foam (which can vary in density) for comfort and pressure relief, and a covering, usually Lycra, Rubatex or Dartex. Positioners such as laterals, abductors, and adductors are easy to mount on these systems.

Contour systems, in contrast to linear systems, conform closer to the actual shape of one's body. When recommending a contour system, close attention should be given to the growth rate and potential medical interventions, as the shape of the contour may not be an appropriate choice. Custom molded systems provide maximal support and should be considered for children with fixed deformities. Molded systems do not change as the child grows, unless remolding is performed, which is potentially time-consuming and costly. Although this system aids in controlling tone and nicely contours to most deformities, it has the reverse effect of limiting the amount of freedom children have in their seating system.

For patients who lack sensation, a variety of cushions exist that assist in alleviating pressure, which will decrease the likelihood of skin breakdown. Cushions fall under several categories, including foam, gel, air, and water (Table 6.4). Cushions should provide pressure relief under bony prominences, provide a stable support surface for the pelvis and the thighs, and function effectively in different climates. They should be lightweight, especially if a person is transferring independently or is a self-propeller, and be durable. Each type of cushion has advantages and disadvantages.

Pressure mapping systems are tools used by clinicians to measure interface pressures between two surfaces, such as a seated person and the cushion he or she is sitting on. (See an example of a pressure mapping system by Vista Medical at http://www.pressuremapping.com/.) A visual output on a computer monitor allows easy viewing and understanding. Using this tool allows clinicians to "diagnose" potential causes of skin ulcers as well as to select a cushion that will provide the most appropriate pressure relief for that patient.

6.4 Cushion Types

FOAM	GEL	AIR
Lightweight	Lightweight	Provides extremely good pressure relief
Provides a stable base of support	Provides a stable base of support	Lightweight
Various densities available that can improve pressure-relieving qualities	Various densities available that can improve pressure-relieving qualities	Can be unstable
Heavy	Heavy	Requires careful monitoring and maintenance
Conforms to individual shape	Conforms to individual shape	

Positioning Components

Within a wheelchair seating system, maintaining proper body alignment is achieved by using various positioning components. Evidence supports that children with cerebral palsy should be fitted for wheelchairs that place them in a functional sitting position (17). Lateral supports can be used to encourage midline trunk position when trunk control is poor. They may also be used to partially correct or delay the progression of scoliosis. Chest harnesses assist in stabilizing the trunk by anterior support as well as by preventing forward trunk flexion.

Positioning belts are used for pelvic alignment and stabilization. An improperly placed pelvic positioner is more detrimental than no positioner at all. The standard angulation of a pelvic positioning belt is at a 45-degree angle to the sitting surface (4). Subasis bars are used primarily for high-tone patients. Proper placement and position of the bar is critical to the success of the product. Improper positioning can potentially lead to skin breakdown.

Additional positioners include abductor pads that reduce or prevent increased adduction and assist in providing proper leg alignment. It should be remembered that abductors are not to be used to block a child from "sliding" out of the wheelchair. This may cause injury to the perineal area (1). Adductors decrease hip abduction and assist in providing proper leg alignment. Shoe holders and ankle positioners help control increased extension or spasms in the lower limbs and correct or prevent excessive internal or external foot rotation.

Head position is important for many reasons, including proper visual input, control of tone, and proper alignment for feeding and swallowing. Headrests provide support and positioning for a patient with poor head control due to low tone, active flexion, or hyperextension. They provide posterior and, if necessary,

lateral support. They also furnish safety in transport. The size and shape of the headrest depend on individual needs. Total head support can be achieved with the same headrest that allows the child to freely move his or her head to explore his or her environment.

When proper seating and positioning components are in place, pediatric wheelchairs provide users with the opportunity to explore and experience the world around them. It encourages social integration as well as enhances the level of involvement in various school and home activities. The majority of wheelchairs can be divided into two main categories: dependent mobility and independent mobility. These categories represent the level of functional mobility the child can achieve. Strollers, recliner wheelchairs, and tilt-in-space wheelchairs typically make up the types of chairs recommended for people who need a temporary means of mobility or who are incapable of independent mobility. Tilt-in-space chairs, such as the Quickie IRIS (see Web site for additional information: http://www.sunrisemedical.com), are recommended for people who need moderate to maximum positioning when there is little tolerance for an upright position. A reduction of pressure readings at the ischial tuberosities with tilt and recline positioning was shown as a general trend in a study by Pellow (15. Tiltin-space chairs provide pressure relief by redistributing body weight. The tilt also can assist the caregiver in properly positioning the child in the wheelchair by allowing gravity to assist. Positioning strollers, such as the KidKart Xpress and the KIMBA (Fig. 6.3) are typically used for younger children in whom independent mobility is less of an issue. Most strollers are also easily transportable.

Independent mobility can be achieved by using a manual wheelchair or a power wheelchair. Functional abilities and mobility goals dictate the type of wheelchair recommended. Manual wheelchairs can range from providing minimal support to complete postural support. Manual wheelchairs are lightweight in nature and have a multitude of features that can be adjusted or added to enhance efficient and effective use. Table 6.5 offers a comparative look at the various wheelchair components. Although this is a list of manual wheelchair components, many features can be considered for power wheelchairs as well.

Power wheelchairs provide independent mobility when manual wheelchairs cannot be used. Independent mobility is believed to be essential for perceptual-motor and social skill development. Self-produced locomotion also is believed to have an impact on cognition, communication, and psychosocial development (11). Technological advances in electronics have enabled people with severe physical disability to operate a motorized wheelchair. Power wheelchairs can incorporate unique features that enhance function critical

Figure 6.3 OttoBock KIMBA.

to health maintenance, as well as social development. The children who received power mobility had significantly greater improvement in receptive language on the Beck Depression Inventory (BDI) and in social-function functional skills and self-care caregiver assistance on the Pediatric Evaluation of Disability Inventory (PEDI) than the children who did not use power mobility (12). Power wheelchairs have pediatric sizes that are capable of raising the child from a seated to a standing position (for an example, see the Permobil Web site at http://www.permobilus.com), as well as elevating in the seated position using a "seat elevator."

Some power wheelchairs lower to floor level to allow the child to socially interact with peers. However, there may be constraints to using a power wheelchair. The family may not have the means to transport the wheelchair, or the power wheelchair cannot be used in the home due to limited physical space and accessibility. Funding may also prohibit the ability to acquire a power wheelchair. Another option for powered mobility for children may lie in three- or four-wheeled scooters. Scooters are usually less expensive than a power wheelchair, but do not offer a great deal of positioning options. Although choices are limited for pediatric-sized scooters, several do exist that can accommodate small children.

6.5 Wheelchair Characteristics

FRAMES

Rigid
(+) Efficient ride
(+) Durable
(+) Lightweight
(–) Decreased shock absorption

Folding
(+) Shock absorption
(+) Ease of transport
(+) Ability to narrow chair
(–) Less efficient propulsion

Hemi-Height
(+) Allows LE propulsion
(+) May make transfer easier
(+) Optimal height for peer interaction
(–) May make transfers difficult
(–) Compromise height at tables

Tilt in Space
(+) Pressure relief
(+) May assist to help balance +/or head control
(+) Change position for respiration
(–) Heavy
(–) Difficult to break down

Recliner
(+) Pressure relief
(+) Seating for hip contractures
(+) Limited tolerance for upright posture
(+) Ease of breathing/ feeding
(–) Difficulty changing position with spasticity
(–) Laterals and headrest move with changing position

One-Arm Drive
(+) One functional UE
(+) Sometimes difficult to manipulate

ARMRESTS

Conventional
(+) Offers protection
(–) Heavy
(–) Hand function
(–) Cosmesis

Height Adjustable
(+) Positioning assist
(+) Offers protection
(+) Ease of transfers
(–) Bulky

Flip-Up
(+) Hand function varies
(+) Remains attached for quick availability
(–) May be in bad position

Swing-Away
(+) Durable
(+) Cosmesis
(+) Easiest to operate
(+) Can change width via cushion
(–) No protection
(–) Must order side guards for protection

Arm Troughs
(+) Alignment of UEs with minimal AROM
(–) Bulky

FOOTRESTS

Hanger Angle

60 degrees
(+) Able to have large casters
(+) Limited ROM
(+) Increase depth without length
(+) Taller person
(–) Increased length of chair

70 degrees
(+) Reduces spasticity problems
(+) Compromise

90 degrees
(+) Reduces turning radius
(+) Reduces chair length

Types

Tapered
(+) Increased accessibility
(+) Positioning
(–) Decreased calf space

Standard
(+) Adequate calf space
(–) Decreased accessibility

Elevating
(+) Positioning—contractures
(+) Edema
(–) Increased chair weight
(–) Increased length
(–) Decreased accessibility
(–) Elevating mechanism
(–) Cumbersome

Continued

6.5 *Continued*

FOOTPLATES

Solid/Platform
(+) Folding frame more stable (+) Durable (–) Must remove to fold on folding frame

FRONT RIGGING

Angle Adjustable	**Hemi Mount**	**Flip-Up**	**Fixed**	**Swing-Away**
(+) Best positioning-ankle contractures (+) Reduce extensor thrust in lower limbs (–) Heavier	(+) Positioning for shorter legs	(+) Easier to move out of way (–) Not as durable	(+) More durable (+) Change seat depth without length (–) Transfers more difficult (–) Cannot reduce chair length	(+) Facilitate transfers (+) Greater accessibility (–) Must manipulate release mechanism

LEG STRAPS

Toe loop, heel loop, calf strap	Shoe holders
(+) Maintain feet on footplates (+) Straps maintain position even with flexor spasticity (+) Straps may be used for WC/floor/WC transfer (–) May make transfer difficult	(+) Control increased extension or spasms in lower limbs (+) Excessive internal, external rotation (+) Prevent aggressive behavior for safety (–) Heavy (–) Cumbersome

CASTERS

Solid	**Pneumatic**	**Semipneumatic**	**Size 6–8 Inches**	**Size 3–5 Inches**
(+) No maintenance (+) Least rolling resistance (+) Energy-efficient	(+) Most shock absorbent (+) Easier to maneuver over small objects	(+) No maintenance (+) A good compromise between solid and pneumatic	(+) Less rolling resistance (+) Increase footplate/ground clearance (+) Good on rough terrain (+) Tilt (+) Rugged terrain (+) Smoother ride	(+) Less shimmy (+) More responsive to quick turns (+) May aid in curb maneuverability (+) Increase footplate/caster clearance (+) Indoor use—tighter turns

AXLES

Axle position

Single Position	*Multiposition*	*Amputee*
(+) Durable (–) No adjustability	(+) Adjustability (–) Decreased durability	(+) Fits special population (–) Decreased durability

Axles

Standard	*Quick-Release*	*Quad-Release*
(+) Threaded (–) Cannot remove rear wheels	(+) Can remove rear wheels (+) Reduce size weight for transportability (–) Need good hand function (–) Durability	(+) Can remove rear wheels (+) Lower hand function (–) Durability (–) May accidentally disengage

Continued

6.5 *Continued*

REAR WHEELS

Spoked	Mag
(+) Shock absorption (+) Lighter (–) Maintenance	(+) No maintenance (+) Decreased chance of finger injury (–) Heavier

TIRES

Urethane	Pneumatic	Kevlar	Knobbie	High Pressure	Airless Inserts
(+) Good Indoors (+) No maintenance (+) Durable (–) Rougher ride (–) Heavier	(+) Rough terrain (+) Good traction (+) Lighter (–) Maintenance	(+) Reinforced tire	(+) All-terrain (+) Increased traction (+) Added flotation (–) Squeaks when new	(+) High pressure (+) Lighter (–) Need Presta valve	(+) Flat-free (+) Compromise (+) Low maintenance (–) 1 pound heavier than pneumatic tires

PUSH RIMS

Aluminum	Friction-Coated	Projection "Quad Knobs"
(+) No friction (+) Fine control (–) Cold in cold weather (–) Slippery if wet	(+) Impaired hand function (–) Chair width increased (–) Slippery if wet (–) Can cause burns (–) Coating wears away	(+) Angle varies (+) Length varies (+) Number varies (–) Angle increases width (–) Decreased efficiency if pegs do not end up in right position (–) Difficult to descend

BRAKES

Push to Lock	Pull to Lock	Scissors	Extensions	Grade Aids
(–) May hit transfer surface and unlock (–) May hit hand when propelling	(+) Not as likely to unlock during transfer (+) Closer for transfers (+) Clear for propulsion	(+) Clear for propulsion (+) Clear for transfers (–) Difficult to manipulate (–) Less surface contact with camber	(+) Easier to reach (+) Easier to operate (–) Decreased brake durability (–) In the way (–) Toggle	(+) Prevents chair from rolling backwards (–) Difficult to propel forward (–) May engage inadvertently (–) Requires treaded tire (–) Prevents recovery from backward fall (–) Low durability

AROM, active range of motion; LE, lower extremity; UE, upper extremity; WC, wheelchair.

Car Seats

Conventional restraint devices may not always be the option for safety in transportation (10). Alternative car seats can be purchased for children with special needs. There are two commonly used types of special needs car seats: the Britax Traveller Plus (for more information, see http://www.snugseat.com/) and the Columbia car seat. Both include seat depth extenders, adequate positioning pads, five-point safety straps, and an appropriate restraint system. The Carrie Car Seat comes complete with head support, harness and safety belt straps, and foot supports. At times, a child is sent home from the hospital in a spica cast or one that limits the fit in a safe manner for travel. The Hippo Car Seat is for transporting children with hip spica casts, broomstick casts, and Ilfeld splints. For those children whose postures require more than a lap belt and shoulder harness, an easy-on vest is recommended. It can be used in upright sitting in the rear seat or in side-lying in the back seat. Models can accommodate ages 2–12, depending on size and weight.

Children with tracheostomies should avoid using child restraint systems with a harness tray/shield combination or an armrest. Upon sudden impact, the child could fall forward and cause the tracheostomy to contact the shield or armrest, possibly resulting in injury and a blocked airway. Five-point harnesses should be used for children with tracheostomies (18).

Transporting wheelchair occupants can be a challenge for many, especially school bus supervisors. Research and accident data show that wheelchair tiedowns and occupant restraint systems (WTORS) can reduce the possibility of injury by preventing the wheelchair occupant's head from hitting the vehicle interior (20) Several commercially available systems exist that secure the wheelchair to the vehicle, including a four-point belt system, a "docking" station, and a "T" bar configuration. It is also recommended that wheelchairs face forward to avoid collapsibility should there be a collision. In addition to the wheelchair seatbelt and shoulder or chest harness, the standard lap and shoulder belt anchored to the vehicle or the restraint system should be used (2,3).

Adaptive Interfaces

With the level of human interaction incorporated into today's technology, interface between a device and the child takes on a new meaning and new challenges. The success of a device is determined by the interface, which takes the form of various technologies. Depending on the physical abilities of the user, the interface between the child and the product can look quite different. A significant proportion of severely disabled people need to use head movements to control assistive equipment such as speech-generating devices, environmental control systems, and powered wheelchairs (8). It could be a palatal orthosis, as in the Tongue-Touch Keypad, used to not only control a wheelchair, but also a computer or home and office devices. Other adaptive interfaces include "sip and puff" systems, chin control devices, and other various switches configured to provide a specific output, depending on the device to be controlled.

Other adaptive interfaces used typically to control one's environment or access to a computer include voice activation and an eye gaze system. Voice recognition may use software such as Dragon Naturally Speaking. Eye gaze technology continues to improve, as in the Tobii Eye Tracking system, a computer hardware and software package for explicitly measuring, recording, and analyzing what a person is doing with his or her eyes. The child can perform a broad variety of functions, including environmental control, playing games, typing, or operating a telephone. As the electronics have advanced, particularly in powered mobility systems, so has the ability to integrate controls. Therefore, it is possible to have a power wheelchair user also control his or her communication device using the same interface that allows him or her to control the power wheelchair. Although these technologies are sophisticated, they offer another means of accessing the environment and maximizing independence. The advantages of integrated control are that persons with limited motor control can access several devices with one access site without assistance, and the user does not need to learn a different operating mechanism for each device (7).

Recreational Equipment

An integral part of a child's life should be learning and self-exploration through recreational activities and play. Many tricycles now fit the needs of some physically challenged children. Special features include hand propulsion, wider seats, seatbelts, trunk supports, and chest straps. The Step-N-Go bicycle allows a rider to stand and pedal, making propulsion easier for children with extensor tone. The Rifton Adaptive Tricycle (see http://www.rifton.com/products/mobility/adaptivetricycles/index.html) provides the user with the ability to sit and pedal. This bike provides multiple positioning supports and the capability to grow. A "roller racer" is a riding toy for children with lower extremity dysfunction. It sits close to the ground and is propelled by moving the handlebars from side to side. Electronic cars can be adapted with switches or a proportional joystick. Scooters can be propelled with arms or legs. Many commercially available mobility devices are on the market today. Further information on recreational equipment is available in the adapted sports and recreation chapter.

AUGMENTATIVE AND ALTERNATIVE COMMUNICATION (AAC) AND COMPUTER ACCESS FOR LEARNING, WRITING, AND LIVING

All children, whether disabled or not, utilize a complex communication system that integrates spoken, written, and pragmatic social language skills. Augmentative and alternative communication (AAC) includes low- and high-technology devices that supplement these skills and facilitate language learning. Augmentative communication options are appropriate for any child whose natural speech and writing does not enable him or her to express himself or herself to all listeners in all environments and for all communication purposes. In addition, they are indicated when natural speech and writing does not sufficiently support continued speech, language, and academic learning and success. The cause of the communication impairment may be a motor speech disorder, such as dysarthria or dyspraxia; a cognitive and language disorder, such as global developmental delay, pervasive developmental disorder, autism, mental retardation, traumatic brain injury, cerebral palsy, or learning disabilities; or a neuromuscular disorder, such as muscular dystrophy or spinal cord injury.

Communication behaviors develop spontaneously in all children, regardless of the severity and multiplicity of their disabilities. Nonverbal communication behaviors may manifest as vocalizations for satisfaction and dissatisfaction; eye gaze and eye contact; looking away from a person, place, or thing; idiosyncratic gestures; and physically leading adults to desired objects and places. Even when such communication behaviors are more "reflexive" or self-directed than intentionally interactive, parents, caregivers, and familiar listeners typically learn to recognize communicative information from their children's behaviors.

The goal of AAC intervention includes introducing communication strategies that help the child develop systematic language and communication behaviors. Systematic communication helps listeners to more readily understand a child's communicative intent, helps to reduce the "20 questions" guesses that parents and caregivers typically engage in, and helps the child and his or her listeners form a communication dyad. With regards to the psychosocial development of children and adolescents with disabilities, the use of a speech-generating device and/or adapted access to computers may enable them to shift social and communication control of interactions from parents, teachers, and caregivers to the child—just as happens with typically developing children.

Not all augmentative communication devices need to be speech-generating. Low-tech aids can include communication notebooks, communication boards, and picture exchange communication displays. They may be even simpler, including no-tech systems, such as refrigerator magnets or homemade picture magnets displayed on the refrigerator or on a cookie sheet for portability.

While it is important that all communicators utilize their residual speech whenever functional, it is especially critical that AAC systems maximize the role of natural speech rather than replace it. Natural speech may be used primarily for initiation and getting attention, with a supplementary device used to communicate specific or complex information. Unaided natural speech may be one's primary communication technique, but supplemented by a speech amplifier or a speech-generating device in noisy environments (7).

Speech-Generating Devices

AAC devices that produce spoken language output are generically known as speech-generating devices. Speech-generating devices (SGDs) fall into different categories (Table 6.6), just as do other orthoses and aids. In the case of SGDs, the categories are based on device features and functions, and are categories that are used as Healthcare Common Procedure Coding System (HCPCS) codes for medical funding.

Recommending and prescribing the most appropriate, least costly, and medically necessary speech-generating device for an individual requires that the clinical evaluation team understands a child's oral speech abilities; language abilities and potentials; visual-motor control and device access skills and needs; and pragmatic language knowledge, skills, and needs with multiple communication partners throughout all domains of his or her life.

Language, Communication, and Literacy

Research into the needs of children with significant disabilities has highlighted the need to consider the role of literacy when evaluating and making recommendations that will enable these children to optimally function socially, educationally, and productively in their lives. "While research has provided ample evidence that individuals with even the most significant disabilities can learn to read and write, 70% to 90% lag significantly behind their peers in literacy learning. More generally, in excess of 20% of American adults read at or below a fifth grade level" (9).

Thus, many children need to augment whatever natural speech they can produce by using low-tech communication boards, manual language signs, or speech-generating devices, and go beyond concretely requesting their immediate wants (such as what they want to eat, when they want to go to the bathroom,

6.6 Speech-Generating Device Categories

CODE	DESCRIPTION
E2500	Speech-generating device, digitized speech, using prerecorded messages, less than or equal to 8 minutes recording time
E2502	Speech-generating device, digitized speech, using pre-recorded messages, greater than 8 minutes but less than or equal to 20 minutes recording time
E2504	Speech-generating device, digitized speech, using pre-recorded messages, greater than 20 minutes but less than or equal to 40 minutes recording time
E2506	Speech-generating device, digitized speech, using prerecorded messages, greater than 40 minutes recording time
E2508	Speech-generating device, synthesized speech, requiring message formulation by spelling and access by physical contact with the device
E2510	Speech-generating device, synthesized speech, permitting multiple methods of message formulation and multiple methods of device access
E2511	Speech-generating software program for personal computer or personal digital assistant

what toy they want to play with, or (when older) what song they want to listen to or DVD they want to watch). Functional communication in a social world requires that children have the ability to communicate for a full range of pragmatic language purposes (Table 6.7).

For instance, an AbleNet Little Step-By-Step Communicator (for more information, see the Web site at http://store.ablenetinc.com/) single-switch SGD that uses digitized, or recorded, speech output and is an E2500 device, can be programmed with a series of messages that enables a person to take active control of his or her personal care and to give caregivers a series of directions of what to do to help with personal care or activities-of-daily-living needs instead of being a passive and dependent recipient of such care. (It is communication control that differentiates a "caretaker," or somebody who takes care of an individual, from a "caregiver," or somebody who gives an individual the care he or she wants and requests.) For adolescents and others, this actually can be an issue of personal safety and privacy.

Similarly, AMDi's Tech/Talk and Tech/Speak are E2500 SGDs that can be programmed with multiple, interchangeable picture overlays and messages that a child can use to make requests, to tell which body parts are sources of pain or discomfort—or to take turns while singing "Hokey Pokey" with siblings, with recorded singing for lines like "Put your right foot in and shake it all about." To allow a nonspeaking child or adolescent to contribute to reading activities at home and in school, overlays can be programmed with pictures and lines of favorite storybooks to help parents read bedtime stories or to give a book report in class.

6.7 Pragmatic Language Functions

Requesting
Preferences and needs for objects and activities
Dislikes
Repetition and termination
Information (incl. about daily routines and schedules; who, what, when, where, etc.)

Giving Information
Personal information
Personal experiences
Active control for personal assistance and care
Replies to questions (who, what, when, where, etc.)

Literacy
Active participation in early literacy learning
Reading for language reception and information development
Reading for spoken output
Writing for written output
Writing for computer access

Social Closeness
Active participation in social interactions
Turn-taking and maintenance of social closeness
Singing and other performances

Social Routines
Politeness
Initiation of communication interactions
Topic initiation and maintenance

AMDi recently expanded the capabilities and memory capacities of their devices by producing its Tech/Smart series—in which an infinite number of overlays can be programmed and inserted into a device with the assistance of interchangeable smart memory cards (such as those used in digital cameras and other devices).

However, such devices are limited not only by their technical features, but also by the way in which their language needs to be programmed. As described in their HCPCS codes, these devices use pre-recorded whole messages and are organized into overlays that need to be physically changed in order to change communication topics or pragmatic language purposes. At the other end of the SGD technology spectrum are E2510 devices. Many of these utilize touch-sensitive screens and organize language through "dynamic display" technology. That is, the "pages" of vocabulary items change dynamically to give a person rapid access to a core vocabulary of the most frequently used words, phrases, and messages; slower access to an extended vocabulary of less commonly used words; and even to keyboards through which a user can spell any word and add it to messages that he or she is spontaneously generating.

E2510 SGDs come in all sizes, shapes, and weights. The smallest are housed in palm-size computers. Two of these include Saltillo's ChatPC (http://www.saltillo.com/products/index.php?product=32) and DynaVox's Palmtop 3 (http://www.dynavoxtech.com/products/palm3/). They can be extremely appropriate for children and adolescents who are ambulatory, have good visual-motor coordination or extremely limited fine motor range of motion or visual fields, and/or who have dynamic language needs about a variety of different topics or in a range of different settings.

All E2510 SGDs can utilize a range of symbols—from digital photographs to more symbolic pictures and multimeaning icons, to printed words and alphanumeric keyboards from which to select or build syntactical messages. Most utilize Universal Serial Bus (USB) drives to transfer digital photographs from digital cameras or computers into the memory of the SGD. Blink Twice's Tango! (Fig. 6.4) was the first to add a digital camera so that new photographs could be taken and new symbols and messages could be programmed wherever a person was and needed new vocabulary.

E2510 devices also come in moderate sizes and weights, such as the Prentke Romich Vantage Plus (Fig. 6.5) and the DynaVox V (Fig. 6.6). Some have larger screens and are heavier, and are appropriate for mounting on wheelchairs, such as the DynaVox Vmax.

SGDs that have the sophistication and features of E2510 devices have been developed to integrate multiple modes of communication, especially spoken and written language expression. All of the devices illustrated here include the ability to be connected to computers so that the same messages that can be sent to the device's built-in speech synthesizer also can be sent directly to the word processing program, e-mail program, or other text-based software on a computer. This enables a person who is unable to read or write orthographically written words to select the same pictured vocabulary items that he or she uses to select, formulate, and speak messages to write the same information on a computer screen and as a printed hard copy. Thus, a child in a typical second grade classroom can use the language and access system of his or her SGD in conjunction with a classroom computer to write daily personal journals and keep up with classmates who do the same activities through handwriting and spelling. What grandparent does not love to get letters written by a grandchild, instead of interpreted and transcribed by a parent? Since more communication (including with grandparents) is occurring through e-mail, connecting the SGD of a child or adolescent with severe disabilities to a computer and opening an e-mail program instead of word-processing software will enable him or her to compose and send e-mail independently and in his or her own words. Some devices, including the Vantage Plus and the DynaVox V, contain additional memory

Figure 6.4 Blink Twice's Tango!

Figure 6.5 Prentke Romich Vantage.

Figure 6.6 Dyna Vox V.

card slots as well as infrared transmitters in order to enable them to become truly multifunctional devices. For instance, young adults who take their SGDs into social situations with peers, who need to be able to contact family members in an emergency wherever they are, or who are preparing to go to college can use their SGDs as cell phones. MP3 cards can be inserted into many of these devices—not just so that children can listen to the music they want to, but so that their SGDs can contain podcasts of news stories, interviews with famous people, and audio books for pleasure and research reading.

Among the newest advances in SGDs in the E2510 category are the devices that are also full-capability Window XP computers—these can run any software that any computer can run. Among these are the DynaVox V and the DynaVox Vmax, Prentke Romich's ECO-14 (see the Web site http://www.prentrom.com/eco), and Tobii ATI's Mercury (http://www.tobiiati.com/corporate/products/merc.aspx).

Physical Access and Access Techniques

E2510 devices include features that permit "multiple methods of message formulation and multiple methods of device access." This means that users do not need well-controlled fine motor skills or functional visual skills. On-screen keyboards can be programmed with different numbers and sizes of keys on a device's touch-sensitive screen for people who have different visual-motor skills and capabilities. In addition, many devices can be programmed with different sizes of keys on different sections of the screen for people who have more controlled fine motor skills in some areas of their range of motion than in others. Many of these devices can also be accessed and controlled through "light pointing" so that head movement or controlled hand movements in space becomes one's access technique. Any other type of mouse or "mouse emulator" can be used as well—including multiple switches that may otherwise control wheelchair driving directionality through the same circuitry. These access modes are typically considered "direct selection" or "directed selection" techniques.

However, people with significant motor impairments can use just one switch, or perhaps two switches, to control all of the same functions of their SGDs—and ultimately their socialization; their face-to-face, written, and electronic communication; their independent participation in literacy activities; their computer use; and eventually their studies and productivity at work. These devices offer a wide range of switch-controlled scanning options. Auditory or spoken language cues for the user, as well as differentiated speech output for the user's listener, are available for people who also have functional visual impairments so that they can better follow scanning auditorily than visually. Consequently, a clinician who is knowledgeable about an individual's unique movement patterns, visual processing, and visual-motor coordination, as well as about the features of different SGDs, can find and customize a device setup to help a client's access be as timely, accurate, energy-efficient, and effective as possible.

Alternative Computer Access

Many of the same features and options that are included in SGD technology are available as computer access

tools for people with significant motor and/or cognitive disabilities but who do not require the assistance of speech-generating devices. These included adapted keyboards that offer either or smaller larger keys and ranges of motion.

The Big Keys Plus USB keyboard not only contains larger keys with more physical separation between the keys than a standard keyboard, but also arrow keys that function to control mouse movements as an alternative to having to move one's hand smoothly through space to control a standard mouse. The TASH USB Mini Keyboard's (Fig. 6.7) overall size is 7.25 by 4.2 inches. It is helpful for people who can use only one hand to type or who have a limited range of motion or field of vision. It also is used by people with a large degree of spasticity whose spastic movements increase as they have to complete larger movements—such as across the range of a standard, full-size computer keyboard.

On-screen keyboards offer efficient access options for people who are more proficient mouse users than keyboard users. Madentec's Screen Doors (see Web site for additional information http://www.madentec.com/products/screendoors.php) and Discover:Screen are two on-screen keyboard options that can be controlled by a mouse or "mouse emulator." These can also be used with a switch-controlled scanning technique rather than with a mouse.

People who have no functional hand control can even use head control to point mouse cursors with the use of a wireless head-pointing system such as Madentec's. The camera is mounted on a computer screen and tracks the person's head movement. It requires the user to wear a small, self-sticking reflective dot on which the camera focuses. The reflective dot can be affixed to the forehead, eyeglasses, cap brim, or sweatband.

Software products like Dragon Naturally Speaking are available to give people completely hands-free access to writing and to mouse control. Naturally Speaking represented a significant technical breakthrough, compared with earlier versions of "speech recognition" software, in that a person can speak at a natural speaking rate, rather than word by word. The computer processor, however, rarely keeps up with real-time speech, so that the fourth word of a sentence may just be showing up on the screen as one finishes dictating that sentence. A speaker also needs to consistently visually monitor the computer's recognition accuracy, since words like "civil" and "Seville" or "print" and "tint" may not have been articulated with sufficient discrimination that the computer enters the intended word.

People who use keyboards but enter words slowly due to either poor motor dexterity or poor spelling skills may find that word prediction software, such as

Figure 6.7 TASH USB Mini Keyboard.

Don Johnston, Inc.'s Co:Writer or QuillSoft's WordQ, can help them become more fluent writers. As one types, these programs display a dynamic list of predicted words based on frequency of use in English, recency of use in one's own writing, spelling completion, and grammar prediction models. Co:Writer also may be set to provide spoken feedback after each keystroke, each word, and/or each whole sentence to help those for whom visual monitoring of one's writing is physically effortful, slow, and/or fatiguing. Word prediction programs may enhance writing speed, efficiency, and fluency by saving numbers of keystrokes or by reducing the cognitive load and time required to spell individual words in a passage correctly.

The goal of all assistive communication technology—for speech output and face-to-face communication, for written language expression, and for electronic communication—is to help people with disabilities overcome these limitations and to become more independent and more efficient, to become faster and to experience more stamina, and to become active members of their social communities. Meeting this goal includes understanding an individual's unique profile of strengths and needs; understanding and demonstrating the growing number of assistive technology options in the marketplace; allowing clients and their families to express their preferences and desires following hands-on trial of appropriate options; and recommending the best match of device features to client skills, needs, and preferences. This process assures that people who require and use such technologies attain an optimally functional outcome.

Resources

Many organizations offer specialized information and resources related to AAC and assistive technologies.

Each offers a different perspective and a different assortment of assistance.

- The International Society for Augmentative and Alternative Communication (ISAAC) offers journals and newsletters as well as information about AAC in countries around the world. ISAAC may be contacted at ISAAC, 49 The Donway West, Suite 308, Toronto, Ontario, M3C 3M9, Canada, 416-385-0351, isaac_mail@mail.cepp.org
- The U.S. Society for Augmentative and Alternative Communication (USSAAC) is the national chapter of ISAAC. Its members include individuals from all professions involved with AAC, including manufacturers and researchers, as well as consumers and family members. USSAAC may be contacted at USSAAC, P.O. Box 5271, Evanston, IL 60204, 847-869-2122, ussaac@northshore.net
- The American Speech-Language-Hearing Association (ASHA) includes a Special Interest Division in AAC for speech and language pathologists. ASHA may be contacted at ASHA, 10801 Rockville Pike, Rockville, MD 20852, 301-897-5700, www.asha.org
- The Rehabilitation Engineering and Assistive Technology Society of North America (RESNA) is an interdisciplinary association for the advancement of rehabilitation and assistive technologies. It includes a special interest group in AAC. RESNA may be contacted at RESNA, 1700 North Moore Street, Suite 1540, Arlington, VA 22209, 703-524-6686, www.resna.org
- Every state has a special project devoted to AAC and assistive technology. These were originally established by federal funding through the Technology-Related Assistance Act. They are known as Tech Act Projects. Directories of them are available from organization such as USSAAC and RESNA.
- The Communication Aid Manufacturers Association (CAMA) offers packets of manufacturer catalogs and series of local workshops on AAC devices and their applications. CAMA may be contacted at CAMA, P.O. Box 1039, Evanston, IL 60204, 800-441-2262, cama@northshore.net
- These and other organizations offer a variety of conferences and publications. *Closing the Gap* offers both; CSUN is an annual assistive technology conference at California State University—Northbridge.

ASSISTIVE ORTHOSES AND ROBOTS

Automated Feeders

Task-specific devices exist for feeding—such as the Winsford feeder, sold through Sammons Preston for approximately $3,800 (Fig 6.8). This is a motorized device intended for people without available arm function. They activate a chin switch, which sends a signal to scoop up the food off a mechanized plate and present it to the user. The Handy 1 device is similar to the Winsford; however, it uses a commercially available robot that is controlled through switch operations (24). The movements are programmed to perform a selection of tasks, such as feeding, applying makeup, and shaving. The food is placed on a custom plate that has different compartments. A scanning system of lights designed into the tray section allows the user to select food from any part of the dish. For other tasks, the user selects similar programmed moves.

Figure 6.8 Winsford Feeder.

The Neater Eater (Neater Solutions, Buxton, UK) is a table-mounted feeding device that comes in two versions. The first is a motorized feeding arm that can be controlled by a user with little arm function, and retails for about $4,000 (Fig. 6.9). It is attached to a tabletop and can be controlled by a foot switch. A manual version is also attached to a tabletop and is for someone with some arm movement but that may be erratic or tremulous. The arm has a built-in damper that filters out unwanted movement.

Gravity-Eliminating Orthoses

A few new devices have become commercially available in this area. What makes this segment unique is that these devices are attached to an appendage (typically the arm) and provide assistance to accomplish activities of daily living. They utilize the remaining residual strength of the individual to allow voluntary movements. These devices act to amplify weak movements of the arm and negate the effect of gravity for the user so that he or she can perform tasks such as feeding easily.

Figure 6.9 Neater Eater.

Among the earliest and most accepted devices is the balance forearm orthosis (BFO), also called the mobile arm support (Fig. 6.10). The BFO (JAECO Orthopedics, Hot Springs, AR), which is a passive (body-powered) device, was developed in 1965. It provides a person with weak musculature with the ability to move the arms in a horizontal plane through the use of two linkages that have joints along the vertical axes. One end of the BFO is attached to a wheelchair; the other end is connected to a trough into which a person places the forearm. The trough uses a fulcrum at the forearm that permits the hand to elevate if the shoulder is depressed. The BFO allows a person to move horizontally, for example, over a lap tray and to use compensatory movements to attain limited movement in the vertical direction. The BFO retails for approximately $350.

The Wilmington Robotic Exoskeleton (WREX) is a body-powered orthosis that is modular and mounted to a person's wheelchair or to a body jacket (Fig. 6.11). It is a two-segment, four-degrees-of-freedom exoskeletal arm, energized by elastic bands that aid in moving the arm in 3-D space. The WREX allows full passive range of motion of the arm and provides a sense of flotation that assists in voluntary movement (25). WREX can easily be adjusted to accommodate subjects of different weights and arm lengths by changing the number of bands or sliding the telescoping links. The device is typically mounted to a wheelchair and intended primarily for people with muscular weakness such as muscular dystrophy and spinal muscular atrophy. It is also being used for children with arthrogryposis who can walk independently by attaching the WREX to a body jacket (26). The WREX was conceived and developed at the Alfred I. duPont Hospital for Children and is now marketed by JAECO Orthopedics, Hot Springs, AR for $2,000.

Two other passive upper extremity orthoses have recently been commercialized, and both emanate from the Netherlands. The first is the Armon made by Micro Gravity Products, which is powered by springs. It is for people with arm weakness. It attaches to the forearm of the user and provides gravity balancing. The device can be attached to the wheelchair or a tabletop. The Armon does not follow the contours of the arm. It can be adjusted by a motor to compensate for the weight of a person. The second device is called the Dynamic Arm Support (DAS) made by Exact Dynamics. It is similar to the Armon, but has a vertical movement that provides the elevation. It, too, can be adjusted for different-sized people with the aid of a motor and can be attached to a wheelchair.

Figure 6.10 Balanced Forearm Orthosis (BFO).

Figure 6.11 Wilmington Robotic Exoskeleton (WREX).

Robots

The Assistive Robotic Manipulator (ARM) (Fig. 6.12) is a six-degree-of-freedom wheelchair-mounted robotic

Figure 6.12 The ARM manipulator.

device developed in the Netherlands by Exact Dynamics, Inc. As a result of its functionality and mobility, the ARM offers users a wide range of manipulation possibilities. Example tasks include eating, pouring and drinking, playing board games, operating switches, and opening doors. The ARM manipulator features a programmable user interface and flexible input/output for interfacing with electrical wheelchairs. It folds into an unobtrusive position at the side of the wheelchair when not in use and folds out when commanded. Its present inputs include a 16-button keypad, trackball, and joystick, which performs individual joint control, integrated hand control, or programmed modes of control. There are currently approximately 100 users of the ARM in Europe, and it costs approximately $40,000.

The Raptor is also a lightweight wheelchair-mounted robot arm that controls each joint individually. It has four degrees of freedom and a gripper. It is sold by Kersten RT in the Netherlands.

Therapy Robots

The term "rehabilitation robot" has been around for a good 30 years, when it was first applied to assistive motorized devices that performed tasks of daily living for people with physical impairments. As shown, these applications are continually being developed; however, the term is being increasingly applied to machines that assist in the recovery from a condition such as stroke. This shift in emphasis from assistive to rehab in robotics is largely driven by an aging population, resulting in a far greater number of potential beneficiaries.

There are approximately 600,000 new cases of stroke in the United States every year. The "graying" of the population is even more pronounced in countries such as Japan. Patients undergo physical therapy to restore lost function. The therapy tends to be repetitive, and evidence suggests that the duration, intensity, and quality of therapy all play a role in recovery. Although functional gains remain small, the potential of machines assisting in therapy is enormous. These machines are ideally suited to the rigorous and repetitive nature of therapy. The following paragraphs describe some of the devices that are currently on the market.

Manually assisted treadmill walking is commonly used for regular therapy for patients with neuromuscular impairments. This type of therapy is performed with some type of harness system that supports the patient's weight. There are two main limitations to this type of therapy: It is labor-intensive, as it requires two therapists to move the patient's legs, which causes therapist fatigue and back pain due to awkward the ergonomic positions. Second, manual therapy lacks repeatability and a way to objectively measure performance. The Locomat (Hocoma AG, Volketswil, Switzerland) is a bilateral robotic gait trainer that is used along with a weight-supported system. It can replace some of the functions of a therapist and free him or her from performing the arduous task of leg movement. The Locomat can provide customized gait training for an individual patient by defining the optimal trajectory of leg movements and creating a specified set of force interactions between the device and the patient. The device has been commercially available since 2000 and is used in numerous clinics for spinal cord injury (SCI), stroke, and traumatic brain injury (TBI) populations. There are about 150 Locomat systems in use worldwide.

InMotion Robots (Interactive Motion Technologies, Inc., Cambridge, MA) are a suite of table-mounted robotic systems that provide therapy for the shoulder, elbow, wrist, hand, and overground ankle training. The robots are combined with a video screen to provide a fun and therapeutic environment for exercise. These robots can be programmed to vary the relative effort between the user and the robot. If, for instance, the user is weak, the robot can do most of the work. As the patient gains strength, the robot's effort can be decreased appropriately. The InMotion system has been developed over the last 15 years, and its strength is that it offers a low impedance system so that the effect of the robot can be imperceptible to the user. It is primarily used for stroke and other neurological disorders.

Another upper extremity robotic-based rehabilitation system is the REO made by Motorika, Ltd., a company established in 2004. REO is an upper extremity device made to apply robotic technology to meet the therapeutic needs of stroke patients. It offers efficient repetitive training activities. "REO Therapy" actively engages a patient in repetitive exercises to improve arm function, while therapists benefit from patient

progress monitoring and practice efficiency. A video screen accompanies the device to provide progress and visual stimulation during exercise. The company also offers a REO Ambulator for lower extremities that is a robotic gait trainer similar to the Locomat. It has been used for a few years in rehabilitation clinics; however, there is still insufficient data to support its findings.

Other robotic therapy devices being developed for the upper extremity include the T-WREX (27), iMove Reacher (iMove Support, Hengelo, Netherlands), and McArm (Focal Revalidatietechniek, Netherlands), HapticMASTER (Moog FCS, Netherlands). A lower extremity device under development is KineAssist (Chicago P, Chicago, IL).

PEARLS OR PERILS

- Multidisciplinary approaches to evaluating a child's needs are most effective. You may have all the tools you need, but the family story is what's important.
- The goal of all assistive communication technology is to help people with disabilities overcome the limitations of those disabilities and to become more independent and more efficient, to become faster and to experience longer stamina, and to be able to become active members of their social communities.

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7

Electrodiagnosis in Pediatrics

Craig M. McDonald

Electromyography (EMG), nerve conduction studies (NCS), and evoked potentials, including somatosensory-evoked potentials (SSEPs) and motor-evoked potentials (MEPs), provide useful information to assist the clinician in the localization of pathology within the lower motor neuron and selected areas of the central nervous system. In the case of acquired or hereditary disorders of the lower motor neuron—anterior horn cell, peripheral nerve, neuromuscular junction (presynaptic or postsynaptic region), or muscle—electrodiagnostic studies are a useful tool as an extension of the clinician's physical examination. The information gained from electrodiagnostic studies may be invaluable in planning subsequent, more invasive diagnostic studies (eg, muscle and nerve biopsy, cerebrospinal fluid [CSF] examination, [magnetic resonance] MR imaging, which at times requires general anesthesia), allow for more cost-effective and specific molecular genetic testing, or aid in the surgical management of peripheral nerve trauma, compressive lesions, or entrapments. In the case of immune-mediated disorders such as myasthenia gravis or Guillain-Barré syndrome, electrodiagnostic studies may permit prompt treatment.

Pediatric electrodiagnosis must be approached with knowledge of peripheral neuromuscular development and thoughtful planning of the study with regard to most likely diagnostic possibilities, developmental status of the child, and the likelihood that the pediatric electrodiagnostic practitioner will be able to provide clinicians and family with useful diagnostic information. The physical examination and developmental level of the infant or child directs the study. The examination requires the patience and technical competence of an electrodiagnostic clinician experienced and skilled in the evaluation of children. This chapter will focus on considerations specific to the electrodiagnostic evaluation of infants and children, with an emphasis on practical suggestions that may facilitate the completion of an accurate pediatric electrodiagnostic examination with a minimum of discomfort and distress to the child, parent, and pediatric electrodiagnostic specialist.

MATURATIONAL FACTORS IN PEDIATRIC ELECTRODIAGNOSIS

The normative neurophysiologic data relating to the maturation of peripheral nerves and muscle in children have been greatly expanded in the recent past (1–10). The reader is referred to the volume by Jones, Bolton, and Harper (8) for an excellent review of neurophysiologic norms in pediatric populations. Peripheral nerve myelination begins at about the 15th week of gestation and continues throughout the first 3–5 years after birth (11). Conduction velocities are determined by myelination, diameter of the fiber, and internodal differences. Myelination occurs at the same rate, whether intrauterine or extrauterine. Conduction velocities are directly related to gestational and postconceptual age,

and are unrelated to birth weight (12,13). Conduction velocities increase in direct proportion to the increase in diameter of fibers during growth. A direct relationship also exists between the diameter of the axon and the thickness of the myelin sheath. The diameter of the fibers at the time of birth has been shown to be one-half of that in the adult. No unusual acceleration of myelination occurs subsequent to birth (14). Peripheral fibers reach their maximum diameter at 2–5 years after birth (14,15). The nodes of Ranvier continue to remodel, with peak internodal distances being reached at 5 years of age.

Nerve Conduction Studies

In general, normal standard adult values for conduction velocities are reached by age 3 to 5. In infancy, upper and lower extremity conduction velocities are similar under age 1. Subsequently, faster conductions are maintained in the upper extremities and comparatively slower conductions in the lower extremities, as with adults. Unique values for expected conduction velocities are observed for specific peripheral nerves.

Motor Nerve Conduction

Motor conduction velocities in infants are found to be one-half of adult values. In infants, conduction studies should be at least greater than 20 m/s. At birth, motor conduction velocity (MCVs) for the median, ulnar, and peroneal nerves are 27 m/s. The median nerve may lag in maturation of conduction velocity (CV) relative to the ulnar and peroneal nerves. Ulnar MCV values reach the lower adult range by age 3 (14). The slight difference between ulnar and median MCV values present in the first 3 years of life disappears in children by 4–5 years of age. Careful and consistent measurements are necessary to achieve reliable and valid data. Normative values or selected motor nerve conduction velocities are shown in Table 7.1.

Distal Motor Latency

Distal motor latencies (DMLs) show maturational changes between infancy and 3–5 years of age, similar to motor conduction velocities. Normative data for distal latencies have generally been more incomplete,

7.1 Normal Motor Conduction Velocities (m/sec)

	MEDIAN (REF.)	ULNAR (REF.)	PERONEAL (REF.)	TIBIAL (REF.)
24–72 hours	25.8 ± 3.40 (9)	28.0 ± 3.38 (9)	26.4 ± 3.58 (9)	23.9 ± 2.73 (9)
7 days–1 month	25.43 ± 3.84 (6) 26.17 ± 2.16 (10)	25.03 ± 2.7 (10)	22.43 ± 1.22 (6) 25.60 ± 3.68 (10)	25.30 ± 1.96 (1) 23.21 ± 2.79 (10)
0–3 months	32.3 ± 3.56 (9)	35.1 ± 3.40 (9)	30.8 ± 2.91 (9)	27.8 ± 3.89 (2)* 27.9 ± 2.27 (9)
4–6 months	37.0 ± 4.38 (9)	40.5 ± 4.24 (9)	36.1 ± 4.67 (9)	36.3 ± 4.98 (2)** 34.7 ± 2.92 (9)
1–6 months	34.35 ± 6.61 (6) 36.35 ± 3.66 (10)	36.33 ± 3.72 (10)	35.18 ± 3.96 (6) 36.69 ± 4.06 (10)	32.55 ± 4.05 (10)
6–12 months	42.3 ± 6.43 (9) 43.91 ± 3.44 (10)	47.2 ± 6.33 (9) 45.02 ± 2.93 (10)	40.8 ± 6.16 (9) 43.11 ± 4.13 (10)	38.5 ± 5.50 (9) 39.45 ± 4.29 (10)
1–2 years	48.23 ± 4.58 (6) 47.81 ± 2.33 (10)	48.95 ± 2.46 (10)	51.42 ± 3.02 (6) 47.43 ± 2.5 (10)	42.6 ± 3.80 (2) 42.42 ± 2.23 (10)
1–3 years	52.7 ± 4.70 (9)	53.8 ± 4.83 (9)	48.7 ± 4.86 (9)	44.9 ± 4.44 (9)
2–4 years	53.59 ± 5.29 (6) 52.71 ± 3.71 (10)	54.19 ± 3.49 (10)	55.73 ± 4.45 (6) 51.21 ± 3.95 (10)	49.8 ± 5.78 (2) 44.81 ± 1.91 (10)
4–6 years	56.26 ± 4.61 (6) 55.0 ± 5.20 (9) 56.48 ± 2.36 (10)	56.9 ± 4.34 (9) 56.51 ± 3.19 (10)	56.14 ± 4.96 (6) 49.6 ± 4.98 (9) 53.99 ± 3.74 (10)	50.0 ± 4.26 (2) 48.6 ± 4.25 (9) 48.43 ± 2.53 (10)
6–14 years	57.2 ± 3.71 (9)*** 57.32 ± 3.35 (6)	58.3 ± 5.76 (9)***	49.6 ± 3.40 (9)*** 57.05 ± 4.54 (6)	48.2 ± 2.76 (9)*** 52.4 ± 4.19 (2) ±

Source: Data are presented as means ± standard deviation.

*1–3 months; **3–6 months; ***7–14 years; ± 6–11 years

with ranges of distances provided (from stimulation to active electrode). While the stimulation distance should always be recorded in the electrodiagnostic report, the specific distal latency is rarely of critical importance in determining a diagnosis in pediatric electrodiagnosis, as distal peripheral entrapments are relatively uncommon. Rather, reported distal latencies that are either unusually fast or unusually slow in the setting of otherwise normal motor conduction velocities should raise a suspicion regarding technical problems and identification of appropriate wave forms.

The *corrected DML* may be used as an alternative in young children using the formula of Slomic and colleagues found in Wagner and Buchthal (16):

$$\text{Corrected DML} = \text{measured DML} - [L - X/MCV]$$

Where L = actual distance between stimulating cathode to the active recording electrode, and X = standard distance (4 cm for nerves of upper limbs and 5 cm for nerves of the lower limbs. Garcia and colleagues (10) have reported the most complete data to date on corrected DML in children—see Table 7.2). Corrected DML in the neonatal group is increased relative to other age groups, decreases over the first 12 months of life, remains unchanged between 12 months and 24 months, and slightly increases later. As most clinicians reading reports are not familiar with corrected DML, an explanation of the calculation and normative interpretation should be included in the report if this data is reported along with the actual DML and distance used.

7.2 Corrected Distal Motor Latency (msec)

	MEDIAN	ULNAR	PERONEAL	TIBIAL
<1 month	3.00 ± 0.18	2.80 ± 0.43	3.33 ± 0.54	3.21 ± 0.36
1–6 months	2.47 ± 0.18	2.20 ± 0.38	2.51 ± 0.22*	2.62 ± 0.41
6–12 months	2.28 ± 0.21	1.98 ± 0.21	2.36 ± 0.31	2.55 ± 0.37
1–2 years	2.34 ± 0.34	1.86 ± 0.16	2.35 ± 0.23	2.45 ± 0.46
2–4 years	2.34 ± 0.23	1.89 ± 0.17	2.57 ± 0.40	2.35 ± 0.24
4–6 years	2.56 ± 0.29	2.03 ± 0.25	3.02 ± 0.48	2.69 ± 0.46

Data are presented as means ± standard deviation.
Corrected DML = measured DML—[L—X/MCV], where L = actual distance between stimulating cathode to the active recording electrode and X = standard distance (4 cm for nerves of upper limbs and 5 cm for nerves of the lower limbs).
Source: Ref. 10.

Compound Muscle Action Potential

Compound muscle action potential (CMAP) amplitudes are important to consider in the evaluation of axonal loss, conduction block, and muscle fiber atrophy. CMAP amplitudes of lower extremity nerves are one-half to one-third adult values in infants, and upper extremity CMAPs may be one-third to one-fourth adult values during infancy. As with motor conduction velocities, CMAP amplitudes increase in size with age, but adult values are generally not reached until the end of the first decade. Normal values for CMAP amplitudes are shown in Table 7.3.

Sensory Nerve Conduction

Modern EMG equipment, which includes amplifiers and signal averaging capability, allows sensory nerve action potentials to be routinely recorded in the absence of peripheral nerve pathology. Maturational changes for orthodromic and antidromic sensory conduction are similar to that for motor fibers (6,17,18). In infants and young children, two distinct peaks are often observed in the sensory nerve action potential (SNAP) with proximal stimulation. This two-peak potential has been attributed to differences in maturation between two groups of sensory fibers (16) and often persists until 4–6 years of age. Sensory nerve conduction velocities may be calculated from single distal antidromic or orthodromic stimulations by measuring the distance from stimulation point to active electrode and the distal latency. Normative values for sensory nerve conduction velocities in selected nerves using orthodromic stimulation and proximal recording are shown in Table 7.4. Normal values for orthodromic and antidromic SNAP amplitudes are shown in Table 7.5.

F-waves

The F-wave is a late response that appears as a supermaximal motor nerve stimulation and arises from the discharge of a small number of motor neurons in response to antidromic stimulation of the motor axon. The F-wave latency is measured from hand and foot intrinsic muscles, and is useful for evaluating the motor nerve conduction velocity and proximal nerve segments. In the F-wave, the speed of motor nerve conduction is measured over a long distance, enhances less subject to errors inherent in the calculation of motor conduction velocities over short distances (10 cm or less). F-waves can be recorded from

7.3 Normal Compound Muscle Action Potential Amplitudes (mV)

	MEDIAN (APB) (REF.)	ULNAR (ADM) (REF.)	PERONEAL (EDB) (REF.)	TIBIAL (AH) (REF.)
24–72 hours	3.60 ± 1.56 (9)	5.42 ± 2.21 (9)	3.43 ± 0.47 (9)	9.29 ± 1.93 (9)
7 days–1 month	3.00 ± 0.31 (6) 1.27 ± 0.74 (10)	1.88 ± 0.92 (10)	3.0 ± 1.26 (6) 1.77 ± 0.62 (10)	4.40 ± 1.73 (10)
0–3 months	4.06 ± 1.49 (9)	6.49 ± 2.83 (9)	4.52 ± 0.85 (9)	13.30 ± 2.86 (9)
1–6 months	7.37 ± 3.24 (6) 2.37 ± 1.27 (10)	3.11 ± 1.45 (10)	5.23 ± 2.37 (6) 2.68 ± 1.04 (10)	6.16 ± 2.44 (10)
6–12 months	5.47 ± 2.01 (9) 2.94 ± 1.17 (10)	6.97 ± 1.89 (9) 2.73 ± 1.09 (10)	5.86 ± 1.12 (9) 2.64 ± 1.32 (10)	14.06 ± 2.58 (9) 6.83 ± 2.69 (10)
1–2 years	8.90 ± 3.61 (6) 4.12 ± 1.90 (10)	4.55 ± 1.53 (10)	5.80 ± 2.48 (6) 3.69 ± 1.27 (10)	9.07 ± 2.12 (10)
1–3 years	5.88 ± 2.51 (9)	7.66 ± 2.23 (9)	6.42 ± 1.92 (9)	15.71 ± 1.79 (9)
2–4 years	9.55 ± 4.34 (6) 5.96 ± 2.01 (10)	5.48 ± 1.42 (10)	6.10 ± 2.99 (6) 4.25 ± 1.59 (10)	9.57 ± 3.54 (10)
4–6 years	10.37 ± 3.66 (6) 6.49 ± 1.83 (9)	8.80 ± 2.35 (9)	7.10 ± 4.76 (6) 3.78 ± 1.23 (10)	9.48 ± 2.39 (10)
6–14 years	12.37 ± 3.66 (6) 8.83 ± 1.87 (9)*	10.27 ± 2/02 (9)*	8.15 ± 4.19 (6) 7.22 ± 1.64 (6)*	15.75 ± 1.77 (9)*

Data are presented as means ± standard deviation
*7–14 years

most limb nerves in newborns and young infants. The minimum F-latency in normal children recorded from hand muscles, with median or ulnar nerve stimulation at the wrist, is generally less than 20 milliseconds in children younger than 6 years of age (6,7,19). In the lower extremities, the F-wave latency recorded from intrinsic foot muscles, with peroneal or posterior tibial nerve stimulation at the ankle, is generally less than 30 milliseconds (4,6). Normal values for F-wave latencies for children are shown in Table 7.6.

H reflex

The H reflex is present in both the upper extremities (median and ulnar) and lower extremities (with posterior tibial stimulation) in infancy. While the tibial H reflex persists into adulthood, the upper extremity H-reflex responses are present in virtually all infants at birth and become suppressed in most children over the course of the first year. Normal values for H-reflex latencies in children are shown in Table 7.7.

Neuromuscular Transmission

The neuromuscular junction shows less stability and reserve in normal newborns. At low rates of stimulation (1–2 Hz), no significant incremental or decremental changes in CMAP amplitude is observed (20). At higher rates of stimulation (5–10 Hz), normal infants may show slight facilitation. Decremental responses averaging 24% have been reported at high rates of stimulation (20 Hz) in normal newborn infants. At 50-Hz stimulation, normal newborns may show decrements on the order of 50% (17). In general, decremental changes of greater than 10% at low rates of stimulation (2–5 Hz) and facilitatory changes of greater than 23% at high rates of stimulation (20–50 Hz) are felt to be significant in the post-term infant (21). Some authors have utilized high rates of stimulation on the order of 50 Hz for ten seconds to document facilitation of greater than 20% to 23% (at times over 100% increments are observed) in infantile botulism (21,22,23).

Electromyography

Motor Unit Configuration and Amplitude

Amplitudes of motor unit action potentials (MUAPs) are lower in infants, with amplitudes ranging from 150 microvolts to approximately 2,000 microvolts. Generally, motor unit action potentials more than

7.4 Normal Sensory Conduction Velocities (m/sec)

	MEDIAN (REF.)	ULNAR (REF.)	SURAL (REF.)
24–72 hours	18.74 ± 2.64 (D2-W) (9)* 21.68 ± 2.43 (D2-E) (9)*	19.13 ± 0.29 (D5-W) (9)* 21.85 ± 1.37 (D5-E) (9)*	17.65 ± 2.43 (6 cm) (9)*
7days–1 month	22.31 ± 2.16 (D2-W) (6) 24.09 ± 2.6 (D3-W) (10)	18.4 ± 3.97 (D5-W) (3)	20.26 ± 1.55 (4–8 cm) (6)
0–3 months	24.20 ± 3.51 (D2-W) (9)* 29.26 ± 4.14 (D2-E) (9)*	25.95 ± 2.49 (D5-W) (9)* 34.42 ± 4.13 (D5-E) (9)*	22.54 ± 2.28 (8 cm) (9)*
4–6 months	29.91 ± 2.17 (D2-W) (9)* 38.44 ± 5.35 (D2-E) (9)*	31.51 ± 2.70 (D5-W) (9)* 44.07 ± 4.12 (D5-E) (9)*	28.78 ± 2.98 (8 cm) (9)*
1–6 months	35.52 ± 6.59 (D2-W) (6) 35.07 ± 4.87 (D3-W) (10)	27.7 ± 6.37 (D5-W;1–3 mo) (3) 37.1 ± 5.25 (D5-W;3–6 mo) (3)	34.68 ± 5.43 (6–8 cm) (6)
6–12 months	40.31 ± 5.23 (D2-W) (6) 32.60 ± 3.15 (D2-W) (9)* 41.14 ± 4.43 (D2-E) (9)* 41.95 ± 2.68 (D3-W) (10)	40.0 ± 5.13 (D5-W) (3) 34.41 ± 3.11 (D5-W) (9)* 44.67 ± 3.45 (D5-E) (9)*	29.40 ± 3.55 (8 cm) (9)*
1–2 years	46.93 ± 5.03 (D2-W) (6) 45.12 ± 2.99 (D3-W) (10)	44.2 ± 7.79 (D2-W) (3)	49.73 ± 5.53 (8–10cm) (6)
1–3 years	36.41 ± 3.93 (D2-W) (9)* 47.23 ± 3.74 (D2-E) (9)*	34.94 ± 2.92 (D5-W) (9)* 45.59 ± 4.26 (D5-E) (9)*	35.37 ± 4.32 (8 cm) (9)* 38.33 ± 4.49 (12 cm) (9)*
2–4 years	49.51 ± 3.34 (D2-W) (6) 48.82 ± 3.02 (D3-W) (10)	48.8 ± 3.01 (D5-W) (3)	52.63 ± 2.96 (8–10cm) (6)
4–6 years	51.71 ± 5.16 (D2-W) (6) 41.04 ± 4.94 (D2-W) (9)* 51.22 ± 5.07 (D2-E) (9)* 50.72 ± 3.6 (D3-W) (10)	47.7 ± 6.75 (D5-W) (3) 42.94 ± 4.55 (D5-W) (9)* 51.58 ± 4.49 (D5-E) (9)*	53.83 ± 4.34 (8–10cm) (6) 39.38 ± 4.58 (8 cm) (9)* 41.49 ± 4.41 (12 cm) (9)*
6–14 years	53.84 ± 3.26 (6) 43.71 ± 3.37 (D2-W) (9)* 53.44 ± 3.19 (D2-E) (9)*	46.6 ± 5.6 (D5-W) (3) 43.92 ± 3.91 (D5-W) (9)* 53.23 ± 3.58 (D5-E) (9)*	53.85 ± 4.19 (6) 40.60 ± 4.79 (8 cm) (9)* 42.75 ± 4.79 (12 cm) (9)* 46.71 ± 4.17 (14 cm) (9)*

Data are presented as means ± SD
*Velocities based on peak latencies for Cai and Zhang (9); others based on onset latencies
D-W = Finger to wrist using ring electrodes with orthodromic stimulation
D-E = Finger to elbow using ring electrodes with orthodromic stimulation
D2 = Index finger for median; D3 = middle finger for median; D5 = fifth finger for ulnar
Sural nerve studies use antidromic with recording electrodes behind the lateral malleolus with stimulus delivered at 6 cm to 14 cm above the malleolus as specified.

1,000 microvolts in 0- to 3-year-old children are rare (24,25). In infants, motor unit action potentials are usually biphasic or triphasic.

Motor Unit Duration

Infantile motor unit action potentials are often shorter in duration. DeCarmo (24) found newborn infants to exhibit durations 17% to 26% shorter than those seen in adults. Durations of motor unit action potentials are often shorter than 5 milliseconds in infants.

Motor Unit Recruitment

In very young infants and children, it is difficult to assess strength of voluntary contraction and determine when the interference pattern is full. In general, as strength of voluntary contraction increases, there is an increase in motor unit action potentials recruited. However, the recruitment pattern in infants may be disordered and chaotic. As with adults, the recruitment frequency, defined as the firing rate of a MUAP when a different MUAP first appears, with gradually increasing strength of voluntary contraction, is helpful

7.5 Normal Sensory Nerve Action Potential (SNAP) Amplitudes in Children (μV)

	MEDIAN (REF.)	ULNAR (REF.)	SURAL (REF.)
24–72 hours	6.76 ± 0.79 (D2-W) (9)	5.26 ± 0.57 (D5-W) (9)	5.29 ± 2.16 (6 cm) (9)
7days–1 month	6.22 ± 1.30 (D2-W) (6) 4.86 ± 2.23 (D3-W) (10)	5.5 ± 3.1 (D5-W) (3)	9.12 ± 3.02 (4–8 cm) (6)
0–3 months	16.74 ± 1.47 (D2-W) (9)	7.83 ± 0.60 (D5-W) (9)	9.97 ± 1.24 (8 cm) (9)
4–6 months	17.72 ± 3.35 (D2-W) (9)	8.26 ± 1.00 (D5-W) (9)	13.58 ± 2.19 (8 cm) (9)
1–6 months	15.86 ± 5.18 (D2-W) (6) 10.66 ± 3.62 (D3-W) (10)	9.4 ± 3.2 (D5-W;1–3 mo) (3) 13.2 ± 3.23 (D5-W;3–6 mo) (3)	11.66 ± 3.57 (6–8 cm) (6)
6–12 months	16.00 ± 5.18 (D2-W) (6) 17.55 ± 1.70 (D2-W) (9) 9.00 ± 3.45 (D3-W) (10)	13.0 ± 5.6 (D5-W) (3) 10.87 ± 2.4 (D5-W) (9)	14.87 ± 4.67 (8 cm) (9)
1–2 years	24.00 ± 7.36 (D2-W) (6) 15.72 ± 4.50 (D3-W) (10)	16.3 ± 2.44 (D2-W) (3)	15.41 ± 9.98 (8–10cm) (6)
1–3 years	19.51 ± 3.99 (D2-W) (9)	12.34 ± 2.1 (D5-W) (9)	18.02 ± 3.83 (8 cm) (9)
2–4 years	24.28 ± 5.49 (D2-W) (6) 12.02 ± 5.89 (D3-W) (10)	16.0 ± 3.6 (D5-W) (3)	23.27 ± 6.84 (8–10cm) (6)
4–6 years	25.12 ± 5.22 (D2-W) (6) 19.78 ± 4.21 (D2-W) (9) 14.04 ± 5.99 (D3-W) (10)	14.2 ± 2.72 (D5-W) (3) 13.15 ± 3.6 (D5-W) (9)	22.66 ± 5.42 (8–10cm) (6) 18.50 ± 3.89 (8 cm) (9)
6–14 years	26.72 ± 9.43 (6) 20.50 ± 3.49 (D2-W) (9)*	13.4 ± 4.2 (D5-W) (3) 14.30 ± 2.5 (D5-W) (9)*	26.75 ± 6.59 (6) 18.67 ± 4.39 (8 cm) (9)*

Data are presented as means ± SD
Amplitudes are determined peak-to-peak from positive-to-negative peak of the SNAP
D-W = Finger to wrist using ring electrodes with orthodromic stimulation
D2 = Index finger for median; D3 = middle finger for median; D5 = fifth finger for ulnar
Sural nerve studies used antidromic stimulation with recording electrodes behind the lateral malleolus with stimulus delivered at 6 cm to 14 cm above the malleolus as specified.

in differentiating a myopathic process (lower recruitment frequency values) from a neuropathic process (higher recruitment frequencies after greater than 20–25 Hz). An example of neuropathic recruitment is shown in Figure 7.1.

TECHNICAL FACTORS WITH INFANTILE NERVE CONDUCTION STUDIES

Temperature

The maintenance of appropriate subject temperature is essential during nerve conduction studies. Neonates generally have difficulty with temperature homeostasis, and low subject temperature may have profound effects on conduction velocities. A skin temperature of 36–37°C produces near-nerve temperatures of 37–38°C and avoids spurious reductions in nerve conduction velocities and prolongation of distal latencies. It is assumed that a 1°C drop in temperature produces a slowing of conduction on the order of 2–3 meters/second. Every attempt should be made to maintain extremity temperature with infant warmers, heating lamps, or warm blankets.

Volume Conduction

Volume conduction is defined as the current transmission from a potential source through a conducting medium, such as the body tissues. This may produce depolarization of peripheral nerves in proximity to the specific nerve being studied, and this is particularly problematic in smaller children with less soft tissue separating nerves. For example, volume conduction can produce simultaneous stimulation of both the median and ulnar nerves at the wrist or at the elbow. Such volume conduction should always be suspected when higher stimulation intensities or durations are utilized and when CMAP configurations show an initial positive deflection or a multiple peak configuration.

7.6 Normal F Wave Latencies in Children (msec)

	MEDIAN (REF.)	ULNAR (REF.)	PERONEAL (REF.)	TIBIAL (REF.)
24–72 hours	19.56 ± 2.44 (w) (9) 16.51 ± 1.74 (e) (9)	19.67 ± 2.74 (w) (9) 16.64 ± 1.30 (e) (9)	27.56 ± 3.82 (a) (9) 24.38 ± 3.74 (k) (9)	26.92 ± 3.27 (a) (9) 23.51 ± 2.45 (k) (9)
7 days–1 month	18.17 ± 2.17 (w) (10)	18.63 ± 1.6 (w) (10)	25.2 ± 4.82 (a) (10)	23.92 ± 1.62 (a) (10)
0–3 months	17.62 ± 1.39 (w) (9) 15.39 ± 1.46 (e) (9)	17.65 ± 1.39 (w) (9) 14.93 ± 1.80 (e) (9)	26.14 ± 2.84 (a) (9) 23.46 ± 2.76 (k) (9)	28.59 ± 2.41 (a) (9) 22.52 ± 2.10 (k) (9)
4–6 months	17.54 ± 1.96 (w) (9) 15.41 ± 1.56 (e) (9)	16.99 ± 1.24 (w) (9) 14.91 ± 1.28 (e) (9)	25.18 ± 4.37 (a) (9) 22.15 ± 2.68 (k) (9)	23.93 ± 1.85 (a) (9) 20.67 ± 2.30 (k) (9)
1–6 months	15.91 ± 1.22 (w) (10)	15.71 ± 1.6 (w) (10)	21.4 ± 1.78 (a) (10)	21.4 ± 1.35 (a) (10)
6–12 months	16.86 ± 1.50 (w) (9) 14.37 ± 1.17 (e) (9) 15.67 ± 0.89 (w) (10)	17.02 ± 1.45 (w) (9) 14.41 ± 0.88 (e) (9) 15.45 ± 1.37 (w) (10)	25.54 ± 2.04 (a) (9) 21.56 ± 3.36 (k) (9) 20.33 ± 1.1 (a) (10)	23.78 ± 1.83 (a) (9) 20.92 ± 1.59 (k) (9) 22.0 ± 2.05 (a) (10)
1–2 years	15.64 ± 1.08 (w) (10)	15.67 ± 0.78 (w) (10)	22.82 ± 1.66 (a) (10)	24.21 ± 1.63 (a) (10)
1–3 years	16.41 ± 1.13 (w) (9) 14.21 ± 0.77 (e) (9)	16.63 ± 1.88 (w) (9) 14.69 ± 1.35 (e) (9)	26.73 ± 2.87 (a) (9) 24.30 ± 2.46 (k) (9)	25.44 ± 2.20 (a) (9) 23.65 ± 1.71 (k) (9)
2–4 years	16.36 ± 1.45 (w) (10)	16.0 ± 1.41 (w) (10)	24.64 ± 2.21 (a) (10)	25.6 ± 2.53 (a) (10)
4–6 years	17.62 ± 1.62 (w) (9) 15.81 ± 1.17 (e) (9) 18.0 ± 1.27 (w) (10)	18.51 ± 1.74 (w) (9) 16.53 ± 1.48 (e) (9) 18.25 ± 1.48 (w) (10)	30.57 ± 3.82 (a) (9) 25.22 ± 3.44 (k) (9) 29.45 ± 2.58 (a) (10)	31.07 ± 3.10 (a) (9) 25.97 ± 2.32 (k) (9) 30.12 ± 2.52 (a) (10)
6–14 years	20.18 ± 1.61 (w) (9) 17.34 ± 1.52 (e) (9)	20.66 ± 1.92 (w) (9) 18.14 ± 1.46 (e) (9)	38.16 ± 4.43 (a) (9) 31.38 ± 4.75 (k) (9)	36.32 ± 3.72 (a) (9) 32.78 ± 3.89 (k) (9)
18–30 years	26.14 ± 3.03 (w) (9) 22.88 ± 1.34 (e) (9)	27.03 ± 2.14 (w) (9) 23.42 ± 1.90 (e) (9)	49.63 ± 7.74 (a) (9) 41.23 ± 7.63 (k) (9)	48.27 ± 3.09 (a) (9) 39.93 ± 2.73 (k) (9)
Side-to-side difference	1.03 ± 0.73 (2.5)	0.94 ± 0.69 (2.3)	1.19 ± 1.17 (3.5)	1.26 ± 1.01 (3.3)

Data are presented as means ± SD
Minimum F-wave latency from 10 recordings
w, wrist stimulation; e, elbow stimulation; a, ankle stimulation; k, knee stimulation
Side-to-side difference shows mean ± SD (upper limits of normal)

Shock Artifact

Shock artifact is a common problem with smaller subjects because of short distances between the stimulator and recording electrodes. This may be particularly problematic with distal stimulation. The ground electrode should be placed between the stimulating and recording electrodes, and, in infants, often a standard 6-mm silver disc or ring electrode can be placed around the wrist or ankle. Alternatively, the ground disc may be taped to the dorsal surface of the hand. Other approaches to minimize shock artifact in young children include the utilization of pumice paste to reduce skin impedance and permit suprathreshold stimulation with lower electrical currents, use of a minimal amount of conduction gel or cream, and rotation of the proximal anode in relation to the distal cathode.

Measurement of Distances/ Measurement Error

Distance measurements must be extremely meticulous during pediatric electrodiagnostic evaluations. Segment studies are often on the order of 6–10 cm in length. A measurement discrepancy of only 1 cm may produce as much as a 10% to 15% conduction velocity error.

Stimulating Electrodes

For neonates and young infants, small stimulators with short interelectrode distances are commercially available and simplify the testing of short nerve segments over small extremities (Fig. 7.2). The stimulation intensity may be reduced by the use of a small monopolar needle electrode as the stimulating

7.7 Normal H Reflex Latencies in Children (msec)

	TIBIAL	MEDIAN	ULNAR
	17.37 ± 1.23	18.67 ± 1.71 (w) (25/25) 15.81 ± 1.15 (e)	18.66 ± 1.48 (w) (25/25) 16.24 ± 0.93 (e)
0–3 months	16.01 ± 1.23	17.15 ± 0.92 (w) (12/20) 14.99 ± 0.94 (e)	17.25 ± 1.93 (w) (9/20) 15.08 ± 1.85 (e)
4–6 months	15.73 ± 1.19	16.95 ± 1.12 (w) (12/20) 14.64 ± 0.82 (e)	16.74 ± 0.77 (w) (7/20) 14.68 ± 0.67 (e)
6–12 months	15.92 ± 1.28	15.75 ± 1.14 (w) (7/20) 14.23 ± 0.51 (e)	16.49 ± 1.01 (w) (4/20) 14.32 ± 0.50 (e)
1–3 years	16.91 ± 1.46		
4–6 years	18.76 ± 1.71		
7–14 years	22.00 ± 1.97		
18–30 years	28.04 ± 1.68		
Side-to-side difference	0.56 ± 0.37 (1.3)		

Data are presented as means ± standard deviation (SD)
The tibial H-reflex was elicited by submaximal intensity of stimulus over the posterior tibial nerve at the knee with recording over the soleus distally measured half the distance from the stimulation point to the medial malleolus.
The median and ulnar H-reflex was elicited in infants with proportions showing a response shown in parenthesis.
Side-to-side difference shows mean ± SD (upper limits of normal)
Source: Ref. 9.

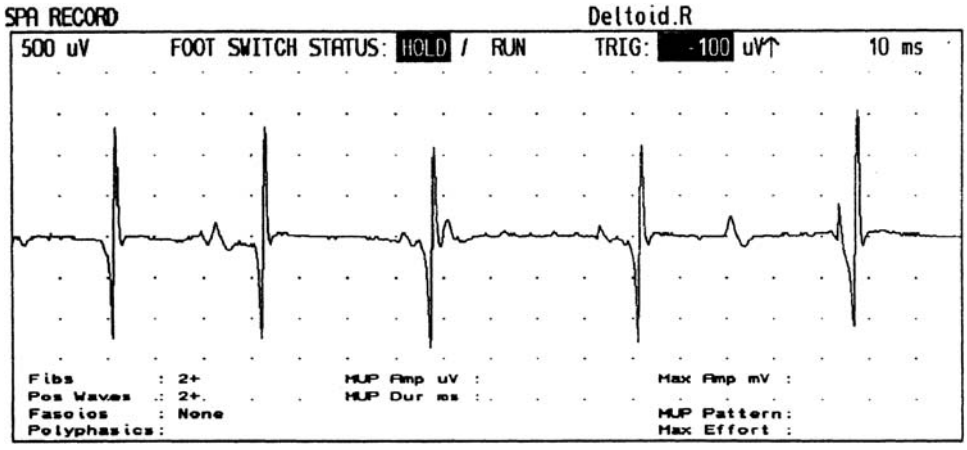


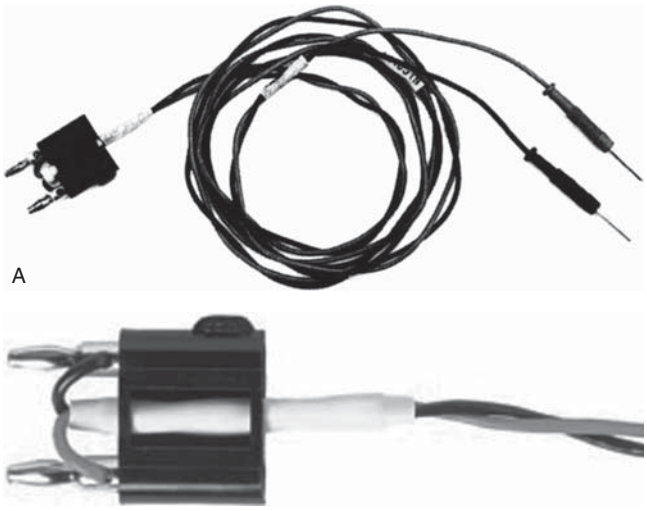
Figure 7.1 Neuropathic recruitment of the deltoid in a 12-month-old child with a brachial plexus injury sustained at birth. The initial recruited motor unit action potential is 2,500 μV, and it is firing at 25 Hz.

cathode, with a more proximal surface anode in close proximity. For example, for ulnar orthodromic sensory studies, the author has utilized ring electrodes on the fifth digit and recording electrodes over the ulnar nerve at the elbow. Generally, a standard bipolar stimulator may be utilized for children 6 months of age and older.

Recording Electrodes

Sensory Conduction

Generally, sensory nerve action potentials are easily recorded in newborns. The standard ring electrodes, needle recording electrodes, and/or pediatric-size finger-clip electrodes may be used. While for adults, a



A

B

Figure 7.2 Pediatric nerve stimulator (A). The interelectrode distance between cathode and anode is less than 2 centimeters (B).

4-cm interelectrode distance is optimal, this is not possible in small children. Hence, the pediatric electrodiagnostic clinician should attempt to obtain as much distance as possible between active and reference electrodes. Every attempt should be made to obtain at least a 2-cm interelectrode distance. Stimulation of the digits, palm, or wrist, with electrodes located more proximally at the elbow for median and ulnar sensory studies provide longer distance and less measurement error. In general, normative data for sensory nerve conduction velocities are more readily available than normative data for distal latencies at specific distances.

Motor Conduction

Generally, standard 6-mm silver disc surface electrodes are used as active and reference electrodes for motor conduction studies. Some electrode diagnosticians prefer the use of ring electrodes on digits as the reference electrode and a standard surface electrode over the Moro point at the muscle as the active electrode (Fig. 7.3). Often, 4–6-cm distances are used from the stimulator to active electrode. Conduction velocities and CMAP amplitudes are generally more relevant data in infants than motor distal latencies because distal nerve entrapments are rare. Thus, the distances used from distal stimulation to active electrode are less critical.

Special Considerations for Nerve Conduction Studies

The best normative data for pediatric nerve conduction studies are available for the median, ulnar,

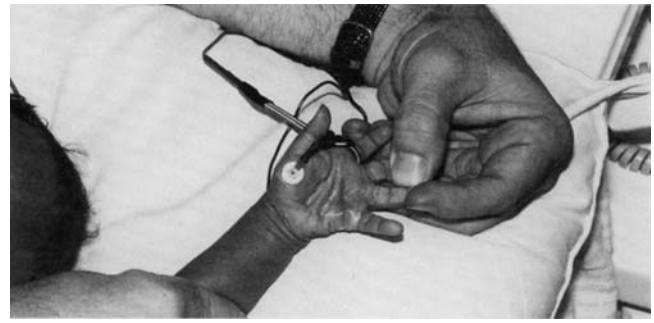


Figure 7.3 Recording electrodes for a median motor nerve conduction study in a small child. The active electrode is placed over the abductor pollicis brevis on the thenar eminence. The recording electrode is a ring electrode placed on the index finger. The ground electrode is a 6-millimeter silver disc electrode placed on the back of the hand.

peroneal, tibial, facial, and phrenic motor nerves and the median, ulnar, and sural sensory nerves. Stimulation of the posterior tibial nerve (recording abductor hallucis brevis) produces a discrete CMAP more commonly than stimulating the peroneal nerve (recording over extensor digitorum brevis). The extensor digitorum brevis (EDB) muscle may be difficult to visualize or palpate in infants. Its CMAP configuration frequently has either an initial positivity or a low broad configuration. In addition, the CMAP amplitude may change substantially with slight changes in position for the active electrode over the extensor digitorum brevis.

The axillary and musculocutaneous motor nerve conduction studies may be helpful in the setting of infantile brachial plexopathy. Care should be taken to minimize volume conduction. Often, the intact side is used for amplitude comparisons.

Evaluations of proximal nerves, such as the axillary spinal accessory musculocutaneous and femoral, are often useful in the evaluation of severe demyelinating neuropathies (Fig. 7.4). The distal latencies of these nerves may be severely prolonged on the setting of severe reductions in the CMAPs of more distal nerves due to conduction block or axon loss.

Percutaneous stimulation of the phrenic nerve is performed with techniques similar to that utilized in the adult, with stimulation performed at the posterior border of the sternocleidomastoid at the level of the thyroid cartilage or alternatively just medial (or occasionally lateral) to the sternal head of the sternocleidomastoid. Recording electrodes may be placed in the fifth to sixth intercostal space 2 cm apart at the anterior axillary line, or alternatively an active electrode may be placed immediately below the costal margin at the level of the nipple with recording electrode at the xiphoid. The active electrode may need to be moved to adjacent

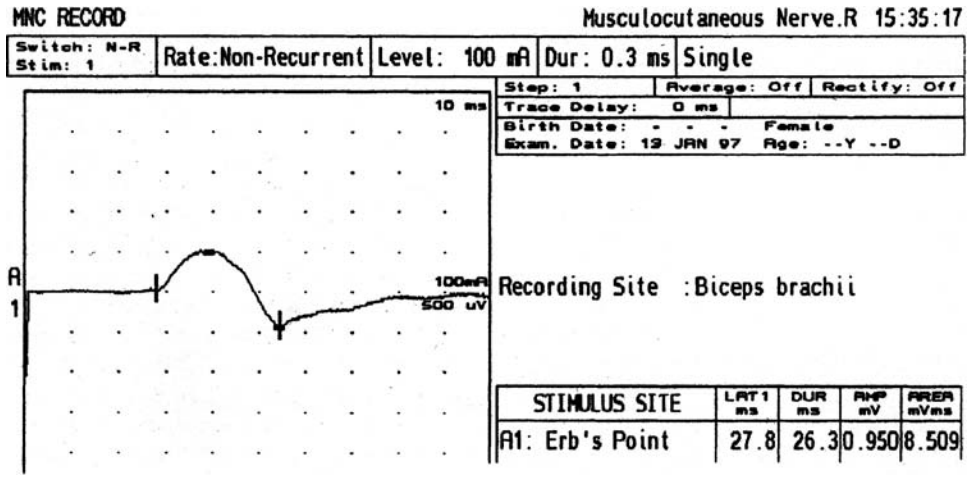


Figure 7.4 Nerve conduction study of the musculocutaneous nerve in Charcot-Marie-Tooth (CMT) type III. The nerve is stimulated at Erb's point and the recording electrode is placed over the biceps brachii. Distal latency is severely prolonged at 27.8 milliseconds. Note the reduced compound muscle action potential amplitude, presumably due to conduction block, and the relative lack of temporal dispersion, which is frequently seen in CMT.

positions to obtain an optimal M-wave (Fig. 7.5). Normative values for phrenic latencies have been reported in children (26,27). The author prefers to use ultrasound visualization of the diaphragm simultaneously with phrenic nerve stimulation to confirm downward deflection of the diaphragm. Volume conduction to the long thoracic nerve may produce a CMAP from the serratus anterior rather than the diaphragm. The downward deflection of the diaphragm spontaneously and with electrical stimulation may be confirmed and distance of diaphragmatic excursion quantitatively measured by ultrasound M-mode.

Repetitive Nerve Stimulation Studies

Every attempt should be made to stabilize the extremity with an infant- or pediatric-size arm board. The author prefers to use a block electrode or surface cathode and anode electrodes taped over the nerve as opposed to a handheld stimulator. This helps standardize each stimulation during a train of 5 stimuli at low or high rates of stimulation. In newborns, the author prefers to stimulate the median or ulnar nerve at the elbow to minimize shock artifact. Care should be taken to obtain a stable baseline between stimulations in a train. Decrements or increments in amplitude should be accompanied by similar decrements or increments in area. If no concomitant area changes occur, technical factors (changing baseline or changing temporal dispersion) may explain a decrement or increment in amplitude.

TECHNICAL FACTORS OF NEEDLE ELECTROMYOGRAPHY

Electrodes

Generally, 26–28-gauge Teflon-coated monopolar electrodes, usually 25 mm in length, are utilized. Some laboratories routinely use disposable concentric facial needle electrodes. These electrodes have smaller calibrated recording areas and hence, provide more stability of MUAP configuration. In addition, concentric needle electrodes are more sensitive to changes in duration and amplitude than monopolar needle electrodes. Use of smaller electrodes (either small monopolar needles or small-diameter concentric needle electrodes originally designed for the examination of adult facial muscles) provides considerable psychological advantages in children of sufficient developmental age to associate needles with pain. The instrumentation utilized for needle EMG of children is essentially the same as that used in adults. In the intensive care unit, electrical interference may necessitate the use of either a facial concentric needle or a needle reference electrode. Long electrodes or long electrode leads can create problems with ambient electrical interference.

Optimal Muscles to Study for Rest Activity

In evaluating an infant or young child for a generalized disorder, specific muscles are chosen to permit

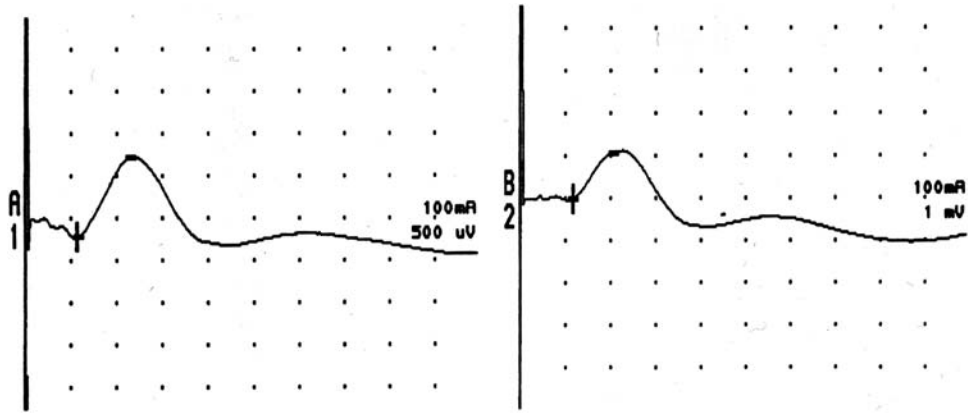


Figure 7.5 Phrenic nerve conduction study in a 13-year-old child with C2 traumatic spinal cord injury. A1 is the compound muscle action potential (CMAP) amplitude obtained on the right side and B2 is the CMAP obtained on the left side. Latencies are approximately 5 milliseconds and amplitudes from baseline to peak 1 mV. The viability of the phrenic nerves allowed placement of a phrenic nerve–diaphragm pacer for ventilation.

evaluation of insertional and spontaneous activity. The distal hand (first dorsal interosseous) and foot muscles of infants usually have minimal voluntary activity due to immature motor control at this developmental age, making them good sites to assess spontaneous activity. In addition, extensor muscles such as the vastus lateralis and gastrocnemius in the legs and the triceps in the upper extremities are useful sites for the evaluation of insertional and spontaneous activity.

In the neonate and young infant, foot and hand intrinsic muscles exhibit high levels of end-plate noise because of the relatively larger end-plate area in the immature muscle. This end-plate activity may be confused with fibrillation potentials. Fibrillation potentials and positive sharp waves are not typically observed in the full-term normal newborn.

Optimal Muscles for Evaluation of Recruitment, Motor Unit Configuration, and Interference Pattern

In general, flexor muscles such as the tibialis anterior and the iliopsoas are useful for the evaluation of MUAPs and recruitment in the lower extremity. These muscles can be activated by tickling or pinching the bottom of the foot, producing a withdrawal response. In the upper extremity, the flexor digitorum sublimis and biceps muscles are often reflexively activated by the newborn or young infant. More proximal muscles can be activated by moving the extremity or positioning it to produce antigravity stabilization of the limb by the firing of proximal musculature. Alternatively, reflex posturing techniques such as the Moro response can be used to activate the shoulder abductors, but are usually not necessary.

Sedation

Pediatric physiatrists and neurologists performing pediatric electrodiagnostic evaluations have noted that extreme behavioral distress most frequently occurs among 2–6-year-olds (28,29). Pain medications are occasionally or always prescribed by 50% of pediatric electromyographers (29). General anesthesia is occasionally utilized by 25% of electrodiagnostic practitioners (29). One study demonstrated that children exhibiting more behavioral distress during pediatric electrodiagnostic evaluations were younger, had been uncooperative with previous painful procedures, were more likely to have had more negative medical/dental experiences, and had mothers who themselves reported greater fear and anxiety about undergoing EMG/nerve conduction studies (28).

While some electromyographers never utilize sedation, there has been more interest in the use of analgesia, conscious and deep sedation, and, more recently, general anesthesia with propofol or inhalational anesthetics. Traditional sedative choices include chloral hydrate (50–100 mg per kg), "DPT" (meperidine hydrochloride, phenylephrine hydrochloride, and chlorpromazine), and midazolam hydrochloride nasal spray. EMLA cream (lidocaine 2.5% and prilocaine 2.5%) has been used during electromyographic evaluations as a topical anesthetic (30). Mean duration of topical application in infants or older children was 45–145 minutes. Greater pain relief was obtained with

use of EMLA over the extensor forearm than the thenar eminence.

While general anesthesia is usually not necessary, the author has increasingly involved critical care and anesthesia colleagues who have utilized either propofol (2,6-diisopropylphenol), an intravenous sedative–hypnotic agent or inhalational anesthetics with laryngeal mask anesthesia (LMA) airways for the electrodiagnostic evaluation of 18-month-old to 6-year-old children who exhibit substantial behavioral distress during an initial attempt at an electrodiagnostic evaluation without sedation. Propofol produces rapid onset of anesthesia (in 1–3 minutes), and sedation is maintained by either a continuous infusion or multiple boluses. Subjects usually awaken in less than 10 minutes of the time the infusion is discontinued. Sedation, analgesia, and particularly general anesthesia have inherent risks and require appropriate monitoring. Propofol should be administered by an anesthesiologist or pediatric intensivist prepared to bag-mask ventilate or intubate the child if necessary. Adequate monitoring generally requires a sedation suite, pediatric intensive care unit (ICU), recovery room, or operating room. The author typically obtains all nerve conduction studies and a thorough examination of multiple muscle sites for abnormal spontaneous rest activity while the subject is deeply sedated or anesthetized with propofol. The level of sedation is then titrated to a point where appendicular movement is elicited with needle insertion or stimulation of the extremity. At this point, under lighter sedation, recruitment pattern and motor unit configuration are assessed. As the child awakens, interference pattern is evaluated with more vigorous motor activity. Children are usually amnestic to the EMG examination subsequent to propofol anesthesia.

The cost of anesthesia must be weighed against the importance of the acquisition of a thorough, technically precise, and accurate electrodiagnostic evaluation. An EMG obtained under anesthesia usually provides a suboptimal evaluation of motor unit configuration, recruitment pattern, and interference pattern, with maximal effort but better evaluation of quiet muscle for spontaneous activity and a more comprehensive acquisition of nerve conduction studies and repetitive nerve stimulation studies.

The key to successful data acquisition in most pediatric electrodiagnostic evaluations remains a well-organized, well-planned approach with distinct diagnostic questions prospectively considered. If the examination is planned to answer a specific question, it is usually possible to proceed expeditiously, completing the examination within a reasonable time (30 minutes). As children approach 6 years of age, it becomes easier to talk them through an evaluation and elicit their participation and cooperation.

Nerve conduction studies are usually better tolerated than needle electromyography, and many pediatric electromyographers perform the nerve conduction studies first. Increased behavioral distress subsequent to a needle examination makes the motor nerve conductions, and particularly the sensory nerve conduction studies, technically difficult due to excessive EMG background noise.

Limitations of Single-Fiber EMG

While normative data for fiber density, mean consecutive difference, and jitter have been reported for different muscles among different pediatric age groups (31), this procedure is difficult to use in younger children with limited ability to cooperate. Alternatively, a stimulated single-fiber EMG study may be obtained under general anesthesia in those suspected of a congenital myasthenic syndrome, and this technique has yielded excellent sensitivity and specificity for identification of a neuromuscular transmission disorder (32–34).

SPECIFIC CLINICAL PROBLEMS IN PEDIATRIC ELECTRODIAGNOSIS

Electrodiagnostic Evaluation of the Floppy Infant

The most common referral for an electrodiagnostic examination in the infant is generalized hypotonia. The most common etiology for infantile hypotonia is central, accounting for approximately 80% of cases. A differential diagnosis of infantile hypotonia is shown in Table 7.8 (35). Electrodiagnostic abnormalities in selected conditions producing infantile hypotonia are shown in Table 7.9.

Neurogenic causes of generalized weakness in infants are more accurately diagnosed with electrodiagnostic studies than are myogenic causes (36–38). A study of the predicted value of the electrodiagnostic examination in the hypotonic infant showed that electrodiagnostic studies accurately predicted the diagnosis in 65% of infants with spinal muscular atrophy and only 10% of infants with myopathy. Seventy-five percent of the electrodiagnostic studies performed on infants with documented myopathies were considered normal (39). The sensitivity of EMG improves after age 2 (38).

In arthrogryposis multiplex congenita and hypotonia, neither muscle biopsy nor NCS/EMG alone had consistently high sensitivities, positive

7.8 Differential Diagnosis of Infantile Hypotonia

Cerebral hypotonia
Chromosome disorders
- Trisomy
- Prader-Willi syndrome

Static encephalopathy
- Cerebral malformation
- Perinatal CNS insult
- Postnatal CNS insult

Peroxisomal disorders
- Cerebrohepatorenal syndrome (Zellweger syndrome)
- Neonatal adrenoleukodystrophy

Inborn errors of metabolism
- Glycogen storage disease type II (Pompe disease)
- Infantile GM1 gangliosidosis
- Tay-Sachs disease (infantile GM2 gangliosidosis)
- Vitamin-dependency disorders

Amino acid and organic acid disorders
- Maple syrup disease
- Hyperlysinemia
- Nonketotic hyperglycinemia
- Propionyl-CoA carboxylase deficiency

Other genetic disorders
- Familial dysautonomia
- Cohen syndrome
- Oculocerebrorenal syndrome (Lowe)

Benign congenital hypotonia

Spinal cord
Trauma (obstetrical, postnatal)
- Hypotonia early with acute paraplegia
- Hypertonia

Tumor or AVM
- Hypertonia may occur later or with slow-growing tumor

Anterior horn cell
- Spinal muscular atrophy type I (Werdnig-Hoffman)
- Spinal muscular atrophy type II
- Distal SMA with vocal cord paralysis and diaphragm weakness
- Poliomyelitis
- Neurogenic arthrogryposis

Polyneuropathies
Congenital hypomyelinating neuropathy
Chronic inflammatory demyelinating polyneuropathy
Acute inflammatory demyelinating polyradiculoneuropathy (Guillain-Barre syndrome)
Hereditary motor-sensory neuropathies
- Dejerine Sottas
- Congenital hypomyelinating neuropathy

Toxic polyneuropathy
Leukodystrophies (Krabbe's, Nieman-Pick)
Leigh's syndrome
Giant axonal neuropathy
Dysmaturation neuropathy

Neuromuscular junction
Presynaptic
- Infantile botulism
- Hypermagnesemia—eclampsia
- Aminoglycoside antibiotics
- Congenital myasthenia
- Choline acetyltransferase (CHAT) deficiency
  - Paucity of acetylcholine synaptic vesicles
  - Congenital Lambert-Eaton–like syndrome
- Decreased quantal release

Synaptic basal lamina defects
- Congenital myasthenic syndrome
  - Endplate acetylcholinesterase (AChE) deficiency

Postsynaptic
- Neonatal (autoimmune)
- Congenital myasthenia
- AChR disorders involving α, β, δ, ϵ receptor subunits
  - AChR deficiency causing kinetic abnormalities in function
  - AChR slow-channel syndromes
  - AChR fast-channel syndromes
  - Endplate rapsyn deficiency

Myopathies
Congenital myopathies
- Nemaline rod
- Central core
- Myotubular (centronuclear)
- Mini-core (multi-core)
- Congenital fiber type disproportion

Congenital myotonic dystrophy (DM1)
Congenital muscular dystrophy
- Fukuyama type (CNS involvement)
- Merosin deficiency (with or without CNS involvement)
- Ullrich'e congenital muscular dystrophy (collagen VI deficiency, scleroatonic)
- Congenital muscular dystrophy with early spine rigidity
- Muscle-eye-brain disease
- Walker-Warburg syndrome
- Undifferentiated

Inflammatory myopathies
- Infantile polymyositis

Metabolic myopathies
- Acid maltase deficiency (type II)
- Muscle phosphorylase deficiency (type V)
- Phosphofructokinase deficiency (type VII)
- Cytochrome c oxidase
- Carnitine deficiency

Endocrine myopathies
- Hypothyroidism

Hypoparathyroidism

AVM, arteriovenous malformation; CNS, central nervous system; SMA, spinal muscular atrophy.

7.9 Infant Hypotonia: Electrodiagnostic Abnormalities

DIAGNOSIS	MOTOR CONDUCTION	SENSORY CONDUCTION	SPONTANEOUS ACTIVITY	MOTOR UNITS
SMA	Decreased amplitude; may show decreased velocity	Normal	Fibrillation ±; spontaneous rhythmic motor unit firing	Decreased number; may show mild increase in amplitude, duration
HSMN III	Markedly prolonged	Prolonged or absent	0	Reported normal
Hypomyelinating neuropathy	Markedly prolonged; markedly decreased amplitude	Prolonged or absent	0	Reported normal or increased amplitude
Inflammatory polyneuropathy	Decreased amplitude; possibly decreased velocity; conduction block	±	Fibrillation may be present	Decreased number
Botulism	Decreased amplitude; normal velocity; decremental response to MNCV; facilitation > 20 Hz	Normal	Fibrillations	Decreased amplitude, duration
Spinal cord injury	Normal motor velocity and amplitudes if nerves tested are not originating from area of injury; F-wave or H-reflex may be prolonged or absent	Normal	Fibrillations may be present in muscles innervated at level of injury	Decreased number at involved muscles; poor motor control below level of injury
Congenital myopathy	Normal velocity; amplitude may be decreased	Normal	Fibrillations may be present (in congemital myotubular myopathy)	Normal to decreased amplitude, durartion; increased polyphasisity
Congenital myotonic dystrophy	Normal	Normal	Absent or few fibrillations	Poor activation; likely normal
Glycogen storage disease	Normal	Normal	Fibrillations (in types II, V, VII); frequency varying; trains of positive waves	Decreased amplitude, duration
Metachromatic leukodystrophy	Decreased velocity; decreased aplitude	Slowed		

HSMN, hereditary sensory motor neuropathy; MNCV, motor nerve conduction velocity; SMA, spinal muscular atrophy.
Source: Adapted from Turk MA. Pediatric electrodiagnostic medicine. In: Dumitru D, ed. *Electrodiagnostic Medicine.* Philadelphia: Hanley & Belfus;1995, 1133–1142.

predictive values, or specificities (40). When the clinical evaluation indicates a specific syndromic, developmental, or exogenous cause, NCS/EMG and muscle biopsy are not helpful and may not need to be performed. When the history, examination, and genetic evaluation are unrevealing, NCS/EMG and muscle biopsy together provide valuable diagnostic information.

In the evaluation of hypotonia, a complete electrodiagnostic evaluation is useful, including motor and sensory nerve conduction studies and appropriate needle examination with the highest yield muscles examined initially, and, if necessary, repetitive nerve stimulation. It should be emphasized that nerve conduction studies and electromyography are an extension of the clinician's physical examination. Electrodiagnostic findings need to be interpreted in light of clinical examination findings. Care should be taken not to overinterpret subtle findings on needle electromyography. Low-amplitude, short-duration, polyphasic motor unit action potentials, which would be considered myopathic in adults, may be normal in young children. Motor unit amplitudes and durations may be reduced in the normal young child and

mistaken for myopathic MUAPs. End-plate noise, abundant in the small intrinsic muscles of the hand and foot, may be difficult to distinguish from fibrillation potentials. Thus, borderline findings on needle EMG should not be overinterpreted in the infant and young child.

Parents should be cautioned prior to an electrodiagnostic evaluation that definitive diagnostic information is often not obtained and the results may help guide further diagnostic studies. For example, results from EMG may help to guide further studies such as muscle biopsy by providing information about the most appropriate muscle site for the biopsy. With spinal muscular atrophy, an electrodiagnostic evaluation can allow the clinician to defer a muscle biopsy and proceed with molecular genetic studies of the survival motor neuron (SMN) gene. Often, the SMN gene test is ordered prior to any electrodiagnostic studies being performed, so fewer studies have been performed on this population over the past decade. Electrodiagnostic studies in patients with hereditary motor sensory neuropathy help to categorize the neuropathy as either primarily demyelinating or axonal, and such information may help focus subsequent molecular genetic analyses. In general, nerve conduction and electromyography still provide a useful tool for the localization of lesions within the lower motor neuron, but fewer studies have been required as genetic studies have become commercially available.

Differential Diagnosis for Early Respiratory Distress in Infancy

The differential diagnosis of lower motor neuron disorders with perinatal respiratory distress is fairly limited. Generally, respiratory distress within the first few days of life can be seen in spinal muscular atrophy type I, congenital hypomyelinating neuropathy, congenital myasthenia, transient neonatal myasthenia, congenital myotonic muscular dystrophy, neurogenic arthrogryposis, and x-linked myotubular myopathy. These disorders are easily differentiated with electrodiagnostic studies and, in some instances, molecular genetic findings. For example, congenital myotonic muscular dystrophy may be definitively diagnosed with molecular genetic studies at the chromosome 19q13.3 locus. In congenital hypomyelinating neuropathy, sensory conduction abnormalities are unrecordable and motor nerve conduction velocities are markedly slowed (2–5 m/s) with temporal dispersion and low-amplitude evoked potentials (Fig. 7.6). Spinal muscular atrophy (SMA) patients show normal sensory conductions, decreased CMAP amplitudes, occasional fibrillations, and decreased numbness of MUAPs. Congenital myasthenia patients show normal sensory conductions, normal motor nerve conduction velocities, and abnormalities on repetitive nerve stimulation studies. X-linked myotubular myopathy patients show profuse fibrillations and myopathic MUAPs on EMG, and diagnosis is confirmed by muscle biopsy.

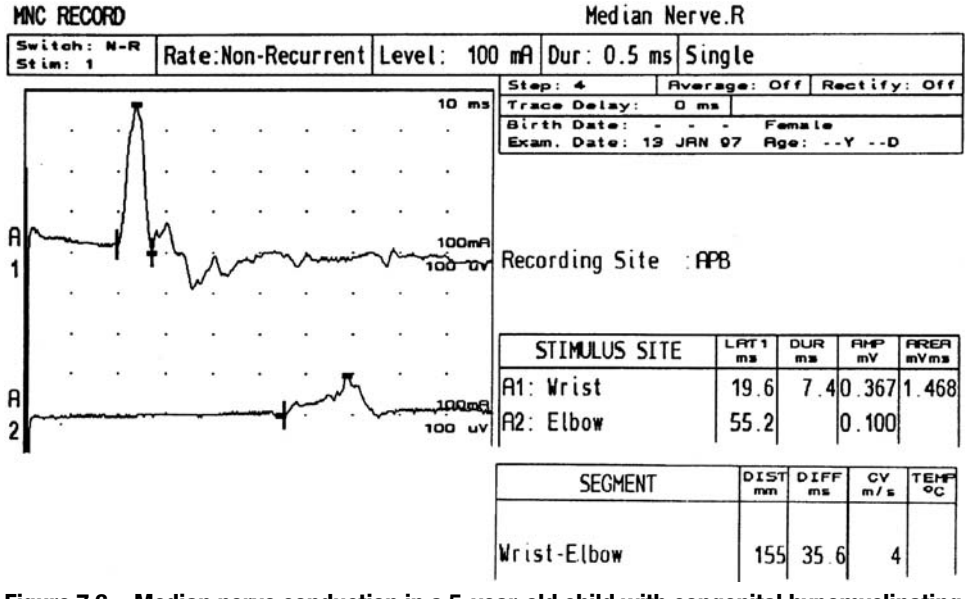


Figure 7.6 Median nerve conduction in a 5-year-old child with congenital hypomyelinating neuropathy documented by sural nerve biopsy and molecular genetic studies of the EGRF 2 gene. Distal latency is markedly prolonged at 19.6 milliseconds. There is reduced compound muscle action potential amplitude, at 0.367 mV, conduction block (note the drop in amplitude from distal to proximal), and conduction velocity at 4 m/s.

Acute Onset Infantile Hypotonia

Acute onset hypotonia in a previously normal infant should warrant an evaluation to rule out acute inflammatory demyelinating polyneuropathy (AIDP), infantile botulism, infantile polymyositis, an infantile form of myasthenia, a toxic process, or acute onset myelopathy. Repetitive motor nerve stimulation studies should be performed under the following circumstances: 1) there is constipation, bulbar involvement, and/or respiratory distress; 2) an infant presents with ptosis or extraocular muscle weakness; 3) CMAP amplitudes are severely reduced; 4) "myopathic" MUAPs are present; 5) a repetitive CMAP is observed after single supramaximal stimulation on routine nerve conduction study, suggestive of a diagnosis of congenital myasthenia with congenital acetylcholinesterase (AChE) deficiency or classic slow channel syndrome.

Motor Neuron Disorders

Spinal muscular atrophy (SMA) is perhaps the most common lower motor neuron disorder causing infantile hypotonia. The predictive value of needle EMG in the diagnosis of SMA has been established (36–39), but the need for electrodiagnostic studies has diminished over the years, given the 95% or greater sensitivity of SMN gene studies. As SMA remains an important consideration in infantile hypotonia, a review of the elecrodiagnostic findings is useful.

The findings in this motor neuron disorder have largely been consistent with motor axonal loss, denervation, and (among persons less severely affected) reinnervation. Traditional electrodiagnostic criteria for motor neuron disease are not suitable for patients with childhood SMA. For example, Buchthal (41) found that many infants with SMA did not meet strict criteria for motor neuron disease. If clinical findings suggest SMA, study of at least two muscles innervated by different nerve roots and peripheral nerves in at least three extremities is indicated (42). In the infant, spontaneous activity may be more readily determined with study of muscles that are not as commonly recruited, such as the vastus lateralis, gastrocnemius, triceps, and first dorsal interosseous. Recruitment and motor unit characteristics can be assessed in muscles that are readily activated, such as the anterior tibialis, iliopsoas, biceps, and flexor digitorum sublimis (42). The paraspinals are usually not studied due to poor relaxation, and the experienced pediatric electrodiagnostic medicine consultant usually defers needle evaluation of the tongue in the hypotonic infant.

Although some authors (43) have described high-density fibrillation potentials in infants with poorer outlook, most studies have not demonstrated abundant fibrillation potentials in the infantile form (42,44,45). In SMA III, the incidence of fibrillation potentials ranged from 20% to 40% in one series (46) to 64% in another (47). The incidence of fibrillation potentials in SMA type III does not approach the level seen in SMA type I. In addition, spontaneous activity has been more frequently observed in the lower extremities than upper limbs and proximal more than distal muscles in SMA type III (46). The degree of spontaneous activity has not been found to be independently associated with a worse prognosis in SMA (39). Fasciculations are uncommonly observed in SMA type I and appear more commonly in SMA types II and III (42,43,45). In younger patients, fasciculations are difficult to distinguish from spontaneously firing MUAPs. In relaxed muscles, some motor units exhibit a spontaneous rhythmic firing (43, 44, 45).

Voluntary MUAPs frequently fire with an increased frequency, although recruitment frequency may be difficult to determine consistently in infants. Compared to age-matched norms, MUAPs show longer duration, particularly in older subjects, and higher amplitude; however, a bimodal distribution may be seen with some concomitant low-amplitude short duration potentials (44). Large-amplitude, long-duration MUAPs may be absent in many infants with SMA type I but more commonly observed in SMA types II and III (42). The percentage of large-amplitude MUAPs increases with the duration of the disease (46). Other signs of reinnervation, such as polyphasic MUAPs, may be observed in more chronic and mild SMA. These polyphasic MUAPs may include late components such as satellites or linked potentials. There may also be temporal instability of the waveform observed in individual MUAPs. Reduced recruitment (an incomplete interference pattern) with maximal effort is perhaps the most consistent finding in all SMA types (Fig. 7.7). In one series (39), the amplitude of MUAPs and degree of decrement in recruitment pattern were not individually associated with worse prognosis.

Motor nerve conduction velocities and CMAP amplitude have been shown to be reduced in many patients with infantile SMA. The degree of motor conduction slowing (if present) tends to be mild and greater than 70% of the lower limit of normal (45,47–50). Reduction of motor conductions to less than 70% of the lower limit of normal is described as an exclusionary criterion for SMA (51). The mild slowing of motor conductions is present to the same degree over distal and proximal segments as determined by M- and F-waves responses (49). The slowing of conduction is generally seen in those with correspondingly low-amplitude CMAPs and is thought to be due to selective loss of the fastest conducting fibers from large motor units. Alternatively, arrested myelination in utero has been proposed to explain this slowing in motor conduction noted in some SMA cases at birth (39). Survival has

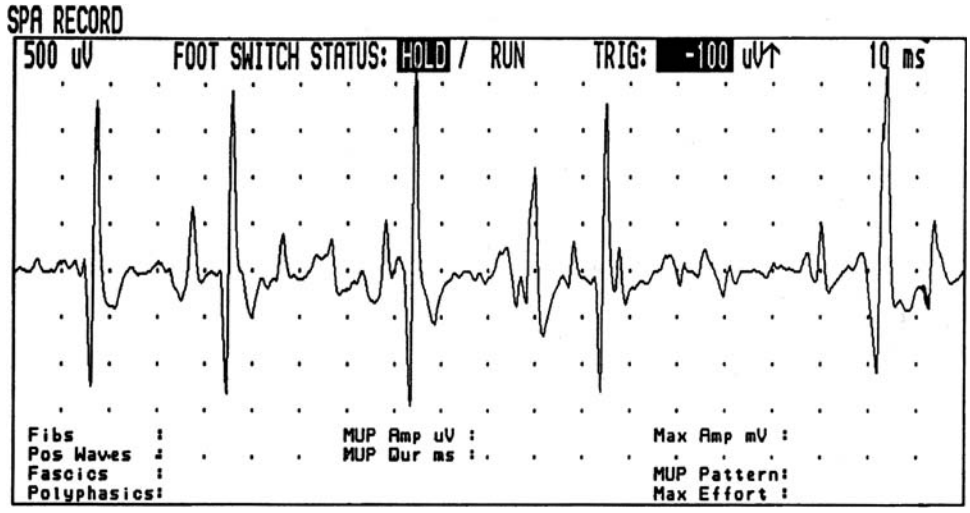


Figure 7.7 Incomplete or reduced interference pattern in spinal muscular atrophy type II. Note the large amplitude motor unit action potential (3,000 μV) firing at 25 Hz.

been found to be longer for those SMA infants with normal motor conduction velocities over a distal segment (39). Significant reductions in CMAP amplitudes have been frequently reported in SMA types I–III (39,42,47). Kuntz (47) reported a tendency toward greater reductions in CMAP amplitude among patients with earlier age of onset and shorter survival.

Sensory nerve conduction studies (NCSs) in SMA show essentially normal sensory conduction velocities and sensory nerve action potential (SNAP) amplitudes. Significant abnormalities in sensory studies exclude a diagnosis of SMA (51), while minor abnormalities in sensory conduction velocities have infrequently been noted in SMA (48,52,53). Such rare sensory abnormalities have not been reported in SMA patients with diagnostic confirmation by molecular genetic studies.

Spinal Cord Injury

Neonatal spinal cord injury may occur as an obstetrical complication or as a result of a vascular insult to the spinal cord. Typical clinical presentation may include findings of diffuse hypotonia, possible respiratory distress, hyporeflexia, and urinary retention. An anterolateral spinal cord injury due to a vascular insult will produce EMG findings of severe denervation in diffuse myotomes. Typically, two to three weeks may lapse before fibrillations and positive sharp waves are elicited. Anterior horn cell and axonal degeneration will typically result in decreased CMAP amplitudes in multiple peripheral nerves. SNAP amplitudes are spared. Somatosensory-evoked potentials may be spared if posterior columns are preserved.

Traumatic spinal cord injury often results in loss of anterior horn cells at a specific "zone of injury." For example, a child with C5 tetraplegia may have denervation present at the bilateral C6 and C7 myotomes. This zone of partial or complete denervation becomes particularly relevant in the evaluation of a patient for possible placement of an implanted functional electrical stimulation system for provision of voluntary grasp and release. Presence of denervation necessitates concomitant tendon transfers with electrical stimulation of the transferred muscle group.

SSEPs may help establish a sensory level in an infant or young child with spinal cord injury, and is also useful in the evaluation of the comatose or obtunded child at risk for spinal cord injury without radiographic abnormality (SCIWORA). Somatosensory evoked potentials are discussed in a following section.

Transcranial electric motor evoked potentials (MEPs) to monitor the corticospinal motor tracts directly are now used routinely in addition to SSEPs for detection of emerging spinal cord injury during surgery to correct spine deformity or resect intramedullary tumors (54–56). Afferent neurophysiological signals can provide only indirect evidence of injury to the motor tracts since they monitor posterior column function. Transcranial electric motor evoked potentials are exquisitely sensitive to altered spinal cord blood flow due to either hypotension or a vascular insult. Moreover, changes in transcranial electric MEPs are detected earlier than are changes in SSEPs, thereby facilitating more rapid identification of impending spinal cord injury.

Brachial Plexus and Cervical Nerve Root Lesions

Traumatic obstetrical brachial plexopathy usually results from traction on the brachial plexus (predominantly

upper trunk) and its associated spinal roots. This can lead to stretching or rupture of the trunks of the plexus and/or partial axonotmesis or avulsion of the spinal roots. The most common cause is a shoulder dystocia of the anteriorly presenting shoulder causing excessive lateral neck traction. Injury to the upper trunk of the brachial plexus and/or C5–6 cervical roots is the more common injury known as Duchenne-Erb's palsy. Damage to the lower trunk and/or C8–T1 cervical roots is referred to as Klumpke's palsy. Severe brachial plexus injuries may involve the entire plexus and C5–T1 nerve roots diffusely. A Horner's syndrome due to injury of the C8 and T1 roots and the superior cervical sympathetic ganglion may be an associated clinical finding. An isolated Klumpke's palsy is rare in the setting of traumatic birth palsy and usually results from a fall onto a hyperabducted shoulder, penetrating trauma, or tumor.

Electrodiagnostic studies help determine the location (root and/or plexus), extent, and severity of the brachial plexus injury. Examination should be deferred until at least three to four weeks after the injury to allow for abnormal spontaneous rest activity (fibrillations and positive sharp waves) to develop in the setting of denervation and axon loss (Fig. 7.8). Complete injuries are characterized electromyographically by absent MUAPs and absent CMAP amplitudes in peripheral nerves supplied by the transected axons. In the setting of total motor paralysis, motor nerve conduction studies with measurement of the amplitude of the CMAPs in distal and proximal muscles provides useful prognostic information. For example, the preservation of the CMAP amplitude 10 days or more after the injury with complete clinical paralysis suggests that the damage is, in part, a neuropraxic injury with better prognosis. In this setting, F-waves are absent. If motor function is absent and no MUAPs are observed, examination of the amplitude of the sensory nerve action potentials in the dermatomal distribution of the branches of the affected brachial plexus trunks

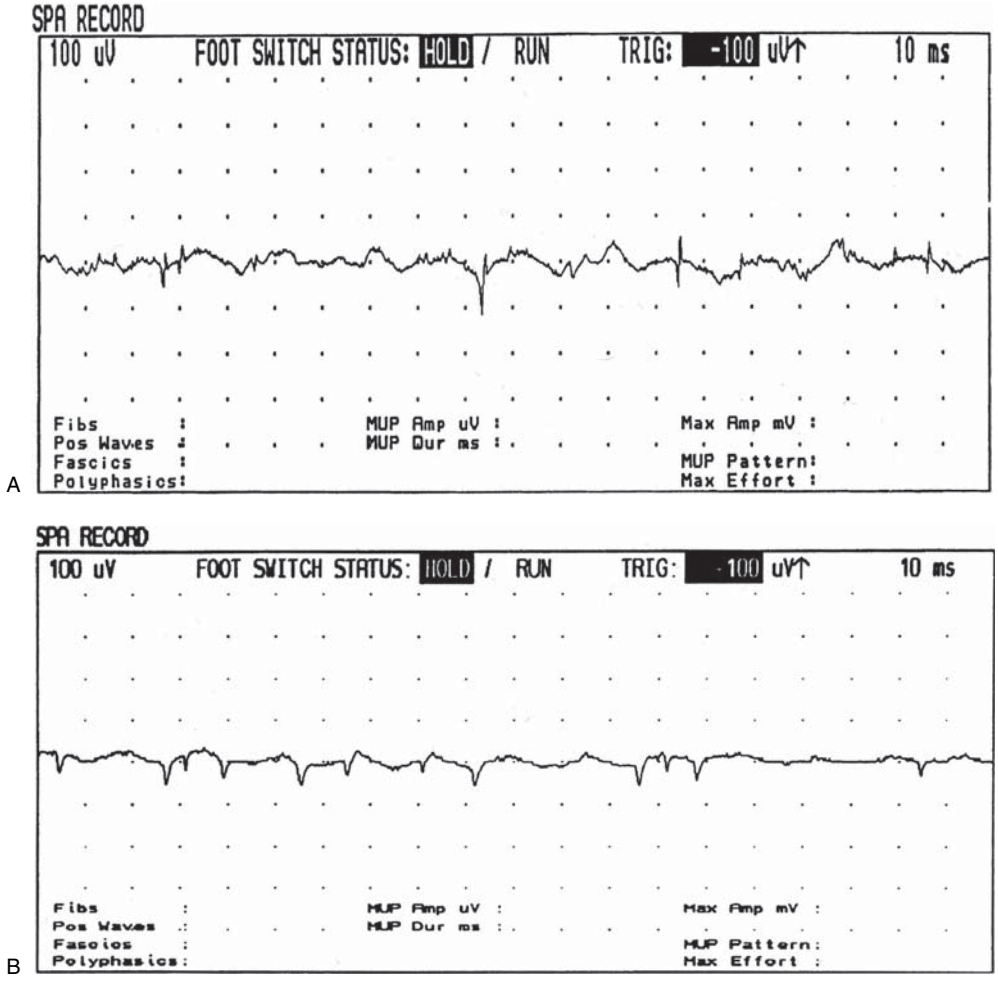


Figure 7.8 Fibrillation potential (A) and positive sharp waves (B) indicative of acute denervation and axon loss.

can help distinguish injuries to the plexus from severe cervical root injuries or avulsions. The sensory dorsal root ganglion lies in the intervertebral foramen distal to the damaged segment with a root injury, leaving the sensory axon projection from the dorsal root ganglion to the limb intact. Thus, the sensory nerve action potential is obtainable in the setting of a root avulsion with absent clinical sensation.

In the setting of Erb's palsy, assessment of a superficial radial sensory or median sensory response to the index finger is useful in making a distinction between a C6 root avulsion and a more distal lesion involving the trunk of the brachial plexus. The median SNAP to the middle finger provides information about the integrity of C7 axon projections distal to the dorsal root ganglion. The presence or absence of an ulnar sensory nerve action potential can help distinguish a lower trunk injury from a C8 nerve root injury.

In perinatal traumatic brachial plexopathy, positive sharp waves and fibrillations, indicative of true denervation, can be found by 14 to 21 days after injury (57). Absence of fibrillations or positive sharp waves after this time frame suggests a neuropraxic lesion with intact axons. In this setting, the prognosis for recovery is favorable. Early in the course of recovery prior to reinnervation, interference pattern usually is reduced or discrete and recruitment frequencies increased into the neuropathic range (often >20 Hz). A follow-up needle EMG evaluation three to six months after the injury is useful to determine subclinical evidence of reinnervation. Such reinnervation is typically characterized initially by "nascent" polyphasic MUAPs (Fig. 7.9). With reinnervation, the numbers of positive sharp waves and fibrillations decreases over time, amplitude of MUAPs increases as collateral spouting occurs, and with evaluation of interference pattern, there is an observed increasing number of voluntary MUAPs.

The author prefers to initially obtain sensory nerve conduction studies (occasionally with sedation) consisting of a median sensory nerve conduction study recorded from the index finger (C6 dermatome), a median sensory nerve conduction study recorded from the middle finger (C7 dermatome), and an ulnar sensory nerve conduction study recorded from the fifth digit (C8 dermatome). Median and ulnar motor nerve conduction studies are useful to evaluate the integrity of axons traveling through the lower trunk. Axillary and musculocutaneous motor nerve conduction studies (with assessment of CMAP amplitudes) are useful if an upper trunk injury is suspected. These CMAP amplitudes may be compared to the intact side, depending on patient tolerance of the study (58). A CMAP amplitude reduction of more than 90%, compared to the unaffected side, predicted severe weakness of the corresponding root level. During the EMG study of the deltoid, the examiner should assess the clinical sensation of the C5 dermatome. The use of dermatomal and mixed-nerve SSEPs in brachial plexus injuries are discussed in a following section.

In addition to a complete needle EMG screen of upper extremity muscles clinically affected, electromyographic examination of the infraspinatus or supraspinatus can help localize an upper trunk injury proximal to or distal to the takeoff to the suprascapular nerve. While the examination of the rhomboid can be difficult in the infant, a finding of fibrillations or positive sharp waves supports the presence of a C5 root injury. While in the adult electromyographic evaluation of the cervical paraspinal muscles may help evaluate the extent and severity of cervical root injuries, generally the cervical paraspinals are extremely difficult to study in the infant due to poor relaxation. In the young child, adequate relaxation of the cervical paraspinals may be obtained with general anesthesia,

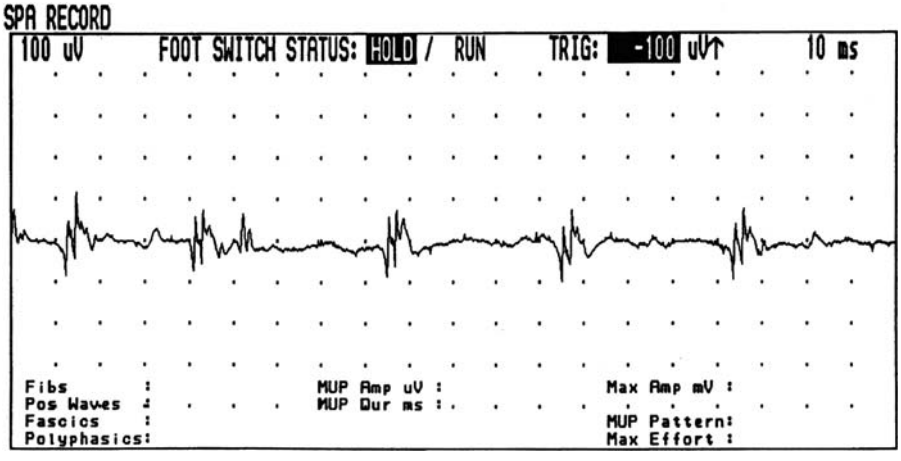


Figure 7.9 Polyphasic motor unit action potential (MUAP) with a neuropathic firing frequency at 25 Hz. These polyphasic MUAPs obtained 4 months after brachial plexus injury are indicative of reinnervation.

but this is usually not necessary and does not influence management. In addition, study of the serratus anterior and rhomboids (typically performed to assess involvement of C5 and C5–C7 roots, respectively) may be technically difficult in the infant due to intact sensation, the presence of the trapezius overlying the rhomboids, depth of the rhomboids and serratus anterior, and the risk that sudden movement may cause penetration of the needle into the pleural space. Usually, a combination of needle EMG evaluation, sensory and motor conduction studies, and F-wave studies allows the electromyographer to determine the location and severity of the injury.

The natural history of conservatively managed brachial plexus birth palsy has been reported (59). Seventy-two percent of those referred for rehabilitation evaluation showed stable functional status at follow-up. There has been a resurgence of interest in surgical exploration of obstetrical brachial plexus palsy with external and internal neurolysis, neurotization, and, in selected cases, nerve grafting (60–66). EMG evaluation at approximately 4–9 months postinjury may support the possible utility of a surgical exploration for neurolysis, neurotization, and/or nerve grafting if there is limited electrophysiologic evidence of reinnervation. Some authors suggest a repeat study within three months of the injury (67). Preoperative electrodiagnostic studies, intraoperative nerve conduction studies, and somatosensory-evoked potentials are helpful in the surgical decision making. Preoperative and/or intraoperative somatosensory-evoked potentials may provide evidence of upper cervical root avulsion versus partial trunk and nerve root integrity, as discussed in a following section.

Facial Paralysis in the Neonate

Facial paralysis or an asymmetric facies is a common finding in the neonate. This may be due to acquired traumatic facial palsy (a common iatrogenic problem with forceps deliveries), central nervous system conditions, congenital facial palsy, and congenital hypoplasia of the depressor anguli oris muscle. Facial nerve conduction studies aid in diagnosis (68). Side-to-side comparisons of amplitudes and latencies are essential. CMAP amplitude reduction and prolonged latency on the involved side indicate facial nerve involvement. Brainstem auditory-evoked potentials and blank reflexes may be helpful in determining central nervous system involvement. Axonal integrity can be determined by electromyographic evaluation for spontaneous activity and motor unit recruitment. Improvement on serial testing provides favorable prognostic information, particularly when improvement occurs over one to two weeks. Normal facial nerve distal latencies in the newborn are $<$12.0 msec; in children 1–12 months, $<$ 10.0 msec; in children 1–2 years of age, $<$6.3 msec; in children 2–3 years of age, $<$4.5 msec; in children 3–4 years, $<$4.0 msec; and $<$5.0 msec in children older than 4 years of age (69).

Common Polyneuropathies

Hereditary Neuropathies (Charcot-Marie-Tooth Subtypes)

Clinical findings associated with hereditary neuropathies and the current classification of these disorders is described in the chapter on pediatric neuromuscular diseases. The demyelinating form (Charcot-Marie-Tooth [CMT] I) typically has onset in early childhood. Marked slowing of motor conduction velocities, usually to less than 50% of normal, is often present in early childhood (70–72). Generally, marked swelling of motor nerve conduction velocities is present by 3 to 4 years of age (70). Distal latencies are usually severely prolonged. There is usually less temporal dispersion than observed in acute inflammatory demyelinating neuropathy (Guillain-Barré syndrome) due to fairly uniform demyelination of all axons. Needle EMG abnormalities include defibrillation with positive sharp waves, decreased interference pattern, and large amplitude polyphasic MUAPs resulting from reinnervation by collateral axonal sprouting.

CMT II is the axonal form. CMAP and SNAP amplitudes may be reduced, but nerve conduction velocities are either low-normal or mildly reduced. Needle EMG shows evidence of chronic denervation and reinnervation. CMT III, also referred to as Dejerine-Sottas disease and congenital hypomyelinating neuropathy, often present in infancy. CMAP amplitudes are reduced due to a combination of conduction block and axonal loss, motor nerve conduction velocities are typically less than 10 meters per second, and latencies may be three times the normal value (73).

Acute Inflammatory Demyelinating Polyradiculoneuropathy (Guillian-Barré-Syndrome)

These children often present with an acute rapidly ascending paralysis initially affecting the lower limbs. While pain is common, sensory symptoms are usually mild, and objective sensory loss is fairly rare. Electrophysiologically, criteria for poor recovery in adults may not apply to children. One study documented good recovery in children with low median CMAPs and fibrillation potentials (74), while another study showed no difference in the incidence of reduced CMAP amplitude among ventilated and nonventilated children (75). Classic electrophysiologic findings in Guillain-Barré include prolonged or absent F-waves early in the course of the disorder, slowing of

conduction velocities (both proximally and distally), prolonged distal latencies, reduced CMAP amplitudes with evidence of conduction block, and significant temporal dispersion (Fig. 7.10). The electrophysiologic findings may lag behind the clinical signs and symptoms. In addition, electrophysiologic recovery may lag behind clinical recovery.

Chronic Inflammatory Demyelinating Polyradiculoneuropathy

This disorder has many features in common with acute inflammatory demyelinating polyradiculoneruopathy. These patients typically show a subacute or chronic onset lasting more than four weeks, and the disorder continues with either a chronic or relapsing course. Electrophysiologic findings generally show more marked slowing of conduction velocity (often below 10 meters per second) and elevated stimulation thresholds. As in AIDP, there is evidence of focal conduction block, temporal dispersion, prolongation of distal motor latencies, and prolonged or absent H-wave and F-wave responses. These late responses may be absent due to proximal conduction block. Needle EMG may show a paucity of abnormal spontaneous rest activity and normal or slightly enlarged MUAPs, which exhibit a neuropathic firing pattern.

Axonal Guillain-Barré/Acute Motor Axonal Neuropathy

In this disorder, children often present with rapid onset, quadriparesis, bulbar dysfunction, and respiratory insufficiency (76). The patients may have inexciteable motor nerves or very low-amplitude CMAPs. The author has observed such a case with clinical findings mimicking cerebral death (77). The child had combined demyelinating and axonal findings and eventually had near complete recovery over 18 months. In general, children with the axonal form of Guillain-Barré are more likely to require assisted ventilation, develop severe quadriparesis, and require a much longer period of time to become ambulatory. *Campylobacter jejuni* has been implicated as a precipitating agent in many cases.

Neuropathies Associated With Central Disorders

A variety of metabolic disorders produce abnormalities of both the central and peripheral nervous system. Abnormalities of lipid metabolism, such as metachromatic leukodystrophy, may produce a severe demyelinating peripheral neuropathy with electrophysiologic findings of high stimulation threshold and low conduction velocities. Somatosensory-evoked potentials may show both central and peripheral delay, and visual-evoked potentials show central delay. Other disorders showing both central and peripheral nervous system involvement include Krabbe disease, Refsum's disease (phytanic acid storage disease), Tangier disease (hereditary high-density lipoprotein deficiency), a-beta lipoproteinemia (a vitamin E deficiency syndrome), Fabry's disease (alpha galactosidase A deficiency), Niemann-Pick disease (a variant of sphingomyelin lipidoses), peroxisomal disorders such as adrenoleukodystrophy, porphyria (which produces axonal degeneration of predominantly motor fibers), and tyrosinemia (which produces

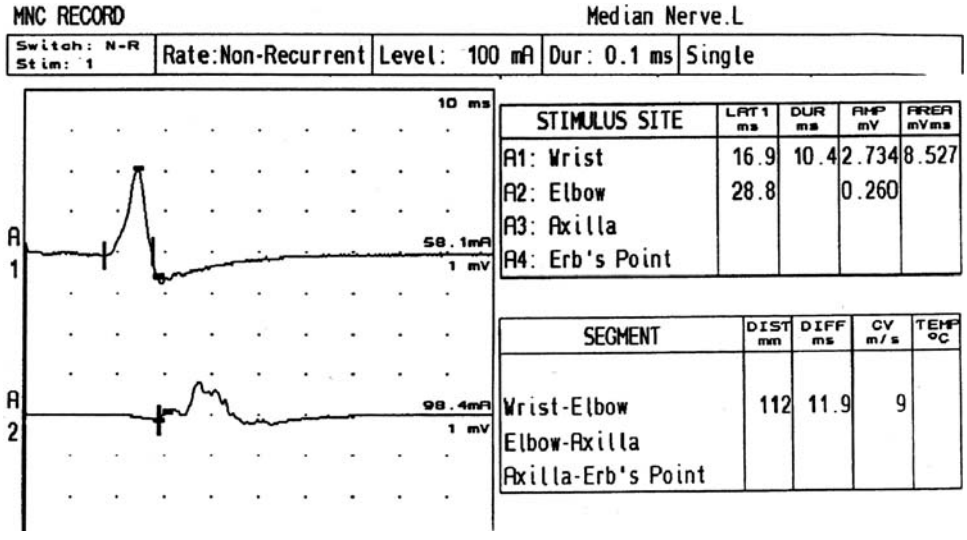


Figure 7.10 Median motor nerve conduction in a 4-year-old child with Guillain-Barré syndrome. Distal latency is prolonged at 16.9 milliseconds, and conduction velocity is slowed at 9 m/s. Note the conduction block (amplitude drop from 2.734 to 0.260 mV) and temporal dispersion.

primary axonal degeneration with secondary segmental demyelination).

Krabbe disease is associated with marked central and peripheral demyelination, and NCS typically show a mixed sensorimotor demyelinating peripheral neuropathy. The peripheral neuropathy occurs early in the neonatal period in Krabbe disease and affects the nerves uniformly. Nerve conduction studies may provide a highly sensitive tool to screen this patient population (78).

In ataxia telangiectasia, there is a loss of large, predominantly sensory, myelinated fibers due to a primary axonal degeneration. In Friedreich's ataxia, an autosomal recessive condition, there is a primary axonal degeneration of peripheral nerve fibers producing reduced or absent sensory compound action potential amplitudes.

Acquired Toxic Neuropathies

Toxic polyneuropathies with predominantly axonal involvement include lead-, mercury-, and vincristine-induced neuropathy, among others. Predominantly demyelinating neuropathies may be caused by organophosphate poisoning and arsenic poisoning. While arsenic poisoning may clinically simulate Guillain-Barré syndrome or chronic inflammatory demyelinating polyneuropathy (CIDP), electrophysiologic studies have shown evidence of both axonal degeneration and severe demyelination.

Burn-Associated Neuropathies

Children and adults with extensive burns are at increased risk for mononeuropathies and/or peripheral neuropathies (79–84). Mechanisms include direct nerve tissue destruction from the burn, extensive edema with compartment syndrome, critical illness polyneuropathy caused by systemic mediators, and entrapment neuropathies caused by scarring during and/or after healing. The incidence of neuropathy exceeds 10% in many series. Burn-associated polyneuropathy (BAPN) is common after thermal injury, and the electrophysiologic manifestations of BAPN are usually present within the first week (81). Thermal injuries may induce an inflammatory cascade that results in alterations of nerve function. In one series, those with severe neuropathy had higher levels of C-reactive protein (81). Other risk factors associated with a significantly higher prevalence of neuropathy include age above 20 years, electric burns, burns involving full thickness of the skin, a surface area of more than 20%, history of alcohol abuse, and number of days in the intensive care unit. In animal models of burn injury, both functional and morphological deficits are produced in peripheral nerve axons at sites well removed from a full-thickness dermal burn injury (85). The neural deficits may contribute to changes in neuromuscular transmission and the development of limb and respiratory muscle weakness that also accompany burn injury. Further animal work has demonstrated that burn wound excision at 30 minutes but not at 3 hours prevented the nerve conduction deficits measured in mice with 20% body surface area burns (86). The cellular basis of burn-induced neuropathy is unknown, but nitric oxide and tumor necrosis factor-alpha appear to play a role.

Diabetic Polyneuropathy

Nerve conduction velocity (NCV) in the distal motor and sensory nerves, the motor nerve distal latency, and the sensory nerve action potential (SNAP) amplitude were impaired in adolescent patients with type 1 diabetes. The deterioration in motor NCV, H-reflex latency, and SNAP amplitude became more conspicuous in late puberty and postpuberty, and was related to poor metabolic control (87). In another study of children 7 to 20 years old with a duration of diabetes of more than 3 years, 57% of the patients had abnormal conduction, which was seen most often in the motor nerves, especially in the peroneal nerve (41%), followed by the median nerve (24%) (88).

Neuropathies Associated With Infections

HIV Infection

Children with HIV may develop a variety of neurologic sequelae, including encephalopathy, progressive multifocal leukoencephalopathy, myelopathy, intractable seizures, optic neuritis, acute vasculitis, hemiplegia, paraspinal lymphoma, and peripheral nerve disease. The peripheral nerve dysfunction may present as distal symmetric sensory or sensorimotor polyneuropathy, carpal tunnel syndrome, lumbosacral polyradiculopathy, motor neuronopathy, AIDP and CIDP, autonomic neuropathy, sensory ganglionopathy, and toxic neuropathy (caused by antiretroviral medications) (89). In addition, polyradiculopathy and multiple mononeuropathies may be caused by other infections (eg, cytomegalovirus, hepatitis B or C, and herpes zoster). In one series, one-third of children 5 to 14 years of age had symptoms and signs of peripheral nerve involvement. Distal paresthesia and/or pain plus diminished ankle jerks and/or diminished vibration sense were the most common clinical findings. Symptoms were chronic and fluctuating, and pain was, in general, not severe. Nerve conduction studies primarily revealed axonal changes (90). The issue of peripheral nerve involvement may be multifactorial. Children with

HIV-1 infection are exposed to antiretrovirals for an ever-increasing length of time throughout postnatal growth and development, and the cumulative toxicities are becoming progressively apparent. Evidence for nucleoside reverse transcriptase inhibitor (NRTI)–associated mitochondrial toxicity is seen in vitro, in animal models, and in NRTI-exposed adults and children (91). Peripheral neuropathy is associated with the chronic use of dual nucleoside reverse-transcriptase inhibitor regimens in HIV-infected children, and regimens containing zidovudine have less toxicity than do those containing d4T (92).

Lyme Disease

Lyme disease is the most common tickborne disease in the United States. Children and those spending extended time outdoors in wooded areas are at increased risk. The spectrum of neurologic manifestations and the relative frequencies of different syndromes associated with North American Lyme disease caused by *Borrelia burgdorferi* infection has been reviewed in a series of 96 children referred for neurologic problems in association with the infection (93). The most frequent neurologic symptom was headache, and the most common sign was facial palsy. Less common manifestations were sleep disturbance and papilledema associated with increased intracranial pressure. Signs and symptoms of peripheral nervous system involvement were infrequent. The most common clinical syndromes were mild encephalopathy, lymphocytic meningitis, and cranial neuropathy (facial nerve palsy). In contrast with adult patients with neurologic Lyme disease, meningoradiculitis (Bannwarth's syndrome) and peripheral neuropathy syndromes were rare in children.

Entrapment Mononeuropathies in Children

Carpal Tunnel Syndrome in Children

Carpal tunnel syndrome (CTS) is a relatively rare complication in children, with mucopolysaccharidosis types I, II, and III (eg Hunter's and Hurler's syndromes) and mucolipidosis being the most common populations to manifest CTS during childhood (94). Treatment of the metabolic disorder does not necessarily reverse the symptoms, and prompt surgical release is necessary. Other uncommon etiologies include hereditary neuropathies such as CMT 1 and hereditary neuropathy with liability to pressure palsies (HNPP), CIDP, treatment with growth hormone, hemophilia with localized bleeding in the region of the carpal tunnel, Schwartz-Jampel syndrome, multiple xanthomas associated with familial hypercholesterolemia, congenital macrodactily in a median nerve territory, fibrolipomas of the median nerve, and Klippel-Trenauny syndrome (95).

Ulnar Mononeuropathies in Children

Ulnar mononeuropathies are the most common upper extremity mononeuropathies seen in children (96). The most common etiology is acute trauma (eg, midshaft or proximal forearm fractures, elbow dislocation, etc.), compression from compartment syndrome, or entrapments in association with HNPP or other anomalous anatomy producing entrapment. Other etiologies include baseball throwing injuries in adolescents, Larsen's syndrome with dislocations, congenital constriction band syndrome, insulin-dependent diabetes mellitus, leprosy, and so on. The location of the neuropathy is most commonly the cubital tunnel, but it may also localize to the forearm, wrist, or hand.

Radial Mononeuropathies in Children

Radial mononeuropathies are rare but do occur in children. In one series, 50% of radial neuropathies, including two in newborns with apparent prenatal onset, were atraumatic, primarily related to compression in six and entrapment in two. The other 50% were traumatic mononeuropathies related to fractures or lacerations (97). Electromyography documented the radial neuropathy to be localized to the proximal main radial nerve trunk in 13%, distal main radial nerve trunk in 56%, and posterior interosseous nerve in 31% of children.

Peroneal Mononeuropathies in Children

The most common entrapment in the lower extremity is peroneal mononeuropathy at the fibular head. Children with peroneal mononeuropathy typically present with unilateral foot drop. Both distal branches are involved in the majority of cases; hence, the level of the lesion is most often the common peroneal nerve at or above the fibular head, followed by the deep peroneal nerve and superficial peroneal nerve (98). Common etiologies include compression from a short leg cast, compression from prolonged surgical positioning, and trauma (eg, distal femoral physeal fractures, proximal tibial fractures, etc.). Contributing factors include hereditary neuropathies (CMT or HNPP) and significant rapid weight loss in an adolescent. Other etiologies may include compression from osteochondromas, neurofibromas, and intraneural ganglions; arthrogenic cyst of the fibula; and stretch during tibial limb lengthening.

Sciatic Mononeuropathies in Children

Sciatic mononeuropathies are uncommon in children. Etiologies in one series included compression, stretch

injuries (eg, during closed reduction of a hip dislocation), lymphoma, vasculitis associated with hypereosinophilia, and penetrating trauma (99). The peroneal division is more commonly affected than the tibial division in the absence of penetrating trauma. The vascular supply to the peroneal division may be more susceptible to compromise from stretch or compression. Axonal sciatic lesions are more common than demyelinating lesions.

Neuropathies With Limb-Lengthening Procedures

Mononeuropathies in the setting of limb lengthening are not uncommon, but are frequently subclinical. Patients undergoing tibial limb lengthening procedures are at risk for peroneal neuropathies in particular and rarely tibial mononeuropathies. Femoral lengthening can place a patient at risk for neuropathies affecting the sciatic nerve (particularly the peroneal division). Humeral lengthening can place upper extremity nerves at risk. Some have monitored for subclinical neuropathy of the upper and lower extremities using mixed-nerve somatosensory-evoked potentials during pin placement and serially during distraction (100,101).

Neuromuscular Junction Disorders

Infantile Botulism

Infantile botulism primarily occurs in infants 2–6 months of age. Clinical findings include diffuse weakness, hypotonia, weak cry, poor feeding, constipation, and occasionally respiratory distress. The onset is fairly rapid. Electrophysiologic studies may show a reduced CMAP amplitude, preserved motor conduction velocities and SNAPs, and abnormal repetitive nerve stimulation findings at high rates of stimulation (Fig. 7.11). One study demonstrated an incremental

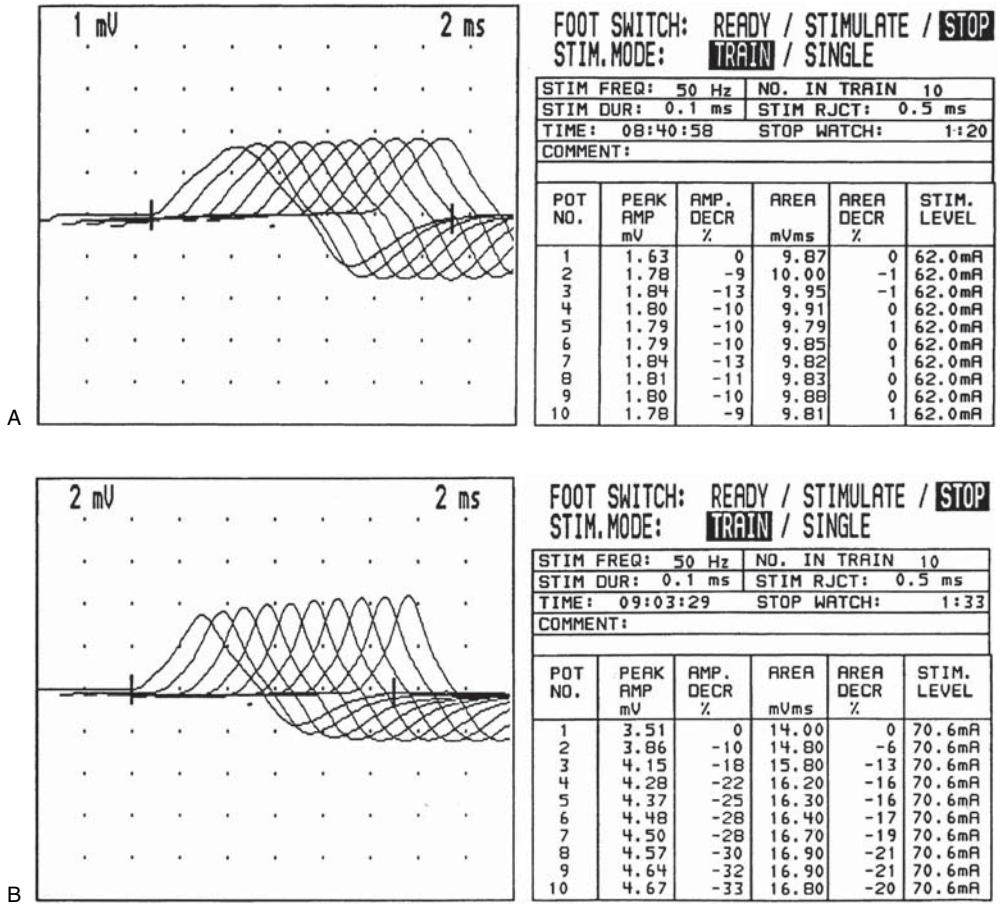


Figure 7.11 High frequency repetitive nerve stimulation in a 7-week-old infant with marked progressive weakness, respiratory failure, and botulism. (A) Several days into the course, the repetitive stimulation study of the ulnar nerve at 50 Hz is normal; however, the compound muscle action potential amplitude is severely reduced (1.63 mV). (B) Twelve days later, the infant is slightly improved clinically. A repeat study of the ulnar nerve at 50 Hz is diagnostic of infantile botulism with a 33% increment obtained between first and tenth stimuli. Clostridium botulinum was isolated from the stool.

response to repetitive nerve stimulation at rates of 20–50 Hz in 92% of infants with infantile botulism (22). The mean increment was 73%, with a range of 23% to 313%. With the lower-frequency stimulation (2–5 Hz), variable changes occurred, but the majority of infants showed decremental responses. A recent study demonstrated that the isolation of Clostridium botulinum from stool obtained by enema effluent was actually more sensitive for the diagnosis of infant botulism than electrodiagnostic studies (102).

EMG in infants with botulism demonstrates abnormal spontaneous rest activity with fibrillation potentials and positive sharp waves and short-duration, low-amplitude MUAPs (22).

Transient Neonatal Autoimmune Myasthenia Gravis

This disorder is caused by passage of antibodies from myasthenic mothers to their fetuses. Infants often present with hypotonia and respiratory distress. The diagnosis may be made by repetitive nerve stimulation studies. Given that normal infants exhibit less neuromuscular reserve than older children or adults, repetitive stimulation studies in this clinical setting utilizes rates of 2–5 Hz almost exclusively. A decrement of greater than 8% to 10% between the first and fifth CMAP in the train is considered positive for myasthenia. The combination of repetitive motor nerve stimulation and edrophonium or neostigmine testing may improve the accuracy of the diagnosis (103). If a decremental response is obtained, the repetitive nerve stimulation may be repeated at 30–120 seconds after administration of edrophonium utilizing a stimulation rate of 2–5 Hz. Near complete repair of the decremental response may be evident in the myasthenic infant (Fig. 7.12). Serologic antibody testing may be helpful if the mother has documented antibodies. Transient neonatal myasthenia gravis is self-limited,

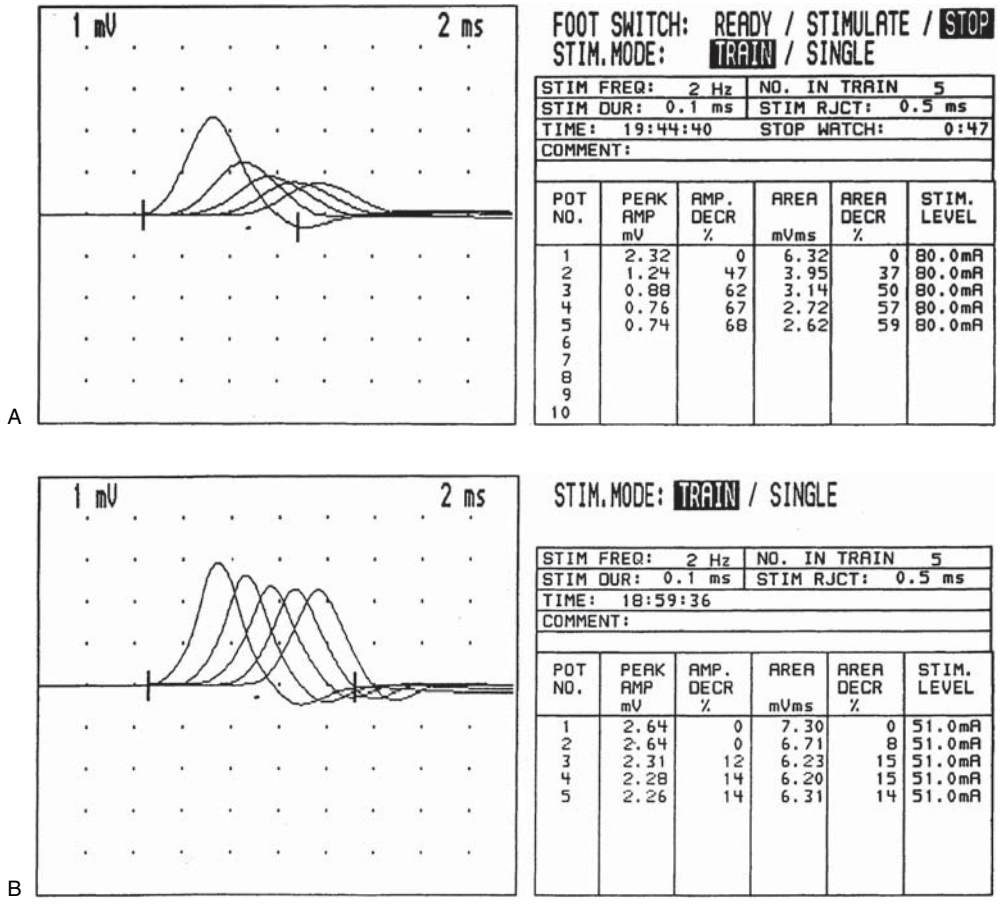


Figure 7.12 Low-frequency repetitive nerve stimulation study of the ulnar nerve in a 2-week-old infant with respiratory failure secondary to congenital myasthenia. (A) At baseline, a 68% decrement in amplitude and a 59% decrement in area is present between first and fifth stimuli with a stimulation frequency of 2 Hz. (B) Twenty minutes after intravenous neostigmine is given, the initial compound muscle action potential has improved from 2.32 to 2.64 mV and the decrement has improved to 14%. The infant was treated with Mestinon and later extubated.

with a reported duration of 5–47 days, with a mean duration of 18 days (104).

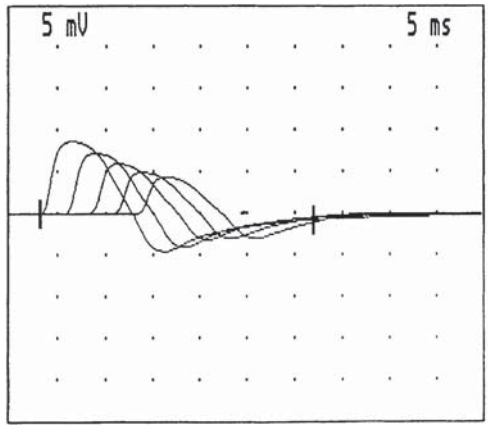
Toxic Neuromuscular Junction Disorders

Medications can interfere with neuromuscular transmission by inhibiting the release of acetylcholine, impairing the function of acetylcholinesterase (AChE), or binding directly to the acetylcholine receptor. Two drugs that may produce clinically significant weakness in normal children are magnesium and organophosphates (105,106).

Congenital Myasthenic Syndromes

Numerous presynaptic and postsynaptic congenital myasthenic subtypes exist, which are described in the pediatric neuromuscular disease chapter. These disorders often show decremental responses at high rates of stimulation, whether they are pre- or postsynaptic. Typically, the decremental responses are greater at higher rates of stimulation. Standard repetitive nerve stimulation studies do not adequately distinguish presynaptic from postsynaptic subtypes, but they do help diagnostically (Fig. 7.13).

Based on clinical findings, repetitive nerve stimulation studies, and/or stimulated single-fiber EMG, a strong clinical suspicion of a neuromuscular junction disorder, such as a congenital myasthenic syndrome, might warrant further elucidation of the specific subtype of presynaptic or postsynaptic abnormality with application of a motor point biopsy. Ultra-structural evaluation of the neuromuscular junction (NMJ) with electron microscopy is usually performed on a biopsy of the deltoid or biceps, including the muscle region containing the-NMJ (the "motor point"). For in vitro



A

FOOT SWITCH: READY / STIMULATE / STOP
STIM.MODE: TRAIN / SINGLE

STIM FREQ: 3 Hz	NO. IN TRAIN 5
STIM DUR: 0.1 ms	STIM RJCT: 0.5 ms
TIME: 09:51:53	STOP WATCH: 0:30
COMMENT:	

POT NO.	PEAK AMP mV	AMP. DECR %	AREA mVms	AREA DECR %	STIM. LEVEL
1	8.85	0	98.90	0	62.8mA
2	7.30	18	79.00	20	62.8mA
3	5.97	33	61.90	37	62.8mA
4	4.98	44	53.00	46	62.8mA
5	4.44	50	48.60	51	62.8mA
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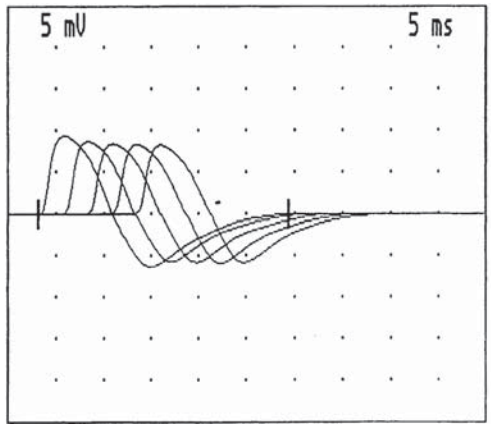


B

FOOT SWITCH: READY / STIMULATE / STOP
STIM.MODE: TRAIN / SINGLE

STIM FREQ: 3 Hz	NO. IN TRAIN 5
STIM DUR: 0.1 ms	STIM RJCT: 0.5 ms
TIME: 09:55:53	STOP WATCH: 0:32
COMMENT:	

POT NO.	PEAK AMP mV	AMP. DECR %	AREA mVms	AREA DECR %	STIM. LEVEL
1	9.49	0	95.80	0	62.8mA
2	8.68	9	84.80	11	62.8mA
3	8.39	12	84.70	12	62.8mA
4	8.32	12	84.50	12	62.8mA
5	8.25	13	83.10	13	62.8mA
6					
7					
8					
9					
10					

Figure 7.13 Low-frequency repetitive nerve stimulation study of the axillary nerve in a 12-year-old child with presynaptic congenital myasthenia. The active electrode is placed over the deltoid with stimulation at Erb's point using a block stimulator. (A) A 50% amplitude decrement is obtained between the first and fifth stimuli with 3 Hz stimulation frequency. (B) After a 30-second isometric contraction of the deltoid, the amplitude decrement has improved to 13%. The child was later confirmed to have a presynaptic congenital myasthenia by motor point biopsy of the anconeus muscle.

electrophysiologic and immunocytic chemical studies of the neuromuscle junction, a short muscle is usually removed from origin to insertion along with its motor branch and NMJ. Muscles obtained have included the anconeus muscle near the elbow, the external intercostal muscle, and the fifth or sixth intercostal space near the anterior axillary line or the peroneus tertius muscle in the lower extremity. Often, patients undergo simultaneous biopsy of the deltoid (for EM) and motor point biopsy of the anconeus or intercostal muscle (for in vitro electrophysiologic studies). The in vitro electrophysiologic studies often allow specific delineation of the congenital myasthenic syndrome into one of the numerous specific subtypes. In recent years, many of the subtypes have been mapped to specific gene loci, and increasingly, molecular genetic studies are being used for diagnostic purposes.

Myasthenia Gravis

Myasthenia gravis presents in adolescents more frequently than younger children. Muscle weakness typically increases with exertion but improves with rest and anticholinesterase medication. The disorder is an autoimmune etiology due to circulating antibodies that bind to the postsynaptic membrane. While elevated acetylcholine receptor antibody levels may be diagnostic, a significant percentage of cases with autoimmune myasthenia gravis may have nondetectable circulating antibodies. Electrophysiologic studies demonstrate abnormal decremental responses at low rates of stimulation (2–3 Hz). The limb is well immobilized. A supramaximal train of three to five stimuli is applied. Typically, patients exhibit a smooth, reproducible decrement of the evoked synapse of greater than 8% to 10%. The defect in neuromuscular junction transmission can be enhanced by exercise, which results in postactivation facilitation. Often, there is an increased decremental response obtained two to four minutes after exercise with low rates of stimulation (2–3 Hz). This is due to postactivation exhaustion (21). Proximal muscles may show increased sensitivity versus distal muscles. Children with ocular myasthenia frequently exhibit normal responses with distal repetitive nerve stimulation studies, and sensitivity of the repetitive nerve stimulation (RNS) study is enhanced by use of a more proximal shoulder girdle muscle (eg, axillary or spinal accessory nerve) or by study of the facial nerve. Combining the diagnostic yield, patient comfort, and technical ease, the choice of muscle for RNS should be ulnar to the abductor digiti minimi, followed by spinal accessory to the trapezius for patients with predominant limb weakness; facial nerve to the nasalis and spinal accessory to the trapezius in oculobulbar; and facial to the nasalis in ocular myasthenia (107).

Lambert-Eaton Syndrome

This presynaptic neuromuscular junction disorder usually found in adults with small cell carcinoma of the bronchus has been described in children. Approximately 5% of all cases occur in children. The amplitude of the single evoked CMAP is low. With low rates of repetitive nerve stimulation, a decremental response is often obtained. After exercise or tetanic contractions, there is facilitation of the potentials by as much as 100% to 200%.

Myopathies

Polymyositis/Dermatomyositis

Polymyositis/dermatomyositis has been described in children ranging in age from infancy to adulthood. Children may result with progressive proximal muscle weakness, dysphagia due to involvement of pharyngeal musculature, dyspnea, and muscle tenderness. A classic skin rash may or may not be present. Creatinine kinase values are often markedly elevated. Classic EMG findings include increased insertional activity with complex repetitive discharges; fibrillations and positive sharp waves; and low-amplitude, polyphasic, short-duration motor unit action potentials recruited rapidly in relation to the strength of contraction.

Congenital Myopathies

Congenital myopathies are a heterogeneous group of disorders usually presenting with infantile hypotonia, normal cognitive status, and primary structural abnormalities of the muscle fibers, which are elucidated on histologic and electron microscopic evaluations of muscle biopsy specimens. Patients usually develop proximal greater than distal muscle weakness that is nonprogressive and static. These myopathies are described in the chapter on pediatric neuromuscular diseases. Nerve conduction studies are generally normal; however, there may be mild reductions in CMAP amplitudes. On needle EMG, findings are either normal or there may be mild, nonspecific changes, usually of a myopathic character (small-amplitude, short-duration polyphasic MUAPs). The only congenital myopathy consistently associated with abnormal spontaneous rest activity is myotubular (centronuclear) myopathy. In this disorder, the EMG reveals myopathic motor unit action potentials with frequent complex repetitive discharges and diffuse fibrillation potentials.

Dystrophic Myopathies

The dystrophic myopathies are extensively described in the chapter on pediatric neuromuscular diseases.

EMG is rarely used at the present for the diagnostic evaluation of a suspected dystrophic myopathy due to molecular genetic testing and the importance of muscle biopsy in differentiating among Duchenne muscular dystrophy, Becker muscular dystrophy, and limb girdle muscular dystrophies. EMG in dystrophic myopathies is characterized by low-amplitude, short-duration polyphasic MUAPs (Fig. 7.14). Recruitment is myopathic in nature with increased recruitment or "early" recruitment demonstrated with slight effort. Interference pattern is usually full. Complex repetitive discharges (Fig. 7.15) and abnormal spontaneous rest activity may be present, reflecting membrane instability.

Metabolic Myopathies

Nonspecific myopathic EMG findings may be demonstrated in metabolic myopathies. For example, absent maltase deficiency shows increased insertional activity; complex repetitive discharges; low-amplitude, short-duration MUAPs; profuse fibrillations; and positive sharp waves. Carnitine deficiency, a disorder of lipid metabolism, demonstrates increased recruitment for effort, decreased amplitudes of MUAPs and occasional fibrillations. EMG may be normal in many metabolic myopathies, such as carnitine palmityl transferase deficiency.

Myotonic Disorders

Myotonic disorders such as myotonic muscular dystrophy and Schwartz-Jampel syndrome may show myotonic discharges with either positive sharp wave or fibrillation configuration and a waxing and waning firing frequency. The myotonic discharges are often described as exhibiting the sound of a "dive bomber." There may be

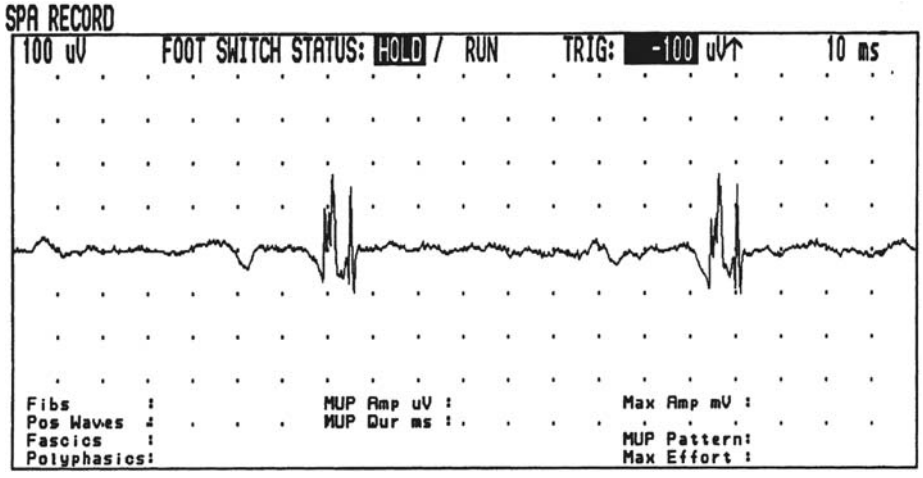


Figure 7.14 Low-amplitude short-duration polyphasic motor unit action potential in a 14-year-old girl with limb-girdle muscular dystrophy.

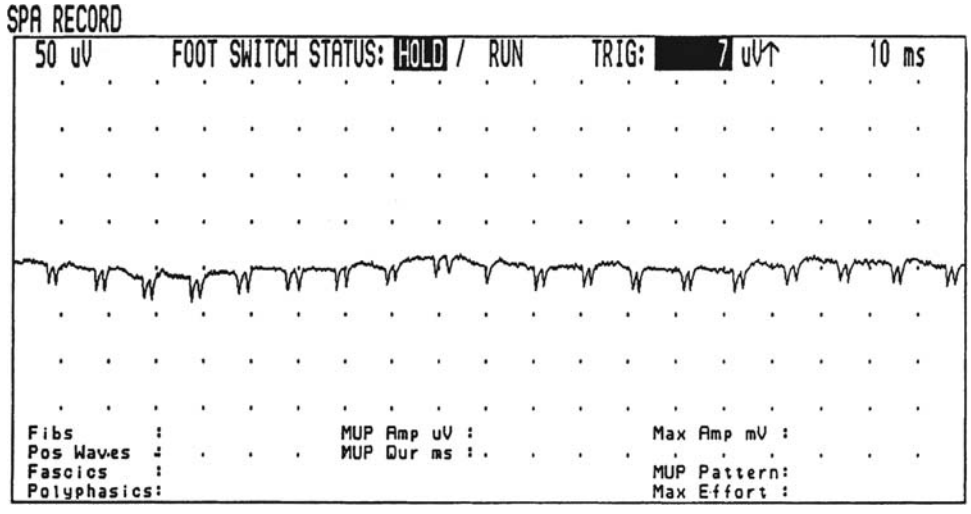


Figure 7.15 Complex repetitive discharges in a dystrophic myopathy.

profuse fibrillations and positive sharp waves. MUAPs are often of low amplitude and short duration. There may be more involvement of distal musculature than proximal musculature in myotonic muscular dystrophy. Again, with a known family history of myotonic muscular dystrophy, confirmation of the diagnosis in an individual with classic clinical features can be expeditiously and cost-effectively confirmed in the EMG laboratory. However, clinical trials frequently require molecular genetic confirmation of myotonic muscular dystrophy (DM1 versus DM2 and other myotonic disorders). so EMG is becoming less utilized diagnostically.

Somatosensory-Evoked Potentials

General Principals

The somatosensory-evoked potential (SSEP) is the sequence of voltage changes generated in the brain and the pathway from a peripheral sensory nerve following a transient electrical stimulus to the sensory cortex. Evidence suggests that these signals are related to large afferent fibers and peripheral nerves, which ascend through the dorsal column pathways of the spinal cord, proceed to the thalamus, and arrive at the somatosensory cortex. These are the same pathways that mediate light-touch two-point discrimination, proprioception, and vibration. Sensitive amplification and averaging techniques enable discrimination between the evoked response and other larger and more random physiologic potentials with which the signal is mixed. As a general rule, SSEP studies may be considered whenever the disease process in question can involve the somatosensory system. SSEPs reflect neurophysiologic activity in the posterior column, medial lemeniscus pathways. They do not reflect activity in the anterolateral column of the spinal cord. Thus, SSEPs correlate better with clinical examinations of proprioception and vibration rather than pain or temperature sensation.

Individual components of the SSEP waveform are identified by their latency (ie, the time at which they occur following a peripheral stimulus), their polarity, their position at which they are observed to be maximal, and, to a lesser extent, by the amplitude and shape of the waveform. Individual components are referred to by a letter and number. The letter (N for negative or P for positive) refers to the polarity of the wave and the number either to the latency in milliseconds of the signal from the time of the stimulus (eg, N20), or alternatively, especially appropriate in pediatric SSEPs, the order in which the component was observed (eg, N1, P2). Examples of median and tibial SSEPs are shown in Figures 7.16 and 7.17.

With mixed-nerve stimulation, recording electrodes are placed over the peripheral nerve more proximally, thoracolumbar or cervical spine, linked mastoids, and scalp. For upper extremity stimulation, the likely generator source for the cervical spine response is the incoming root, as well as postsynaptic excitatory potentials generated at the dorsal root entry zone (108). For the lower extremity, the lumbar spine responses are similarly a reflection of the root or cauda equina activity and the postsynaptic activity of the cord. The linked mastoid response is generated at the brainstem level. The difference in the latency of scalp N1 and the cervical spine response with median nerve stimulation gives a central conduction time. Similarly, the difference in latency between scalp P1 for posterior tibial nerve stimulation and the spinal potential generated over T12 or L1 gives a central conduction time.

Filter settings vary from a low-frequency filter of 3–30 Hz to a high-frequency filter of 1.5–3 KHz. The peripheral nerve is typically stimulated with a rate of 3.1 Hz. Our lab utilizes a stimulation intensity of 1.5 times motor threshold for mixed-nerve stimulation and 2.5 times sensory threshold for dermatomal stimulation. Electrodes are positioned according to a modified international 1020 electrode system.

SEP latencies decrease with age until well into childhood (108–111). The maturation with growth of SSEPs is mainly associated with cell-growth processes such as myelination and with cell differentiation and synaptic development. Conduction velocity along the central pathways progressively increases until 3–8 years of age, remains constant between 10–49 years of age, and slows thereafter. The N1 scalp latency of the median SSEP decreases until 2 to 3 years of age (owing to peripheral myelination) and then increases with body growth until adulthood. The cervical spine latency is relatively stable during the first two years (due to concomitant peripheral myelination and body growth), and then increases with age from 2 to 3 years until adulthood. The median SSEP central interpeak latency between cervical spine latency and scalp N1, which reflects central conduction time, decreases from a mean of 11.6 milliseconds at 4 to 8 months of age to a mean of 7 msec at 6 to 8 years of age, and remains constant between 6.9 and 7.0 msec until adulthood (112,113).

Among infants less than 4 months of age, sleep can affect the cortical components and is best performed on the awake infant. With children greater than 4 months of age, sleep or sedation usually has little effect on the SEP waveform when performing mixed-nerve stimulation. Indeed, the author has had no difficulty obtaining median nerve scalp responses in the pediatric ICU in comatose children with head trauma or those heavily sedated. Dermatomal SSEPs, on the other hand, are state-dependent responses affected by both sleep and sedation.

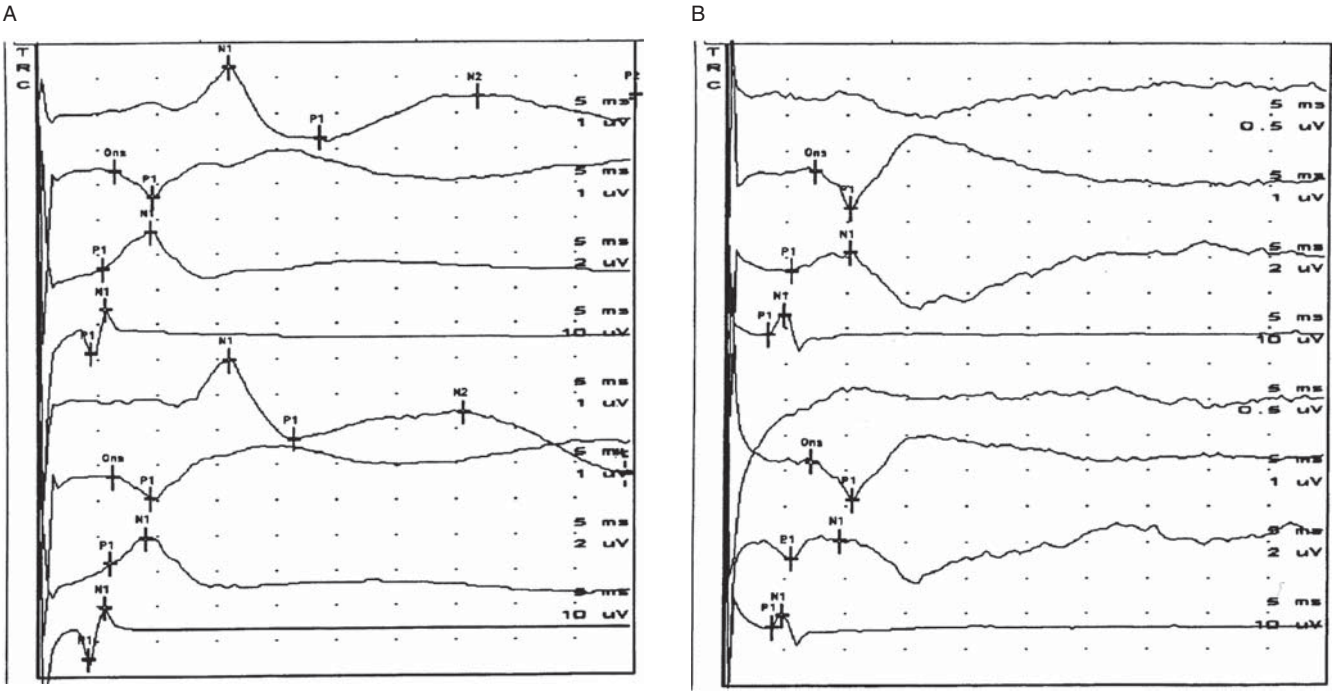
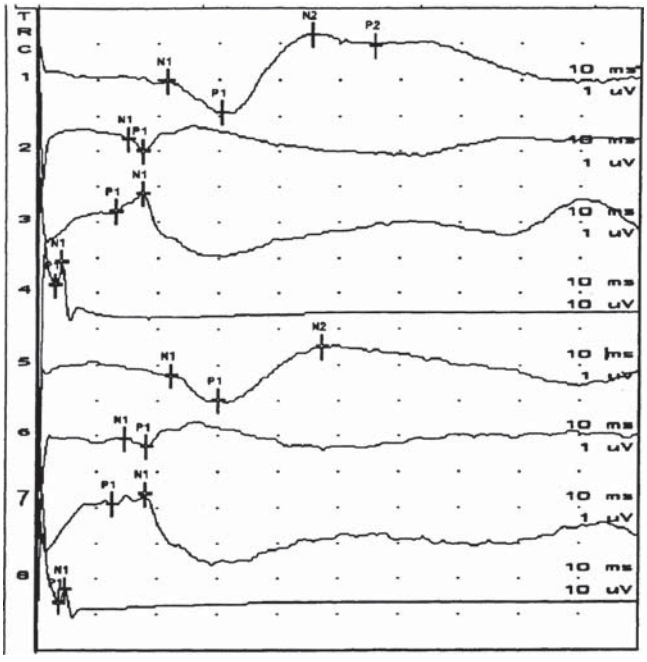


Figure 7.16 Median nerve somatosensory-evoked potentials (SSEPs) obtained in the pediatric intensive care unit. Channels 1–4 are responses with left median nerve stimulation, and channels 5–8 are responses with right median stimulation. Channels 1 and 5 are scalp responses (C4′ and C3′ referenced to Fz); channels 2 and 6 are brain (C4′ and C3′ referenced to linked mastoids); channels 3 and 7 are lower cervical spine responses (C7 spine referenced to Fz); channels 4 and 8 are peripheral responses obtained at the axillae. (A) Normal median SSEP responses obtained from a child with an epidural hematoma who was paralyzed with vecuronium for intracranial pressure control. There is no evidence of myelopathy. The child later recovered with minimal sequelae. (B) Abnormal median SSEP responses in a comatose child with severe brain injury and C1—C2 vertebral injuries. Note the bilaterally abnormal scalp reponses. Brainstem, C7 spine, and peripheral responses show no evidence of a spinal cord injury affecting posterior column pathways.

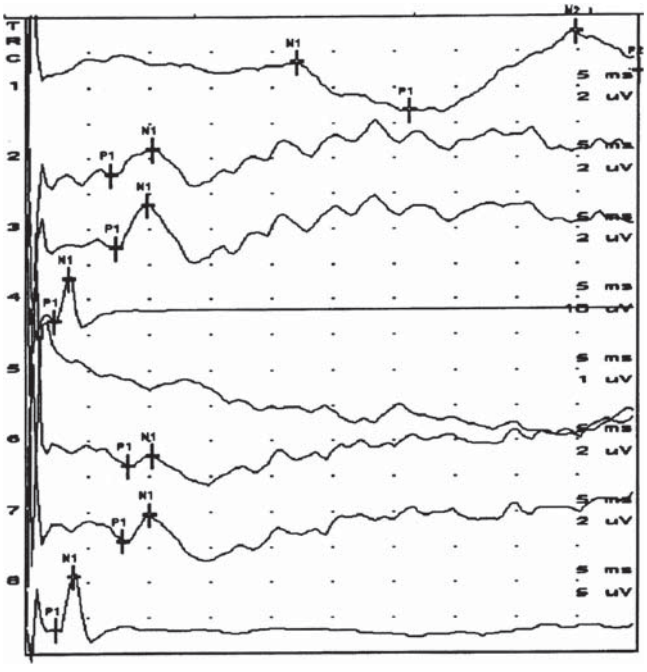
Clinical Applications of SSEPs in Children

Brain Injury in SSEPs. Abnormalities of median SSEPs can be predictive of poor prognosis in the situation of brain injury due to head trauma or hypoxia. A loss of bilateral SSEP scalp waveforms, as shown in Figure 7.16A, portends a poor prognosis in comatose children (114–119). Asymmetric scalp responses in a comatose child may be associated with the development of motor abnormalities such as hemiparesis because of the proximity of the sensory cortex to the motor cortex (Fig. 7.17B). A recent study compared the predictive powers of clinical examination (pupillary responses, motor responses, and Glasgow Coma Scale [GCS]), electroencephalography (EEG), and computed tomography (CT) to that of SSEPs in a systematic review. SSEPs appear to be the best single overall predictor of outcome (118). Posterior tibial nerve SSEPs performed on neonates at high risk of future neurodevelopmental impairment have demonstrated a highly significant relationship between bilaterally abnormal posterior tibial nerve SSEPs and the presence of cerebral palsy at 3 years of age (120). Normal posterior tibial nerve SSEPs were associated with a normal outcome in 24 of 25 infants. In this study, posterior tibial nerve SSEPs were more predictive than cranial ultrasound. Another study of 43 children with hemiplegic cerebral palsy found a positive correlation between median nerve SSEPs and the affected side using the amplitude of the responses rather than the latency (121). Other studies have confirmed the prognostic value of SSEPs in infants at risk for neurodevelopmental impairment (122–125).

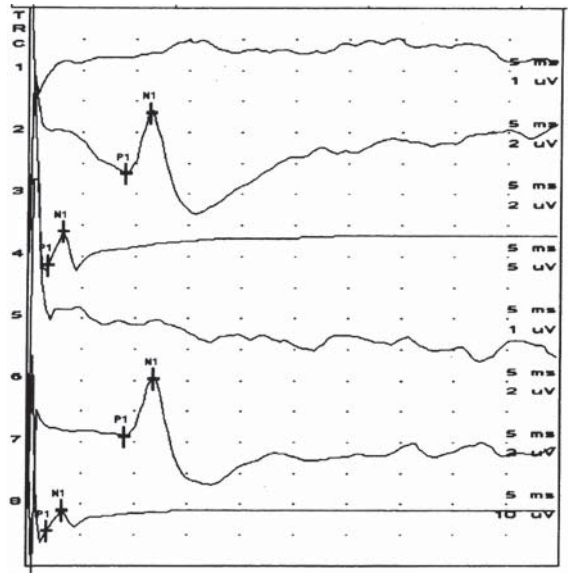
Traumatic Spinal Cord Injury. SSEP results combined with early American Spinal Injury Association (ASIA) motor scores have been shown to predict ultimate ambulatory capacity in patients with acute spinal cord injury (126,127). Other authors have shown that SSEP improvement over a one-week interval during the first three weeks after spinal cord injury was associated with motor index score improvement over a six-month period (128). Both ASIA scores and MEP recordings are similarly related to the outcome of ambulatory capacity and hand function in patients with SCI. Dermatomal somatosensory-evoked potentials have also been shown to be more sensitive for the detection of sacral sparing and of more prognostic value



A



B



C

Figure 7.17 Tibial somatosensory-evoked potentials (SSEPs) obtained in the pediatric intensive care unit. Channels 1–4 are responses with left tibial stimulation, and channels 5–8 are responses with right tibial stimulation. Channels 5–8 are responses with right tibial stimulation. Channels 1 and 5 are scalp responses (C2' to Fz); channels 2–7 are spine responses (L2 spine referenced to flank); and channels 4 and 8 are peripheral responses obtained at the popliteal fossa. (A) Normal tibial SSEP study. (B) Abnormal tibial SSEPs in a child with left hemispheric brain injury. Peripheral and lumbar spine (L2 and T12 level) responses are normal bilaterally. The scalp response is normal with left tibial nerve stimulation (channel 1), but absent with with right tibial nerve stimulation (Channel 5). (C) Abnormal tibial SSEPs bilaterally in an awake 4-year-old with low cervical spinal cord injury without radiographic abnormality. Peripheral (channels 4 and 8) and L2 spine (channels 2 and 7) responses are normal. Scalp responses (channel 2 and 5) are absent as a result of the low cervical spinal cord injury.

than mixed-nerve somatosensory-evoked potentials (129). However, somatosensory-evoked potentials and dermatomal SSEPs have been shown to add little or no useful prognostic information to the initial physical examination in either complete or incomplete spinal cord injury patient groups (130).

The author has a great deal of experience utilizing somatosensory-evoked potentials in the pediatric intensive care unit to evaluate for spinal cord injury without radiographic abnormality (SCIWORA) (131) in the situation where children are comatose or too obtunded to cooperate with the examination, or the child's age precludes a detailed sensory examination. Figure 7.17A shows an example of a normal tibial SSEP, whereas Figure 7.17C demonstrates the impaired posterior column conduction between the lower cervical spinal cord and brainstem with a SCIWORA injury sustained by a 4-year-old child.

Tethered Cord Syndrome. Posterior tibial SSEPs have been shown in some studies to be a sensitive indicator of declining neurophysiologic status and a more sensitive diagnostic tool than the clinical testing of sensation in patients with tethered spinal cord post-myelomeningocele repair (132–135). In addition, improvement of the evoked potentials has been documented subsequent

to untethering (132,133,135). In the author's experience, the spine response is often caudally displaced in myelomeningocele. Absent- or reduced-amplitude lumbar spine potentials or prolonged lumbar spine or scalp latencies with tibial nerve stimulation in the setting of normal median somatosensory-evoked potentials (normal spine latencies and amplitudes with median nerve stimulation, normal cervical-to-brain central conduction time, and normal median scalp latencies) have been suggested to be indicators of electrophysiogic impairment due to tethered cord syndrome.

In the most comprehensive study to date, 90 children were followed with serial peroneal SSEPs after a repair of their spinal dysraphic lesions with the objective of evaluating whether SSEPs were a useful way of monitoring these children to facilitate early detection of clinically significant retethering. Three hundred and nine studies were performed on these children, yielding a mean of 3.4 studies per patient. The median time between SSEP studies was 13 months. A clinical examination was performed at the time each SSEP was done. There was a false-positive rate of 71% and a false-negative rate of 43%. It was concluded that serial SSEPs do not correlate well with clinical status and are not a useful modality for monitoring patients at risk for retethering (136). The author has followed a large population of children with myelomeningocele for decades and similarly has not found mixed-nerve SSEPs to be useful in the evaluation of secondary tethered spinal cord after myelomeningocele repair.

Intraoperative Spinal Monitoring. There are many reports detailing the usefulness of intraoperative SSEP monitoring during scoliosis surgery (137–140), as well as during other surgical procedures of the spine. The limitation of SSEPs is that they only monitor afferent pathways in the dorsal columns. Over the past decade, intraoperative spinal monitoring has evolved to include monitoring of the motor pathways. The corticospinal tracts are now being routinely monitored intraoperatively using transcranial electrical stimulation of the motor cortex (141), with motor-evoked potentials recorded from either peripheral motor axons or as a CMAP from innervated muscles. Transcranial electric MEPs to monitor the corticospinal motor tracts are now used routinely in addition to SSEPs for detection of emerging spinal cord injury during surgery to correct spine deformity or resect intramedullary tumors (54,55,56).

Brachial Plexus Injury. The dermatomal SSEP can be a useful supplement to the assessment of the child with a brachial plexus injury (142). The child needs to be awake during the study. The C5 and C6 dermatomal SSEPs are generally most useful in the author's experience. The C5 dermatome is stimulated over the lateral proximal shoulder, using a proximal disk as cathode and distal disk as anode. Intraoperative SSEPs with direct stimulation of exposed nerves may demonstrate incomplete injuries of upper cervical roots, a proximal stump of the ruptured C5 root with functional central continuity (thus, potentially suitable for grafting), or complete root avulsion. Preoperative diagnostic SSEPs, while a useful adjunct to conventional electrodiagnosis, do not enable one to discriminate incomplete cervical root avulsion from intact roots (143).

Demyelinating Diseases. Both SSEPs and brainstem auditory-evoked potentials have been reported to be abnormal in children with or carriers of leukodystrophy (144,145). Peripheral and/or central abnormalities have been documented in metachromatic leukodystrophy, Pelizaeus-Merzbacher disease, Krabbe disease, adrenoleulodystrophy, Canavan disease, Alexander disease, and multiple sulphatase deficiency (146).

Pediatric multiple sclerosis (MS), while relatively rare, does occur in preadolescents and adolescents (147). MRI has been shown to be slightly more sensitive than multimodal-evoked potentials in confirming the clinical diagnosis of childhood MS. However, in suspected or probable MS, both SSEPs and visual-evoked potentials may contribute to the determination of clinical diagnosis because of their capacity to demonstrate asymptomatic involvement in central somatosensory and central optic nerve pathways (148,149).

Acute transverse myelitis often results in severe myelopathy due to inflammation and demyelination. SSEPs have been shown to be abnormal in this condition and may provide prognostic information regarding ultimate outcome (150).

The extent and location of nerve involvement in demyelinating peripheral neuropathies has been evaluated with SSEPs; however, SSEPs do not usually provide necessary additional information to standard nerve conductions. Hereditary motor sensory neuropathy type I shows impaired peripheral conduction in both proximal and distal nerve segments with normal central conduction. AIDP patients have been shown to exhibit prolonged posterior tibial peripheral SSEP latencies in addition to prolonged or absent median F-waves. However, posterior tibial F-wave latencies and median nerve SSEPs were less sensitive studies for the detection of demyelination in AIDP (151). SSEP can detect an abnormality and thus support the clinical diagnosis of Guillain-Barré syndrome in the acute stage when the results of more conventional tests are inconclusive (152).

Conclusion

Pediatric electrodiagnostic studies are a useful diagnostic tool that aid in the localization of abnormalities

within the lower motor neuron, and often providehelpful prognostic information. Electrodiagnostic studies have been less utilized in the diagnosis of many myopathic disorders and anterior horn cell diseases due to the importance of molecular genetic studies and/or muscle biopsy for determination of disease subtypes. However, there remains a use for EMG and nerve conduction studies in many focal and generalized lower motor neuron conditions. For children suspected of having hereditary neuropathies with no family member possessing genetic confirmation, a directed nerve conduction study may guide the acquisition of more specific and less costly molecular genetic studies. In other conditions, such as Guillain-Barré syndrome, or focal neuropathic conditions, electrodiagnostic studies remain critical for diagnostic confirmation.

Practical suggestions relating to the pediatric electrodiagnostic evaluation have been provided. Study results must be interpreted in light of developmental and maturational issues affecting both clinical findings and electrophysiological processes. A skilled electrodiagnostic evaluation utilizes careful strategic planning to provide the most important diagnostic information needed in an expeditious manner, with the least distress possible to the child and parent. Ongoing electrodiagnostic experience with the pediatric population provides increasing diagnostic acumen regarding pediatric lower motor neuron disease processes and sufficient technical skills to provide the referring physician with accurate diagnostic information.

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8

Cerebral Palsy

Mary McMahon, David Pruitt,
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Cerebral palsy (CP) is defined as "a group of disorders of the development of movement and posture, causing activity limitations that are attributed to nonprogressive disturbances that occurred in the developing fetal or infant brain" (1). There are three major criteria for diagnosis of cerebral palsy: a neuromotor control deficit that alters movement or posture, a static brain lesion, and acquisition of the brain injury either before birth or in the first years of life. Due to the breadth of these criteria, cerebral palsy is an extremely heterogeneous diagnosis in terms of clinical presentation, etiology, and pathology. Although the brain lesions that result in cerebral palsy are not progressive, the clinical picture of CP may change with time as the affected individual grows and develops.

EPIDEMIOLOGY AND RISK FACTORS

CP is the most common motor disability of childhood, affecting approximately 3.6 per 1,000 school-age children (2) with at least 8,000 new cases each year in the United States (3). The population of children with CP may be increasing due to premature infants who are surviving in greater numbers (4), higher incidence in normal-weight term infants (3), and longer survival overall. The proportion of CP that is most severe is also increasing, with as much as a third of all children with CP having both severe motor impairments and mental retardation (5).

The etiology of CP is often not well understood. The majority of cases in term infants do not have an identifiable etiology (6). Factors that may contribute to brain injury and CP include prematurity, infection, inflammation, and coagulopathy (7). There is also considerable interest in the contributory roles of various biomolecules and cytokines that accompany infectious or inflammatory processes (8).

The greatest risk factor for the development of CP is prematurity. Premature infants (born earlier than 37 weeks gestation) are much more likely to develop the condition than term infants, and incidence rates are highest in the very earliest infants (9,10). Rates of CP in premature and low birth-weight infants vary from 40 to 150 per 1,000 live births (11), with some reports suggesting increasing (4) or decreasing rates (11,12) in the last decade or more. Figure 8.1 (5) demonstrates the role of prematurity in CP. The vertical bars represent raw numbers of children with CP and demonstrate that the largest numbers of children with CP were born at term. The horizontal lines represent the rates of development of CP at roughly 2/1,000 live births for term infants, 5/1,000 for infants born at 33–36 weeks gestation, and 30/1,000 live births for infants born prior to 28 weeks gestation. These rates demonstrate the profound effect of prematurity as a risk factor for CP.

Prenatal risk factors for CP include being small for gestational age (13), being of low or very low birth weight (14), developing infection (especially chorioamnionitis and cytomegalovirus) (15), having evidence of stroke (16), or having neonatal encephalopathy (17).

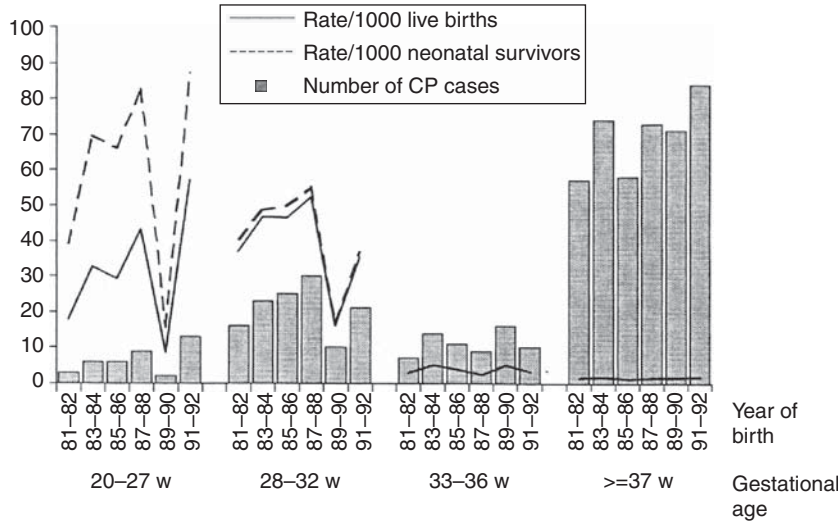

Figure 8.1 Cerebral palsy numbers and rates (excluding cases due to postneonatal causes) by gestational age in western Australia, 1981–1992. (Reprinted with permission from: *Cerebral Palsies: Epidemiology and Causal Pathways*. London: MacKeith Press; 2000; 151:26.)

Maternal risk factors for CP include chorioamnionitis (18,19) or fever during labor, coagulopathy or bleeding (20), placental infarction, and thyroid disease (21). Postnatal risk factors for CP are often related to social disadvantage, and include trauma in developed nations (22) and infection in developing nations (23). Additional risk factors for CP include kernicterus (24), methyl mercury exposure (25), and genetic causes (26).

Severe birth asphyxia in term infants is not a major cause of CP. Less than 10% of children with the condition had asphyxia, in contrast to prematurity, which is associated with up to half of all cases of CP. Nonetheless, for children who have true birth asphyxia, the risk of CP is increased (27). Fetal monitoring in the United States has probably increased the rate of cesarean section deliveries, but has not been associated with any decline in rates of CP (28). Term infants described as having birth asphyxia often manifest certain signs, including acidosis, bradycardia, or neonatal encephalopathy. Intrauterine exposure to infection or a coagulation disorder can cause a similar clinical picture at birth and may be mistaken for complications of birth asphyxia. Birth asphyxia by itself accounts for a small minority of cases of CP (29). Neonatal encephalopathy generally is diagnosed in neonates with significant neurologic dysfunction, including respiratory difficulties, altered tone, low consciousness, or seizure activity. It is the best predictor of CP in term infants, regardless of the cause of the encephalopathy.

CLASSIFICATION

CP has traditionally been classified by type of movement disorder and anatomic distribution.

Movement patterns include spastic, dyskinetic, hypotonic, ataxic, and mixed forms. The most common movement pattern is spastic, with a minority of cases being primarily dyskinetic, ataxic or hypotonic (2). The distinction between spasticity and dystonia is not always clear. An interdisciplinary group developed a consensus statement on the definition of each term. Spasticity was defined as hypertonia in which one or both of the following signs are present: a) resistance to externally imposed movement increases with increasing speed of stretch and varies with the direction of joint movement, and/or b) resistance to externally imposed movement rising rapidly above a threshold speed or joint angle (30). Dystonia was defined as a movement disorder in which involuntary sustained or intermittent muscle contractions cause twisting and repetitive movements, abnormal postures, or both (Fig. 8.2) (30). Hypotonic and ataxic forms of CP are rare and, therefore, any child suspected of having either of these diagnoses should receive a thorough diagnostic evaluation for other neurologic conditions.

The anatomic distribution of motor problems in CP is the primary means of classification. The three categories of hemiparesis, diparesis, and quadriparesis occur with fairly equal frequency (2,5). Hemiparetic CP affects only one side of the body and typically demonstrates greater impairments in the upper extremity (Fig. 8.3). Diparetic CP affects the lower extremities more than the upper extremities (Fig. 8.4). Spastic quadriparetic CP affects the entire body, including the axial as well as appendicular skeleton (Fig. 8.5).

An interest in classifying children with CP based on function in addition to the distribution of motor impairment resulted in the development of the Gross Motor Function Classification System (GMFCS). The

Figure 8.2 A child with dystonic cerebral palsy.

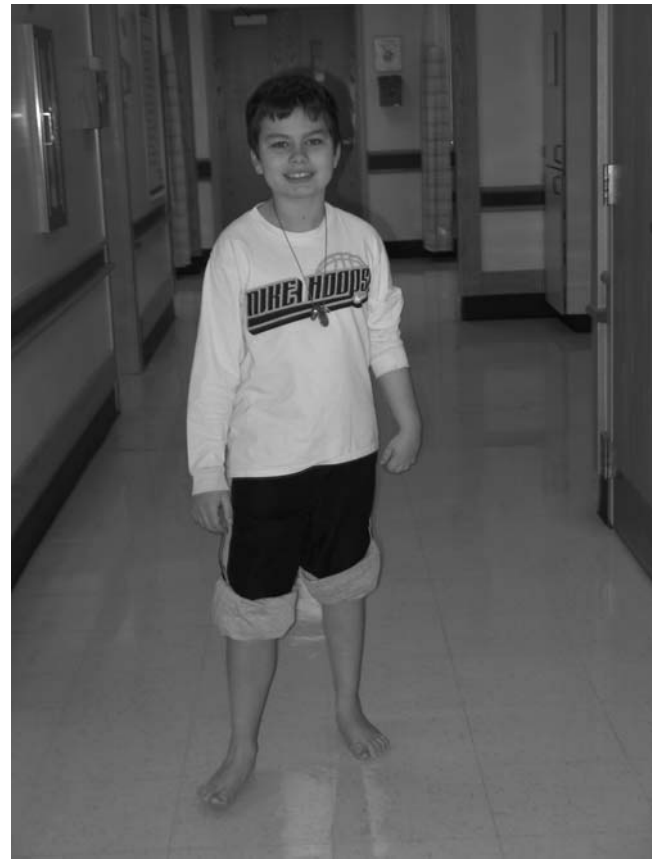

Figure 8.3 A child with hemiparetic cerebral palsy.

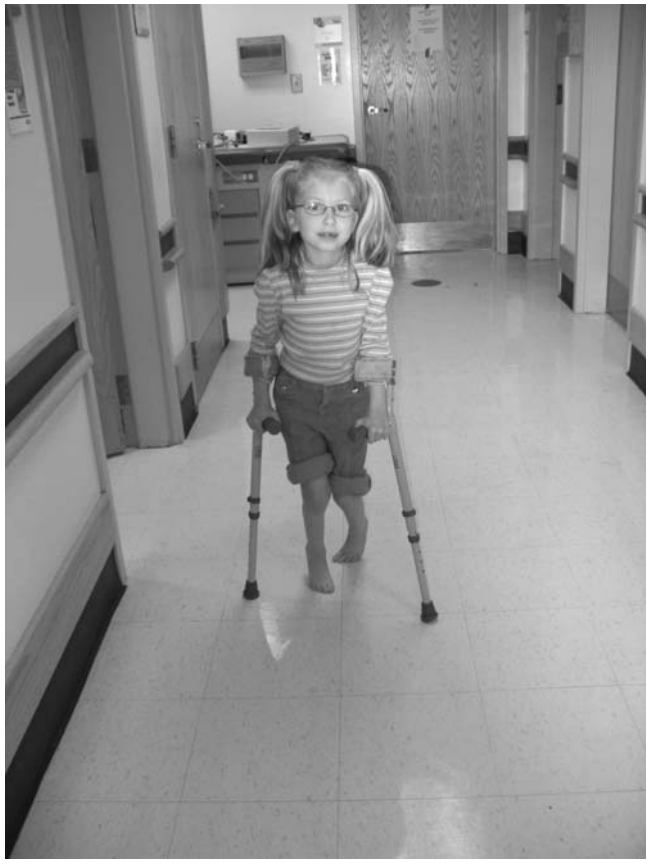

Figure 8.4 A child with diparetic cerebral palsy.

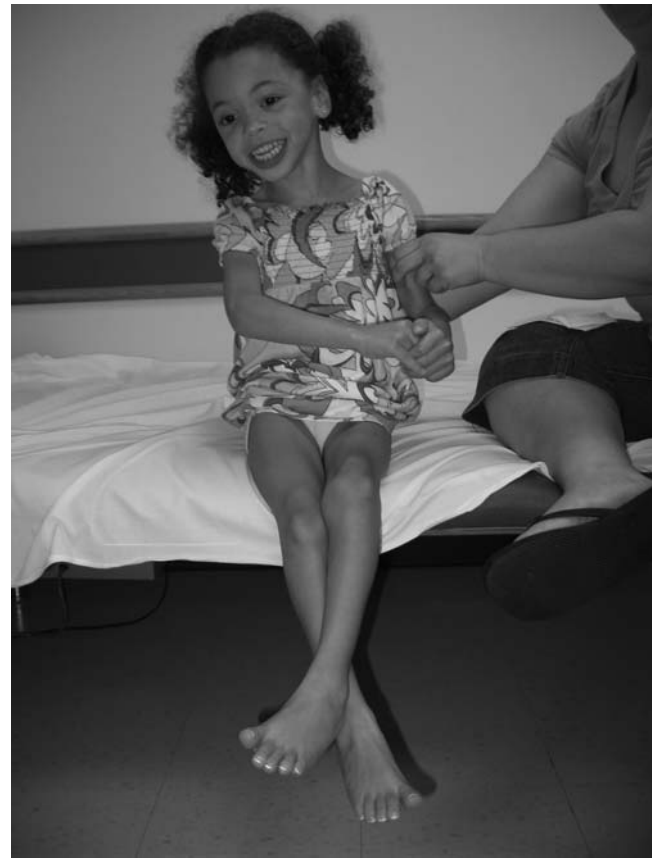

Figure 8.5 A child with quadriparetic cerebral palsy.

GMFCS stratifies children with CP into five groups based on gross motor skills (31) (Fig. 8.6). In this system, specific descriptions of mobility functions, based on age, allow each child with CP to be categorized. In gross motor function classification (GMFCS) I children walk indoors and outdoors and climb stairs without limitation. Children who are GMFCS II walk indoors and outdoors and climb stairs holding onto a railing but experience limitations walking on uneven surfaces and inclines. Children who are GMFCS III walk indoors or outdoors on a level surface with an assistive mobility device. Children may climb stairs with a railing or propel a manual wheelchair. Children who are GMFCS IV may walk short distances with a device, but rely more on wheeled mobility at home and in the community. Children at GMFCS V have no means of independent mobility. A related classification system for upper extremity function, the Manual Abilities Classification System, permits categorization by fine motor performance (32).

A more comprehensive rubric for the classification of CP has recently been proposed. Ideally, each individual with CP will be classified in four dimensions, including motor abnormalities, associated impairments, anatomical and radiological findings, and causation and timing of injury (1). Currently, quantitative tools to describe the clinical and radiographic features of cerebral palsy are being developed and refined, which will improve the robustness of CP classification.

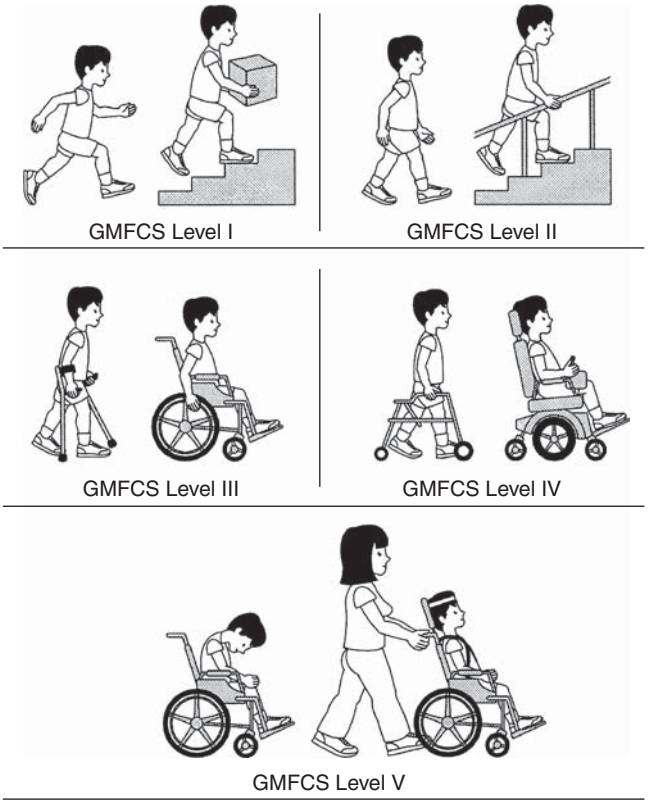

Figure 8.6 The Gross Motor Classification System for children aged 6 to 12 years. (Reprinted with permission from: Graham HK. Classifying cerebral palsy. *J Pediatric Orthop*. 2005; 25:128.)

PATHOLOGY

More than 80% of children with CP will have abnormal findings on neuroimaging (33–35). These abnormal findings can provide valuable clues to pathogenesis.

The most common abnormality on neuroimaging is found in the white matter near the lateral ventricles, often termed periventricular leukomalacia (PVL), with reports of up to 56% of all cases of CP demonstrating abnormalities in this location (34) (Fig. 8.7). PVL occurs much more commonly in premature infants than in term infants (90% vs 20%) and is a common outcome of intraventricular hemorrhage in premature infants (34). Because the corticospinal tract fibers to the lower extremities are medial to those of the upper extremities in the periventricular white matter, children with PVL typically have spastic diparesis. One large study found that PVL was present in 71% of the children with diparesis, 34% of those with hemiparesis, and 35% of those with quadriparesis (33).

Deep grey matter lesions to the basal ganglia and thalamic region are mainly associated with dystonic CP, and have been found in approximately 12% of children

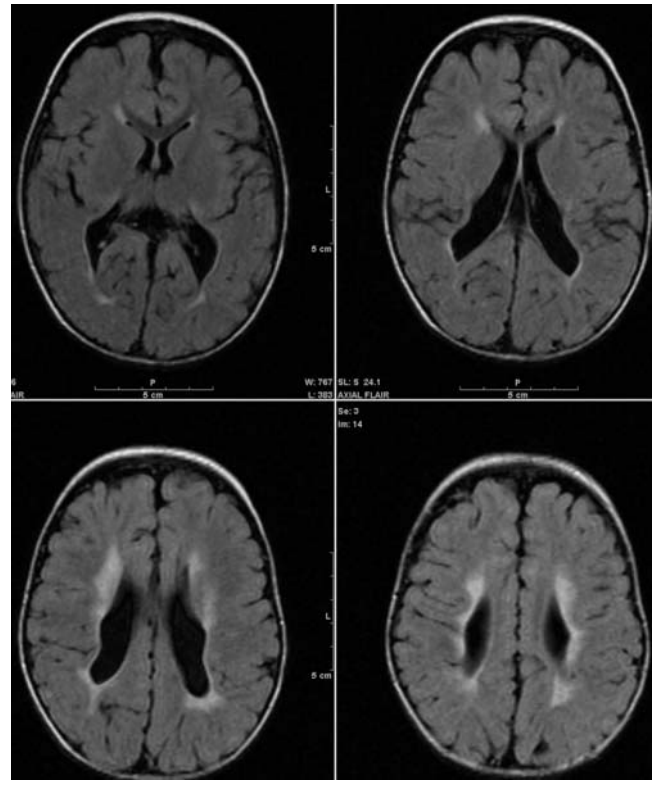

Figure 8.7 Periventricular leukomalacia.

with the condition (33). Historically, large numbers of children acquired athetoid CP following a diagnosis of kernicterus, due to concentrated damage to the basal ganglia with bilirubin encephalopathy. These cases are far less common with advancements in the treatment of neonatal jaundice.

Focal cortical infarcts involving both the grey and white matter are found almost exclusively in patients with hemiparesis, and are typically related to middle cerebral artery strokes. In a group of children with hemiparetic CP, 27% were found to have a focal infarct on imaging (33).

Brain malformations can be found on neuroimaging in approximately 10% of children with CP (33–35). Neuronal migrational disorders early in pregnancy can result in lissencephaly, polymicrogyria, schizencephaly, or holoprosencephaly. Some in utero infections, such as those caused by cytomegalovirus, can also cause distinctive brain malformations (33). Brain malformations are more commonly found in cases of term infants and hemiparesis (35).

Children who sustain diffuse brain insults demonstrate more extensive injury on neuroimaging. Infection and ischemia are two of the more common causes of generalized encephalomalacia. A wide range of findings may be present on magnetic resonance imaging (MRI), including multiple cysts, cortical thinning, white and grey matter loss, and microcephaly. Children with diffuse brain lesions or anomalies typically demonstrate spastic quadriparesis and are at high risk for additional medical and cognitive problems.

INITIAL EVALUATION AND CLINICAL FINDINGS

Signs and Symptoms

Early identification of children who have CP allows for early therapeutic intervention and screening for associated conditions. Because CP is a descriptive term that does not infer a single etiology, pathology, or prognosis, there is no specific diagnostic test. It is a diagnosis of exclusion based on a careful history and physical exam. It can be difficult to make a definitive diagnosis in infants less than 6 months old. Prior to this time, the infant has a limited repertoire of volitional movements, which makes milder delays in motor development difficult to detect. In addition, abnormalities in tone and reflexes are often subtle in early infancy. As the cortex matures in the second half of the first year, the diagnosis typically becomes more apparent.

The first step in the evaluation for suspected CP is a comprehensive history, including a detailed account of potential risk factors and family history. A thorough history of developmental milestones is also important. Often the parent's initial concern is a significant delay in attaining motor milestones. Prematurity must be considered when evaluating development because milestones are generally corrected for the degree of prematurity. A discrepancy between motor and cognitive milestones should always raise suspicion for CP. Certain deviations in developmental milestones are associated with CP. For example, early hand preference or asymmetric use of the extremities may be the first indication of hemiparesis. Early head control, rolling, or rigid standing are all associated with abnormally increased tone and/or exaggerated primitive reflexes. The parent may also describe unusual means of mobility, such as bunny hopping, combat crawling, or bottom scooting. The most important aspect of the developmental history is to confirm that the child has not lost any skills or milestones, as this would suggest a neurodegenerative disorder.

Following a detailed history, a thorough physical examination should be performed. A careful neurologic exam is an essential piece of the evaluation. In infancy, the neurologic exam focuses on tone and infantile developmental reflexes. Deep tendon reflexes, plantar responses, and the presence of clonus are more informative in the older child. Tone should be assessed by gently moving the infant's joints through their appropriate range of motion and evaluating the amount of resistance. Careful observation will also provide information about an infant's tone. Infants with severe hypotonia will lay in a frog-leg position with their hips abducted, flexed, and externally rotated. Their arms will lie limply at their sides. Persistent fisting or scissoring may be observed with increased tone. Most infants will undergo an early stage of mild or moderate hypotonia prior to more traditional signs of CP. A prolonged period of hypotonia or fluctuating tone is more typical of dyskinetic CP. In general, however, longer periods of hypotonia and severe hypotonia are associated with more severe motor deficits, regardless of the type of CP.

The earliest indication of CP may be a delay in the disappearance of primitive infantile reflexes. Commonly examined primitive reflexes include the Moro reflex, palmar grasp reflex, asymmetric tonic neck reflex, and tonic labyrinthine reflex. During the first six months of life, maturation of the cortex gradually overrides these primitive responses, and voluntary motor activity should increase. Persistence of these primitive reflexes past six months of age, asymmetry of the response, or an obligatory response at any age should be considered highly suspicious for a significant motor impairment. As the primitive reflexes become suppressed, postural or protective reactions such as the parachute and the equilibrium or tilting reactions should emerge. In children with CP, postural reactions may be less effective, appear later than usual, or fail to develop.

A definitive diagnosis of CP should be made cautiously, especially in the first six months of life. Infants who are suspected of having CP should be followed closely with serial developmental evaluations and physical exams until the diagnosis is clear. Further evaluation, including neuroimaging, should be considered to help clarify the diagnosis.

Imaging

Neuroimaging can be helpful in determining the etiology of CP and the timing of the insult. The Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society published two practice parameters that address the use of neuroimaging in the neonate and the child with suspected CP (36,37). Recommendations for imaging in the preterm neonate include a screening cranial ultrasonography on all infants <30 weeks gestation between 7 and 14 days of age and again between 36 and 40 weeks' postmenstrual age (36). This recommendation was based, in part, on the fact that a 10-fold elevation in the risk of adverse outcome for the very low birth-weight infant was identified with ultrasound evidence of grade 3 or 4 intraventricular hemorrhage, periventricular cystic lesions, or moderate to severe ventriculomegaly.

In term infants with neonatal encephalopathy, the practice parameter recommends a non-contrast computed tomography (CT) to detect hemorrhagic lesions when there is a history of birth trauma, low hematocrit, or coagulopathy (36). If the CT is inconclusive, MRI should be performed between day of life 2 to 8 in order to assess the location and extent of injury. Abnormalities of the thalamus and basal ganglia were associated with increased neurodevelopmental disability at 1 to 2 years of age.

Neuroimaging can also be useful in determining an etiology in children suspected of having CP outside of the neonatal period. The practice parameter on the diagnostic assessment of the child with CP found that an abnormal MRI scan was found in the majority of children with CP (average 89%) and that MRI was more likely to show an abnormality when compared to CT (average 77%) (37). The practice parameter, therefore, recommends neuroimaging in the evaluation of a child with CP if the etiology has not been established and MRI is preferred to CT (Fig. 8.8).

Laboratory Findings

Metabolic or genetic causes for CP are unusual, and laboratory studies to investigate these conditions are not routinely recommended. Metabolic or genetic testing is recommended in the following conditions: if neuroimaging does not determine a specific structural abnormality or if it reveals a developmental malformation, if there is evidence of developmental deterioration, or if there is a family history of a childhood neurologic disorder associated with a diagnosis of "cerebral palsy (37)." The practice parameter also recommends consideration of diagnostic testing for a coagulation disorder in children with an unexplained cerebral infarction on neuroimaging (see Fig. 8.8).

DIFFERENTIAL DIAGNOSIS

Young infants with CP often present with hypotonia. The differential diagnosis for the floppy infant is vast. The most common etiologies include central nervous system disorders such as CP, neuromuscular disorders, genetic disorders, and metabolic disorders. Clues to a neuromuscular disorder include diminished deep tendon reflexes, weakness (which may result in absent infantile reflexes), or a positive family history. Dysmorphic features may suggest a genetic cause for hypotonia, such as Down's syndrome, Prader-Willi syndrome, or Angelman syndrome. Metabolic disorders may present at any age, but are most likely to present in infancy. Metabolic disorders should be considered if a previously healthy child presents with an acute encephalopathy without an adequate explanation. Metabolic acidosis, hypoglycemia, hepatic involvement, or cardiac involvement should also prompt consideration of a metabolic disease. Dystonia and spasticity are present in a number of metabolic disorders, including mitochondrial disorders, glutaric aciduria type I, Lesch-Nyhan syndrome, and homocystinuria. A diagnosis other than CP should always be sought in children who have evidence of progressive disease or loss of previously obtained milestones.

ASSOCIATED DISORDERS

Sensory Impairments

CP is defined as a disorder of movement control and posture, and therefore sensory impairments are easily overlooked. Deficits in two-point discrimination, proprioception, and stereognosis have been described (38–40). Sensory deficits are believed to be most common in children with hemiparesis. A study of children with spastic hemiparesis found that 97% of the spastic limbs had a stereognosis deficit, 90% had a two-point discrimination deficit, and 46% had a proprioception deficit, and these sensory deficits were more commonly present in limbs with a greater size discrepancy (38). Sensory deficits can also be found in the limbs that do not appear to be affected by CP. Bilateral sensory deficits were found in 88.8% of children with hemiparesis in one study (39). Stereognosis and proprioception were the most common bilateral abnormalities, and the extent of sensory loss did not mirror the motor

deficit. Another study identified abnormalities of tactile spatial discrimination in the hands of children with spastic diparesis with apparent normal motor function in their upper extremities (40). Sensory deficits are important to recognize because they can significantly affect functional use of the extremity.

Visual Impairments

Visual impairments are common in children with CP, with a reported prevalence of 39% to 100% (41). The inherent difficulty in doing an ophthalmologic exam on children with varying degrees of cognitive and motor impairments makes it difficult to determine the precise incidence of visual disorders. Strabismus is the most commonly reported visual disorder, but a wide variety of other disorders have been described. Some visual deficits demonstrate a relationship to the underlying etiology, such as retinopathy of prematurity in premature infants, cortical visual impairment in hypoxic ischemic encephalopathy, and homonymous hemianopsia in hemiparesis. One study demonstrated a

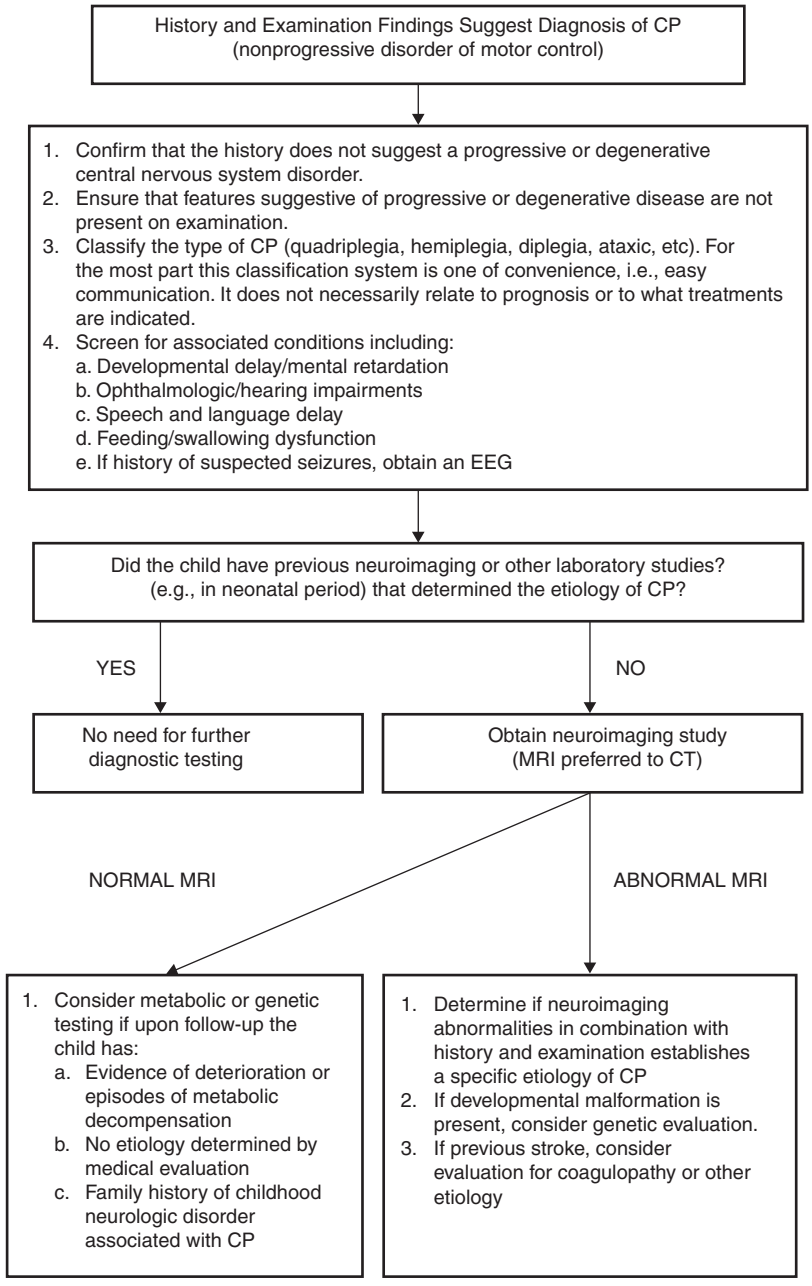

Figure 8.8 Algorithm for the evaluation of the child with cerebral palsy. (Reprinted with permission from: Aswal S et al. Practice parameter: Diagnostic assessment of the child with cerebral palsy. Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology*. 2004;62:851–863.)

relationship between visual deficits and severity of CP as measured by the GMFCS (42). In this study, children in each level of the GMFCS had visual deficits 10- to 70-fold higher than those reported in the general age-matched pediatric population. Children with milder CP, GMFCS level I to II, had visual deficits that resembled neurologically normal children with strabismus and amblyopia. Children with the most severe CP were at greatest risk for high myopia, absence of binocular fusion, dyskinetic strabismus, severe gaze dysfunction, and optic neuropathy or cortical visual impairment.

Hearing Impairments

Hearing impairments are relatively rare in CP. Sensorineural hearing loss is most commonly associated with congenital TORCH (toxoplasmosis, rubella, cytomegalovirus, and herpes) infections, bacterial meningitis, and ototoxic drugs. In the past, kernicterus was a relatively common cause of sensorineural deafness in athetoid CP.

Cognitive Impairments

Cognitive impairments are common in CP. It is difficult to make generalizations about the specific relationship of CP and cognitive function because CP is a heterogeneous disorder and the available literature often does not differentiate between the various types. In addition, assessment of intellectual functioning can be difficult in patients with severe motor and communication difficulties, which may lead to an underestimation of cognitive function. An overestimation of cognitive function can occur in patients who are socially responsive. The overall frequency of mental retardation, defined as an IQ score of 69 or below, is reported to be 50% to 70% (43). In general, patients with more severe neuromuscular impairments are at greater risk for cognitive impairments, but some patients with severe motor impairments can have normal cognition. For example, a patient with athetosis secondary to a discrete lesion in the basal ganglion is likely to have normal intelligence. It is important to attempt an accurate assessment of intelligence in order to assist in appropriate educational and vocational plans.

Psychological Impairments

The prevalence of emotional and behavioral problems in different populations of children with CP is reportedly 30% to 80% (44), but in general, it has not been well defined in the literature. A wide variety of behavior and emotional disorders are possible, including attention deficit disorder, passivity, immaturity, anger, sadness, impulsivity, emotional lability, low self-esteem, and anxiety. A population-based analysis of behavior problems in children with CP identified problem behaviors in 25% of the children as assessed by parent report (44). Specific behaviors that were most common in this population included dependency, being headstrong, and hyperactivity. An additional population-based study in Europe found a similar prevalence of significant emotional and behavioral symptoms in 26% of children with CP (45). The most common problems identified were in peer relationships (32%), hyperactivity (31%), and emotion (29%). Difficulty with peer relationships has been found even in children with milder CP (GMFCS I). Compared to their classmates, children with mild CP were found to have fewer reciprocated friendships, fewer sociable and leadership behaviors, and were more isolated and victimized by their classmates (46). Professionals and parents need to be aware that children with CP are at higher risk for psychological impairments than their nondisabled peers and that consideration should be given to a referral to a mental health specialist for evaluation and treatment.

Epilepsy

The overall occurrence of epilepsy is reported to be between 15% to 55% in a mixed population of children and adults with CP (47). A wide variety of types of seizures are possible, and a clear correlation between various risk factors and seizure frequency or type has yet to be established. Seizures are more common in children with more severe CP and in children with quadriparesis and hemiparesis versus diparesis (48).

Oromotor Impairments

Oromotor impairments are associated with more severe CP. A weak suck, poor coordination of the swallowing mechanism, tongue thrusting, and a tonic bite reflex may all lead to feeding difficulties and increased risk for aspiration. Speech disorders range from mild articulation disorders to anarthria, and are most commonly seen in children with spastic quadriparesis or athetosis. Oromotor dysfunction may also lead to difficulty controlling oral secretions and drooling, which may negatively affect social interactions. Oromotor impairments are associated with dental malocclusion and difficulty with oral hygiene, leading to an increased risk of periodontal disease.

Nutritional Disorders

The assessment of growth and nutrition in children with CP can be difficult due to the lack of a reliable means of measuring stature in children with contractures and scoliosis and the lack of appropriate reference data or growth curves specific to CP (49). Population-based growth patterns of CP have been published (50), but they probably include many children with conditions affecting growth and feeding, and therefore

should not be considered prescriptive of how children with CP should grow (49).

Poor oromotor skills, gastroesophageal reflux, and the inability to self-feed or communicate hunger can all increase the risk for malnutrition in children with CP. The North American Growth in Cerebral Palsy Project (NAGCPP) is a population-based study that identified the presence of feeding problems in 58% of children with moderate to severe CP. In addition, children with a pattern of severe feeding dysfunction were described as having the greatest risk for poor nutritional status and health, but even those with only mild feeding dysfunction were identified as being at risk for poor nutritional status. Subjects who were enterally fed were taller and had greater body fat stores when compared to subjects with similar motor impairments who were exclusively fed by mouth (51). Data from the NAGCPP also revealed that children with the best growth had better health and social participation (52).

Although malnutrition is a primary concern, children with CP are also at risk for overfeeding and obesity. Children with more severe CP have a lower total energy expenditure and higher body fat content than age- and sex-matched children without disabilities, placing them at risk for overfeeding with energy-dense enteral feeds (53). A study of ambulatory children with CP showed an increase in the prevalence of obesity from 7.7% to 16.5% over a 10-year period, an increase similar to that seen in the general pediatric population in the United States (54).

Genitourinary Disorders

The development of urinary continence is typically delayed in children with CP. A study of 601 children with cerebral palsy found that by the age of 6, 54% of children with spastic quadriparesis and 80% with spastic hemiparesis or diparesis had gained urinary continence spontaneously (55). The most important factors associated with urinary incontinence were quadriparesis and impaired cognition. Incontinence was the most common complaint, but frequency, urgency, hesitancy, and urinary retention may also be present. Frequency and urgency are often associated with spasticity of the detrusor muscle, causing small, frequent voids. Detrusor overactivity and a small bladder capacity were the most common findings on urodynamic studies in children referred for voiding dysfunction, but a minority were also found to have detrusor sphincter dyssynergia (56,57).

Respiratory Disorders

Children with CP are at increased risk for respiratory illnesses. Impaired control of respiratory muscles, ineffective cough, and aspiration due to an impaired swallow; gastroesophageal reflux; or seizures all increase the risk for chronically increased airway secretions. Increased airway secretions may lead to wheezing, atelectasis, recurrent aspiration pneumonia, restrictive lung disease, or bronchiectasis. Bronchopulmonary dysplasia in an infant born prematurely will also increase the risk for respiratory disorders.

Bone and Mineral Density Disorders

Decreased bone mineral density (BMD) and increased risk of fracture with minimal trauma is common in patients with moderate to severe CP, especially those who are nonambulatory. By the age of 10 years, most nonambulatory children have osteopenia, as defined by BMD *z* score of <-2.0 in their femur (58). Data from the NAGPP revealed that increasing severity of neurologic impairment, increasing difficulty feeding the child, use of anticonvulsants, and lower triceps skin fold *z* scores all independently contribute to lower BMD *z* scores in the femur (58). Longitudinal data from this project revealed that BMD *z* scores typically decrease with aging in CP, in spite of increases in BMD that average 2% to 5% per year in the distal femur and lumbar spine. Changes in BMD were quite variable, however, ranging from +42% to –31% (59).

Musculoskeletal Disorders

Foot/ankle

Equinus deformity, due to increased tone or contractures of the gastrocsoleus complex, is the most common musculoskeletal deformity in CP. Equinovarus foot deformity is primarily due to a combination of spasticity of the posterior tibialis muscle and the gastrocsoleus complex, resulting in inversion and supination of the foot and a tight heel cord (Fig. 8.9). This deformity is most common in a child with hemiparesis. Equinovalgus foot deformity is due to spasticity of the gastrocsoleus complex and the peroneal muscles, as well as weakness in the posterior tibialis muscle. This deformity is most common in older children with spastic diparesis and quadriparesis. Hallux valgus deformities are associated with valgus deformities of the foot, which may lead to a painful bunion at the head of the first metatarsal.

Knee

Knee flexion contractures are common due to spasticity in the hamstring muscles and static positioning in a seated position. If a severe knee flexion is present, hip flexion will be limited, resulting in lumbar kyphosis in the seated position. Flexion contractures at the knee are associated with hip and ankle flexion contractures and patella alta. Genu valgus may also occur, and is most commonly associated with excess femoral anteversion.

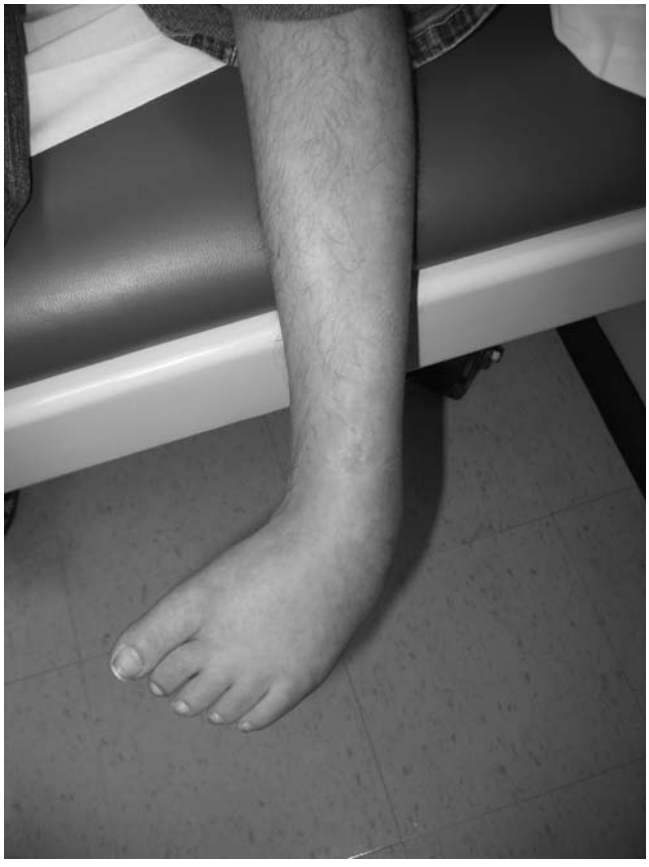

Figure 8.9 Equinovarus foot in a child with cerebral palsy.

Hip

Acquired hip dysplasia is common in cerebral palsy and often leads to progressive subluxation and possible dislocation. Hip subluxation can begin as early as age 2 years (60) and should be monitored closely by exam and serial radiographs. On exam, passive hip abduction of less than 35 degrees and a hip flexion contracture of more than 20 degrees are concerning signs of hip instability (61). On x-ray, hip subluxation is typically defined as a migration percentage greater than 30%. Close surveillance of hip migration with intermittent serial hip radiographs is recommended once hips have subluxed (62).

The reported incidence of dislocation in untreated hips varies, but 25% to 35% is the average estimate from most large series (62,63). Causative factors include persistent excessive femoral anteversion, a dysplastic acetabulum, and muscle imbalance from overactive hip adductors and flexors. These factors cause the hip to be adducted, flexed, and internally rotated, placing it at risk for posterior dislocation. A large population-based sample of children revealed a linear relationship between the incidence of hip displacement and level of gross motor function on the GMFCS (62). The incidence of hip displacement for each GMFCS level was as follows: I 0%, II 15%, III 41%, IV 69%, and V 90%. The natural history of hip dislocation has not been well described. Early osteoarthritis and difficulty with positioning and hygiene are not uncommon. The reported incidence of pain associated with a dislocated hip varies, but is commonly felt to be present in at least 50% of patients with dislocations (63).

Children with CP may also develop a "windswept deformity" of their hips, described as an adduction deformity of the elevated hip and an abduction deformity of the opposite hip, which also tends to be externally rotated and commonly results in pelvic obliquity (Fig. 8.10). The hip on the elevated side is at significant risk for dislocation, and positioning can be challenging. Hip dislocation with pelvic obliquity is often associated with scoliosis, but any potential causative relationship remains unproven.

Spine

Spinal deformities, including kyphosis, lordosis, or scoliosis, are common in children with CP. Kyphosis is often seen in conjunction with significant weakness of the spinal extensor muscles and tightness in the hamstrings, leading to a posterior pelvic tilt. Lordosis is frequently associated with hip flexion contractures.

Figure 8.10 Windswept hip deformity in a child with cerebral palsy.

The likelihood of scoliosis increases with the severity of CP. An overall incidence of approximately 20% (64) has been reported, with an incidence as high as 68% in children with spastic quadriparesis (65). Curves greater than 40 degrees tend to progress, regardless of the patient's skeletal maturity (65). The risk of progression is greatest for patients with quadriparesis, increased spasticity, a larger curve, a younger age, poor sitting balance, or pelvic obliquity (61).

Upper extremity

Spasticity and muscle imbalances can often lead to joint deformities in the upper extremity. The shoulder is often positioned in an adducted and internally rotated position. Spasticity in the biceps, brachioradialis, and the brachialis frequently result in elbow flexion contractures. Elbow flexion contractures less than 30 degrees rarely have functional significance. Forearm pronation deformities are common and can significantly affect functional use of the hand. The most common deformity of the wrist is flexion, typically with ulnar deviation (Fig. 8.11). The most common finger deformities are flexion and swan neck deformities due to hand intrinsic muscle spasticity. A thumb in palm deformity is commonly seen with adduction at the carpometacarpal joint, which may be associated with hyperextension of the metacarpophalangeal and interphalangeal joints.

Gait Impairments

A wide variety of gait classification systems have been developed to assist in diagnosis, clinical decision-making, and to facilitate communication among health care providers. A systematic review of the literature, however, concluded that no single classification system appeared to reliably and validly describe the full magnitude or range of gait deviations in CP (66).

The following is a description of the more common gait deviations associated with CP (Table 8.1) At the hip, increased hip adduction tone can cause scissoring and difficulty advancing the limb in swing phase. Increased tone in the iliopsoas can lead to increased hip flexion, resulting in an anterior pelvic tilt and a crouched gait. Increased femoral anteversion can contribute to in-toeing. At the knee, tight hamstrings can inhibit the knee from extending during stance phase, further contributing to a crouched gait. Spasticity of the rectus femoris may limit knee flexion during the swing phase, causing a stiff-kneed gait pattern. At the ankle,

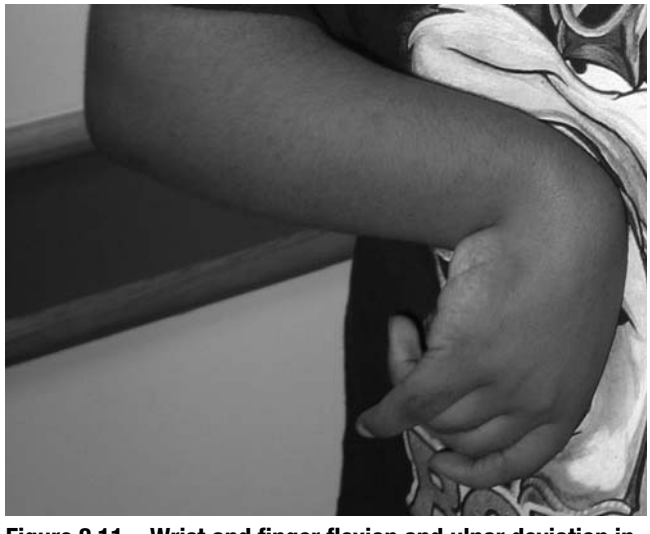

Figure 8.11 Wrist and finger flexion and ulnar deviation in a child with cerebral palsy.

8.1 Common Gait Deviations in Cerebral Palsy

LOCATION	IMPAIRMENT	POTENTIAL EFFECTS
Hip	Increased adductor tone	Scissoring; difficulty advancing leg in swing phase
	Increased iliopsoas tone	Anterior pelvic tilt; increased lumbar lordosis; crouched gait
	Increased femoral anteversion	Intoeing; false genuvalgus; compensatory external tibial torsion
	Abductor weakness	Trendelenburg gait
Knee	Decreased hamstring range of motion	Crouched gait
	Hamstring/ quadriceps co-contraction	Stiff-kneed gait
Ankle	Increased gastrocsoleus tone or contracture	Toe walking; genu recurvatum; difficulty clearing foot during swing
	Internal tibial torsion	Intoeing; ineffective push-off
	External tibial torsion	Out-toeing; ineffective push-off
	Varus	Increased ankle supination in stance or swing
	Valgus	Increased pronation in stance or swing; midfoot break

spasticity of the plantarflexors can lead to toe walking, difficulty clearing the foot during swing phase, or genu recurvatum (due to limited dorsiflexion in stance phase creating an extension moment at the knee). Spasticity of the ankle invertors, most commonly seen in spastic hemiparesis, can lead to supination of the foot and weight bearing on the lateral border of the foot. Weight bearing on the talar head is more common in spastic diparesis or quadriparesis, and is associated with an equinovalgus deformity. Malrotation of the leg can interfere with stability during stance phase and effective pushoff. Internal rotation is more common with a varus deformity and external rotation with a valgus deformity.

TREATMENT

General Principles

The treatment of a child with CP requires a multidisciplinary approach. Once the diagnosis is made, the infant or child should be evaluated by a comprehensive rehabilitation team. The members of this team will vary, depending upon site and availability. Potential team members may include a physiatrist, developmental pediatrician, orthopedist, neurologist, physical therapist, occupational therapist, speech and language pathologist, therapeutic recreation specialist, orthotist, psychologist, social worker, and a nutritionist. The team should work with the child's caregivers to develop short- and long-term goals that address neuromuscular concerns such as maintaining range of motion and tone control, as well as functional goals related to self-care skills, mobility, and communication. Goals related to increased societal participation should also be included. Goals should be routinely reassessed to ensure that they continue to be valid as the child grows older, and the child should be encouraged to take an active role in goal setting when appropriate.

Once the goals are determined, the family and the team must determine the most appropriate therapeutic approach. Although there are many treatment options to choose from, little scientific evidence exists on which to base one's treatment decisions. The heterogeneity of CP, in addition to the lack of controls and disease-specific outcome measures, all contribute to this lack of evidence. In general, treatment should always start with the least invasive means with consideration of the cost-effectiveness of treatment options.

Physical and Occupational Therapy

Therapy Methods

Physical therapists and occupational therapists working with children with CP may choose from a variety of therapy methods, including neurodevelopmental therapy, Vojta, Peto, and Rood. There is, however, no clear scientific evidence to support the superior effectiveness of any one particular approach. Often, therapists will use a combination of these therapeutic methods in association with an emphasis on functionally based therapies. The ideal duration and frequency of therapeutic programs is also not clear. There has been a recent interest in intermittent high-frequency therapy models, but controlled studies have failed to demonstrate any advantage to this approach (67,68).

Stretching

Children with CP are at significant risk for contracture formation due to muscle imbalances and static positioning. Contractures can interfere with comfortable positioning, functional activities and care needs, such as dressing, bathing, and toileting. After an initial assessment of baseline range of motion, institution of a daily home exercise program with repetitive stretching exercises is usually recommended, although there is no clear evidence to support its efficacy or provide guidance in regards to the ideal frequency or duration. There is some evidence to suggest that a sustained stretch is preferable to manual stretching (69). Positioning techniques, orthotic devices, splints, and casting are often recommended to provide a more prolonged stretch. Serial casting is a technique where a series of successive casts are applied in the hopes of progressively increasing the range of motion with each cast. It is used most frequently at the ankle joint, often in conjunction with botulinum toxin serotype A (BoNT-A), in order to improve dorsiflexion range of motion. Systematic reviews of the literature reveal little evidence to suggest casting is superior to no casting, primarily due to the lack of randomized controlled trials (70). The evidence does suggest a short-term effect on improved range of motion (71) and stride length during ambulation (72). Although a number of small randomized controlled trials (RCTs) have compared BoNT-A and casting, there is no strong and consistent evidence that casting, BoNT-A, or the combination of the two is superior to the others (70). Lack of evidence was primarily attributed to methodological limitations of the available studies.

Strengthening

Formalized strength testing in ambulatory children with spastic diparesis or hemiparesis has confirmed greater weakness in all muscles tested using age-matched controls (73). Weakness was more pronounced distally, as expected, and hip flexors and plantar flexors were relatively stronger than their antagonists when compared to the strength ratios of the control

group. Strength of the uninvolved side in children with hemiparesis was also weaker than age-matched controls (73). Deficits in voluntary muscle contraction in CP are felt to be due to decreased central nervous system motor unit recruitment, increased antagonist coactivation, and changes in muscle morphology, including muscle fiber atrophy and increased fat and connective tissue (74). This weakness is thought to be a large contributor to functional deficits in children with CP, but historically, strengthening programs were not recommended due to concerns of increasing spasticity. A number of studies have shown, however, that strengthening programs can increase strength without adverse effects such as increased spasticity, resulting in an increased interest in strengthening programs for children with CP (75,76).

Although strengthening has the potential to positively affect children with CP in many areas of the International Classification of Functioning, Disability, and Health (ICF) model, most studies have focused on changes in strength alone. Recent studies have begun to evaluate changes in gross motor function related to increased strength. Improved gross motor function, as measured by the Gross Motor Function Measure (GMFM), has been reported following a 6- to 8-week program of strengthening (77–79). Not all studies have demonstrated a positive effect with strengthening. An RCT evaluating the effects of a 9-month strength training program in addition to conventional physical therapy, versus therapy only following orthopedic surgery, did not demonstrate any improved function in the treatment group (80). Although not typically measured, increased participation and self-esteem have also been associated with participation in a strengthening program (81,82). Strengthening appears to be a promising intervention for children with CP, but future studies are needed to determine the effect of contextual and individual patient factors on a wide variety of potential outcomes, including societal participation.

Partial Body Weight Support Treadmill Training (PBWSTT)

PBWSTT reduces the amount of weight required to support patients ambulating on a treadmill by utilizing a postural control system consisting of a harness. It has been effectively used in adults with diparesis and hemiparesis, and its use is gaining popularity in children with CP. Current theories of motor learning suggest that task-specific repetitive practice can improve activities, including walking, in people with neurologic disorders such as CP (83–85). The theoretical basis of this treatment is an activation of spinal and supraspinal pattern generators described in animal experiments with subsequent development of locomotion patterns (86). PBWSTT in nonambulatory subjects with cerebral palsy has demonstrated significant improvements in the standing and walking sections of the GMFM and functional gains, including the ability to transfer from a sitting to standing position without use of the arms, walking and stopping, and climbing stairs in some patients (87). An additional study, using a matched-pairs design, evaluated the effects of PBWSTT conducted twice weekly for six weeks in order to evaluate the walking speed and endurance of children with CP, with a GMFCS level of III or IV and revealed a significant increase in self-selected walking speed (83).

PBWSTT enabled by a driven gait orthosis (DGO) utilizes two mechanically driven leg orthoses, resulting in a kinematic pattern resembling normal walking. This allows for an intensification of locomotor training by increasing the amount of stepping practice, as well as altering the amount of body weight support being provided while decreasing the therapist's manual assistance. To date, few studies have reported on the effects of DGO in children. A study of 10 children with CP demonstrated a significant increase in gait speed, as well as markedly improved GMFM scores in Dimensions D (standing) and E (walking) following 10 to 13 sessions of using a DGO (88).

Constraint-Induced Movement Therapy (CIMT)

CIMT was developed for treating adults with hemiparesis or "learned nonuse" following a stroke (89). The therapy includes intensive motor practice or shaping of the paretic upper extremity combined with restraint of the uninvolved extremity. CIMT is defined as restraint of the unaffected limb in conjunction with at least three hours per day of therapy for at least two consecutive weeks, whereas modified CIMT requires restraining the unaffected limb for fewer than three hours per day with therapy. Forced-use therapy involves restraining the unaffected limb with no additional therapy (90).

Children with hemiparetic CP have been described as having a "developmental disregard" for their impaired upper extremity (91). The favorable reports of CIMT in adults with stroke have resulted in an interest in applying the technique to children with hemiparetic CP. Preliminary results of controlled studies on a small number of subjects have revealed improved functional use of the affected extremity following CIMT (91), modified CIMT (92), and forced use (93,94). Cortical reorganization was also demonstrated by functional MRI and magnetoencephalography in case report of a child with hemiparetic CP following modified CIMT (95).

The preferred frequency, duration, or method of CIMT has yet to be determined. A variety of methods have been used to restrain the unaffected arm, including a long-arm bivalved cast, a short-arm cast, a sling, and a fabric glove with built-in stiff volar plastic

splint. The child who is most likely to benefit from this therapy has also yet to be identified. In general, it is believed that the child must have the cognitive ability to understand and follow directions, the ability to at least grossly grasp and release an object, and have adequate balance to not be at substantial risk for falls when wearing the restraint (96). The ideal age for CIMT is unknown, but one study comparing CIMT in children ages 4 to 8 versus 9 to 13 years showed equal efficacy in either age group (97). Because the time involved in carrying through with a CIMT program can be difficult for parents and constraint of a child's good limb has the potential to lead to frustration on the part of the child, further carefully designed studies need to be undertaken to answer these important questions.

Electrical Stimulation

Interest in the use of electrical stimulation in CP is growing. Proponents of electrical stimulation suggest that it increases strength and motor function, and it is an attractive alternative for strengthening in children with poor selective motor control (98).

Neuromuscular electrical stimulation (NMES). NMES utilizes electrical current to produce a visible muscle contraction. The results of two small case series found increased active and passive range of motion at the ankle after stimulation of the anterior tibialis (99) and improved sitting balance following stimulation of the abdominal and posterior back muscles (100). Two RCTs failed to identify any statistically significant improvement in strength or function following NMES of the quadriceps (101) or gluteus maximus (102), but both of these studies were underpowered.

Functional electrical stimulation (FES). If NMES is used to make a muscle contract during a functional activity, it is termed FES. FES is commonly used at the anterior tibialis muscle to increase dorsiflexion during ambulation. A small case series documented improvement in heel strike and ankle dorsiflexion following FES (103). Another study identified clinically significant improvements in gait in only 3/8 subjects, as measured by a three-dimensional gait analysis (104). One proposed reason for lack of response was spasticity of the antagonist muscles limiting range and speed of movement.

Threshold electrical stimulation (TES). TES is a low-level electrical stimulus, often applied during sleep, that does not result in a visible muscle contraction. The proposed mechanism of TES is that increasing blood flow during a time of heightened trophic hormone secretion results in increased muscle bulk (105). There have been four RCTs evaluating TES to date, and three of them failed to show any improvement in strength or function (101,106,107). The parents, however, reported a perceived positive effect of treatment in two of the studies (106,107), and a decreased impact on disability as measured by the Lifestyle Assessment Questionnaire was found in the third (101). In the only positive RCT, children with spastic diparesis with prior selective dorsal rhizotomy were found to have improved GMFM scores following TES, despite a lack of significant improvements in strength, range of motion (ROM), or tone (108).

A systematic review of electrical stimulation in CP concluded that the scarcity of well-controlled trials makes it difficult to support definitively or discard the use of this therapy (98). In addition, the authors concluded that the available literature appears to provide more evidence to support the use of NMES than TES. Further studies with more rigorous designs, longer follow-up, larger sample sizes of more homogenous subjects, and clarity in the reporting of stimulation parameters are recommended to clarify the age and type of patient most likely to benefit from this intervention (98).

Speech Therapy

Involvement of a speech and language pathologist is useful in the assessment of children prior to early intervention or early childhood educational planning. Many children with CP have oromotor deficits, dysphagia, dysphonia, and/or articulation and language deficits. It is essential to recognize these deficits promptly and enroll these children into speech therapy services to address treatment strategies in an effort to correct or improve these concerns.

Hypertonia Management

Hypertonicity affects the majority of children with CP (109,110). It may occur focally in distinct muscle groups, as is often the case in diparesis or hemiparesis, or more globally, affecting the majority of axial and appendicular skeletal muscles. Hypertonicity can result in a number of negative effects. It can interfere with positioning, contribute to the formation of contractures and musculoskeletal deformities, and be a source of discomfort. It can also negatively affect function and make caregiver tasks, such as transfers and dressing, more difficult. Increased tone can sometimes assist with function. For example, increased extensor tone in the lower extremities may assist with standing and transfers.

A wide variety of treatment options for hypertonicity are available, including oral medications, nerve blocks, and surgery. Determining whether abnormal

tone is present globally or focally and the magnitude of its effect on an individual's musculoskeletal system, function, and comfort should guide one's treatment plan. The specific goals of tone reduction should always be determined prior to any intervention. The first-line approach should always include stretching, splinting, and positioning as appropriate. Other medical or surgical interventions can be can be used in conjunction with these when further reduction in abnormal tone is desired.

Chemical Denervation

Chemical denervation should be considered for the treatment of significant focal increases in tone.

Alcohol Blocks. Alcohol nerve and motor point blocks have been used for many years to reduce focal increases in tone. Phenol injections, at 3% to 5% solutions, either at motor points of selected muscles or perineurally, denature proteins and disrupt efferent signals from hyperexcitable anterior horn cells by inducing necrosis of axons (111–113). Alcohol blocks have the potential to cause painful dysesthesias (113). Nerves that are more commonly treated with phenol include the musculocutaneous and obturator nerves, given the reduced sensory function of these nerves and the lower risk for dysesthesias. The low cost of phenol, coupled with reports of duration of action exceeding 12 months (114), render phenol injections an attractive treatment option in selected patients with focal spasticity (111). They are frequently done under general anesthesia, however, adding additional risks and costs.

Botulinum Neurotoxin (BoNT). BoNT is a protein composed of a heavy chain, which binds nerve terminals at the neuromuscular junction, and a light chain, which is transported into the nerve terminal blocking the release of acetylcholine presynaptically and thereby weakening the force of muscle contraction produced by the hyperexcitable motor neurons. BoNT exists in seven serotypes, designated A through G. Serotypes A and B are approved by the Food and Drug Administration (FDA) for the treatment of dystonia in adults. The FDA has not approved BoNT for the treatment of spasticity in children. BoNT-A is marketed as Botox in the United States and Dysport in Europe. BoNT-B is marketed as Myobloc.

Muscles commonly treated with BoNT include the gastrocsoleus complex, hamstrings, hip adductors, and flexor synergy muscles of the upper extremity. Intramuscular injections can be localized by surface landmarks, electromyographic guidance, and/or ultrasound. Following injection, muscle relaxation is evident within 48 to 72 hours and persists for a period of 3 to 6 months (115). Dosing is based on units derived from the mouse lethality assay and is not equivalent among the various brands. It is dependent upon both body weight and size of the target muscle(s). Universally accepted dosing guidelines do not exist, but a consensus statement (116) and systematic reviews (117,118) of dosing and injection techniques are available for guidance. Injections are typically spaced a minimum of three months apart due to concerns of antibody formation in an estimated 5% of patients, resulting in potential resistance (111,119).

Many studies in the literature describe the effects of BoNT-A in children with CP. A systematic review of the literature summarized 17 controlled trials (120). The literature supports improvement in gait over the one to three months following injections into the gastrocnemius muscles for spastic equinus (121–125). Two small open-label studies found modest improvements in either gait kinematics or muscle length following injection into the hamstrings (126,127). Several small trials evaluating the effectiveness of casting of the ankle in addition to BoNT-A failed to show any additional benefit (128–130). Injections into the hip adductors resulted in improved range of motion (131) and decreased postoperative pain in children undergoing adductor lengthenings (132) in two RCTs. Two small RCTs addressing the use of BoNT-A in the upper extremities described modest improvements in tone and ROM, without a significant change in function. The authors of the review concluded that more research needs to be done to determine the optimal choice of muscles, the most appropriate dose and number of injection sites, the safety of repeated and long-term injections, and the risk of development of secondary resistance to BoNT due to antibody formation (120).

Side effects are rare with BoNT, but may include pain during injection, infection, bleeding, a cool feeling in injected limbs, rash, allergic reaction, flulike symptoms, excessive weakness, and fatigue (123,133,134). Reports of serious or potentially life-threatening side effects from BoNT are extremely rare. The FDA issued a statement on February 8, 2008, identifying cases of respiratory failure and mortality in children with CP linked to injection with botulinum toxin serotypes A and B (135). The FDA stated that "posting the information does not mean [the] FDA has concluded that there is a causal relationship between the drug products and the emerging safety issue (135)." In addition, rare cases of serious systemic effects have been reported in the literature in children receiving higher doses of BoNT (136,137). Caution is recommended when injecting children with pseudobulbar palsy.

Oral Medications

Oral medications are often used as an early treatment strategy for global spasticity. Medications that are most

frequently used include baclofen (Lioresal), dantrolene sodium (Dantrium), clonidine, diazepam (Valium), and tizanidine (Zanaflex). All of these medications work through the central nervous system, with the exception of dantrolene sodium and, therefore, have the potential for sedation (Table 8.2). None of these medications have been found to be universally effective in relieving spasticity (138), and evidence related to functional improvement is extremely sparse. The choice of medications is, therefore, often based on the impact of potential side effects on the individual patient.

8.2 Medications Used to Treat Spasticity in Children

DRUG	MECHANISM OF ACTION	SIDE EFFECTS AND PRECAUTIONS	PHARMACOLOGY AND DOSING
Baclofen	Binds to receptors (GABA) in the spinal cord to inhibit reflexes that lead to increased tone Also binds to receptors in the brain leading to sedation	Sedation, confusion, nausea, dizziness, muscle weakness, hypotonia, ataxia, and paresthesias Can cause loss of seizure control Withdrawal can produce seizures, rebound hypertonia, fever, and death	Rapidly absorbed after oral dosing, mean half-life of 3.5h Excreted mainly through the kidney Dosing: in children start 2.5–5 mg/d, increase to 30 mg/d (in children 2–7 years of age) or 60 mg/d (in children 8 years of age and older)
Diazepam	Facilitates post-synaptic binding of a neurotransmitter (GABA) in the brain stem, reticular formation and spinal cord to inhibit reflexes that lead to increased tone	Central nervous system depression causing sedation, decreased motor coordination, impaired attention and memory Overdoses and withdrawal both occur The sedative effect generally limits use to severely involved children	Well absorbed after oral dosing, mean half-life 20–80 h Metabolized mainly in the liver In children, doses range from 0.12–0.8 mg/kg/d in divided doses
Clonidine	Alpha2-agonist. Acts in both the brain and spinal cord to enhance presynaptic inhibition of reflexes that lead to increased tone.	Bradycardia, hypotension, dry mouth, drowsiness, dizziness, constipation, and depression These side effects are common and cause half of patients to discontinue the medication	Well absorbed after oral dosing, mean half-life is 5–19 h Half is metabolized in liver and half is excreted by kidney Start with 0.05 mg bid, titrate up until side effects limit tolerance May use patch
Tizanidine	Alpha2-agonist Acts in both the brain and spinal cord to enhance presynaptic inhibition of reflexes that lead to increased tone	Dry mouth, sedation, dizziness, visual hallucinations, elevated liver enzymes, insomnia, and muscle weakness	Well absorbed after oral dosing, half-life 2.5 h Extensive first pass metabolism in liver Start with 2 mg at bedtime and increase until side effects limit tolerance, maximum 36 mg/d
Dantrolene sodium	Works directly on the muscle to decrease muscle force produced during contraction Little effect on smooth and cardiac muscles	Most important side effects is hepatotoxicity (2%), which may be severe Liver function tests must be monitored monthly, initially, and then several times per year Other side effects are mild sedation, dizziness, diarrhea, and paresthesias	Oral dose is approximately 70% absorbed in small intestine, half-life is 15 hours Mostly metabolized in the liver Pediatric doses range from 0.5 mg/kg, bid, up to a maximum of 3 mg/kg, qid

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Benzodiazepines. Benzodiazepines have an inhibitory effect at both the spinal cord and supraspinal levels mediated through binding near but not at the gamma-aminobutyric acid (GABA) receptors and increasing the affinity of GABA for GABAA receptors (139). Diazepam is the most frequently used benzodiazepine and oldest antispasticity medication that is still in use (140), but like other oral medications in CP, its effectiveness has not been well evaluated. It is rapidly absorbed, reaching peak drug levels an hour after drug administration. The positive effect of diazepam may be related to general relaxation that permits improvements, especially in those individuals with athetosis and spasticity (141,142).

Baclofen. Baclofen is a GABA analogue that acts at the spinal cord level to impede the release of excitatory neurotransmitters implicated in causing spasticity (143). Low lipid solubility impedes passage through the blood–brain barrier with more than 90% of the absorbed drug remaining in the systemic circulation (144). As a result, large doses may be necessary to achieve an effect, which may result in dose-related side effects such as drowsiness. Very few studies have been published regarding the use of oral baclofen in CP. Two small double-blind, placebo-controlled, crossover trials produced differing conclusions regarding the effectiveness of baclofen in reducing spasticity, but neither employed validated outcome measures (145,146). Additional studies assessed the effect of oral baclofen for reduction of spasticity and improved function in small numbers of subjects with moderate to severe spasticity. One study showed possible deleterious effects on motor function (117), while the other demonstrated no difference with placebo except in goal attainment (147).

Dantrolene Sodium. Dantrolene sodium is unique in that it works primarily through actions on the skeletal muscle and not through central nervous system pathways. It inhibits the release of calcium from the sarcoplasmic reticulum, thereby uncoupling electrical excitation from muscle contraction and reducing contraction intensity. It is well absorbed within three to six hours after ingestion and is metabolized in the liver to 5-hydroxydantrolene, with peak effect in four to eight hours (148). Doses in children range up to 12 mg/kg/day (142). It is often suggested that dantrolene be considered for the treatment of spasticity of cerebral origin because its mode of action is not central nervous system–mediated and it is less likely to be sedating (140,142,149). Side effects from treatment, however, can include mild sedation as well as nausea, vomiting, and diarrhea. Use of dantrolene is also associated with hepatotoxicity (148,150). Liver function studies should be done prior to instituting treatment and periodically while on maintenance therapy (140). There are a few published trials of Dantrium in CP. One report of long-term use of dantrolene in children with spastic diparesis indicated that young children achieved greater levels of function than predicted prior to dantrolene administration and older children were able to move more easily and maintain their highest level of function (151).

Additional oral medications used to treat spasticity in children with CP include alpha$_2$-adrenergic agonists, such as clonidine and tizanidine, as well as certain anticonvulsants, including gabapentin (Neurontin). The alpha$_2$-adrenergic agonists result in decreased motoneuron excitability by decreasing the release of excitatory amino acids (150). The side effects associated with these agents are frequently the cause of their more limited use and include nausea, vomiting, hypotension, sedation, dry mouth, and hepatotoxicity. In addition, reversible liver enzyme elevations have been noted in 2% to 5% of patients (140). Gabapentin is structurally similar to GABA, readily crosses the blood–brain barrier, and is not protein-bound. It does not activate GABA, but results in increased brain levels of it (140). Reports of its use in children with spasticity are not available as of yet.

Intrathecal Baclofen (ITB)

ITB was first described by Penn and associates in 1984 and was FDA-approved for the treatment of spasticity of cerebral origin in 1996. Baclofen is delivered directly to the cerebrospinal fluid via a catheter connected to an implanted device in the abdomen. The device contains a peristaltic pump, a battery with an operational life of four to seven years, a reservoir for baclofen, and electronic controls that permit regulation of the pump by telemetry (143) (Fig. 8.12). This feature allows baclofen infusion rates to be either continuous throughout the

Figure 8.12 Synchromed II programmable pump.

day or at varied dosages in order to accommodate the patient's specific needs. By infusing baclofen directly into the subarachnoid space around the spinal cord, potentiation of GABA-mediated inhibition of spasticity can be achieved while minimizing side effects related to high levels of baclofen in the brain (111). Administration of intrathecal baclofen produces levels of baclofen in the lumbar cerebrospinal fluid that are 30-fold higher than those attained with oral administration (111). The half-life of intrathecal baclofen in the cerebrospinal fluid is five hours (152).

Candidates for ITB have severe, generalized tone that has not been successfully managed with oral medications and other more conservative measures. The increased tone must have a significant effect on function, ease of care, or comfort. Intrathecal pumps can be implanted in children generally greater than 15 kg in body weight (111,153). Prior to surgical implantation, a test dose of 50–100 μg of intrathecal baclofen is typically given, via lumbar puncture, to verify a reduction in tone. Occasionally, a repeat test dose at a higher dose is necessary if results are inconclusive.

Once implanted, the intrathecal pump is typically programmed to deliver baclofen at a continuous rate, typically at a daily dose similar to the dose given during the trial. The dose is not related to age or weight (152), and intrathecal baclofen dosages typically increase over the first year of treatment and then stabilize (143). Refills of intrathecal baclofen are generally needed every one to six months, depending on baclofen infusion dosage, the size of the pump, and the concentration of the baclofen being used.

Complications from ITB can result from programming error, pump failure, catheter failure, and infection. The majority of these problems involve breakage or disconnection of the catheter, but can also include blockage and kinking (140,154). The most common postoperative complications are pump pocket collections and infections (111). Infection may remain isolated to the pump pocket or may track along the catheter, resulting in meningitis (152,154). Pumps have also been reported to flip, requiring either manual flipping to allow refill or surgical correction of the problem (140).

Catheter or pump dysfunction can result in decreased baclofen delivery and baclofen withdrawal. Intrathecal baclofen withdrawal can also be seen in cases of battery failure without low battery alarm warning (140). Early symptoms of withdrawal include pruritis, dysphoria, irritability, increased spasticity, tachycardia, fever, and changes in blood pressure (155). If not recognized and managed optimally, baclofen withdrawal may progress to serious and life-threatening complications, including severe hyperthermia, seizures, rhabdomyolysis, disseminated intravascular coagulation, altered mental status, psychomotor agitation followed by multisystem failure, and death (156,157). Immediate treatment with high-dose oral baclofen and referral to an emergency room setting is recommended in these scenarios. Investigations into the causes for withdrawal should then ensue, including plain radiographs to assess pump and catheter placement in comparison to previous radiographs. Further studies may include dye or isotope studies to assess for catheter placement, leakage, and kinking.

Treatment for withdrawal can include any combination of oral baclofen, intravenous diazepam, or infusion of intrathecal baclofen through use of a lumbar drain (158). Cyproheptadine, a serotonin antagonist, has also been used as an adjunct to baclofen and diazepam for treatment of severe intrathecal baclofen withdrawal (159,160). Dantrolene sodium use should also be considered in patients with suspected rhabdomyolysis as a result of withdrawal.

Overdoses have been reported, typically as a result of human error in programming or refill procedure (140). Symptoms can include nausea, vomiting, respiratory depression, and reversible coma. In such cases, the pump is stopped through programming and respiratory support is provided until the effects of baclofen have worn off. Intravenous physostigmine or withdrawal of 30 to 40 mL of cerebrospinal fluid can be tried in severe overdoses (155).

A number of studies have reported on the outcomes of ITB. Randomized controlled trials have shown a significant decrease in spasticity (154,161). Noncontrolled trials have demonstrated improvements in joint range of motion, reduced pain, ease of care, and function (162–166). Treatment with intrathecal baclofen is also associated with an increase in weight gain velocity (167). Retrospective studies in children with cerebral palsy receiving ITB document varying effects on scoliosis, including rapid progression (168,169) and/or no significant effect on curve progression, pelvic obliquity, or the incidence of scoliosis when compared with matched controls (170).

Selective Dorsal Rhizotomy (SDR)

SDR is a neurosurgical procedure that involves partial sensory deafferentation at the levels of L1 through S2 nerve rootlets (171). Operative technique involves the performance of single or multilevel osteoplastic laminectomies, exposing the L2–S2 roots (111,172). Motor and sensory roots are separated to allow for electrical stimulation of individual sensory roots. The selection of rootlets for cutting is based on the lower extremity muscular response to electrical stimulation of the rootlets. Although there is variability in percentages of rootlets cut, in general, a maximum of 50% of the sensory rootlets at any level are cut (173). Following the procedure, the reduction in spasticity often unmasks a significant amount of lower extremity weakness. As a

result, an extensive amount of intensive therapy is necessary to guide the patient through appropriate motor patterns and strengthening programs. Ideal candidates for SDR include children between the ages of 3 and 8 years of age who are GMFCS level III or IV (174).

A meta-analysis of three randomized controlled studies comparing SDR plus physical therapy with physical therapy alone has been completed (174). Findings included a clinically important decrease in spasticity, as well as a small but statistically significant advantage in function (GMFM-88) with SDR plus physical therapy. The subjects in these studies were primarily ambulatory children with spastic diparesis; those with dystonia, athetosis, and ataxia were excluded. An additional larger nonrandomized controlled study compared SDR with physical therapy to physical therapy alone in children with spastic paraparesis, GMFCS levels I to III (175). Results of this study were similar to studies in the meta-analysis, including gains in strength, gait speed, and overall gross motor function in children who received SDR plus physical therapy (175).

Although immediate perioperative complications are not uncommon with SDR, long-term complications such as sensory dysfunction, bowel or bladder dysfunction, or back pain are infrequent (176). The risk of subsequent spinal deformities may increase after laminectomies or laminoplasties done in conjunction with SDR, although this may be less of a problem in the lumbar or lumbosacral area than higher in the spinal column (177). Decreased spasticity and alterations in the balance of muscle tone in the trunk and hips may also influence the development of spinal deformities (177). A retrospective review of patients who underwent SDR reported a 32% incidence of new spinal deformity at five years after multilevel laminectomies, including scoliosis, hyperlordosis, and hyperkyphosis (178). SDR may reduce the need for subsequent orthopedic surgical interventions (179,180).

Orthopedic Surgery

Orthopedic surgery is most often recommended in children with muscles that are dysphasic, firing out of phase, or those muscles that show excessive activity while working in phase, thereby overpowering their antagonist and thereby inhibiting smooth joint motion (181). The combination of this muscular imbalance with the lack of stretching of the muscles in the relaxed state leads to contracture formation as the muscle–tendon unit fails to keep up with the skeletal growth of the child, and may lead to bony changes as well as fixed deformities (182). The usual goal of surgery is to weaken these dysphasic muscles and reduce potential contracture formation and spasticity. The muscles that are most frequently addressed surgically are those that cross two joints, including the hip adductors, hip flexors, hamstrings, rectus femoris, and gastrocsoleus complex. Rotational osteotomies are occasionally done to correct femoral anteversion or tibial torsion that results in significant gait disturbances.

When improved function is the goal of surgery, multiple muscles and joints may be targeted because they are all interrelated in specific movement patterns; therefore, a single multilevel surgical procedure is more common than multiple staged surgeries (183–185). A common multilevel soft tissue surgical approach includes three procedures: the hamstring lengthening, rectus femoris transfer, and gastrocsoleus lengthening (186). Assessment of mobility after multilevel surgery for CP with use of a functional walking scale was performed in 85 nonambulatory children who were able to attain independent sitting balance by the age of 5 to 6 but who did not have access to previous spasticity management (187). Significant improvements in joint contractures were noted in addition to the fact that all patients gained walking capabilities, including one-third of the patients ambulating community distances (187).

Orthopedic surgery is ideally delayed until the age of 4 to 7 years, due to the high risk of recurrence of tightness and contracture formation in younger children (182,183,188). In a retrospective study, a recurrence rate for Achilles tendon lengthening was found in 18% of children with diparesis and 41% with hemiparesis (188). Children older than 6 years of age at the time of initial operation were not found to commonly have recurrence.

Postoperative care should include aggressive pain management to minimize pain-related muscle spasms, which may further increase discomfort. Rapid mobilization with minimal casting is also recommended, usually with only a two- to three-day period of recumbency following surgery. The need for physical therapy should be assessed and started as soon as possible if necessary to minimize postoperative weakness and disuse atrophy, as well as improve muscular reeducation and training in those muscles or tendons that were manipulated.

Surgical spinal fusion is not uncommon in CP. Indications for surgical management may vary between centers, but, in general, curvatures greater than 40 degrees in skeletally immature persons and greater than 50 degrees in skeletally mature persons are recommended for evaluation and consideration of possible fusion surgery (189). Before pursing spinal fusion, the child should receive careful preoperative evaluation and preparation, including close monitoring of nutrition and respiratory status in order to reduce postoperative complications. Goals of surgical intervention include prevention of curve progression with subsequent pulmonary and skin complications,

as well as improved sitting balance, positioning, and comfort.

Orthoses

Many children with cerebral palsy utilize orthotic devices for maintaining or increasing range of motion, protection or stabilization of a joint, or promotion of functional activity. Orthoses can be expensive, and with a wide variety of designs to choose from, care should be taken to provide the appropriate design to meet the child's needs.

Upper Extremity (UE) Orthoses

Static wrist hand orthosis (WHO) are commonly used in CP to improve hand position for functional activities and to maintain range of motion. Dynamic WHO are much less commonly used because children are often reluctant to use them for functional activities, in part due to the decreased sensory feedback caused by the orthosis. The use of either type is not well studied in CP, but a small controlled study of 10 children revealed increased grip and dexterity with the use of dynamic splint (190).

Lower Extremity (LE) Orthoses

Many different types of LE orthoses are utilized in the management of CP, including supramalleolar orthotics (SMOs), solid ankle foot orthotics (AFOs), hinged AFOs, posterior spring-leaf AFOs, and ground-reactive AFOs. Knee ankle foot orthoses and hip knee ankle foot orthoses are rarely used in CP. In spite of many published studies on the effectiveness of LE orthotics in CP, precise indications have yet to be established. A systematic review of 27 studies (191) resulted in the following recommendations: a) Only orthoses that extend to the knee and have a rigid ankle, leaf spring, or hinged design with a plantarflexion stop can prevent equinus deformities; b) SMO designs with tone-reducing features (or dynamic ankle foot orthotics) do not prevent equinus; c) preventing plantarflexion or equinus has been shown to improve the temporal parameters of gait, such as walking speed and stride length for the majority of children, and thereby improved gait efficiency; d) children with less severe impairments often performed better on stairs and moving from sitting to standing in less restrictive hinged, leaf spring, or SMO designs.

Rotational-control orthoses, both twister cables and rotation straps, are also used occasionally in children with cerebral palsy. Twister cables have a pelvic band with attached cables of twisted spring steel, with torque typically applied to provide an external rotation force by attaching to the shoes or AFOs. Rotation straps are elastic and attach to buckles on AFOs or to an eyelet attachment on shoestrings, and can provide internal or external rotation forces depending on the application of wrapping the straps around the lower extremities. While these orthoses can help to control rotation, especially in younger children, families often complain that they are cumbersome and often prefer not to use them.

Spinal Orthoses

The role of spinal orthoses in children with CP and scoliosis has not been well studied. There are no RCTs, and there is no agreement as to whether spinal orthoses can prevent the progression of scoliosis. There is general agreement that if bracing controls the progression of scoliosis, it will not work in every patient (192) and it at best is only likely to slow progression, delaying surgery until a more ideal time (193). Regardless of its effect on curve progression, a positive effect on sitting stability and function has been reported by parents and caregivers (194), but this also has not been well studied.

Adaptive Equipment

The goal for the use of adaptive equipment is to improve positioning either in the supine or sitting position, or to improve level of function in self-care skills, including in the home, school, or community. These devices include, but are not limited to, seating or support systems, mobility devices, augmentative communication devices, computer or computer aids, and environmental control devices. Assessment by a team, including physicians and therapists to assess physical capabilities, as well as to develop and refine appropriate goals, is essential to address and optimize adaptive equipment needs for children with cerebral palsy.

Alternative Therapy

The use of complementary and alternative medicine (CAM) in cerebral palsy is not uncommon. CAM has been defined by The American Academy of Pediatrics as "strategies that have not met the standards of clinical effectiveness, either through randomized controlled clinical trials or through the consensus of the biomedical community (195)" and by the National Center for Complementary and Alternative Medicine "as a group of diverse medical and health care systems, practices, and products that are not presently considered to be part of conventional Western medicine (196)." It is not surprising that caregivers would be attracted to therapies that promise significant functional improvement when traditional medicine may appear to have little to offer. CAM is more commonly used in children with

chronic diseases such as CP despite lack of substantiating evidence (197). CAM is often used in addition to orthodox medicine, but often its use is not discussed with the child's treating physician secondary to a feared negative response (197). Several studies have documented increased use of CAM in children placed in higher GMFCS categories (198,199). One study found that 56% of families surveyed had utilized at least one CAM therapy for their child with CP (198). The most commonly utilized therapies were massage therapy (25%) and aquatherapy (25%). The most significant predictors of use were the child's age (younger), lack of independent mobility, and parental use of CAM (198). Other CAM therapies utilized by children with CP include conductive education, patterning, hyperbaric oxygen therapy, Adeli suit therapy, acupuncture, craniosacral therapy, chiropractic manipulation, and many others (Table 8.3).

Hyperbaric Oxygen Therapy (HBOT)

Proponents of HBOT propose that "dormant areas" can be found surrounding injured areas in the brains of children with CP and that high levels of oxygen in the brain reactivate, or "wake up," the cells of this dormant area (200). Delivery of hyperbaric oxygen typically consists of treatments with pressures of 1.5 to 1.75 atmospheres for one hour per session, sometimes as often as five to six times per week, for up to 40 treatment sessions in a phase of treatment. A blinded, randomized, controlled clinical trial of 111 children with cerebral palsy compared treatment with hyperbaric oxygen at 1.75 atmospheres with a control group that received air at a pressure of 1.3 atmospheres (201). Both groups demonstrated significant functional improvements, but no differences were found between the groups. While some authors have argued that this demonstrates the value of elevated oxygen, even at minimal levels (202), others argue that the effect demonstrates a "powerful clinical trials effect (203)," with the effect primarily due to highly motivated parents spending many hours with the children in an intensive setting, knowing that developmental outcomes would be evaluated (200). A systematic review of the evidence revealed that there is inadequate evidence to establish a significant benefit of HBOT or for identifying potential adverse effects of HBOT in children with CP (204).

Conductive Education (CE)

CE is a combined therapeutic and pedagogic program for children with CP developed by the Hungarian child neurologist Andras Petö in the 1940s that has been given increased attention in Western countries in recent years with the main elements being task-oriented learning within highly structured programs; facilitating and commenting on motor actions by rhythmic intending, for example, rhythmic speaking or singing; integration of manual abilities into the context of activities of daily living; and child-oriented group settings to facilitate psychosocial learning to increase the level of participation (205). In this program, the "conductor" is trained in special education and therapy and administers the conductive education program. As CE has spread from Hungary to other countries, it has been packaged in an array of delivery models, making it difficult to ascertain specific criteria that define CE as a program (206). The use of adaptive equipment such as splints, walkers, and wheelchairs in the classroom is generally discouraged (200). An American Academy of Cerebral Palsy and Developmental Medicine (AACPDM) Treatment Outcomes Committee Evidence Report was conducted to evaluate the current state of evidence regarding CE and found that the present literature base does not provide conclusive evidence either in support of or against CE as an intervention strategy, primarily due to the limited number of studies and their weak quality (206).

Adeli Suit Therapy (AST)

AST was introduced in 1991 and incorporates a prototype of a device developed in Russia in the late 1960s to maintain neuromuscular fitness during weightlessness experienced by cosmonauts. The treatment is based upon three principles: the effect of the suit (working against resistance loads, increased proprioception, and realignment), intensive daily physical therapy for one month, and active motor participation by the patient (207). The suit consists of a vest, shorts, knee pads, and specially connected shoes; pieces of the suit are connected by hooks, rings, and elastic bands that are adjusted to optimally position limbs and joints. The bungeelike cords are adjusted by therapists to mimic normal flexor and extensor patterns of major muscle groups in an attempt to correct abnormal muscle alignment (208). The theory is that once the body is in proper alignment, aggressive movement therapy can be performed that will reeducate the brain to recognize correct movement of the muscles (208). It is also felt that deep pressure at the joints improves the sensory and proprioceptive information at that joint, enhances the vestibular system, and improves coordination (200). Treatment is typically given at a higher intensity, at one to two hours per day, multiple times per week, for a four- to six-week period. One randomized, controlled, clinical trial compared the efficacy of AST in children with CP to neurodevelopment treatment (NDT) (207). Both groups received the same intensity of treatment, totaling 20 sessions in four weeks, and were evaluated with the GMFM-66

8.3 Summary of Selected Complementary and Alternative Treatments for Cerebral Palsy

THERAPY	THEORY/BENEFITS	ADVERSE EFFECTS	EVIDENCE	COMMENTS
Hyperbaric oxygen	Awakens dormant brain tissue surrounding the original injury	Ear trauma, pneumothorax, fire and explosions	Uncontrolled studies show improvements in the treated children. Controlled study showed improvement in Treated and controls	More evidence is required before recommendations can be made; eg, what is the role of increased pressure without supplemental oxygen?
Adeli suit	Resistance across muscles can improve strength, posture, and coordination	Discomfort from suit; expense for intensive therapy and for travel to centers that prescribe the suit	No conclusive evidence either in support of or against the use of the Adeli suit	
Patterning	Passively repeating steps in normal development can overcome brain injuries	Time, energy, and expenses required for treatment	Results of uncontrolled studies are inconsistent; controlled trials show no benefits	Cannot be recommended
Electrical stimulation				More evidence is required before recommendations can be made
Threshold electrical stimulation	Increased blood flow from electrical current will lead to stronger muscles	Expense for unit; generally safe	Some uncontrolled trials show subjective improvements; controlled trials are inconclusive	
Functional neuromuscular stimulation	Increased muscle contraction will improve strength and function	Expense; infection from needles; discomfort	Evidence somewhat more positive than for threshold stimulation but still inconclusive	
Conductive education	Problems with motor skills are problems of learning; new abilities are created out of teaching	None known	Uncontrolled trials show benefit; controlled trials are mixed	Conductive education is implemented in many different ways making generalizations from a single program difficult
Hippotherapy	Riding a horse can improve muscle tone, head and trunk control, mobility in the pelvis, and equilibrium	Trauma from a fall; allergies	Uncontrolled and controlled trials show beneficial effects on body structures and functioning	Horseback riding also increases social participation
Craniosacral therapy	Therapy is used to remove impediments to the flow of cerebrospinal fluid within the crainum and spinal cord	None known	No studies showing efficacy in CP; some question the basis of the intervention	
Feldenkrais	Change of position and directed attention can relax muscles, improve movement, posture, and functioning	None known	No studies showing efficacy in CP; studies in other conditions are equivocal	
Acupuncture	Acupuncture can help to restore the normal flow of Qi, or energy	Forgotten needles, pain, bruising, and infection	Uncontrolled studies show improvements in several areas; two controlled trials also showed improvements	Appears promising, but more studies are required before specific recommendations can be made

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at baseline, after one month of AST or NDT therapy and again nine months later after they had returned to their baseline therapies. When administered with equal intensity, the AST did not show superior motor skills retention in comparison with NDT (207).

Additional Therapies

Patterning (Doman Delacatto method), hippotherapy, craniosacral therapy, Feldenkrais, and acupuncture are additional CAM therapies that are sought out by parents of children with CP. In regards to patterning, the American Academy of Pediatrics concluded that "patterning treatment continues to offer no special merit, [and] that the claims of its advocates remain unproved . . . (209)" There are a few uncontrolled and controlled studies revealing improvements in GMFM scores as well as other benefits in regard to decreasing muscle tone, improving head and trunk postural control, and developing equilibrium reactions in the trunk from hippotherapy (210–212). No published studies are available on the use of craniosacral therapy or the Feldenkrais method in children with CP. Most studies published in English regarding acupuncture are uncontrolled and primarily case series.

The American Academy of Pediatrics Committee on Children with Disabilities published recommendations for counseling families on CAM, which includes the following: maintaining a scientific perspective, providing balanced advice about therapeutic options, guarding against bias, and establishing and maintaining a trusting relationship with families (195). Ethically, families have the right to use alternative medicine therapies for their children as a matter of autonomy, but they also have the duty not to harm their children (213). The care of patients should be based, to the greatest extent possible, on existing sound evidence revealing that the therapy recommended is effective in reducing morbidity; the benefits outweigh the risks; the cost of the treatment is reasonable compared to its expected benefits; and the recommended therapy is practical, acceptable and feasible (200).

COURSE AND PROGNOSIS

Outcome Measures

Children with cerebral palsy often change over time, due either to growth and development or as a result of treatment. Various means of determining change may be employed. Subjective evaluations that ask the child, parent, or therapist their opinion are most commonly used. Occasionally, more quantified techniques are employed, particularly in research settings, although clinical use also occurs.

Outcome measures may best be classified by the domains they seek to measure and the methods of assessment. Using the International Classification of Functioning, Disability, and Health—Children and Youth Version (ICF-CY) (214), measures can be divided into those that define body functions and structures, activity, or participation (Table 8.4).

Body Structure and Function

When considering children with cerebral palsy, few outcome measures directly relate to body structure. Imaging such as functional MRI or physiologic measures like transcranial magnetic stimulation or electromyography could be considered in this domain. Because very few interventions for cerebral palsy are expected to alter body structures, such as brain tissue, these types of outcome measures are seldom employed. Many outcome measures for cerebral palsy address body function. Body function is assessed with spasticity measurement (Ashworth, modified Ashworth, and Tardieu scales, or specialized measurement systems), strength measurement (muscle grading or dynamometry), or range of motion.

Activity

Because many interventions for cerebral palsy are intended to reduce activity limitation, a wide range of outcome measures are specific for this ICF domain. Common assessments of gross motor function and walking include the Gross Motor Function Measure and Gross Motor Performance Measure as well as gait analysis, ranging from observational scales (Physicians Rating Scale) to instrumented digital kinematic analysis. Fine motor may be assessed with the Quality of Upper Extremity Skills Test, Assisting Hand Assessment, Jebsen-Taylor Hand Function Test, and Melbourne Assessment of Unilateral Upper Limb Function, among others. More global functional measures include the Functional Independence Measure for Children (WeeFIM), the Pediatric Evaluation of Disability Inventory, the Pediatric Outcomes Data Collection Instrument, and the Bruininks-Oseretsky Test of Motor Proficiency. Assessment of energy expenditure or efficiency, timed walking tests, and movement monitors are also used to assess the domain of activity in children with cerebral palsy.

Developmental assessments are generally wide in scope and used more frequently in younger children. These include the Peabody Developmental Motor Scales, Battelle Developmental Inventory, Denver II, Bayley Scales of Infant Development, and Revised Gesell Developmental Schedule.

8.4 Outcome Measures Used in Cerebral Palsy

	BODY STRUCTURE	BODY FUNCTION	ACTIVITY	PARTICIPATION	QUALITY OF LIFE OR NON-ICF DOMAINS
Administered by questionnaire or self-report			Pediatric Outcomes Data Collection Instrument (230)	Children's Assessment of Participation and Enjoyment and Preferences for Activities for Children (231) Assessment of Life Habits for Children (232)	Cerebral Palsy Quality of Life Questionnaire for Children (233) PedsQL (234) Child Health Questionnaire (235)
Measured by trained investigator or with specialized equipment	Fuctional MRI (236) MRI (35) Diffusion tensor imaging (237) Transcranial magnetic stimulation (238) PET scan (239)	Spasticity [*Ashworth scale* (240), *Modified Ashworth scale* (241), *Tardieu scale* (242), specialized systems (243)] Strength [muscle *grading* or dynamometry (244)] Range of motion (245) Electromyography (246)	Gross Motor Function Measure (215) Gross Motor Performance Measure (247) Gait Analysis [observational *scales* (248,249) *to instrumented digital* analysis (216)] Quality of Upper Extremity Skills Test (250) Assisting Hand Assessment (251) Jebsin-Taylor Hand Function Test (252) Melbourne Assessment of Unilateral Upper Limb Function (253) Functional Independence Measure *for children* (*WeeFIM*) (254) *Pediatric Evaluation of* Disability Inventory (255) *Canadian Occupational Performance Measure* (256) Bruininks-Osteretsky Test of Motor Proficiency (257) Energy expenditure/efficiency (258), movement monitoring (259) *Timed walking* (260)	*Canadian Occupational Performance Measure* (256) Pediatric Evaluation of Disability Inventory (255)	*Goal Attainment Scaling* (261)

Italicized measures are commonly used in clinical settings and may also be used for research, whereas other measures are predominantly research tools.

Participation

Participation for children with cerebral palsy is most often assessed with the Children's Assessment of Participation and Enjoyment and the Preferences for Activities for Children. The Activities Scale for Kids and Assessment of Life Habits for Children are also employed in this domain. The Pediatric Evaluation of Disability Inventory and the Canadian Occupational Performance Measure assess both activity and participation realms. Some instruments address health status or quality of life, and may be placed in the domain of participation, while other instruments assess environmental factors. Common outcome measures in this group include the Child Health Questionnaire or other generic pediatric measures, the Cerebral Palsy Quality of Life Questionnaire for Children, and Goal Attainment Scaling.

Gross Motor Function Measure (GMFM)

The GMFM is a functional outcome tool that was developed specifically for use in cerebral palsy (215). Widely used in research settings, the GMFM is also employed clinically for evaluation of children with cerebral palsy. The GMFM consists of a broad range of gross motor tasks, which a trained evaluator observes a child attempting to complete over a 45- to 60-minute time interval. Five dimensions of function (lying and rolling; sitting; crawling and kneeling; standing; and walking, running, and jumping) are examined. Specific scoring algorithms result in a score that can be used as an interval measure.

Gait Analysis

Instrumented gait analysis is another objective functional measure that is widely used in cerebral palsy (216). Many centers do not use gait analysis; other centers rely upon it heavily, particularly in guiding treatment decisions such as orthopedic surgery. This technique can only be employed for children who have some ability to walk, even if they require gait aids. Gait analysis involves having a child walk in a specialized laboratory wearing markers and muscle activity sensors. Using sophisticated computers, cameras, and force plates implanted on the floor surface, the child's movement patterns can be analyzed in great detail. Information about movement patterns in all planes, kinetics, and kinematics are generated. Although some controversy exists as to the reproducibility of gait analysis results and the means by which gait analysis should be employed to guide surgical decision-making (217), gait analysis remains a common tool for evaluation of cerebral palsy.

Quality of Life

Children with CP experience limitations in mobility and are at risk for lower participation in leisure and social activities, and therefore, there is a perception that they have a lower quality of life (QOL). The WHO defines QOL as "an individual's perception of their position in life in the context of the culture and value systems in which they live, and in relation to their goals, expectations, standards and concerns (218)." QOL is, by definition, subjective, yet most of the literature to date looking at QOL in CP has used data from parents rather than the children themselves. The literature also has tended to focus on functional skills and their role in QOL, and little attention has been paid to other important contextual factors such as environment and family functioning, which are felt to be important determinants of QOL (219).

Recent literature has begun to focus on the child's self-report of QOL and contextual factors outside of physical functioning. In a population-based study of 217 children with CP ages 6 to 12 years, the authors found that the QOL was highly variable, but about half experienced a QOL similar to typically developing children (219). Children were less likely to rate themselves low for psychosocial well-being when compared to their parents' report. Functional limitations were good indicators for physical but not psychosocial well-being, and family functioning, behavioral difficulties, and motivation were all found to be important predictors of social–emotional adaptation.

Two more recent studies of self-reported QOL found no difference in QOL between children with CP and typically developing children (220,221). One of these studies was a large population-based study of 1,174 children between 8 to 12 years in Europe (220), and the other looked at a convenience sample of 81 children 10 to 13 years with GMFCS Level I–III (221). The finding that many children perceive their QOL as similar to their peers is encouraging and suggests that children who grow up with an impairment incorporate it into their sense of self from birth and it is possible for them to embrace growth, development, and living with the same excitement as most children (220). Future large population-based studies would be helpful to validate these findings and to look more closely at the contextual factors that may affect QOL. Longitudinal studies would also be helpful to determine potential changes in QOL over time.

Prognosis for Ambulation

Shortly after caregivers are given the diagnosis of cerebral palsy, they will often want to know if their child will walk. Many studies have been published on this subject, and the best predictors of eventual ambulation

appear to be persistence of primitive reflexes, gross motor development, and type of cerebral palsy. The persistence of primitive reflexes or the absence of postural reactions at age 2 years is associated with a poor prognosis for ambulation (222). A longitudinal study of 233 children with mixed types of CP found that all of the children who were sitting by the age of 2 years eventually ambulated and that only 4% of the children who were not sitting by 4 years ever gained the ability to ambulate (223). Prognosis for eventual ambulation is also closely related to the type of cerebral palsy. Children with spastic hemiparesis have the best prognosis for ambulation, with nearly 100% achievement. More than 85% of children with spastic diparesis will eventually ambulate. The likelihood for ambulation is much less with spastic quadriparesis, but the studies have revealed a wide range of eventual ambulation of 0% to 72% (222). This wide range is likely due to differences in the population of children studied and the definition of ambulation that was used. The presence of severe intellectual impairment also is a poor predictor for walking. A large population-based study in Europe found that a severe intellectual impairment increased the risk of being unable to walk 56 times in hemiplegic CP and 9 times in bilateral spastic CP (224). If one takes into account all of these potential predictors, it possible to make a relatively accurate prognosis for ambulation by the age of 2 to 3 years. This will help the child's caregivers set realistic goals and guide appropriate therapeutic intervention.

Aging With Cerebral Palsy

The United Cerebral Palsy Association has estimated that there are approximately 400,000 adults with cerebral palsy living in the United States (225). It is expected that this number will grow due to improvements medical care. A number of studies have published data on the life expectancy of persons with cerebral palsy. A population-based Health Surveillance Registry in British Columbia was utilized to study a cohort of 3,189 persons with cerebral palsy born between 1952 and 1989 (226). Overall survival rate at 30 years was estimated to be at least 87%. Mental retardation and epilepsy were determined to have a negative effect on survival. The projected life expectancy of children who currently have cerebral palsy is unknown, as these surveillance studies are based on medical practices from previous decades.

Musculoskeletal symptoms are commonly identified complaints in adults with CP, even at a relatively young age. Issues that are commonly identified include cervical pain, back pain, and hand paresthesias (225). Other concerns include maintenance of mobility, availability of adaptive aids, incontinence, and lack of appropriate preventative medical care (225). A large number of adults with CP do not obtain regular general health evaluations or rehabilitative care. This is largely due to the lack of adult physicians with an interest and knowledge of medical issues in persons aging with cerebral palsy and the lack of an organized system of care similar to what is currently available for children with cerebral palsy.

Information on education and employment in adults with CP is limited. Reported competitive employment rates vary from 24% (227) to 53% (228). The only population-based study took place in Europe and found 33% of young adults participated in higher education (vs 77% of controls) and 29% were competitively employed (vs 82% of controls) (229). Proposed reasons for lower education and employment rates include impaired cognition, employment policies, inadequate accessibility, attitudes towards individuals with CP in the workplace, or impaired social functioning (229).

It is clear that more attention needs to be paid to issues related to aging in cerebral palsy. Adult medical care providers need to be identified and educated, and a routine means of transitioning care needs to be in place. An early emphasis should be placed on independent living skills. Adaptive equipment needs to be routinely reassessed for its appropriateness. Adults with cerebral palsy need to be aware of the community support services available to them and learn to advocate for themselves. Active vocational counseling should begin in high school. Hopefully, the growing awareness of this population will lead to improved quality of life and increased functional independence.

PEARLS OR PERILS

1. Although prematurity is a major risk factor for CP, most children with CP were not premature infants.
2. Hand preference prior to the age of 18 months may be an indication of hemiparetic CP.
3. Additional workup for an etiology other than CP should be undertaken in any child who has lost developmental milestones.
4. Sensory impairments, especially in hemiparesis, can be an important contributing factor to decreased functional hand use.
5. Children with severe motor impairments related to CP can have normal cognition.
6. Periods of rapid growth in children with CP may be associated with worsening contractures because spastic muscles fail to grow as quickly as bones.
7. Children who have a sudden increase in spasticity should be evaluated for constipation, urinary tract infection, esophagitis, musculoskeletal pain, or other potential sources of noxious stimulation.
8. Oral baclofen should be titrated up slowly to minimize sedation and titrated off slowly to minimize

the likelihood of withdrawal symptoms, including increased tone and seizures.

9. When evaluating toe walking due to equinus, always evaluate and address spasticity and contractures of more proximal muscles, in particular, the iliopsoas and the hamstrings.
10. Not all children who walk on their toes have CP. Toe walking can also be idiopathic or due to proximal muscle weakness, as is the case with Duchenne muscular dystrophy.
11. Children with CP who sit independently by age 2 years are likely to be functional ambulators, while those who fail to walk by age 4 years are unlikely to be a functional ambulatory (223).

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Spina Bifida

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Spina bifida is the second most common disability in children. The National Spina Bifida Association documents more than 70,000 individuals in the United States living with spina bifida. This is a small fraction of all those affected worldwide. Spina bifida is a complex disorder that has physical, psychological, and social implications. Medical professionals treating these individuals should have a thorough understanding of the spectrum of the disability.

EPIDEMIOLOGY

According to estimates by the Centers for Disease Control (CDC), spina bifida and anencephaly, the two most common neural tube defects (NTDs), affect approximately 3,000 pregnancies yearly in the United States. These NTDs vary in prevalence, depending on race and ethnicity, with women of African American and Asian descent having the lowest, while the highest is noted among women of Hispanic ethnicity.

It is notable that affected pregnancies in both Hispanic and non-Hispanic whites have declined significantly since the mandatory fortification of grain products in the United States with folate (see the discussion in the section "Genetic Influences"). Several trials have shown that folic acid fortification can alter a woman's risk of an NTD-affected birth from 50% to 70%. Although the impact of folic acid on NTDs does not appear to be influenced by race or ethnicity, the disparity between Hispanics and other races and ethnicities remains, and the causes are unknown at this time. It is hypothesized that differences in eating habits, supplement-taking practices, and in general an awareness of how nutrition affects pregnancy outcomes has a major impact. Indeed, differences in social structure may play a role. Studies have demonstrated also that other risk factors such as maternal obesity, nutrient intake, and supplement use are different, depending on racial/ethnic grouping. Genetic factors, some of which are discussed in a later section, have a major direct impact on NTD incidence and serve to alter susceptibility to numerous environmental influences. It is this interaction that requires continued study in order to discern how racial and ethnic factors change over time and affect NTDs.

The current American Academy of Pediatrics (AAP) guidelines for folic acid supplements are:

- All women of childbearing age: 400 micrograms or 0.4 milligrams/day
- Women with a previous NTD pregnancy: 4000 micrograms or 4 milligrams/day one month prior to conception and through the first trimester
- High-risk pregnancies (such as a mother who is taking valproic acid or has maternal diabetes): 4 milligrams/day (1)

Understanding the multifactorial etiology of this disorder is complex, but the underlying genetic risks are well established. Although recurrence risk varies around the world, most children are born to families without a prior affected child (0.1%–0.2%). The risk for recurrence in a family with one child with NTD

is 2% to 5% and increases to the 10% to 15% range if two siblings are affected. If one parent has spina bifida, the risk is 4% of having a child with a similar disorder. For a recent review, see Deak et al. and the NTD Collaborative Groups work (2).

ETIOLOGY

It is important for the clinician to understand the embryogenesis of neural tube defects. Clearly, spina bifida is a complex, heterogeneous disorder whose etiology in humans appears to be multifactorial. In simple terms, however, spina bifida, classically defined as meningocele and myelomeningocele, is the consequence of neural tube closure failure during embryonic development. The following section discusses normal central nervous system (CNS) embryogenesis and the pathological differences associated with these neural tube defects (3–6).

Normal Development

During the first two weeks, postfertilization embryonic development involves repeated cell division and organization, resulting in a blastocyst, an embryo with two layers: the epiblast and the hypoblast. The epiblast layer consists of the dorsally oriented cells adjacent to the amniotic cavity. The hypoblast layer consists of the ventrally located cells adjacent to the yolk sac. At the end of this period, on days 13–16, a primitive streak forms that begins caudally and progresses towards the rostrally located prochordal plate. The prochordal plate and the development of the primitive streak are the beginnings of the rostral-to-caudal orientation of the embryo.

The development of the primitive streak is followed by invagination of epiblast cells, forming a trough along the midline. Subsequent movement of different populations of epiblast cells remodels the embryo (ie, gastrulation) into a three-layered structure comprised of ectoderm, mesoderm, and endoderm, the precursors of all tissue types and body structures. As the primitive streak regresses, presumptive notochord cells migrate through a structure at the rostral end known as Hensen's node. These cells align themselves along the midline of the embryo between the underlying endoderm and the overlying ectoderm (presumptive neuroderm and overlying surface ectoderm). The exact process by which this occurs varies among different species, and it has not been clearly defined in humans.

In humans, the formation of the neural tube begins around Day 16, when the neuroectoderm and the laterally adjacent cutaneous ectoderm can be seen overlying the notochord in a "platelike" structure along the midline groove of the embryo. Direct cell–cell contact by the notochord is required for neural plate induction as well as the production of diffusible factors. By about day 21, the plate bends as the groove deepens, and its walls and their adjacent cutaneous epithelium begin to oppose one another.

The eventual closure of the neural tube proceeds over a period of four to six days and typically involves primary closure of the cutaneous ectoderm. This is first followed by the neuroectoderm, which subsequently separates from the overlying cutaneous ectoderm, resulting in a closed tube. Closure begins at a point just caudal to the developing rhombencephalon and proceeds via several waves rostrally rather than in the continuous "zipperlike" fashion previously envisioned. Spinal closure appears to proceed in a continuous fashion from the initial rostral closure point caudally to the end of the neural tube. There is, however, an alternative view proposed by Van Allen and colleagues that describes several closure initiation sites over the same period of time (7,8). Regardless, closure of the primary neural tube is typically complete around developmental day 27. This process, primary neurulation, completes the presumptive spinal cord down to the lower lumbar and/or upper sacral levels.

A secondary wave of neurulation begins around day 25 from a collection of remaining primitive streak cells and mesoderm located along the midline axis from the caudal end of the primary neural tube to the cloaca. These collections of cells form cavities that coalesce to form a tube that eventually becomes continuous with the primary neural tube. This process completes the formation of the sacral levels of the spinal cord and terminal filum, and is species-specific. The specific process by which secondary neurulation occurs and merges with the primary neural tube is uncertain in humans.

Understanding the process of primary and secondary neurulation is of paramount importance in comprehending the pathogenesis of spina bifida. The process of neurulation is completed by the end of the first month of embryonic development.

Expansion of the cranial brain structures via development of a primitive ventricular system is thought to be accomplished by temporary occlusion of the caudal (spinal) neural tube (days 23–27), which creates a rostral-enclosed fluid-filled space, thus providing pressure to expand the cranial lumen, providing the impetus for brain enlargement. Theory suggests that, in part, failure of this expansion pressure is a cause for Chiari malformation (9).

Neural crest cells, precursors to cell types such as melanocytes, Schwann cells, dura matter, and dorsal root, as well as autonomic ganglion, are thought to arise during this same time from the neural tube near

the junction between neuroectoderm and cutaneous ectoderm.

Pathology

Spina bifida is typically considered a primary failure of neurulation. Failure of neurulation and, thereby, loss of neural tube closure, prevents the mesoderm adjacent to the notochord from forming muscle and bone (ie, via somitic mesoderm), which normally forms around the tube to protect it. Therefore, the mechanisms involved in this process are suspect in the pathology of this disorder. Although this is the most popularly accepted theory, there are other proposed mechanisms. Dias and colleagues have discussed the idea that several forms of myelomeningoceles are not failures of neurulation, but a failure of Henson's node to lay down the notochord correctly—in other words, a failure in gastrulation (10,11) that causes significant errors in induction of the neural tube. Further research is necessary to elucidate and verify currently proposed theories.

The mechanisms by which the neural tube is formed and closed are varied. Morphogenic changes in cell populations such as wedging result in the shaping of the neural plate into a tubelike structure early in neurulation (12–14). Several mechanisms are proposed for closure of the neural tube, such as interaction between various glycoproteins and cell adhesion molecules (CAM), multiple roles for various signaling protein/receptor interactions, the interlinking of numerous cell filopodia, and formation of intercellular junctions. The current view suggests that the process likely involves all of these and perhaps others not yet visualized.

Failures of induction of NTD by the notochord can result in incomplete CNS development and/or overgrowth of CNS precursors. Indeed, NTDs are described not only as failures of neural tube closures, but as failure to properly induce the development of mesenchymal and neuroectodermal structures. Neural induction involves numerous soluble, diffusible factors produced by a variety of genes (eg, sonic hedgehog), specific cells-surface signaling molecules important for appropriate migration of cells within the developing neural tube, and direct cell–cell signaling (eg, CAMs).

Genetic Influences

Genetic mutations can certainly have a significant impact on all of the previously mentioned processes and have been both demonstrated experimentally in rodents and documented clinically in humans. Alterations in genes that affect metabolism, nucleotide synthesis, cell programming, and cell–cell signaling can all affect aspects of neural development, ranging from the signaling aspects of the induction of neural tube formation initiated by the notochord to alterations in programmed cell death. This affects overall CNS development.

Induction of the neural plate is controlled by a variety of genes. Sonic hedgehog (SHH) is a vertebrate gene expressed by cells within the notochord that—in conjunction with the Patched (PTC) gene—produce proteins that are involved in the induction of the floor plate during embryogenesis, the proliferation of neuronal subtypes such as motor neurons, and the beginnings of somite development. Early work in *Drosophila* and then in avian systems have described how the proteins produced by these genes induce the expression of various signaling proteins on the surfaces of cells, allowing for the sequential transmission of signals regulating the cells' fate.

PTC is a gene that functions downstream of SHH. Its function is hypothesized to serve as a negative feedback to SHH, thereby regulating the induction of numerous cell types in the developing neural tube. Failure of this system and its feedback loops and/or overexpression of one portion of the process could easily be involved in neural tube development failure. Although not currently implicated by empirical data, much research is currently in place to elucidate the impact of this system on human NTDs.

Genes associated with folate metabolism and methyltransferase reactions associated with methionine and homocysteine metabolism are both of major interest. Folate serves as a cofactor for enzymes that participate in nucleotide synthesis as well as being important in methylation processes. Evaluations of folate levels of mothers with NTDs shortly after birth have produced equivocal results, suggesting that absolute folate deficiency is rare. Indeed, disturbances in the metabolic pathways that utilize folate may predispose to NTDs. This could conceivably be corrected by supplementation with folate. Metabolism of folate and homocysteine is interdependent, and the risks associated with alterations in their metabolism are thought to be connected. Indeed, elevated homocysteine levels in pregnant women are a known risk factor for NTDs. Mutations/polymorphisms in the enzyme 5,10-methylenetetrahydrofolate reductase (MTHFR) have been associated with diminished plasma folate levels, with commensurate elevated homocysteine levels. These alterations have been identified in patients with spina bifida as well as their mothers and fathers. In addition, using cultures of fibroblasts from NTD-affected patients, homozygosity for defects in the MTHFR gene have been shown to have a 7.2-fold increased risk for neural tube defects. The prevalence of these defects appears to vary by race. Homozygosity for the C677T MTHFR mutation is a known risk factor for upper-level spina bifida lesions in Hispanics. The MTHFD1

1958G>A polymorphism is also associated with NTDs in those of Irish descent.

It is well known that in early stages of nervous system development, more cells are produced than needed and that the process of apoptosis and autophagy are coordinated during development to yield a well-defined and functioning nervous system. Apoptosis, the most studied of these processes, is modulated by various members of the Bcl2 gene family, the caspase family of cysteine proteases, and other genes which produce proteins that are necessary intermediators. Expression of these genes at different times and in different combinations essentially controls the development of specific populations of cells within the CNS. Several mouse models have shown that altering the expression of these genes (ie, knockout experiments) results in neural tube defects similar to that identified in humans. Such evidence strongly suggests their involvement in human neural tube pathology. Autophagy, an autodegenerative cell process, has a significant impact on the recycling of cellular components in the cytoplasm as a result of cellular organelle damage. This process can also be affected by nutritional stresses. A variety of genes that affect this process have been investigated using mouse models. Loss of Beclin 1 and Ambra 1 expression has been noted to result in overgrowth of the developing CNS. Therefore, identifying the human equivalent of these and other similar genes could yield information as to cause of various NTDs and provide information for new therapeutic targets.

Environmental Influences

The external environment has a significant impact on embryonic development and the incidence of NTDs. This has been documented in several ways. Hyperthermia during early pregnancy—the first 28 days during which neurulation occurs—has been shown to increase the incidence of NTDs. Specifically, maternal febrile events as well as sauna/hot tub use has increased the risk of NTDs (15–19).

Parental occupation has been demonstrated to have a definitive influence on the risk for neural tube defects. Increases in risk for NTDs have been noted for occupations involving exposure to solvents (eg, painters, industrial process workers, etc.). The health care profession has also been seen to impart an increased NTD risk. Also, agricultural workers, along with those involved in the transportation industry, have been noted to have an increased risk for NTDs. The exact etiology behind these changes in risk can only by hypothesized at this time.

Nutritional influences have a broad impact and interact in many ways with environmental as well as genetic influences. A primary example is folate metabolism. As indicated previously, folate is a cofactor for the enzymatic process involved in purine and pyrimidine synthesis and is also important in facilitating the transfer of methyl groups during the metabolism of methionine and homocysteine. Taken together, alterations in these folate-sensitive processes can have an impact on cellular proliferation. Lowered intake of foods containing folate in the diet is associated with an increase in the risk for NTDs. Also, as one can imagine, disorders of absorption of folate in the intestine can significantly affect folate levels and potentially affect NTD risk. However, studies of folate receptor/carrier densities in the intestines of women with NTD offspring or their progeny do not have abnormally low receptor levels. A significant number of studies, both in the United States as well as Europe, have shown that supplementation can alter this risk. Indeed, mandatory supplementation of folate in grain products in the United States has caused a steadily declining incidence of NTDs since it was initiated in 1996. Since the introduction of this program, it has been estimated that the number of pregnancies affected by NTDs has declined from approximately 4,000 to 3,000 per year. In fact, studies have shown that the risk for recurrence of NTDs can be decreased approximately 50% by taking recommended folate supplementation.

The risk for NTDs varies for couples, depending on whether there is a prior history of such defects. U.S. couples with a prior history of NTD births have an increased risk for recurrence (2%–5%) (2). Because of this, the U.S. Public Health Service and the CDC have two separate recommendations for supplementation based on prior NTD histories. Elevated supplementation is appropriate for couples with a prior NTD birth. Limited studies have also identified zinc as a nutritional entity that can also elevate NTD risk. It was discovered that women with the genetic disorder of zinc metabolism acrodermatitis enteropathica are at high risk for NTDs and that supplementation can lower those risks.

Maternal obesity and associated diabetes have been found to be associated with increases in risk for NTDs. Specifically, women with a pre-pregnancy body mass index (BMI) suggestive of obesity (>29 kg/m) are more inclined to give birth to children with NTDs. This holds true for women with diabetes, although the etiology of this association may be linked to alterations in glucose metabolism during organogenesis. It is notable that experimentally manipulated glycosylation in rodents results in birth defects not unlike those seen born to mothers with diabetes. Risks for NTD-affected births has been estimated at 2% here in the United States and as high as 7% in England. These risks include spina bifida as well as other significant NTDs such as anencephaly. The NTD recurrence risk for mothers with diabetes in the United States is around 4%, which is similar to that found for mothers without diabetes.

Teratogenic influences from the environment—such as the consumption of prescribed drugs—have been associated with neural tube defects, particularly myelomeningocele. Valproic acid taken for seizures during pregnancy has been shown to increase the incidence of neural tube defects. Mechanistically, it appears to work by disrupting folate metabolism, thereby inhibiting neural tube closure. Alterations in folate-dependent methylation of regulatory proteins is theorized to be the cause. Regardless, administration of folate during pregnancy counteracts valproic acid–associated neural tube defects.

The rising use of highly active antiretroviral therapy (HAART) in the treatment of human immunodeficiency (HIV) disease has increased the incidence of women exposed to these drugs entering and during pregnancy. A variety of case reports as well as animal studies have suggested an association between antiretroviral drug use and NTDs (20). Drug-induced interference with DNA synthesis during development would likely have an impact on gastrulation and neurulation. Other drugs are also associated with NTDs, such as isotretinoin (Accutane), which is used for acne treatment; etretinate (Tegison), which is a psoriasis treatment; and anticancer agents such as methotrexate. Indeed, even fetal alcohol syndrome has an association with increased risk for abnormal CNS development, including NTDs.

Some chromosomal disorders that have multivariate etiologies and presentation are known to have an association with increases in risk for NTDs. Trisomy 21 (Down's syndrome) and trisomy 13 (Patau syndrome) are notable examples. Although the incidence is relatively small, studies have shown that various NTDs, including spina bifida but not anencephaly, have been found upon autopsy of definitively karyotyped infants. Interestingly, trisomy 21 has been shown to be associated with genetic polymorphisms involved in homocysteine/methionine methylation (see the previous discussion on folate metabolism) and has a noted familial clustering with NTDs.

PRENATAL SCREENING

Prenatal screening is recommended for pregnant women to detect not only NTD but also to screen for Down's syndrome and related disorders. A simple blood test known as the quad screen is done in the second trimester. The elements of the test include alpha feto-protein (AFP), human chorionic gonadotropin (HCG), estriol, and inhibin A. Elevated levels of AFP suggest that a NTD is present and further testing is indicated. This includes high-resolution ultrasounds and amniocentesis. Ultrasound can detect a splaying of the pedicles and the classic "lemon and banana signs." The lemon sign relates to the shape of the head, and the banana sign is related to herniation of the cerebellar vermis through the foramen magnum, which appears to be banana-shaped. What is critical about prenatal diagnosis is the ability to plan ahead. Fetal surgery is available for families on a research basis through the Mothers of Meningomyelocele (MOM) program. The optimal delivery options should include cesarean section in a high-risk center with a neurosurgeon available.

CLINICAL TYPES OF NEURAL TUBE DEFECTS

Spina Bifida Occulta

- Bony defect with no herniation of meninges or nervous elements
- Incidental finding in 5% to 36% of adults; a small percent can develop clinical findings (21,22)
- Can be associated with pigmented nevus, angioma, hairy patch, dimple, and dermoid sinus
- Usually found in the lumbosacral/sacral segments
- Can have associated tethered cord with development
- May have bowel and bladder involvement
- No hydrocephalus or Chiari malformation

Spina Bifida Cystica

- Bony defect with herniation of spina canal elements.
- Meningocele-herniation of the meninges, but does not contain neural tissue
  - Usually normal neurological exam
  - No association with hydrocephalus or Chiari malformation
  - Uncommon—occurs less than 10%
- Meningomyelocele-herniation of meninges and neural elements
  - Most common
  - Associated with hydrocephalus and Chiari type 2 malformations
  - Abnormal motor and sensory exam
  - Neurogenic bowel and bladder
  - 75% in the lumbosacral segment

Caudal Regression Syndrome

- Absence of the sacrum and portions of the lumbar spine
- Associated with maternal diabetes
- Associated findings include syringomyelia, anorectal stenosis, renal abnormalities, external genital abnormalities, and cardiac problems
- Motor and sensory abnormalities

CLINICAL SIGNS AND COURSE

The spinal cord defect associated with spina bifida is often associated with other malformations. This results in a multisystemic process that leads to a variety of health problems and potentially life-threatening complications. Motor and sensory deficits vary according to the level and extent of spinal cord involvement (23–25).

In the care of spina bifida patients, two levels are often described: the anatomic level of the lesion and the neurologic level of functional involvement. In terms of the level, it is the neurologic or functional level that gives health care providers prognostic information with respect to long-term expectations and functional outcomes. Spinal cord involvement may result in asymmetric motor and sensory deficits. Sensory deficits usually follow a dermatomal pattern and may not affect all sensory modalities equally (23,24).

Neurogenic bladder and bowel dysfunction may be present in all patients because of the distal level of innervation of the bladder and bowel. This is true even if there is no apparent motor involvement/deficit in the legs.

In the following discussion, clinical signs of muscle weakness are described. These levels are functional neurologic levels and may not directly reflect the anatomic level of the malformation.

Musculoskeletal deformities related to muscle imbalance may present serious clinical concerns. Deformities may be static deformities present at birth or may develop over the years.

Figure 9.1 summarizes segmental innervation, preserved muscle function, and musculoskeletal complications typical of various levels of spinal cord malformation. Providers must keep in mind that the overall functional outcome for the individual is related in part to neurologic level, in addition to other associated central nervous system and medical issues.

Thoracic Lesions

Thoracic-level malformations spare the upper extremities, with the exception of decreased ability to abduct the fifth digit (thoracic level 1 = T1). There is usually partial innervation of the abdominal and intercostal musculature, which may result in respiratory dysfunction or insufficiency. Kyphosis and kyphoscoliosis may result from trunk weakness and be more prominent in individuals with vertebral anomalies (26). The lack of volitional movements combined with the effect of gravity lead to lower extremity deformities. The usual lower extremity posture in the supine position is partial hip external rotation, abduction, and ankle plantarflexion. Deformities develop from sitting, hip flexion, knee flexion, and equinus. Hip flexion contractures with compensatory lumbar lordosis increase any preexisting kyphosis or kyphoscoliosis.

L1–L3 Segment

Hip flexors and hip adductors are innervated at the L1–L2 levels. With L2 sparing, knee extensors have partial innervation but are not at full strength. Distal lower extremity muscle strength is absent. The distribution of muscle imbalance—hip flexion and hip adduction with absent hip extension and hip abduction—leads to the development of contractures and early paralytic hip dislocation. Pelvic obliquity seen in asymmetric hip pathology enhances scoliosis. Gravity-related foot equinus deformity may develop.

Ambulation during young childhood is typical with the use of bracing and assistive devices. Long-term ambulation through adulthood is less likely as priorities change and there are further increases in the already high-energy demands of walking (25). The extent of bracing necessary to achieve ambulation is usually related to the amount of active knee extension.

L4–L5 Segments

Innervation of the hip flexors, hip adductors, and knee extensors are usually complete; however, hip abductors and hip extensors remain weakened. Coxa valga and acetabular dysplasia are still a concern. Typically, hip dislocation occurs later at the L4–L5 segmental levels. Newborns with a well-defined lesion sparing L4 lie in a typical position of hip flexion, hip adduction, and knee extension. When the L5 segment is spared, the gluteus medius, gluteus maximus, and hamstrings have partial strength and knee extensor contracture is less likely. Because the tibialis anterior is unopposed by its plantarflexion and everter antagonists, a calcaneovarus foot deformity develops. If the peroneus muscles are spared, the varus is eliminated. Although the plantar flexors are partially innervated, they are not strong enough to counter the strong force of the ankle dorsiflexors.

Sacral Segments

Active plantarflexion is present and some toe movements are present. Intrinsic foot muscles remain weak and may result in a cavus foot deformity with clawing of the toes.

Sensory Deficit

Partial or complete absence of different sensory modalities predisposes individuals with spina bifida

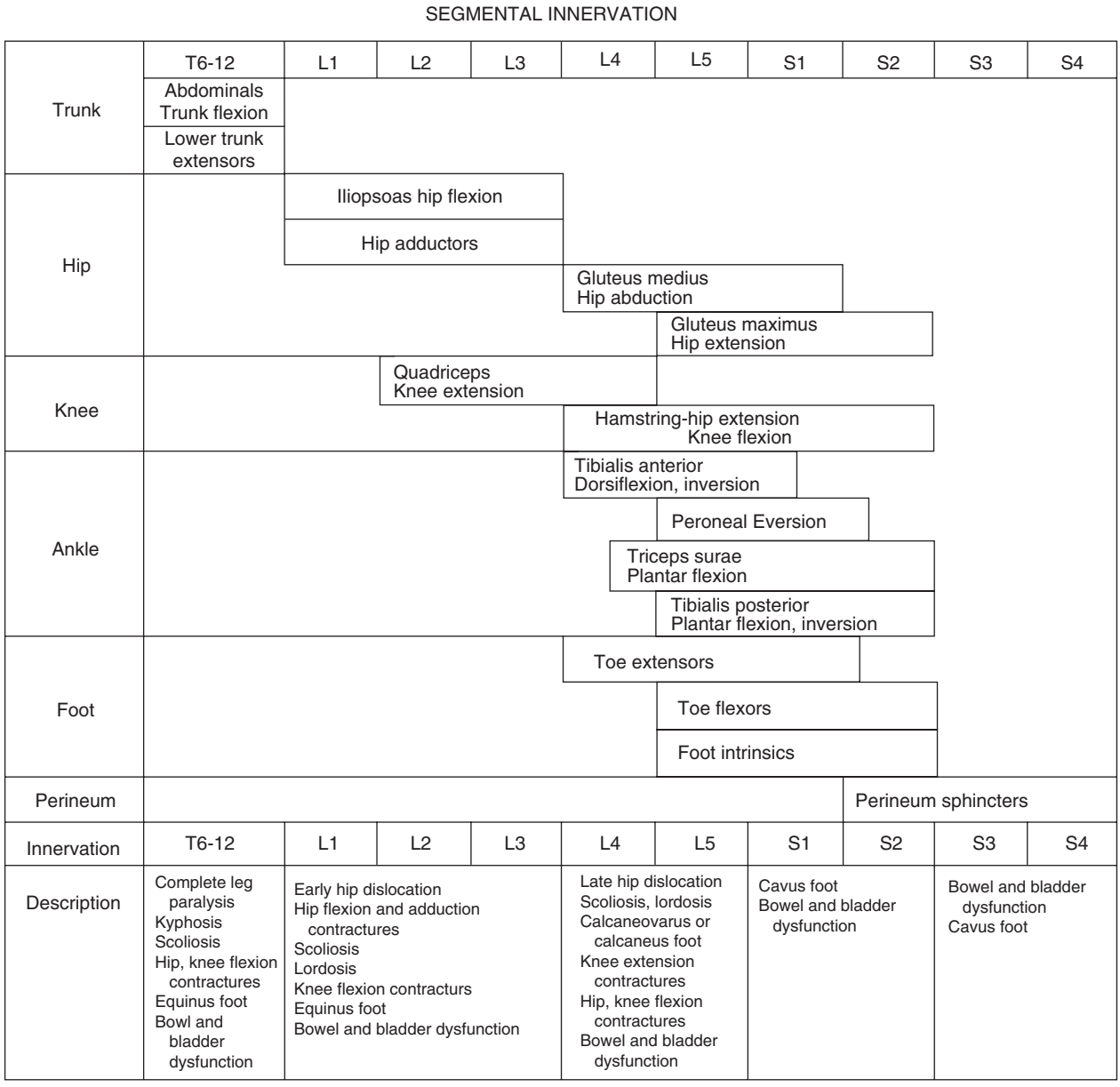


Figure 9.1 Musculoskeletal, sensory, and sphincter dysfunction by segmental level.

to skin injuries because of decreased ability to perceive pressure, pain, trauma, or heat (23,24,25,27). Skin breakdown tends to occur over areas of prominence and weight bearing. The lower back, intergluteal, perineum, feet, heels, and toes are the sites of predilection, but any area with sensory loss may be affected. Scoliotic and kyphotic prominences are areas prone to breakdown (26). Pressure ulcers often heal slowly, tend to get infected, and often recur. A pressure ulcer may be a symptom of a tethered cord. Long-standing ulceration with deep tissue necrosis may spread to bone and lead to acute or chronic osteomyelitis.

Other complications of denervation include vasomotor instability, neuropathic Charcot joints, and osteoporosis in individuals with extensive lower extremity weakness (25,26,27,28).

The spinal cord defect usually results in a lower motor neuron process. Spasticity is present in most individuals with spina bifida across their lifetime (29). The presence or gradual development of spasticity above the level of the spinal cord lesion may be related to tethering of the spinal cord, Chiari type II malformation exerting pressure on the cervical spinal cord, decompensating hydrocephalus, ventriculitis, syringohydromyelia, or coexistent encephalopathy sustained at birth (30–33).

ASSOCIATED CENTRAL NERVOUS SYSTEM MALFORMATIONS

Extensive neuropathologic studies have demonstrated that neural tube defects are associated with a high incidence of gross and microscopic malformations of the forebrain and hindbrain (34). Additional anomalies in the spinal cord may complicate the original local dysraphic defect (23,24,27,35,36). Table 9.1 lists associated anomalies and malformations by location.

Spinal Cord

Tethered cord refers to an abnormal attachment of the spinal cord at its distal end (27). Under normal circumstances, the conus medullaris ascends from its distal position to the L1 to L2 vertebral level during the first year of life (37). Focal abnormalities—including thickened and shortened filum terminale, supernumerary fibrous bands, persistent membrane reunions, dural sinus, diastematomyelia, entrapment by lumbosacral tumors, and adhesions in the scar tissue of the repaired myelomeningocele—interfere with this process (27). All children born with spina bifida have a low-lying cord on magnetic resonance imaging, and approximately one-third develop neurologic, urologic, or orthopedic complications or symptoms (38) (Fig. 9.2).

Tethering of the spinal cord is the second most common cause of neurologic decline in a child with myelomeningocele (38). The most common clinical signs or symptoms of a tethered cord include spasticity in the lower extremities, decline in lower extremity strength, and worsening scoliosis. Other signs and symptoms that strongly suggest tethering of the spinal cord include back pain, changes in urologic function, changes in gait, and development of lower extremity contractures. In patients who are suspected of having a symptomatic tethered cord, the function of their shunt needs to be evaluated prior to proceeding forward with surgical management (39).

The reported functional outcome of surgical management of a tethered cord is variable. One study reported improvements in gait in almost 80% of patients following untethering, whereas other studies report improvement in as few as 7% (40). (Note: All cords tether to some extent following repair.) Less than 20% of children with a tethered cord experience back pain. However, this is the symptom most likely to improve with surgery (30,41).

Diastematomyelia is a postneurulation defect that results in a sagittal cleavage of the spinal chord, most commonly affecting the lumbar and thoracolumbar levels of the spinal cord. It is more common in females (42,43). Diastematomyelia may have both neurologic and orthopedic presentations. Orthopedic symptoms include scoliosis, Sprengel's deformity (especially when associated with Klippel-Feil sequence), hip subluxation, and lower extremity limb-length discrepancies (43,44).

9.1 Associated Central Nervous System Malformations

Spinal cord
- Tethering
- Distal focal abnormalities
  - Thick, short filum terminale
  - Supernumerary fibrous bands
  - Lumbosacral tumors (lipoma, fibrolipoma, fibroma dermoid, epidermoid cyst, teratoma)
  - Bony vertebral ridge
  - Diastematomyelia, diplomyelia, split cord

Brainstem
- Arnold type II malformation
  - Kinking, inferior displacement of medulla
  - Herniation into cervical spinal canal
- Abnormalities of nuclear structures
  - Dysgenesis, hypoplasia, aplasia, defective myelination
- Hemorrhage, ischemic necrosis
- Syringobulbia

Cerebellum
- Arnold-Chiari type II malformation
  - Elongated vermis, inferior displacement
  - Herniation into cervical spinal canal
- Abnormal nuclear structures
  - Dysplasia, heterotopia, heterotaxia

Ventricular system
- Hydrocephalus
- Aqueductal stenosis, forking, atresias

Forebrain
- Polymicrogyria
- Abnormal nuclear structures
  - Heterotopia (subependymal nodules)
  - Heterotaxia
- Prominent massa intermedia
- Thalamic fusion
- Agenesis of olfactory bulbs and tracts
- Attenuation/dysgenesis of corpus callosum

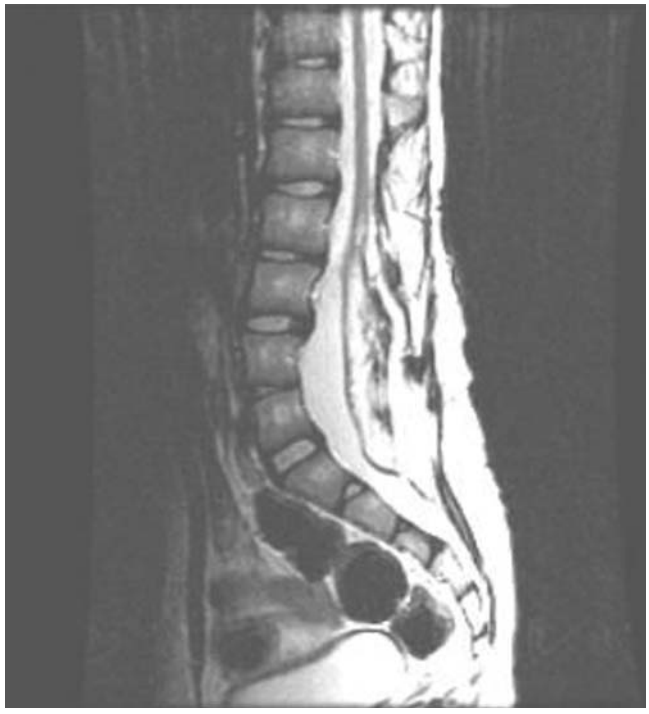
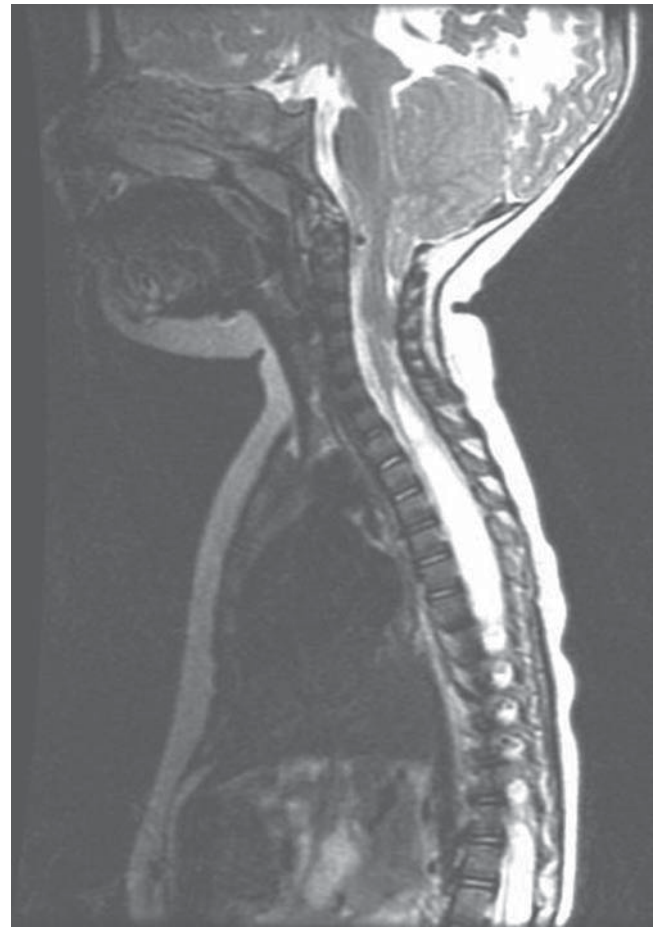


Figure 9.2 T2-weighted magnetic resonance image of tethered cord. There is tethering of the spinal cord with conus seen down to the L5 vertebral level, heterogeneous signal intensity characteristics, and areas of fibrofatty tissue.

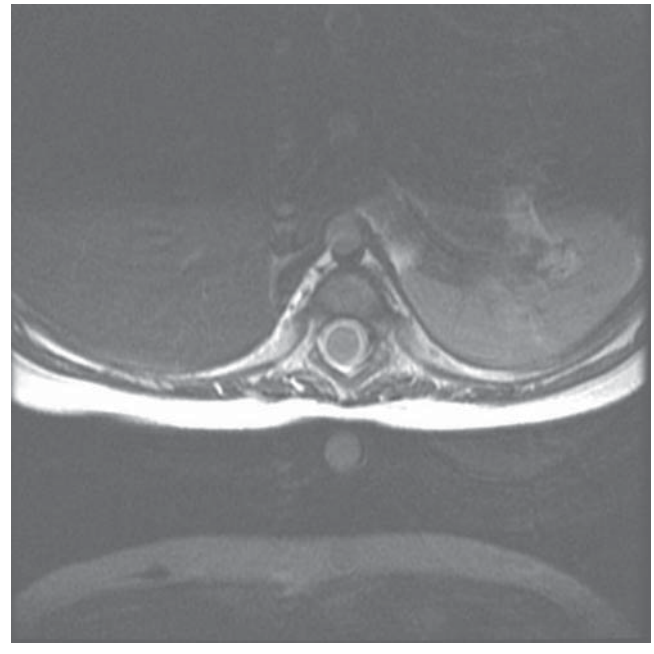
Neurologic symptoms include gait abnormalities, asymmetric motor and sensory deficits of the lower extremities, and neurogenic bladder and bowel (45). Symptoms of diastematomyelia may present in childhood or, less commonly, in adulthood (46).

It is not uncommon for individuals to develop syringomyelia—a tubular cavitation in the spinal cord parenchyma extending more than two spinal segments (47). Syringomyelia is present in up to 40% of individuals with myelomeningocele (48). The syrinx may be located anywhere along the spinal cord, medulla, or pons, but is most common in the cervical region (23,24,49). Magnetic resonance imaging (MRI) is used to detect syringomyelia (50) (Fig. 9.3).

Often, a syrinx is of little clinical significance; however, if a patient develops decreasing function above the level of their lesion, syringomyelia must be considered in the differential diagnosis. Although shunt malfunction and cord tethering are more common complications, symptomatic hydromyelia may explain a slower-than-expected progression through gross motor and fine motor developmental milestones or a decrease in strength/function. Early progression of scoliosis above the initial neurologic level may be the earliest sign of a syrinx. A shunt malfunction



A



B

Figure 9.3 T2-weighted magnetic resonance image showing sagittal (A) and axial (B) views. There is a large syrinx present, beginning at the mid portion of C6 and extending to L4.

may contribute to a symptomatic syrinx, and shunt function should be evaluated. Placement of a syringopleural shunt may be necessary to decompress the syrinx.

Cerebellum and Hindbrain

The most common hindbrain abnormality in neural tube defects is Chiari type II malformation, seen in 80% to 90% of individuals with myelomeningocele (23,24,27,51).

This malformation results in caudal displacement or herniation of the medulla, lower pons, elongated fourth ventricle, and cerebellar vermis into the cervical spinal cord (Fig. 9.4). This often interferes with cerebrospinal fluid outflow and is, therefore, almost always associated with hydrocephalus. Caudal displacement of the medulla may occur and result in traction neuropathies of the lower cranial nerves. Signs of bulbar compromise arise from compression of the herniated hindbrain.

A broad spectrum of clinical symptoms is seen in individuals with this malformation. However, only 20% will develop clinical signs of brainstem dysfunction, with most occurring in the neonatal period (52,53). Symptoms may be evident at birth or present within the first two to three months.

The most severe symptom is respiratory compromise, which may be both central and peripheral in etiology. Individuals may experience stridor, laryngeal nerve palsy with vocal cord paralysis, upper airway obstruction, periodic breathing, central or obstructive sleep apnea, or aspiration. Dysphagia and extraocular motion abnormalities may also be seen related to other cranial neuropathies. Dysphagia may be severe enough that gastrostomy tube placement is required. Airway compromise may necessitate tracheostomy.

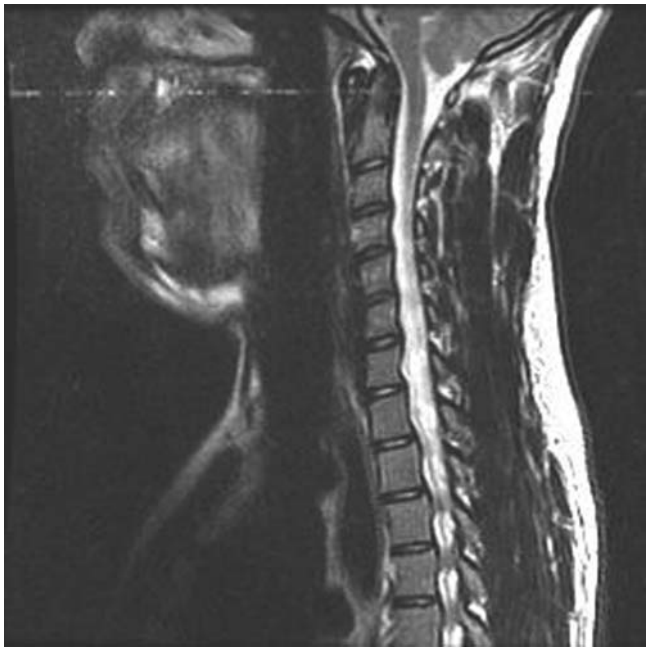


Figure 9.4 T2-weighted magnetic resonance image of the cervical spine. The posterior fossa is crowded. There is cerebellar tonsillar herniation, with the cerebellar tonsils lying 9 millimeters below the foramen magnum. This is the expected finding for a Chiari II malformation.

In the presence of brainstem compromise, hemiparesis or tetraparesis may be seen (this is more common in older children or adults than infants). Impairment of fine motor hand function is well documented and is seen in more than half of individuals with thoracic-level lesions and approximately one-fourth of individuals with lumbosacral lesions.

Control of ocular motility is related to cerebellar function (saccadic eye movements, visual fixation, and pursuit). There is a high rate of visual problems in individuals with spina bifida. Fewer than one-third have completely normal visual function (54,55).

Despite successful initial treatment with surgical decompression, problems may recur. Typically, vocal cord paresis in the first two months of life is a sign of irreversible damage, and surgical decompression is unlikely to result in clinical improvement (56).

Ventricles

Hydrocephalus is a significant problem in the majority of patients with myelomeningocele. The pathogenesis of hydrocephalus is multifactorial and is related to aqueductal stenosis, occlusion of the foramen of Luschka and Magendie, hindbrain herniation, obliteration of the subarachnoid spaces at the level of the posterior fossa, compression of the sigmoid sinuses with consequent venous hypertension, and fibrosis of the subarachnoid spaces (57,58). The prevalence of hydrocephalus in individuals with myelomeningocele is reported to be as high as 95%, with shunt rates ranging from 77% in the 1980s to 58% in more recent years (59). Hydrocephalus rates are closely associated with the level of the spinal dysraphism. In one cohort, 100% thoracic, 87% lumbar, and 67% sacral myelomeningocele patients required shunting (60). In all cases of symptomatic hydrocephalus, surgical management is recommended.

Symptoms of hydrocephalus include those that are classic for increases in intracranial pressure—this varies based on the presence or absence of an open fontanelle. In an infant, signs of increased intracranial pressure include lethargy, decreased feeding, bulging fontanelle, increasing head circumference (greater than expected for age), poor developmental progress, and "sun downing." In patients with a closed fontanelle, signs of increased intracranial pressure include headache, vomiting, drowsiness, changes in behavior, changes in personality, irritability, diplopia, and

papilledema. With the sudden onset of increased intracranial pressure, Cushing's triad may be seen. Cushing's triad consists of progressively increasing systolic blood pressure, bradycardia, and irregular respirations.

At present, placement of a shunt is standard of care for surgical management of hydrocephalus. Shunting has many complications, including both mechanical and infectious. Up to 95% of adult patients with myelomeningocele have required at least one shunt revision. The rate of shunt infection is between 5% and 8% per procedure (38,61–65).

Endoscopic management of hydrocephalus is being increasingly presented as an alternative to shunting. Endoscopic third ventriculostomy (ETV) provides direct communication between the third ventricle and the subarachnoid space by way of interpeduncular and prepontine cisterns. The success rates for ETV as the sole management for hydrocephalus in infants with myelomeningocele range from 12% to 53% (66–70). In most infants with myelomeningocele, ETV alone is not an effective treatment for hydrocephalus. More recently, ETV has been combined with choroid plexus cautery (CPC). This has resulted in an improved success rate for treatment of hydrocephalus in infants, with a success rate of more than 70%. If an ETV combined with a CPC fails, it will typically do so during the first three months (71).

Endoscopic third ventriculostomy may also be an option in the setting of a shunt malfunction in the older child. In one study reported by Teo and Jones, the majority of ETV failures were during the first six weeks postoperatively. However, failures were seen as late as five years postoperatively (70). Longevity of the ETV/CPC for treatment of hydrocephalus beyond two or three years has yet to be determined. It is not known if there is a difference in neurocognitive outcomes in patients treated with an ETV/CPC (shunt-independent) as compared with individuals who are shunt-dependent. Although not yet considered "standard of care," ETV in combination with CPC holds promise for surgical management of hydrocephalus without creating shunt dependency and the complications associated with it (71).

Forebrain

Malformations of the forebrain are broad, and range from gross anatomic malformations to microscopic anomalies. Polymicrogyria are increased numbers of small-sized cerebral gyri with shallow disorganized sulci, and this is seen in up to 65% of individuals (72). Heterotopias are aberrant neural tissues in the form of subependymal nodules. They are present in approximately 40% of cases (34). Microscopic studies have demonstrated disordered cortical lamination, neuronal hypoplasias of the thalamus, and complete or partial agenesis of the olfactory bulbs and tracts. Dysgenesis or agenesis of the corpus callosum may be seen and may also be associated with a malformed cingulated gyrus and septum pellucidum (72). The contribution of these forebrain malformations to the development of cognitive and perceptual dysfunction remains unknown.

Other Malformations

Neural tube defects are also associated with an increased rate of malformations unrelated to the central nervous system. Vertebral anomalies are not uncommon and contribute to progressive kyphosis and scoliosis. Thoracic deformities may result from rib deformities, including absence, bifurcation, or reduction of the ribs. Malformations of the urinary system may be present and result in accelerated deterioration of renal function.

Neural tube defects have been associated with genetic abnormalities, including trisomy 18, trisomy 13, Turner's syndrome, Waardenburg's syndrome, renal aplasia and thrombocytopenia syndrome, nail–patella syndrome, deletion 13q syndrome, and others (73).

TREATMENT

Team Approach

A team approach is an important part of the care of the individual with congenital spinal dysfunction. The multidisciplinary team often includes neurosurgery, orthopedic surgery, urology, rehabilitation medicine, physical and occupational therapy, social work, nutrition, and nursing. Coordination of all modes of treatment is important for a successful rehabilitation plan. Primary care for the usual childhood illnesses and health maintenance should remain the responsibility of the pediatrician.

After birth, parents and families of individuals with spina bifida need to be informed about their child's diagnosis and its implications. A prenatal visit with the neurosurgeon and other medical specialists may be beneficial. Parents often ask questions regarding anticipated functional abilities and limitation, including self-care and ambulation. Cautious predictions based on the current functional level may be given. Medical providers should be frank in their discussion of the problems that the parents and child will face, but this should be done with cautious optimism. Discussions and instructions about the child's care and handling at home may require several sessions so that the family members are not overwhelmed by the

amount and complexity of the information. Families should be informed of the many issues involved and the need for seeing several medical specialists. Frequent follow-up after discharge from the neonatal unit is often necessary and typically involves visits every three to four months for a couple of years and then every six months thereafter (74).

Neurosurgical Treatment

Neurosurgeon involvement in the care of the individual with spina bifida begins with a prenatal visit. Studies regarding prenatal surgical closure of a neural tube defect are underway. To date, intrauterine repair has not been shown to decrease the motor deficits associated with myelomeningocele, but in some series it has been demonstrated to decrease the degree of associated Chiari type II malformations and the need for shunting procedures for hydrocephalus in the first year of life (75–78).

Neurosurgical repair of an open neural tube defect, such as a cystic lesion, is usually performed on the first day of life. If hydrocephalus is present at birth, surgical management may be necessary. Ninety-five percent of children with spina bifida are likely to have hydrocephalus, and 75% to 85% require surgical management. The average revision rate is 30% to 50% (79), and after 2 years of age there is a 10% per year risk of failure (64). Most neurosurgeons believe that a child with hydrocephalus that required shunting will remain shunt-dependent (65,80). These statistics may change as endoscopic third ventriculostomy with choroid plexus cautery is performed with increasing frequency.

Neurosurgical follow-up is required, even after the neonatal period, to monitor for symptomatic hydrocephalus, shunt malfunction, and other neurosurgical complications. Pediatric patients with myelomeningocele should be followed routinely, usually on an annual basis.

NEUROGENIC BLADDER

Physiology

The fundus is made up of three layers of crisscrossing smooth muscle, called the detrusor. These three smooth muscle layers extend down the posterior urethra (where there is also skeletal muscle) toward the external sphincter region. T10 to L1 supplies the sympathetic innervation for the bladder; this causes the detrusor to relax and the bladder neck and posterior urethra to contract. S2–S4 provides the parasympathetic innervation to the bladder and primarily supplies the fundus. The neurotransmitter is acetylcholine; this causes contraction. The sympathetic innervation is active during bladder filling, and the parasympathetic innervation is active during urination. Somatic nerves via the pudendal (from sacral cord) innervate the skeletal muscle component of the external urethral sphincter; this leads to relaxation of the external urethral sphincter (81).

Bladder Capacity

The prediction of normal bladder capacity aids the diagnosis of abnormal voiding patterns. It is typically accepted that the bladder capacity of a baby during the first year equals the weight of baby in kilograms times 7–10 milliliters. A study with 200 children (132 with normal voiding, 68 frequent and infrequent voiders) demonstrated that from approximately 1–12 years of age, that age plus 2 equals the bladder size in ounces ($\times 30$ = volume mL). After that, the teenager assumes an adult-size bladder, typically around 400 cc. Clinically infrequent voiding causes an increase in bladder size. Clinically frequent voiding causes a decrease in bladder size (82). Post-void residual is generally accepted as 10% of bladder capacity, taking into account the appropriate bladder capacity for age.

Diagnostics

Checklist for Diagnosing Neurogenic Bladder

- Are the bladder and kidney studies up to date?
- If voiding on own, is it overflow incontinence?
- Is bladder size and bladder compliance appropriate for age?
- Is post-void residual (PVR) appropriate?
- Is the sphincter mechanism competent?
- Is the current management preserving the kidneys?

Diagnostic Tests

- US—Ultrasound of kidneys and bladder to determine any structural abnormalities.
- VCUG—Voiding cystourethrogram to detect vesicoureteral reflux (VUR), evaluate the bladder contour, and evaluate the urethra. The first VCUG study is a contrast VCUG for boys and girls. Subsequent VCUG studies, for boys and especially girls, should be nuclear cystograms, as the radiation is markedly reduced.
- UDY—Urodynamics to determine detrusor leak point pressure, uninhibited bladder contractions, detrusor sphincter dyssynergia, bladder capacity, post-void residual, and bladder compliance and sensation. The basic urodynamic formulas are:

Pressure detrusor = pressure vesical (bladder)—
Pressure abdominal (rectum)

Bladder compliance = change in bladder volume / change in pressure

It is recommended that these tests (US, UDY, and VCUG) be performed in the neonatal period, as newborns with spina bifida need baseline studies. As growth of the infant is rapid in the first 12 months, abnormal studies may require two subsequent studies in the first year of life. It is generally recommended that bladder and renal ultrasounds be repeated at three-month intervals in the first year and then twice yearly the second year and then yearly. The UDY and VCUG is repeated at 3 months of age, at 1 year, then at 2–3 years of age, and then repeated every other year (83). Small-for-age and/or high-pressure bladders may need studies more frequently. Abnormalities on ultrasound will likely lag those found on UDY. Studies should be repeated with significant clinical changes in bowel or bladder incontinence, infections, or gait.

Other Studies. The excretory urethrogram (EXU) and intravenous pyelography (IVP) tests detect urinary tract stones, anatomic abnormalities, and obstruction. Diethylene triamine acetic acid (DTPA) and mercaptoacetyltriglycine (MAG3) evaluate urinary tract (UT) drainage/obstruction. The DTPA and MAG3 attach to a radioactive tracer and are processed by the kidneys. While MAG3 is expensive, it can also be used to assess renal cortex functioning. Cystoscopy for bladder cancer surveillance is typically performed for the first time 10 years after bladder augmentation surgery or after the initialization of long-term indwelling catheter use. It is then performed yearly thereafter. Consolation with an urologist in the case of either bladder augmentation surgery or long-term indwelling catheter use is advised for current recommendation (84). Technetium 99m dimercaptosuccinic acid (DMSA) is the best test for determining the functioning areas of the renal cortex and those areas with scarring. This test should be done when there is abnormality on a renal ultrasound, a history of multiple urinary tract infections (UTIs), or pyelonephritis.

Urinalysis. The nitrate test indirectly detects urine bacteria with enzymes that reduce nitrate to nitrite in urine (eg, *Klebsiella, Enterobacteriaceae, E. coli,* and *Proteus*).

The Leukocyte Esterase Test. While leukocytes in the urine can disintegrate and disappear rapidly, leukocyte esterase persists.

Figures 9.5 and 9.6 show a normal urodynamic study and a urodynamic study reflecting spastic bladder detrusor and sphincter dyssynergia.

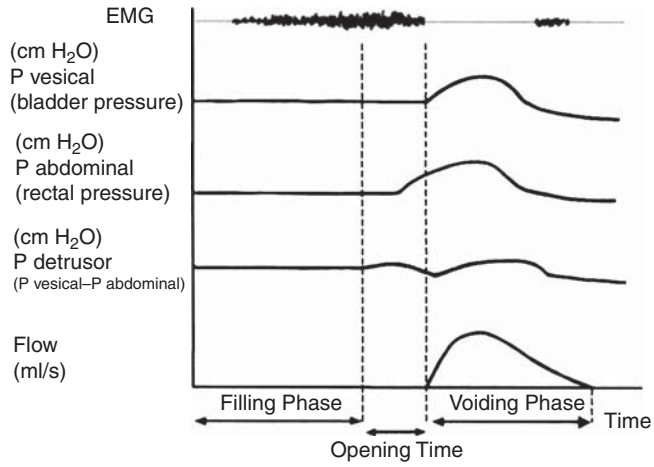


Figure 9.5 Normal urodynamic study.

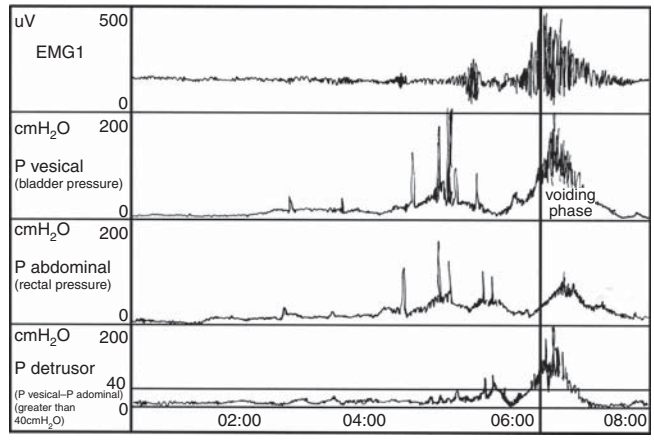


Figure 9.6 Urodynamic study reflecting spastic bladder detrusor and sphincter dyssynergia.

Risk Factors for Upper Tract/Kidney Deterioration

- Leak point pressures >40 cm H_20
- Vesicoureteral reflux
- Detrusor sphincter dyssynergia
- Poor bladder compliance
- Bladder hyperreflexia
- Increased post-void residual (greater than 10% of the total bladder capacity)

Treatment

Goals

- Preservation of renal function
- Age-appropriate social continence
- No significant urinary tract infections
- Normalized lifestyle

Treatment of Storage Dysfunctions

Detrusor hyperreflexia is decreased with these anticholinergic medications: propantheline bromide or

oxybutynin chloride. A study found that oxybutynin tablets, syrup, and extended-release tablets are safe and effective in children with neurogenic bladder dysfunction. [Note: The youngest child in the study was 6 years old (85).] Ineffective closure of the internal urethral sphincter mechanism may be improved by the following alpha-sympathetic stimulation medications: phenylephrine, ephedrine, and imipramine. External urethral sphincter closure problems may require neuromuscular reeducation or surgical treatment.

Treatment of Emptying Dysfunctions

The typical day-to-day management of the neurogenic bladder is clean intermittent catheterization every four hours while awake to keep bladder volumes within normal limits for age. In 1972, Lapides was the first to state that the sterile single-use catheter is unnecessary in the management of persons with neurogenic bladders because it does not reduce bacteriuria (86). This continues to be substantiated in the pediatric population (89). If this intervention is unsuccessful, various pharmacological and urologic surgical procedures may be explored. Crede's manuver should be used with extreme caution. Valsalva or Crede's maneuvers to empty the neurogenic bladder that has detrusor sphincter dyssynergia will likely raise the intravesicular pressure to greater than 40 cm H_20, thus putting the kidneys at risk. Bethanechol is rarely used to treat weak expulsive force of the detrusor. Hyperactive internal sphincter mechanism may be treated with alpha-adrenergic blockers. Hyperactive external urethral sphincter may be treated with baclofen, neuromuscular reeducation of the pelvic floor, Botox injections (87), or surgery.

Botox A injections to the external sphincter have shown promise for decreasing the resistance of the external urinary sphincter (87). The Mitrofanoff procedure, first introduced in this country in the 1980s, uses the appendix to create a catheterizable conduit, typically between the bladder and the umbilicus. A flutter valve can help prevent external leakage; however, leakage may be problematic in a small percentage (88). A Mitrofanoff procedure may be useful in females who may have more difficulty cauterizing than males. A vesicostomy may be a temporizing measure for older children and adults. The ileal conduit was the first urinary diversion procedure, but follow-up studies showed a disappointingly high rate of renal deterioration, calculosis, hydronephrosis, and the need for reversal of the procedure (74). Artificial sphincters have been found to be helpful in some, but can have infection, erosion, and mechanical problems.

Primary Care Treatment of Children Managed With Clean Intermittent Catheterization

Routine urinalysis (UA) and urine culture (UC) are not recommended during well-child check-ups if the child looks well. If bacteriuria is detected in the urine, it is important to determine whether it represents a clinical infection or colonization of the bladder. Only clinical UTIs should be treated (89). Prophylaxis in the absence of VUR is not routinely recommended.

Antibiotic Prophylaxis and Bacteriuria Treatment

A number of studies were done on antibiotic prophylaxis and bacteriuria treatment with individuals with neurogenic bladders. Kass found that if there is no VUR, bacteriuria is innocuous; in his study, 17 hydronephrotic kidneys showed significant radiographic improvement since starting clean intermittent catheterization (CIC) (90). Ottolini found that asymptomatic bacteriuria requires no antibiotic therapy in the absence of VUR (91). Van Hala found that there is no correlation between number of UTIs, the type of catheter used, or the use of prophylactic antibiotics (92). Johnson et al found that nitrofurantoin is an effective prophylactic agent during a three-month period for bacteriuria (93). Schlager et al found that asymptomatic bacteriuria persists for weeks in children with neurogenic bladders with normal upper urinary tracts managed with CIC (94). The asymptomatic bacteriuria is different from the symptomatic bacteriuria. Jayawardena et al found that patients with spinal cord injury (SCI) frequently have asymptomatic bacteriuria without data to support treatment and that routine urine cultures should not be done at annual evaluations (95).

(Note: It may be appropriate for a pediatric patient without a neurogenic bladder and with frequent UTIs secondary to dysfunctional voiding to receive prophylactic antibiotics for a time. Patients with VUR and with or without a neurogenic bladder routinely receive prophylactic antibiotics.)

Neonatal vs Childhood Treatment

Early proper management is imperative for the preservation of renal function (96). Kidney damage was found to be approximately 1 in 4 without proper management of the neurogenic bladder (97). On urodynamic testing, subtracted detrusor leak point pressure (p vesical-p abdominal) greater than 40 cm H_2O, with a bladder capacity less than 33% of expected, was associated with renal damage (97).

Treatment of neurogenic bladder dysfunction due to myelomeningocele in neonates is recommended. A study of 98 individuals (46 started CIC in first year of

life, 52 began CIC after four years of life) reviewed the charts of those using CIC who were believed to be at risk for renal deterioration. The mean follow-up of this study was 4.9 years, and the average age of the patient at the last follow-up was 11.9 years. The study found that neonatal treatment enabled UDY to identify those infants at risk for upper tract deterioration, which was prevented by the start of Ditropan (oxybutynin chloride) and CIC. There was a similar improvement in UTI rate, hydronephrosis, and reflux. The percentage of patients with worsening hydronephrosis and persistent high intravesical pressures who needed bladder augmentation was 11% in the earlier treatment group versus 27% in the later treatment group, $p < 0.05$ (98).

Further Surgical Management for Reflux and Small Neurogenic Bladders

Ureteral reimplantation may be necessary for reflux; however, most people with neurogenic bladders have reflux from a high-pressure bladder and not from an ureterovesical junction that is dysfunctional. Bladder augmentation may be considered for a small bladder. Bladder augmentation increases the risk of bladder cancer, rupture, and stone, and mucus may be excessive in the urine, obstructing CIC (99). For cancer surveillance in those with augmented bladders, cystostomy is recommended annually starting 10 years following the bladder augmentation (84). Augmentation should be explored only after pharmacological management has failed and the system continues to be a high-pressure system, thus putting the kidneys at risk. The complication rate has been found in a recent study to be approximately 1 in 3 (100).

In conclusion, those individuals with a normal neurological exam with sacral-level spina bifida likely have a neurogenic bladder and need appropriate management. This point is demonstrated in a study of bladder dysfunction and neurological disability at presentation in closed spina bifida. There were 51 individuals in the study, with a mean age of presentation of 3.3 years. Of these patients, 25 had urinary tract disturbance, 12 had neurological problems, 33 had normal neurological exam, 21 had normal renal ultra sound (RUS), and 31 had abnormal video-urodynamics, despite normal neurological exam and RUS (101). Although the majority of individuals with spina bifida have neurogenic bowel and bladder, even if there are no motor signs of weakness, infants found to have perineal sensation are likely to be continent as well as having decreased renal complications and improved survival long-term (102).

Social Aspects

The bladder focus may put considerable strain on the family (103). Children and adolescents with neuropathic bladder using intermittent catheterization have worries about peers discovering CIC use to empty their bladder and leakage. However, urinary incontinence does affect self-esteem, and it is important to aim medical management at continence for psychological (104) as well as physical well-being. Urinary continence is an important developmental milestone in individuals with and without spina bifida (104). As mentioned in the previous edition of this book urinary continence, although important, should not occur at the expense of the kidneys.

NEUROGENIC BOWEL

Neurogenic Bowel Dysfunction

The colon, rectum, and internal anal sphincter are controlled by autonomic nerves. Parasympathetic innervation is from S2–S4, whereas sympathetic fibers arise from the lower thoracic and lumbar segments. Voluntary somatic motor and sensory nerve supply for the external anal sphincter is from S2–S4 through the pudendal plexus. Coursing through the spinal cord, these nerves have direct connections with the integrating supraspinal centers in the pons and cerebral cortex (74).

Colon peristalsis propels feces into the rectum. The gastrocolic reflex increases peristalsis for about 30 minutes after food intake. Rectal fullness initiates an autonomic stretch reflex, with relaxation of the internal anal sphincter, and creates a sensation of predefecation urge. In contrast, the voluntarily controlled external sphincter remains contracted to retain feces. When the situation warrants, this action is further enhanced by voluntary contraction of the levator ani, gluteal, and other thigh muscles. Defecation occurs when the external sphincter is voluntarily relaxed (74).

Most children with spina bifida have a patulous anus, absent cutaneous reflex response, and perianal sensory deficit. This indicates that S2–S4 segments are involved and leads to fecal incontinence. With lesions about L2, there can be an intact reflex arc to maintain perineal sphincter tone, despite absent rectal sensation.

Bowel Management

Fecal incontinence and constipation are common problems with spina bifida. The external anal sphincter can be weak and patulous. The reflex arc can be interrupted, causing an interruption of innervation to the anal sphincter.

Bowel management is essential for a successful bladder program. Daily bowel movements should be a goal. Aspects of daily bowel care include drinking

enough fluids and using an osmotic laxative such as polyethylene glycol, a high-fiber diet to bulk up the stool, and preferably digital rectal stimulation or a glycerin suppository in infants and younger children and a bisacodyl suppository 5 mg rectally in younger children and 10 mg rectally in those at least older than 2 years (usually school age for the higher dose). Although the use of the gastrocolic reflex is questionable in the spina bifida population, it is still advised to try having the bowel movement approximately 20 to 30 minutes after the nightly meal. (The morning or afternoon meal are both okay, too, but secondary to schedules, it may be difficult to embark on a bowel program just before school or work or during school or work.)

If the bowels are void of constipation, accidents of stool and urine are less likely. With severe constipation seen on an abdominal film, or with palpable stool still in the abdomen, the previous procedure should be followed along with an enema. Anatomic bowel obstruction should be ruled out by abdominal x-ray in severe constipation before a colonic cleansing enema is performed. A surgical procedure may be necessary, such as a catheterizable appendicocecostomy through the abdominal wall to flush the large intestine from the proximal end with an enema (105).

Anorectal manometry and biofeedback in the presence of intact or partial rectal sensation anocutaneous reflex offer encouraging results. Rectal sensation is considered normal when a rectal balloon inflated with 10 mL of water or less is perceived. The external sphincter activity can be recorded with surface electrodes. Repeated sessions of inflating and deflating the balloon comprises the biofeedback training (74). This can be done during a urodynamic procedure.

ORTHOPEDICS

Overview

Children with spina bifida are prone to multiple orthopedic issues during the course of their lifetime. Many of the problems can be predicted by understanding the effects the neurologic deficits will have on normal motor control and development. The goal of the multidisciplinary team is to anticipate these orthopedic issues and discuss options with the family and the person with spina bifida.

Orthopedic complications of spina bifida are common and have predictable patterns based on the neurosegmental level. Treatment needs to be developed in partnership with the family and child. Realistic goals of the interventions need to be discussed up front and the post-treatment plan of care developed. Surgeries often improve range of motion only to be short lived for lack of compliance with the postoperative plan. The plan often includes twice daily stretching and daily bracing and positioning.

Clinical Case. JR is a three-year-old with a history of a meningomyelocele at an L3 level. The family came in today to discuss the orthopedic issues. They feel that his hips are popping and he has a difference in his leg lengths. They also want to know if he is going to develop a scoliosis.

Spine

Spine deformities are common in this population and can be grouped as congenital or paralytic in nature (106). The common descriptions of spine deformities are classified as kyphosis, lordosis, and scoliosis. The probability of development of scoliosis tends to follow the neurologic level. Historically, those with thoracic lesions have an 80% to 100% chance of developing scoliosis, followed by lumbosacral levels, with a 5% to 10% risk. Stratification of spine risk defined by neurologic level makes intuitive sense. Glade et al grouped children into four groups to predict spine deformities: Group 1 is L5 and below, Group 2 is L3–L4, Group 3 is L1–L2, and Group 4 is T12 and above (107). Based on these categories, Group 1 tends to have a low probability of developing spine deformities, Group 2 has a medium risk, and Groups 3 and 4 have a high probability of developing spinal deformities (108). Scoliosis tends to progress most rapidly during growth periods, especially during puberty. The effects scoliosis can have on the individual include changes in sitting balance, abnormal weight distribution and increased risk for pressure ulcers, compromised respiratory capacity, functional changes, pain, body image changes, and impact on ambulation.

Treatment of the different spine deformities are grouped into observational, nonsurgical, and surgical. Nonsurgical options include bracing, seating, therapy, and complementary techniques. Spine orthotics and braces used are mainly thoracic lumbar spine orthoses (TLSO) and incorporate three points of pressure to maintain alignment of the spine. Wheelchair seating can be incorporated to optimize spine position using molded systems or lateral support. However, molded seating systems often encourage spinal curve and have to be redone more frequently than those that are noncontoured. Therefore, a TLSO is a good option to encourage the spine to be in as straight a posture as possible, especially during those activities when the pressure on the spine is the greatest. The pressure on the spine is greatest in sitting, followed by standing, and least in the supine position. Surgical options should be considered when spine curves are above 45 degrees and the child is at an appropriate developmental level.

Spinal deformities in this population present some unique challenges. The child with a high lesion in the thoracic area may be born with a congenital kyphosis and Gibbus deformity (Fig. 9.7). These structural abnormalities not only cause seating and mobility issues, but also present the clinician with challenges in maintaining skin integrity. The deformity can affect the development of the chest and has cosmetic implications. Treatment options include conservative management with bracing and seating modification to much more aggressive approaches. Kyphectomy and posterior fusion done at an early age is one option. This surgery generally includes not only bony procedures, but also may include transection of the spinal cord. These surgeries are known for their high complication rate (89%), but have an average kyphosis correction of 81.9 degrees. However, 22% of the cases in this series required shunt revision within six weeks due to surgically induced altered cerebrospinal fluid (CSF) dynamics (109) (Fig. 9.7).

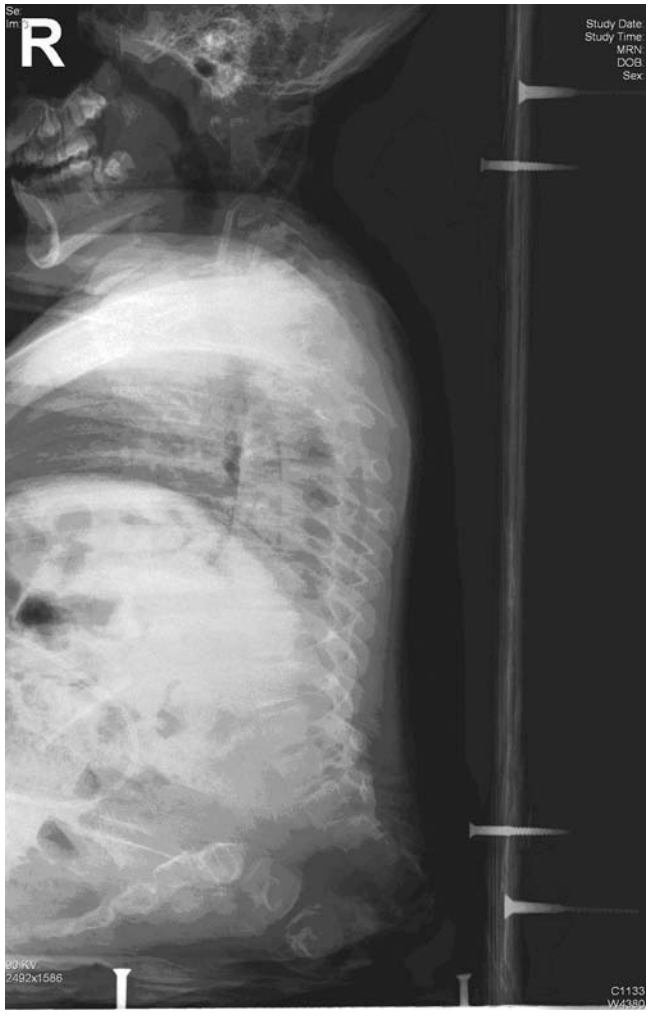


Figure 9.7 Congenital structural kyphosis with a sharply angled curve, or Gibbus deformity, associated with thoracic-level spina bifida.

Once a definite procedure is required, more traditional techniques for spine fusion are done. Surgery timing is based on degree of curvature and is generally considered at 45 degrees. Different approaches for fusion include anterior, posterior, and endoscopic options. Realistic goals of the procedure, along with potential complications, should be discussed prior to surgery. These include improved sitting, reduced pelvic obliquity, impact on functional status, and ambulation. To maintain ambulation and control pelvic rotation, discussions about fusion to the pelvis are important considerations (110). However, fusion to the pelvis can interfere with sitting. The impact on function and self-perception after surgery remains controversial (111). Complications after surgery are common and include a high risk of infections, pseudoarthrosis, and instrument failure. It has been noted that it takes several months to get back to presurgery ambulation baseline.

Newer surgical treatment for neuromuscular scoliosis has evolved over the last decade. Techniques to deal with the growing child have encouraged development of fusionless surgeries. Specific goals of these techniques are to delay definitive surgery until the child has reached a more optimal size, allow chest development and improve lung capacity, and sometimes to avoid surgery. These techniques include growing rods, intervertebral stapling, and use of vertical expandable prosthetic titanium rods (VEPTR) (112,113) (Fig. 9.8).

Hips

The development of the hip and associated problems is related to the neurologic level. Broughton found that by age 11 years, children with thoracic-level lesions had a 28% risk for dislocation, L1–L2 had a 30% risk, L3 had a 36% risk, L4 had a 22% risk, and L5 had a 7% risk. The development of hip flexion contractures was highest in the thoracic- and high lumbar–level lesions (114). Children with thoracic-level lesions have no muscle influence on hip stability and may or may not develop hip dislocation. These children tend to frog-leg (hip abduction and external rotation) when lying down and develop contractures of the hip flexors and external rotators. In addition, the tensor fascia lata becomes contracted and may need to be surgically lengthened if it affects positioning. Children with high lumbar lesions have an imbalance of muscle activity around the hip joint. The active hip flexors and adductors (L1–L2) with unopposed abduction and extension tend to result in persistent coxa valga and development of acetabular dysplasia. These forces can result in hip subluxation and dislocation. This process occurs not only in the higher lumbar levels, but also in the mid- and lower lumbar segments. Weak or absent hip extension and abduction are directly related to hip dislocations. Unilateral hip

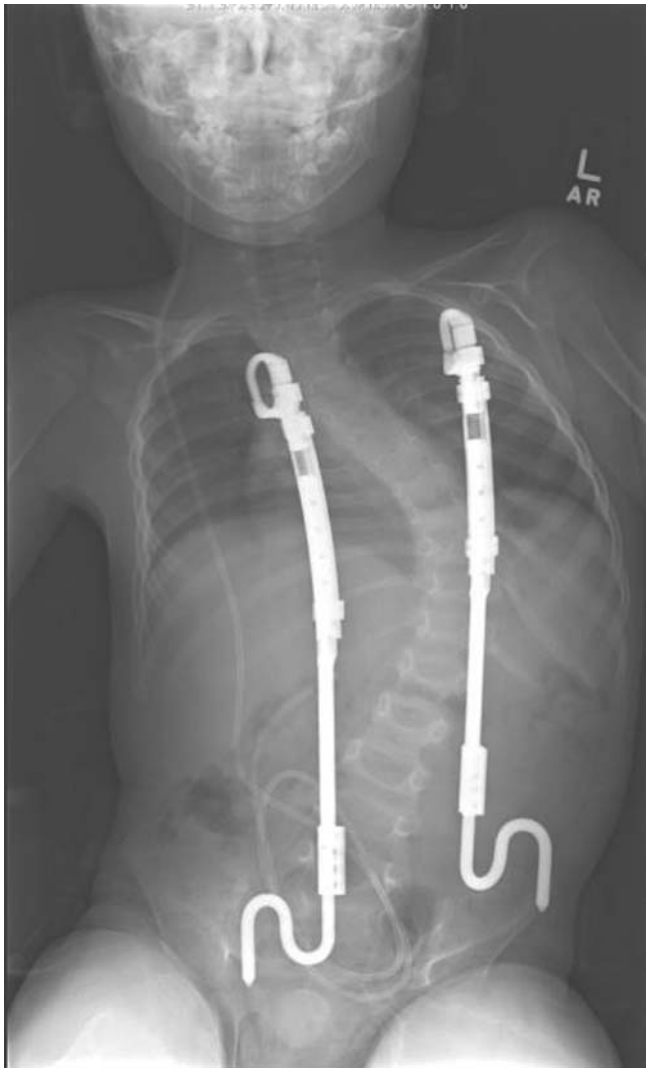


Figure 9.8 Child with spina bifida and scoliosis treated with vertical expandable prosthetic titanium rib (VEPTR). Note the spinal dysraphism with increased intrapedicle width and ventriculoperitoneal shunt.

dislocations tend to cause pelvic obliquity, and surgery has been advocated. Bilateral hip dislocations generally do not require surgical interventions. Heeg et al found that it was more important to have a level pelvis and good range of motion for ambulation then to have located hips (115).

Hip flexion contractures can be treated with soft tissue releases. Nonsurgical interventions include lying prone for 30 minutes daily. Other soft tissue surgeries designed to correct muscle imbalances have been employed. Transfers of the iliopsoas to the greater trochanter in association with adductor releases are designed to improve the hip abduction and flexion motion. Osseous surgeries may need to be done to correct acetabular dysplasia and rotational deformities (Fig. 9.9).

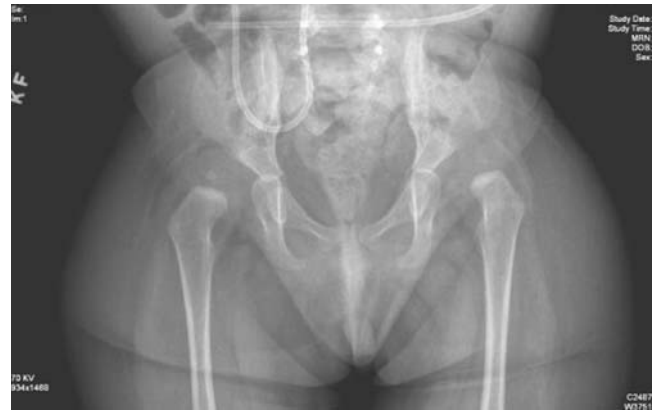


Figure 9.9 Bilateral hip dysplasia. Note the dysplastic acetabulum, femoral head migration, and broken Shenton's line. Ventriculoperitoneal shunt is in place.

Knees

The knee motion is influenced by the muscular control of the quadriceps and hamstrings. Knee flexion contractures are a common occurrence at all neurologic levels, but are seen in a higher frequency in thoracic and high lumbar lesions. Weak quadriceps and positional factors, along with fractures and spasticity, have been proposed as the etiology.

Treatment is geared toward preventive strategies of stretching and standing. When all else fails, surgical interventions are indicated. Contractures of greater than 30 degrees often require surgery. Aggressive posterior capsule release is used in thoracic and high lumbar lesions and soft tissue releases in lower levels (116). Recent techniques have evolved in the treatment of knee flexion contractures, such as the "guided growth" approach developed by Klatt and Stevens. By surgically placing anterior tension band plates, gradual correction of the deformity is achieved by the use of tension forces to guide bone development in growing children (117). Knee hyperextension can be seen in the L3 level from unopposed contraction of the quadriceps. Serial casting and capsule releases may be required. Abnormal gait patterns have been identified and include genu valgus. This can result in knee pain and may require more aggressive bracing as a preventative strategy.

Tibia

Rotational deformities in the tibia are fairly common and can have a functional impact on ambulation. Internal and external tibial torsion can both affect gait patterns. In-toeing is often seen in L4/ L5 neurological levels and is related to muscle imbalances, particularly in the hamstrings. The medial hamstring is much stronger than the lateral and may internally rotate the

leg. Derotational surgeries should be used only in those who are ambulatory in the community (118).

Feet

Foot deformities are fairly common in children with spina bifida. In fact, it is felt that almost 90% have some abnormality. Foot management is based on developing a plantar-grade foot and to protect vulnerable soft tissues. The clubfoot (talipes equinovarus) deformity in these children can be more rigid than in other populations. The foot classically has hind foot varus and equinus; the forefoot is supinated and adducted and is rigid. Nonoperative management involves early casting and splinting. Conservative methods often have suboptimal effects and need to be done cautiously in insensate feet. Surgery should be scheduled when the child becomes weight bearing to optimize effects. Congenital vertical talus deformity or rocker bottom foot is a nonreducible dislocation of the navicular on the talus. The talus is in equinus, and the Achilles tendon is short. The talus on radiographs is vertically positioned, and clinically the talus is medially located. Muscle imbalances are the implicated forces in this deformity.

Serial casting is often not effective, and surgical intervention is often required. Timing for surgery is before age 2 years. Complex tendon releases and bony interventions are done. Salvage procedures include triple arthrodesis and the Grise procedure. Calcaneus deformities occur when the anterior tibialis, toe extensors, and peroneal muscles are unopposed. This is seen in those with L4-level spina bifida. The calcaneal deformities affect the gait pattern and can cause the skin over the heel to break down. Stretching is not effective, and surgery is indicated. This includes tendon transfers of the anterior tibialis and anterior capsule release. Even though some power can be generated in plantarflexion, this is generally not enough to walk without braces. Equinus deformities generally require an Achilles lengthening procedure. Cavus foot deformity is found in sacral-level injuries. Intrinsic muscle abnormalities lead to high arches and toe clawing. These deformities can cause areas of increased pressure and the risk for skin breakdown. Orthotics and extra-depth shoes may reduce pressure points. Surgery is indicated if these measures fail. Plantar fascial release and multiple bony surgeries can be done. Toe deformities such as hammer toes often require tendon procedures and fascial release.

Clinical Case: JR is an L3 level, which means he has strong hip flexors, quadriceps, and adductors. We know this places him in Group 2 related to risk of spinal deformities. This suggests he has a medium risk for developing scoliosis and lordosis. His level places him in a very high risk for hip subluxation/dislocation (36%), and we know that he may develop knee contractures. Leg length problems will be based on dislocations. He will not have any foot control and could have congenital foot abnormalities.

CLINICAL PEARLS

- Scoliosis associated with spina bifida can occur at any neurologic level, but is most common in the higher lesions. Thoracic and high lumbar levels almost always develop these spinal deformities.
- Hip dislocations are most common in the L3 level based on muscle imbalances of hip flexion and adduction being present while the opposing muscles are weak or absent.
- Foot deformity treatment is geared toward developing a plantar-grade foot and minimizing pressure areas.

Figure 9.10 shows scoliosis noted in a child with lumbar level meningomyelocele.

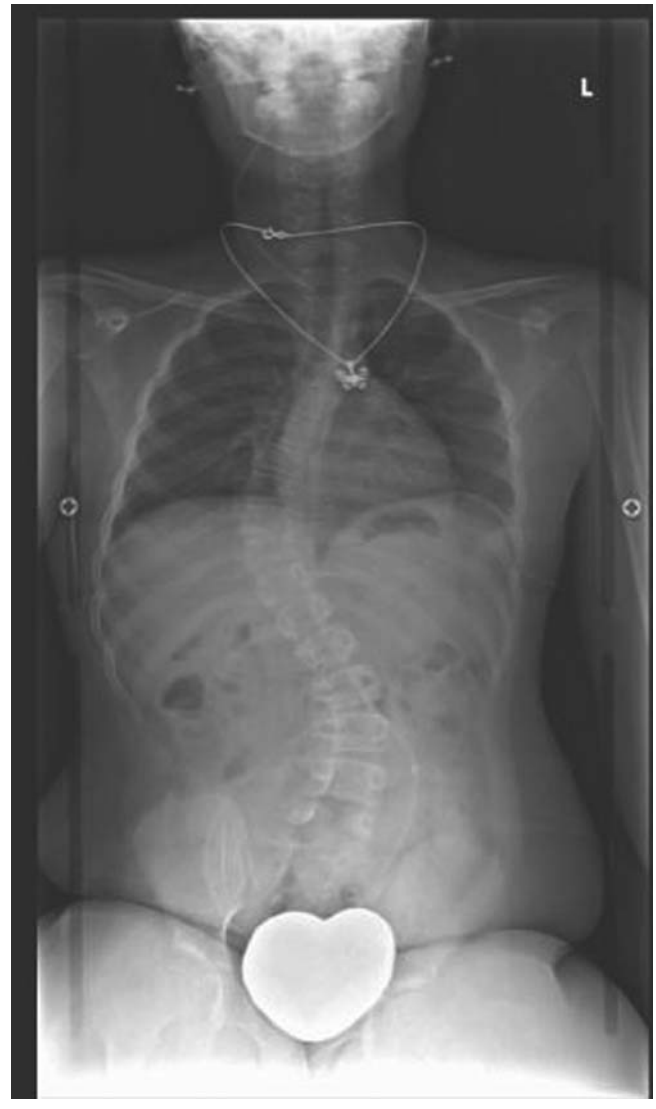


Figure 9.10 Scoliosis noted in child with lumbar-level meningomyelocele.

REHABILITATION

The role of the rehabilitation specialist with neural tube defects is specifically to understand the complex nature of this group of disorders and apply sound principles in defining a plan. The plan will change based on the level of the lesion, the developmental age of the individual, family resources, and community resources. This plan should be family-centered and include all pertinent disciplines.

Musculoskeletal

Conservative management of potential or existing musculoskeletal deformities begins in the newborn and should continue as part of daily care thereafter. Passive range-of-motion exercise (PROM) is applied to all joints below the level of paralysis, with special emphasis on joints with evident muscle imbalance. The infant should not lie constantly in one position, but should be moved and turned frequently. This practice must be taught to parents, not only to mitigate contractures, including those related to gravity, but also to avoid breakdown of the anesthetic skin. For the same reason, splints must be used with great precaution, removed frequently to check for skin irritation, and adjusted or discontinued if such problem occurs. PROM and splints are advisable after surgical correction of deformities to maintain joint mobility gained by the procedure. Strengthening exercises are sometimes beneficial for partially innervated muscles or after surgical muscle transfer for improving strength or function. They are also part of ambulation training with upper extremity assistive devices.

Examination of motor function in the neonate is based primarily on observation of spontaneous movements, presence or absence of deep tendon and infant-like reflexes, habitual postures, passive joint motion, and tone. For example, consistently maintained hip flexion, particularly when passive extension is incomplete, is a sign of hip extensor weakness. Palpation of muscle bulk is helpful because atrophy may be evident with severe or complete paralysis in particular muscles. In assessing motor or sensory function, the presence of spinal reflex withdrawal or triple flexion of hip, knee, and ankle should not be mistaken for voluntary motion and preserved sensation, particularly in high spinal lesions. A normal asymmetric tonic neck reflex elicited in the arms without response in the legs suggests lower extremity paralysis (74).

Development

Development is the natural and predictable sequence that an individual progresses through to attain skills in multiple domains. Children with physical disabilities may not be able to accomplish these tasks, given their physical and cognitive limitations. The impairment will affect activities and participation. Motor acquisition can be predicted based on the level of the lesion, which affects normal balance, coordination, and postural control.

First Six Months of Life

Most children do follow normal development, attaining head control, fine motor skills, and language. This can be disrupted in light of hydrocephalus, medical complications, and severe cognitive involvement (see Table 9.2).

Six to Twelve Months of Age

This is a critical time for gross motor development, where most typically developing children are sitting, crawling, and walking. Predictably, children with spina bifida can be expected to have delays in this domain. The residual motor function will allow the medical team to discuss realistic expectations for family members. Early mobility mirroring normal development should be incorporated into the rehabilitation plan. Lack of environmental experiences can lead to sensory/motor deprivation and affect developmental potential. Children are amazing at learning substitution patterns to compensate for these neurologic losses.

Head control is a crucial milestone and prerequisite for emerging skills. Most children achieve this skill irrespective of level of lesion. Delays are mainly central in etiology. Children with high thoracic lesions lack adequate trunk and abdominal muscles to get and maintain sitting balance. Compensatory strategies include prop sitting, rolling to side, and pulling up. Sitting is necessary for play and hand skills, and appropriate equipment should be used. This skill may be delayed in children with mid-lumbar and lower lesions, but they will achieve this skill. Rolling is always delayed in children with thoracic and high lumbar lesions. To roll, a child uses head, trunk, and legs. Thoracic and high lumbar muscle weakness delay this skill until the child can figure out adapted motions, including using momentum to propel the legs. Most have learned this skill by 18 months of age.

Floor mobility is a way for a child to move from place to place. It is needed for environmental exploration, and different neurologic levels have different methods. Children with high-level lesions tend to roll, and in sitting, lean forward over the legs and combat-crawl. Crawling is really not a viable option unless the child has hip flexor strength and knee extension.

Ambulation/Mobility

As stated, the job of a child is to explore the environment. The ability to ambulate and gait abnormalities

9.2 Gross Motor Skills Acquisition

LEVEL OF LESION AND SKILL	T12 AND ABOVE	L1/L2	L3/L4	L5/SACRAL
Rolling over	Delayed, but can be achieved by compensatory means at around 18 months	Delayed, but can be achieved by compensatory methods	Delayed	Minimal delay
Sitting	Delayed, but can sit with propping and equipment	Delayed but can achieve sitting, may have some balance issues	Delayed but able to sit	Minimal delay
Floor mobility	Rolling, combat crawling, bottom scooting	Rolling, combat crawling and bottom scooting	Modified crawling	Crawling
Ambulation	With adapted equipment, orthotics, poor probability of ambulation HKAFO, KAFO, RGO, dynamic and static standers	With adapted equipment and orthotics, household ambulation KAFO, RGO, dynamic standers	With orthotics, household and community ambulation KAFO, floor-reaction AFO, AFO, walkers, and crutches	Community ambulation AFO, UCB

Source: HKAFO, hip knee ankle foot orthosis.

have a direct relationship to the neurologic level of the spina bifida. Mobility can be achieved through various means, including self-propulsion, adapted equipment, and orthotics. Introduction of equipment should follow developmental sequences. Children are pulling to stand at around 1 year and walking by 18 months. Introduction of dynamic standers can be done early in thoracic and high lumbar levels. These include mobile prone stander, Parapodium, and swivel walkers. If the latter is used, you may also need to incorporate a reverse walker. The advantage of using this type of equipment is not only mobility, but also passive stretch of the joints in the lower extremities and a different orientation to the environment.

Orthotics are used in all levels of spina bifida. The child with a thoracic and high level requires much more sophisticated bracing than the lower lumbar levels. Hip knee ankle foot orthosis (HKAFO) and knee ankle foot orthosis (KAFO) stabilize the joints in the lower extremities to allow upright positioning. HKAFO is used when hip instability interferes with knee alignment. With the HKAFO, a child must use a walker and move the brace forward by either leaning or lifting to achieve ambulation. It is a difficult skill to use and is why many children abandon this as they grow. Use of reciprocal gait systems includes a cross-linked hip orthosis, a reciprocal gait orthosis (RGO), or a free hinged gait orthosis such as a hip-guided orthosis (HGO). The isocentric RGO system uses a cabling system. The brace provides structural stability during the stance phase on one side while the opposite side advances. Simply putting hip flexion on one side causes hip extension on the opposite through the cabling system.

Hip-guided orthosis or the Orlau ParaWalker is also an RGO-type system. It does not employ the use of cables, but uses joint stabilization and a rocker foot plate. There are some advantages to using a walking system in young children, as Mazur's study showed fewer fractures and pressure ulcers when comparing those who strictly use a wheelchair to those using a walking system (120). Implementation of these types of braces is best employed when children are around 3 years of age.

Mid-lumbar lesions have knee extension muscles that have a great impact on ambulation. It is imperative that hip flexion and knee flexion contractures be addressed, as these affect upright position. Options include KAFOs and, in some cases, floor-reaction AFOs to assist with knee extension. The majority of these children can hope to have household ambulation with limited community ambulation. All children with spina bifida can be expected to have a delay in ambulation even at the lower sacral levels. In children

with the lower sacral levels, parents can expect ambulation by age 2 years. Typical gait patterns include a Trendelenburg associated with weak hip abduction and steppage gait associated with weak dorsiflexors. Bracing includes AFOs and floor-reaction AFO. Floor reaction is used to assist knee extension and prevent the crouching patterns seen in stance phase. The foot must have some flexibility to accommodate this brace. Community ambulation is possible in these individuals.

Bracing studies have shown that the use of ankle foot orthosis (AFO) in children with L4-sacral–level lesions had improved energy expenditure. Walking speed and stride length increased, while energy costs decreased, using braces compared to not using braces. This is surmised to be related to stability that the braces provide (121). If crutches will be used, most children cannot learn the skill until at least 2 to 3 years of age. Walkers can be used at earlier ages, and dynamic standers can be used when children should be upright.

Ambulation is always one of the first questions parents will ask a health care provider: Will my child be able to walk? To address this question, one needs to look at the whole child and all the factors involved. Swank found that sitting balance and neurologic level were good predictors of ambulation potential (122). A study by Williams et al tracked 173 children with spina bifida (123). Thoracic level was found in 35 children, and only 7 walked at 4.5 years. The study followed 10 children with L1/L2 lesion—and 5/10 walked by 5 years. They followed 15 children with L3, and 9/15 walked at 5 years. There were 45 children in the L4/L5 group, and 38/45 walked at almost 4 years. The 68 children with sacral level were able to achieve ambulation by 2 years. Walking was delayed in all groups, and the higher levels abandoned walking earlier than was previously documented. With development, those with a sacral level do not lose ambulation skills (123). Success in maintaining ambulation has been associated with muscle function of the hip abductors and ankle dorsiflexors (124).

This may be a paradigm shift toward earlier acceptance of wheelchair mobility as a viable option. The demands of walking increase as the person grows taller and requires more energy. Spine deformity has been well documented to have an impact on ambulation. Scoliosis surgery can change ambulation patterns.

Wheelchair mobility should be introduced to all children who will potentially use this as a primary or secondary option. We introduce it at a fairly young age and have found children as young as 1 year can efficiently push a wheelchair. This allows them independence to explore the world around them. Wheelchairs should be appropriately configured to meet the needs of the child. A child is not a small adult and should not be placed in a wheelchair they can grow into. Seating will be adjusted based on neurologic level, posture, and balance. Proper cushions, the seat back, seat, and foot rests should be positioned to prevent pressure areas from developing.

LATEX ALLERGY

Today, latex allergy and latex precautions in the spina bifida population are well-known issues. Prior to the 1980s, latex allergy was a largely unknown entity. Allergy to latex and the potential for anaphylactic allergic reactions came to medical attention in the 1980s in increasing numbers, with the increase usage of latex gloves for barrier protection from hepatitis and HIV. With increased awareness since that time, clinical medical facilities typically take precautions with items containing latex and frequently do not allow products that have high loads of allergen, such as latex gloves (especially those with powder), latex balloons, rubber plungers, blood tourniquets, and rubber dams for dental procedures. It is also recommended that toys and other items with latex be avoided.

Clinical signs of latex allergy are skin rash, angioedema, and, in severe cases, bronchospasm and other symptoms of anaphylactic reaction (74). The prevalence of latex sensitivity (ie, positive IgE skin testing) has been reported as high as 72% in the spina bifida population (125). Spina bifida patients with latex sensitivity are at high risk for anaphylactic response to latex-containing products. The propensity for latex allergy in the spina bifida population is increased by early exposure, specifically on the first day of life, and family of origin atopy. Neurosurgical procedures appear to be correlated with increased latex sensitization; intra-abdominal procedures are not (126,127). Recent studies continue to demonstrate that children with spina bifida have an increased propensity for latex sensitivity and allergy than those who have had multiple surgeries for other diagnoses, implicating that there is something inherent in the condition that predisposes to this allergy (127–129).

In the 1990s, latex-fruit syndrome—most frequently involving the banana, avocado, kiwi, and chestnut cross-reactivity—was reported. Papaya, mango, bell pepper, fig, tomato, celery, and potato are other foods that are potentially problematic too. This list, although comprehensive, may not include all problematic foods. The cross-reactivity is exhibited on radioallergosorbent test (RAST). There can be allergen cross-reactivity between latex and the proteins in these foods. Latex sensitivity and allergy develop over time; therefore, negative RAST tests are not definitive for future allergic reactions. Furthermore, negative

skin tests may or may not be reliable and may depend on the source of the allergen. It is not always clear whether latex sensitization precedes or follows the onset of food allergy (130).

A detailed history regarding latex sensitivity and allergy is important. The management of this condition includes a MedicAlert bracelet and education regarding cross-reactivity between latex and foods in the spina bifida population. Avoidance of these foods is important. Avoidance of latex even as early as day one of life and an anaphylaxis kit are recommended (131). Potential risks must be discussed at each visit. Latex immunotherapy may be a treatment in the future, but currently it is not available secondary to adverse reactions (131).

SELF-CARE

Children with spina bifida should be encouraged to acquire independence in age-appropriate activities of daily living (ADLs) at an early age. Fine, gross, and visual motor skills are rarely significant enough to account for delays in ADLs. Despite adequate intelligence and upper extremity function, delays in ADLs are often appreciated. Family members should be instructed to proceed with age-appropriate expectations. If there are continuing problems and extensive lower extremity paralysis, an occupational therapy consultation is necessary; this should include education of the child and the parents (74).

OBESITY

Similar to the general population (132), excessive weight can be problematic in individuals with spina bifida. Secondary to paralysis and wheelchair mobility, obesity increases the risk of decubiti. In addition, there is increased stress with physical activities on the upper extremities. Self-image, social adaptation, and acceptance are also compounding factors with obesity and spina bifida. The development of positive self-image is greatly affected by social relationships (133).

Body mass index is not as useful, as height calculations may be difficult to do accurately. Subscapular skin-fold thickness is more reliable to assess for obesity. Opportunities for physical exercise are fewer than those for age-matched peers without disabilities. Therefore, preventative anticipatory guidance regarding weight and exercise should be part of the comprehensive care and education for individuals with spina bifida from an early age, as weight loss may be difficult once the child/adolescent or adult is overweight. It may be difficult for children and adolescents to grow into their weight secondary to shortened stature (see the following section). Recommended diet guidelines include decreased caloric intake by 10% to 20%, and a diet low in fat and carbohydrates and high in protein and fiber, with proper vitamin supplementation (74).

PRECOCIOUS PUBERTY

Precocious puberty traditionally is Tanner stage II breast development before age 8 years and testicular enlargement before age 9.5 years (134). Precocious puberty is associated with an accelerated growth velocity and early epiphyseal fusion (134).

Individuals with spina bifida and precocious puberty can have marked short stature if untreated. Their short stature results from abnormalities of the hypothalamic-pituitary axis, the Chiari II malformation, and hydrocephalus (135). These abnormalities are thought to cause premature pulsatile secretion of gonadotropin-releasing hormone (GnRH) (134).

Screening lab tests for girls include a luteinizing hormone and estradiol or testosterone level. For boys, morning testosterone values in the pubertal range are diagnostic with an elevated luteinizing hormone level. For both boys and girls, if the luteinizing hormone level is not clearly elevated, this should be retested following stimulation with a GnRH agonist before treatment is begun (134). Bone age should also be tested and will likely be advanced.

Treatment with growth hormone leads to desensitization of the pituitary gonadotrophs, decreasing the release of luteinizing hormone. Treatment may be associated with menopausal symptoms such as hot flushes and may be associated with headaches. In a recent study in the spina bifida population, near adult stature, improved BMI, better reported self-esteem, and better gross motor skills were reported after treatment with GnRH (135).

OSTEOPOROSIS

Osteoporosis is the pathologic reduction of bone matrix and minerals, whereas osteopenia is a reduced density of bone. Osteoporosis has been identified as a medical problem in the adult with myelomeningocele (136). Although typically considered an adult disease, osteoporosis is a disease that starts in childhood (137,138). The age at which abnormalities in bone mineral density (BMD) first present in the spina bifida population is not known. It has been reported that children with myelomeningocele have a higher fracture risk and that those individuals who fracture have a lower bone density than age-matched peers (139). In patients with myelomeningocele, fractures typically occur in the long bones of the lower extremity, most commonly

in the femur and less so in the tibia (140,141). Recent data suggest that fractures are present at all levels of spina bifida, with an annual incidence of about 3%. The age for first fracture was around 11 years, with the tibia and femur most involved. Of those with fractures, 1 out of 4 reported multiple fractures (119).

Most contemporary studies of osteoporosis utilize dual energy x-ray absorptiometry to assess bone density. Non-weight bearing conditions such as cerebral palsy (CP), Duchenne muscular dystrophy (DMD), and spinal cord injury have been shown to be associated with decreased BMD that can result in fractures, even in the pediatric population (142–144). It has been shown that BMD of the lumbar spine and proximal femur in children often correlates poorly, particularly if BMD is low (145). Studies currently have found utility in assessing the BMD in the lateral distal femur, as the lower extremities are a common site of fracturing (146). Recently published studies in patients with myelomeningocele attempt to describe the effect of non-weight bearing on BMD; however, these studies have been limited by small sample size, inclusion of a limited number of pediatric patients with myelomeningocele, and older technology (138,139,147–149).

There may be technical difficulties in obtaining adequate lumbar spine and proximal femur assessments due to vertebral abnormalities and hip deformities (150).

Treatment

Toddlers (age 1–3 years) require about 500 mg of calcium each day. Preschool and younger school-age children (age 4–8 years) require about 800 mg of calcium each day. Older school-age children and teens (age 9–18 years) require about 1300 mg of calcium each day. This guideline is set by American Academy of Pediatrics (AAP) to meet the needs of 95% of healthy children (151,152).

There are limited natural dietary sources of vitamin D, and adequate sunshine exposure for the cutaneous synthesis of vitamin D is not easily determined for a given individual. In addition, sunshine exposure increases the risk of skin cancer, and decreased sun exposure is not uncommon for individuals with disabilities. The recommendations from the AAP have been revised to ensure adequate vitamin D status. It is now recommended that all infants and children, including adolescents, have a minimum daily intake of 400 IU of vitamin D beginning soon after birth. The current recommendation replaces the previous recommendation of a minimum daily intake of 200 IU/day of vitamin D supplementation beginning in the first two months after birth and continuing through adolescence. These revised guidelines for vitamin D intake for healthy infants, children, and adolescents are based on evidence from new clinical trials and the historical precedence of safely giving 400 IU of vitamin D per day in the pediatric and adolescent population (153).

In the setting of osteopenia or osteoporosis, individuals' vitamin D status and dietary history should be evaluated. Any deficiencies should be treated. Weight-bearing activities should be encouraged; however, there has been little to no data in regard to stander use in the spina bifida population. Standing weight-bearing exercises or activities can apparently increase BMD in the lumbar spine or femur in children with cerebral palsy (154,155).

Treatment for pathologic fractures supports the use of medication such as bisphosphonates. Prevention is key, and careful attention to daily calcium and vitamin D intake, as well as a standing or walking program for those that are nonambulatory, is essential to minimize the reduction in bone density and the fracture risk (156).

More aggressive pharmacologic therapies have been used in other pediatric patient groups for the treatment of osteoporosis. The current treatment garnering most interest is the bisphosphonates. Bisphosphonate use has not been studied in the spina bifida population, but there has been increased use in pediatrics (157,158).

COGNITIVE FUNCTION

Neuropsychology and Learning Problems Associated With Spina Bifida

Spina bifida has long been associated with specific neuropsychological characteristics marked by deficits in nonverbal learning abilities, including math concepts, visual–spatial perception, spatial reasoning, and time concepts (159). Recent studies have revealed specific weaknesses in processing speed, organization, and personality traits. In addition, verbal skills—once thought to be a strength due to the precocious development of conversational speech in young children with spina bifida—are now known to be weak in complexity, organization, and abstract content (160). Recently, neuropsychological studies have investigated the development of executive control processes in individuals who have spina bifida. This line of study has proven beneficial in understanding the essential underlying neuropsychological characteristics associated with this syndrome.

Children with spina bifida can manifest several types of complex learning disorders and neuropsychological sequelae. Intellectual function is historically defined as the intelligence quotient, or IQ. When individuals with meningomyelocele have been compared

to typically developing peers, a shift to the left is present. Although most fall in the average range, the level of the lesion appears to have some impact on this parameter. Thoracic-level lesions trend toward a lower average IQ, while sacral levels cluster in the opposite direction and tend to have higher IQs.

The development of hydrocephalus is a key component in the spectrum of cognitive impairment. A recent study by Lindquist compared children with hydrocephalus to those with hydrocephalus and spina bifida. Both groups had impaired learning, memory, and executive function, suggesting that hydrocephalus is a major factor in these deficits (161). When individuals with spina bifida were stratified to those with and without hydrocephalus, those without hydrocephalus had relatively normal neuropsychological testing scores, while those with hydrocephalus showed impairments, especially in executive functioning (162). The combination of spina bifida and hydrocephalus has also been implicated in deficits of working memory and processing speed, along with retrieval problems (163,164). The level of the spina bifida in association with hydrocephalus has been implicated in additional difficulties with learning. Higher-level lesions above T12 with hydrocephalus showed more severe structural brain anomalies and a poorer cognitive outcome (165). The structural abnormalities were noted in the midbrain, tectum, pons, and splenium; the cerebellum was not noted to be involved. Most recently, newer imaging technology has been better at defining structural differences in the brains of individuals with spina bifida and hydrocephalus, implicating myelinization impairments and abnormal white matter tracts along with a decrease in the grey matter and caudate nucleus structure (166). The clinical implications are emphasized in a recent study done by Matson, which tracked individuals through multiple shunt revisions with neuropsychological testing. The testing revealed lasting cognitive effects after hydrocephalus in verbal IQ, processing speed, organization, and response inhibition (167). Recently the Chiari II malformation has been implicated as affecting specifically verbal memory and fluency (168).

When looking at the trends in testing those with spina bifida, there is a discrepancy between verbal and performance IQ scores. Verbal scores tend to be higher, and the classic "cocktail party syndrome" is frequently encountered in this population. The cocktail party syndrome describes a speech pattern characterized by repeating phrases, using common phrases, and talking about unrelated topics. This pattern creates the impression of high intellectual functioning to the untrained observer.

One of the most common identified educational issues is a nonverbal learning disorder (NVLD). There are three areas of difficulty in those with NVLD: motoric, visual–spatial organization, and social (169). Neuropsychology testing identifies problem areas to be impulsivity, difficulty with staying on task, memory, sequencing, organization, higher reasoning, mental flexibility, and visual perceptual skills. A typical child with spina bifida may have early success in preschool because of verbal skills, but begin to have difficulties once the academic demands become more challenging. In areas of self-care, they often are not at the level of their peers. These children and adults have problems with developing and maintaining bowel and bladder programs. The difficulties are not only in the sequencing, but also in realizing the social implications. NVLD has consequences in school, such as problems with cognitive and educational goals. These individuals have problems with sequencing and memory, along with special visual problems. This can eventually lead to problems with homework and school performance. Social skills acquisition can be affected by such simple things as understanding complex conversations, plots of books, and social jokes.

Selective memory disorders have been identified. This is the ability to sort out information and prioritize it in higher ranking order. This is important in the classroom to sort out irrelevant material from extraneous information. If you cannot select information, you can get lost in the details (170). Adolescent studies have shown impairments in attention and executive function (171). There is evidence that attention deficit problems are more common in children with spina bifida and that it tends to be more related to inattention as opposed to hyperactivity (172). These problems tend to persist into adulthood, and testing shows persistent deficits in the areas of reading and writing. This continues to have an impact on employability and self-care skills as one ages with spina bifida (173).

The learning disorders that influence executive control have a great impact on education and social interactions. Executive control processes are closely associated with the development and functioning of the frontal lobes. Studies of fetal neurological development indicate elementary differentiation of neural cells and migration to the anterior region of developing brain structures as early as 24 days after conception. During development after birth, executive control processes become differentiated and refined in tandem with progressive myelination of the developing brain.

Studies of adolescents who have spina bifida have revealed generalized difficulties with all of these abilities, highlighting significant deficits in initiation, mental flexibility, and organization (174). This same constellation of weaknesses in executive control processes has been discovered in preschool-age children with spina bifida. Abnormal executive control functions may be one of the major factors, explaining the surprising failure of children who have spina bifida to

achieve the typical functional adaptive competencies of their same-age peers, despite adequate intellectual abilities.

Deficits in executive control processes—especially initiation, organization, and mental flexibility—are highly likely to be associated with reduced acquisition of social competencies (dating, living independently, "motivation" for independence, employment). It is also plausible that these neuropsychological challenges contribute to reduced acquisition of functional daily living skills (self-catheterization, independent care of personal hygiene, execution of household chores, making and keeping appointments). Weak executive control capacities may also underlie the mental health problems so often seen in adolescents and young adults with spina bifida. Despite the availability of psychotherapy, they often have difficulty putting talked-about goals and plans into action.

So what is the bottom line with regard to cognition in children and adults with spina bifida? We can assume that most will have some type of learning problem. Those who have hydrocephalus and multiple shunt revisions may have more impairments than those who do not. We know that deficits can be in multiple domains and include visual-spatial, perceptual motor, organization, executive function, sequencing, memory, attention, or just about any other type of learning problem. We feel that early identification and intervention programs are important in this population.

There are some basic principles in treating individuals with a nonverbal learning disorder that apply to this population:

- Identification of the learning disorder is critical. Testing should be done prior to entrance into school.
- Modified program to address these specific needs. Some children will need 504 and Individualized Education Plans (IEP), which are individualized school plans for children with special needs.
- Providing structure and direction for education. Be specific and repetitive.
- Teach step-wise and sequentially (baby steps).
- Make sure to teach social education, as these children may not pick up social cues.
- Use multiple sources available on NVLD for guide in education, self-skills training, and social integration.

LONG TERM

Aging With a Neural Tube Defect

Adults with spina bifida have the normal aging medical problems in addition to those associated with their disability. Age-associated changes can have an impact on medical and functional systems. In treating the adult patient, one has to evaluate the usual age-related medical problems as well as those unique to this population. Medical complications and cardiovascular disease may present at an earlier age. Successful transition to adult-based clinics appears to be based on a few key factors and include preparation, flexible timing, care coordination, transition clinic visits, and interested adult-centered health care providers (175). Adults with chronic conditions generally require more medical visits yearly and have an admission rate nine times more than the nondisabled. Adults with spina bifida in general are satisfied with life, but the area of largest concern is in self-care ability and partner relationships (176,177).

Spina bifida is associated with abnormalities in the brain and spinal cord. Approximately 90% of adults will have ventricular–peritoneal shunts. Shunt malfunctions can occur at any age and present with the classic symptoms of chronic headaches, vomiting, personality changes, concentration difficulty, and other neurologic changes. Shunt malfunction can lead to significant morbidity, mortality, and sudden death (178,179). Treatment is geared toward reducing pressure within the ventricular system either by shunting or ventriculostomy. Adult-onset tethered cord should be considered in a deterioration of neurologic status, bowel or bladder changes, increasing orthopedic deformities, and gait deviations.

There are several age-related musculoskeletal and orthopedic complications. Spinal deformities, including scoliosis, kyphosis, and lordosis, can increase over time and cause back pain. Chronic lack of sensation and muscle imbalances can lead to Charcot joints. Overuse syndromes are common in wheelchair and crutch users. Wheelchair mobility tends to cause stress on the upper extremities, while community ambulators develop knee and hip pain. Carpal tunnel and rotator cuff disease are well documented in wheelchair users (180). Gait abnormalities from underlying muscle weakness can cause undue stress on joints in the lower extremities.

Neurogenic bowel and bladder function is an important component of adult medical care. It is a rare individual with spina bifida who has a completely normal urinary system. Despite these abnormalities, more than 80% of adults are able to develop social bladder continence (38). Methods to achieve this goal include all those previously discussed. In the past, renal damage leading to renal failure and death was a major contributor of morbidity and mortality in adults with spina bifida; although this is much improved, it still remains a problem (181,182). There is also an association between the presence of a neurogenic bladder and the development of bladder cancer (183). Recent data

suggest that this occurs at a young age in the population that develops bladder cancer, with variable pathology and has a poor prognosis (184). Neurogenic bowel function can change over time. Gastric motility seems to decrease with age and affects bowel programs. Treatment needs to be adjusted for these changes and includes different medication, dietary modifications, and newer surgical interventions.

The development of chronic skin problems is inherent in those with insensate skin. Aging causes changes in fat and muscle distribution, which can affect pressure ulcer formation. In the lower extremities, bracing can cause pressure and shear over bony prominences. Burns and abrasions can occur in unprotected skin. The wheelchair seated position results in pressure in the ischial and sacral areas. Prevention is imperative to avoid these secondary complications. The economic burden, along with psychological and functional impact, can be devastating (185,186).

Adults with latex allergies may have a higher rate than children for reactions, including anaphylaxis (187,188). This is probably related to repeated exposure to latex over the years, along with the increasing presence of latex in the environment.

Obesity is a health-related problem for both able-bodied and disabled adults. Nutritional studies indicate a decreased caloric expenditure with the disabled adult. Metabolic syndrome is more common in those with obesity and places these individuals at risk for coronary artery disease, diabetes, and hypertension. Interventions include nutritional counseling and healthy eating, exercise and fitness, and weight reduction Interestingly, most researchers focus on obesity, although eating disorders also occur in the disabled population (186).

Sexuality and sexual function is often overlooked in the disabled population. It is a huge disservice not to address these issues. Current data shows that the majority of males and females with spina bifida have a desire for intimate relationships, including sexual contact (189). Recent data suggest 24% of adults have an active sex life and gender, and continence did not factor into this statistic (190). Men with spina bifida report ability to achieve erections in 72%, and 67% experience ejaculation, but only one-third are happy with the amount of rigidity (191,192). Sildenafil (Viagra) may improve erectile function in 80% of men (193). Fertility is impaired, as only 14% of men report fathering children, and neurologic level is an important factor (192). Women with spina bifida generally have normal menstruation, and 88% have adequate vaginal secretions during intercourse (191). Women with spina bifida are able to conceive and have children (186). Sexual counseling should inform individuals about risk for pregnancy, sexually transmitted diseases, and contraception advice. Risk and benefits of Gardasil human papillomavirus vaccine should also be provided prior to sexual contact.

Vocational Counseling

Vocational counseling is an important aspect of transitional care of the individual with spina bifida, and current information in this area is limited. Recent data from the Netherlands reports a work rate of 62%, although 22% were in a sheltered environment. The definition of employment was based on at least one hour of paid wages per week. The best predictor of employment was level of education. This, along with gender and ability to care for self, were important predicators of full-time employment (194).

Functional vocational planning should be started early in secondary school, assessing career interests, skills, and aptitude. The potential for success in a postsecondary school program should be explored along with vocational job training. A positive realistic approach may provide the best solution in planning for adult employment options.

CONCLUSION

The successful treatment of spina bifida requires a multidisciplinary team approach. Education of the child and family regarding lifelong expectations are a critical part of multidisciplinary management. Knowledge of all the different systems involved—including genetic propensity to latex and fruit allergy, neurological, urological, gastrointestinal, orthopedic, endocrinological, skin, psychosocial, and rehabilitation issues—are essential for comprehensive care. Daily range of motion programs to avoid joint contractures, daily bowel and bladder programs to maintain bowel and bladder health and continence, and independent mobility will promote emotional and social well-being and aid towards educational and vocational advancement.

The Spina Bifida Association of America (SBAA) can be contacted at 4590 MacArthur Boulevard NW, Suite 250, Washington, D.C. 20007–4226, by phone on 202–944-3285, or on the Web at www.spinabifidaassociation.org.

The SBAA has a one-year college scholarship program, established in 1988, to assist persons with spina bifida in pursuing higher education.

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10

Traumatic Brain Injury

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EPIDEMIOLOGY

Traumatic brain injury (TBI) is a major cause of death and disability in children. It is the leading cause of death in those over 1 year of age. In 2004, the Centers for Disease Control (CDC) reported that TBI resulted in 216,000 emergency department visits, 18,000 hospitalizations, and 1,035 deaths in the 0–4 age group and 18,800 emergency department visits, 24,000 hospitalizations, and 1,250 deaths for those between the ages of 5 and 14 years of age (1). The incidence of pediatric TBI peaks at two separate periods: below age 5 and in mid- to late adolescence. The incidence of hospitalization for TBI has been reported to be 125 per 100,000 children per year in the 15–17-year age group (2,3). Males are more likely to sustain TBI than females, at a ratio of approximately 60% to 40% (4).

Children with a history of attention-deficit hyperactivity disorder (ADHD) are at a greater risk to sustain TBI than those without it. ADHD affects approximately 6% of children, has a male predominance, and a hereditary tendency. Of children who sustain TBI, prevalence of preinjury ADHD is noted to be between 10% and 20% (5).

Some authors have also evaluated the incidence of TBI in the United States by race. Langlois et al. (6) evaluated information from the National Center for Health Statistics. They reported a significantly higher rate of both hospitalization and death due to traffic/motor vehicle-related causes in children ages 0–9 in blacks compared to whites. Another group reported their experience in a regional trauma center and concurred that traffic/motor vehicle-related accidents were more frequently seen in minority children; however, there was no difference in death rates or the severity of brain injury (7).

Costs of Injury

The costs associated with pediatric TBI are significant. In a study of hospital resource utilization for pediatric TBI in the year 2000, Schneier et al. (3) reported that more than $1 billion in hospital charges was generated for TBI patients <17 years of age. A survey study of needs after hospitalization reported that at 3 months after injury, 62% of children hospitalized for at least one night after TBI received at least one outpatient health care service during the interval since injury and 26% had unmet needs. At 12 months, 31% were reported to have unmet needs (8). The cost of TBI to families is something that is difficult to quantitate. However, Hawley et al. (9) published a report concerning parental stress after TBI in children and adolescents. The Parenting Stress Index and General Health Questionnaire results of parents of children with hospitalization of greater than 24 hours for TBI were compared to the same measures administered to a control group of parents that was identified by

the subject parents. 44.3% of families reported loss of income due to the TBI. For those with a child with a severe TBI, it was 69%. Also, parents of children with TBI were found to have significantly greater stress and poorer psychological health than the comparison parents. Parents of children with TBI were noted to have clinically significant levels of stress in 41% of the cases (9).

Causes of Injury

The cause of injury differs by age. Nonaccidental trauma is responsible for 17% of brain injuries in infants and 5% in those aged 1–4. It causes a disproportionate percentage of severe TBI, resulting in 56% and 90% of severe injury in these two age groups (10). Motor vehicle–related injuries are more common in adolescents than young children, accounting for 66% and 20% of TBIs in the respective age ranges (2). Falls cause 39% of TBI in those under age 14, being especially common in those under age 5 (4). Falls are the leading cause of injury in children under age 4 (1).

Association With Other Injuries

It is common for TBI to occur in association with other injuries. Children with more severe injury are more likely to have been injured in a traffic-related accident and to have associated injuries (8). It has been reported that about 50% of children with TBI have other injuries as well (11). The presence of chest and abdominal injuries has been associated with decreased survival (12,13). In one study, undetected fractures during the acute care stay were found in 16 of 60 children with TBI, some having more than one fracture (14).

PATHOPHYSIOLOGY

Primary Injury and Secondary Injury

It is likely that the mechanism and consequences of TBI in children differ from those in adults for both primary and secondary injury. Children have a relatively large head and weak neck musculature, higher brain water content, and lack of myelination (15). Primary injuries related to impact and deceleration and rotational forces can be influenced by these factors. It has been suggested that forces could be more easily transmitted to deeper brain structures as a result of lack of myelination and higher brain water content (15). Primary injury related to mechanical forces includes contusions on the surface of the brain, where the brain can impact against the inner surfaces of the skull (usually focal gray matter injury) and the shearing-type injury that is associated with deceleration and rotational forces (usually diffuse white matter injury or at gray-white interfaces). Primary injury results from mechanical disruption of membranes and axons (16,17).

Secondary injuries occur due to complications or other events after the initial trauma. Potential causes of secondary injury include hypotension, hypoxia, vasospasm, infarction, prolonged seizure activity, and diffuse edema, resulting in increased intracranial pressure and a decrease in cerebral perfusion pressure (16,18). Early management of TBI has a goal of preventing secondary injury. Unfortunately, there are no guidelines concerning cerebral perfusion pressure and intracranial pressure targets for children with TBI. Values are thought to be age-dependent (19).

Contributing to both primary and secondary injury in TBI are cascades of biochemical events. Injury evolves as the cascade is initiated and progresses. Mechanisms initiating these cascades include cellular power failure, acidosis, overstimulation of excitatory neurotransmitter receptors, lipid membrane peroxidation, increase in intracellular calcium, and cellular damage by free radicals (2,16). With increasing knowledge about the biochemical processes involved, researchers are attempting to identify biomarkers in serum and cerebrospinal fluid (CSF) that will assist in diagnosis and prognostication regarding outcome of TBI (19–22). Likewise, additional information is being sought utilizing magnetic resonance (MR) spectroscopy. Babikian et al. (23) found that N-acetyl aspartate (NAA) on MRS scans acquired 2–10 days after TBI correlated moderately to strongly with cognitive testing at 1–4 years post-injury. Also, mean NAA/creatine ratio explained more than 40% of the variance in cognitive scores. They hypothesize that these values might be of assistance in predicting long-term outcome soon after injury when length of unconsciousness is not as yet known.

Diffuse Swelling and Second Impact Syndrome

It is more common for children to experience diffuse cerebral swelling than adults (19,24,25). This could be due to increased diffusion of excitotoxic neurotransmitters through the immature brain, an increased inflammatory response in the developing brain, or increased blood–brain barrier permeability after injury in the immature brain (19). When a lucid interval is noted in children prior to deterioration in neurological functioning post-TBI, it is likely due to the development of cerebral edema, in contrast to this phenomenon in adults being most commonly related to a focal mass lesion (2,25). This diffuse cerebral swelling is associated with a poor outcome (26). Children may experience impaired cerebral autoregulation after severe TBI (27,28). Cerebral blood flow varies with age, being

approximately 24 cm/s in healthy newborns, 97 cm/s in children aged 6 to 9 years, and then decreasing to the adult value of approximately 50 cm/s (27). Some studies have suggested that children with TBI have a lower middle cerebral artery flow rate and therefore hypoperfusion is common (27).

Another phenomenon associated with cerebral swelling is called second impact syndrome, and is said to occur after repeated concussion in children and adolescents. Brain swelling can be severe, even fatal, and develops after seemingly minor head trauma in an athlete who is still symptomatic (though at times subclinically) from a previous concussion (29). Second impact syndrome is a theoretical condition with only a few case reports available. The theory describes an initial injury (the first concussion), which deranges the brain's autoregulatory and metabolic systems enough to produce vascular engorgement and poor brain compliance. This allows marked changes in intracranial pressure with small changes in intracranial volume (29). Second impact syndrome presumes that the brain cells are in a vulnerable state after the initial concussion. Minor changes in cerebral blood flow during the second concussion result in an increase in intracranial pressure and ultimately apnea due to herniation, cerebral ischemia, and brain death (30,31). Also, there have been reports of diffuse cerebral swelling after mild TBI in sports, usually occurring in male adolescents (32).

Nonaccidental Trauma

Nonaccidental TBI is a special subset of TBI in children. It has been described as having a clinical triad of subdural hemorrhage, retinal hemorrhage, and encephalopathy, and is commonly associated with the history given, being incompatible with the severity of the injuries and the injuries being unwitnessed and inflicted by a solitary care provider (33). Classically, this so-called shaken baby syndrome has been described as being due to shaking alone causing tearing of bridging veins and rotational forces causing diffuse brain injury. More recent studies have indicated that there is most likely an impact in addition to the shaking episode(s). Often, nonaccidental brain injury in young children is also accompanied by a delay in seeking medical attention, potentially resulting in a hypoxic component to the mechanism of injury (18,21,33).

Implications of Plasticity

One must consider the effect of normal developmental activities of the immature brain on the mechanisms of developing damage after TBI. Apoptotic death of neurons is a part of plasticity and normal brain development. Does this result in the developing brain being more susceptible to activating the apoptotic cascade than the adult brain (15,19,34)? If so, this could help to explain the poorer prognosis for functional outcome for those injured at a very young age (34). In one animal study of posttrauma apoptosis, for a specific developmental age, the areas that had the highest density of programmed cell death were also noted to have high numbers of apoptotic cells in general (15). It may also be possible that excitatory neurotransmitter release could result in excessive stimulation of some pathways and stimulate the development of abnormal connections or that decreased excitatory activity could decrease connections (34). This implies that the relatively high plasticity of the developing brain could actually have a negative impact on the overall outcome after diffuse TBI and be at least partially responsible for the poorer outcomes seen in those injured at a very young age.

Growing Skull Fracture

A rare complication of skull fracture in children is a growing skull fracture. It is reported to occur when a linear skull fracture in a child under age 3 is accompanied by a dural tear and a leptomeningeal cyst develops. Fluid pulsations result in bone erosion and a palpable skull defect that requires surgical repair (35–37). A series of eight children with growing skull fractures had MRI evidence of a zone of signal intensity similar to brain contusion or CSF through the margins of the fracture, leading to their conclusion that MRI can be useful in diagnosing growing skull fracture early after injury (37).

NEUROIMAGING

Computerized tomography (CT) scans are typically obtained early after significant TBI. This relatively rapid imaging study is helpful in evaluating whether there is a condition that requires prompt neurosurgical evaluation and intervention (38–41). Specifically, it is helpful in detecting extra-axial hemorrhage, fractures, acute hydrocephalus, or parenchymal hemorrhages that are relatively large. However, the presence of a skull fracture is not indicative of intracranial pathology (39).

Magnetic resonance imaging (MRI) is more sensitive for the detection of intraparenchymal lesions than CT scan, but takes longer than CT and often cannot be done early post-injury due to the child's medical instability and need for supportive interventions. It is advisable, however, to obtain MRI when the child's condition allows it. Different MRI techniques can be used to evaluate for specific abnormalities (41). For example, susceptibility-weighted imaging was shown to identify a greater number of lesions than other

techniques in one study comparing outcome from pediatric TBI and imaging findings. In this study, children were grouped according to normal, mild impairment, and poor outcome and different imaging modalities were compared. CT did not demonstrate a difference between groups for lesion count or volume. Susceptibility-weighted, fluid-attenuated inversion recovery (FLAIR), and T2-weighted MRI all demonstrated significant difference between the normal versus mild impairment and mild versus poor outcome groups for both volume and number of lesions. They also reported that normal CT scans were seen in 40% of the poor outcome group (38). Others have also reported association between the volume of lesion and severity of injury (42).

Other authors have compared neuropsychological outcomes and imaging findings longer term after injury. One study of 14 children aged 10 to 18 years 6 to 12 months after mild to moderate TBI and a matched comparison group used diffusion tensor imaging (DTI) to evaluate white matter. Authors reported that the groups had no difference in overall intelligence, but did demonstrate differences in processing speed, working memory, executive function, and behavioral problems. Also, the TBI group had lower fractional anisotropy (FA) in three white matter regions: inferior frontal, superior frontal, and supracallosal. FA in the frontal and supracallosal regions correlated with executive function. Supracallosal FA also correlated with motor speed and behavior problems (43). Another group reported DTI findings in an acutely injured child with normal CT imaging. DTI demonstrated temporary marked increase in anisotropy in large areas of the cortical and subcortical right hemisphere at 18 hours after injury. At 135 hours post-injury, subtle changes in anisotropy were present (44).

Late after injury, several different imaging findings can be used to assess global change in the brain. These include cerebral diffusivity, corpus callosum volume, and volumes of brain and ventricles. Increased diffusivity is thought to be related to an increase in the extracellular space. In young children who experience TBI, late cerebral atrophy or decreased total brain volume could be related to tissue loss due to the injury itself or impaired brain growth. In typically developing individuals, white matter is reported to increase by 12.4% from age 4–22 (17). One study of children and adolescents at least 6 years after TBI found a correlation between corpus callosum volume and processing speed and visuospatial abilities. Ventricular volume did not correlate as well with results of neuropsychological testing. Corpus callosum is reported to continue to increase in size in typically developing individuals into early adulthood (45). It is imperative to evaluate scans over time to see the full extent of damage (40).

ELECTROENCEPHALOGRAPHY

Electroencephalograms (EEG) are commonly obtained for children who have sustained TBI. In the practice parameter developed by the American Academy of Neurology concerning antiepileptic drug (AED) prophylaxis in severe TBI, the authors note that in their review of studies, they did not find sufficient data to be able to make a recommendation concerning the use of EEG (46). In one report of 22 children between the ages of 1 week to 14 years at the time of TBI, the degree of EEG abnormality (mild, moderate, or severe) combined with admission Glasgow Coma Scale (GCS) were predictive of outcome in the young children. This was not the case for older children. Degree of EEG abnormality was statistically significantly correlated with full-scale intelligence quotient (IQ), attention and executive function, and memory (47). Additional evaluation of the usefulness of EEG in predicting outcome is needed.

INJURY SEVERITY

The main tools used for classification of brain injury severity are the GCS, length of posttraumatic amnesia (PTA), and duration of unconsciousness. Each has its merits and drawbacks.

Glasgow Coma Scale

The GCS has found wide clinical application since it was first published in 1974 (48). It rates a person's verbal, motor, and eye-opening responses on a scale of 3 to 15. It has the advantages of being simple, having a relatively high degree of interobserver reliability, and the ability to be determined shortly after injury (49). A score of 8 or less is considered to be coma and classified as severe injury, 9–11 as moderate injury, and 12–15 as mild injury. There have been studies that indicate that a GCS score of 5 or lower instead of 8 or lower should be considered as severe injury in children, as scores lower than 5 have been associated with a good outcome (12,50–52). Although the GCS was initially formulated to aid in acute triage and in neurosurgical management, many studies have correlated outcome with initial scores. There is, however, wide patient-to-patient variability. Some have noted that the GCS in the field is more predictive of survival (13,53), and GCS later in the post-injury course (particularly the motor component at 72 hours after injury) is a better predictor of disability (13,53). Adaptations of the GCS have been made to facilitate evaluation of children (54,55). Other refinements of the scale include the number of days until a patient returns to a GCS of 6 or 15.

Posttraumatic Amnesia and Children's Orientation and Amnesia Test

The duration of PTA is another commonly used indicator of injury severity. There is general agreement that the duration of PTA is directly correlated with the severity of injury (56–58). Compared with GCS, PTA has the merit of a longer period of observation. However, there is no generally accepted and easily applied method for determining the duration of PTA, especially in children. Assessments must be adapted, as appropriate, according to an individual's age (58). The Children's Orientation and Amnesia Test (COAT) has been helpful in evaluating length of PTA. It was designed to assess cognition serially during the early stage of recovery from TBI in children. The COAT is composed of 16 items evaluating general orientation, temporal orientation, and memory. The duration of posttraumatic amnesia is indicated by the number of days COAT scores are in the impaired range (59). Although this test should be useful in prospective outcome studies of children without profound injury, it has a major disadvantage because it takes 5 to 10 minutes to administer and, therefore, has not become a routine assessment on most clinical services. It has also been shown to be sensitive to nontraumatic impairment. For example, children receiving special education services fall within the impaired range, and the COAT, therefore, should be interpreted with caution (60).

Duration of Unconsciousness

Duration of unconsciousness is another measure of severity and has the advantage of longer observation than GCS. It is also easier to recognize than the duration of amnesia in children and is more easily determined in retrospective chart reviews. Unconsciousness has been defined as the inability to respond to the environment in any adaptive, meaningful way. Children can have sleep–wake cycles and still be considered unconscious (61). This is the most appropriate measure in series of more severely injured children who are unconscious for many weeks, many of whom never regain recent memory. A study conducted by Massagli and colleagues (53) concluded that there was a strong correlation between length of time to reach GCS of 15 and early and late outcomes.

Although most outcome studies have correlated outcome with only one index of brain injury severity (62,63), McDonald and colleagues (57) compared 10 measures. In their report, the number of days to reach age-adjusted 75% performance on the COAT, the number of days to GCS 15, and the initial GCS scores were most predictive of outcome across all neurobehavioral and functional measures when measured early and at 1 year post injury. The intercorrelations of these brain injury indexes were also quite high. In general, these indexes could be used interchangeably and a single measure of severity predicted most outcomes almost as well as multiple measures. Severity ratings as determined by these alternative criteria are summarized in Table 10.1.

In summary, it is important to use these tools and correlate them with clinical findings to make an assessment of severity of injury and therefore possible long-term outcome. Although useful, these assessment tools do have limitations in determining outcome, and a clinician's clinical impression is also important.

COMMON MOTOR DEFICITS

A wide spectrum of motor deficits is seen after TBI. This spectrum results from the variable nature of the injury and the combination of focal and diffuse damage.

Focal Damage

Isolated focal brain injuries can occur from a variety of causes, including brain tumor resections, gunshot wounds, and other foreign body penetrations.

10.1 Rating of Brain Injury Severity

	MILD	MODERATE	SEVERE	PROFOUND
Initial Glasgow Coma Scale	13–15 with no deterioration	9–12 with no deterioration	3–8	
Posttraumatic amnesia	<1 hour	1–24 hours	>24 hours	
Duration of unconsciousness	<15–30 minutes	15 min–24 hr	1–90 days	>90 days

The cognitive and motor deficits may vary because of differences in brain injury loci. Obviously, if there is a unilateral penetrating or focal injury involving the motor area, a hemiparesis may result. Depending on the precise location of the damage, hemiparesis may be more pronounced in the upper or lower extremity. The long-term outcomes in motor, cognitive, and behavioral functioning may be better in focal injuries versus diffuse injuries given the isolated nature of the brain damage (64).

Diffuse Damage

The diffuse nature of TBI has resulted in a constellation of motor impairments that is familiar to clinicians who work with these problems. These include difficulties with balance, coordination, and speed of response. Despite these impairments, however, a significant number of children achieve functional mobility. In a study by Boyer and Edwards (65), at 1 year after injury, 46% of their patients walked independently without assistive device and 27% walked with an orthosis or an assistive device. Overall, 79% had independent mobility.

Swaine and Sullivan (66) have examined early motor recovery after TBI in 16 adolescents and adults who had a GCS score of 8 or lower for at least 6 hours. Assessments included evaluation of muscle tone, range of motion, abnormal and voluntary movement, primitive reflexes, equilibrium and protective responses, and specific motor skills. There were differential patterns of recovery and differential rates of recovery among the subjects, which is to be expected considering the heterogeneous nature of TBI.

Chaplin and colleagues (67) evaluated motor performance in children after TBI. Fourteen children with TBI who were unconscious for 24 hours or longer were compared with 14 age- and sex-matched children. The Bruininks-Oseretsky Test of Motor Proficiency was administered at least 16 months after injury. Children with TBI scored significantly poorer on the Gross Motor Composite, including all subsets: running speed, balance, bilateral coordination, and strength. Also, they scored lower on the fine motor subsets for upper limb speed and dexterity. Most of these subtests involve timed tasks. Chaplin and colleagues also found a correlation between the Gross Motor Composite score and the time since injury. They concluded that this correlation supports continuing long-term improvement in skills after TBI.

Kuhtz-Buschbeck and colleagues (68) looked at gait, gross motor proficiency, and hand function in 23 children after a TBI, severe in 17 and moderate in 6, during their five months of inpatient stay. They were compared with age- and sex-matched healthy controls. Children with TBI showed marked reduction in gait velocity, stride length, cadence, and balance. Deficits in fine motor skills, speed, and coordination were noted on hand function tests. Hand function skills improved less than gait; degree of impairment was noted to increase with severity of injury. Younger age at injury was not associated with better recovery. It has also been noted that the absence of spasticity is a good predictor of ambulation recovery by discharge (69,70).

Others have also noted impaired fine motor skills after TBI. Again, the speed component of the assessment on these tasks may account for some of the impairments that were observed. Long-term impairment of finger tapping has been described (71). Practice of activities requiring fine motor coordination improves skills, even long after injury (72).

Balance

Balance is frequently found to be abnormal after TBI, as it involves effective integration of the sensory, motor-programming, and musculoskeletal system (73). Cochlear and vestibular function may be impaired. True vertigo may be present. The clinical exam could be normal, despite children being symptomatic (73). Blocking visual input during quiet standing is a simple and sensitive test for postural instability (74). Gait analysis and vestibular testing may be necessary to evaluate subtle changes leading to imbalance (73). When postural instability is assessed quantitatively, long-term impairment of static and dynamic control of posture is often found after TBI (74,75). It may be related to latency of response and asymmetric stance (76). Treatment options include oral medications, visual therapy, vestibular balance rehabilitation therapy (VBRT), and surgery (77). Oral medications, including meclizine and scopolamine, should be used sparingly, as they could slow the natural compensatory process (77). Specific training with VBRT exercises that promote habituation and/or adaptation and/or substitution can be used (77).

Tremor

Another motor impairment that is seen is tremor, which frequently is more pronounced proximally and increases with effort and movement. Lesions have been noted in varying areas. Treatment with medications typically used for tremor may be of benefit (78,79) Andrew and colleagues (80) report stereotactic surgery to be effective in management of tremors.

Tone Abnormalities

Muscle tone abnormalities, including spasticity, dystonia, and rigidity, are common after TBI. The types

of problems noted vary, depending on the time since injury as well as the severity of injury. Cause of acquired brain injury also influences the type of problem that is most commonly noted. Spasticity has been noted in 38% and combined spasticity and ataxia in 39% of children and adolescents 1 year after injury (63). Rigidity or dystonia is especially common when there has been secondary injury due to hypoxia or ischemia (81).

Spasticity

Spasticity results from an upper motor neuron injury and is manifested by increased deep tendon reflexes and velocity-dependent resistance to movement (82,83). Several different scales are available to evaluate spasticity, but they are all subjective (77), and available quantitative tests are time-consuming (84).

Physical Management. It is important to begin treating spasticity in the acute care setting to prevent contracture development (85). Treatment approaches include range of motion, stretching, casting and splinting, medications, and surgical interventions used alone or in combination to manage spasticity. Range of motion itself may be helpful to reduce tone temporarily (86). Also, one may begin with positioning options, including but not limited to, splinting and weight bearing, if tolerated, as well as the use of neutral warmth, gentle shaking, and reflex inhibition (87). If a child has a tendency to assume a total extension posture, positioning in side-lying with hips flexed beyond 90 degrees and neck flexion may assist in interrupting the extension pattern. If active posturing is present, one must be careful in the use of splints and casts because constant pressure against the splint or cast may result in the development of an ischemic ulcer (87). Stretching should always be included in any treatment protocol for spasticity (77).

Pharmacologic Management. Medications for treatment of spasticity can be oral, intrathecal, or injectable. Enterally administered pharmacologic agents may be beneficial in decreasing abnormal muscle tone and posturing. Their potential side effects may limit their effectiveness in this population. This is especially true of the sedating effects of baclofen and benzodiazepines. Dantrolene sodium causes sedation, despite its action at the sarcolemma. Alpha-adrenergic agonists, such as clonidine and tizanidine, have also been reported to decrease tone (77). The effectiveness of all of these medications is variable.

Early after injury, when posturing may be a problem, chlorpromazine has been of assistance. It has the significant potential to cause sedation (82). Bromocriptine has also been effective in reducing posturing early post injury.

Injectable medications include botulinum toxin and phenol motor point blocks. They can be used in combination with positioning, splinting, and casting. Early after injury, with severe posturing and intolerance of splinting, botulinum toxin may be a helpful adjunct in attempting to maintain range of motion. It is reversible, so if there is significant motor recovery, there is no permanent effect of the injection. Functional gains have been noted with the use of botulinum toxin (88–91). Phenol blocks tend to be used later after injury when there is residual difficulty with increased tone. Phenol and botulinum toxin injections can be used concurrently to treat severe spasticity and to increase the number of muscles treated at one time. If severe deformity develops, surgical tendon or muscle lengthening may need to be considered (81).

Intrathecal baclofen (ITB) infusion using a programmable pump has been shown to be effective in the treatment of spasticity of cerebral origin, particularly cerebral palsy (92,93). Studies have also shown functional improvement in gait (94–96) with the use of ITB infusion in patients with acquired brain injury. Francisco (97) and colleagues also noted improvement in activities of daily living (ADLs) and decrease in pain. Two studies have shown caregiver and patient satisfaction in individuals treated with continuous infusion of ITB by an implanted programmable pump (98,99). ITB by an implanted programmable pump should be considered if severe systemic spasticity persists (100–102). Doses can be changed, depending on the patient's progress.

Dystonia

Dystonia is defined as a disorder in which involuntary sustained or intermittent muscle contractions cause twitching and repetitive movements, abnormal postures, or both (83). It has been reported as a rare motor impairment and is more commonly seen in those injured as children rather than as adults (103,104). Interval between injury and onset of dystonia varies. No consistent picture is seen on neuroimaging study. Medications such as trihexiphenidyl hydrochloride, carbidopa/levodopa, and bromocriptine are used in treating dystonia. ITB infusion has also been used effectively to treat dystonia (101,102).

Rigidity

Rigidity is the resistance to an externally imposed joint movement, with an immediate resistance to reversal of the direction of the movement, and the limb therefore does not tend to return to a particularly fixed posture (83). Management of rigidity is similar to the management of spasticity and dystonia; however, it is often more refractory to intervention.

COMMON SENSORY DEFICITS

Olfactory Dysfunction (Anosmia)

Olfactory dysfunction is a common consequence of TBI, most frequently associated with severe injury, and has also been seen with PTA of more than 5 minutes (105). Bakker and colleagues (106) report an association between severity of anosmia and executive function in children. The incidence of anosmia varies from 5% to 65%, depending on the type and severity of the brain injury (107). Olfactory dysfunction can be a partial loss of the sense of smell (microsomia) or a complete loss of sense of smell (anosmia) (108). In a study carried out by Yousem and colleagues (109) to locate and quantify the deficits using radiographic studies, most patients with impaired olfaction showed damage to the olfactory bulbs and tracts, followed by the inferior frontal lobes and volume loss in the olfactory bulbs and tracts. Both patients and their parents are seldom aware of their deficits (110) and therefore formal testing should be done in children with TBI. The three-screen test can be used for quick, gross identification, but the University of Pennsylvania Smell Identification Test (UPSIT) is more reliable in identifying all patients with deficits (111). There is usually poor recovery from anosmia in comparison to parosmia (107). Impairment in the sense of smell may have social and safety implications (108). Those with anosmia must be cautioned to use other senses to look for dangers, such as a gas burner left on, fire hazard, or similar problems. Teenagers and young adults may need to be advised about the use of fragrance when they cannot receive any feedback about its strength.

Hearing Impairment

Hearing impairments and impairments of vestibular function are also commonly noted. Hearing impairment may occur secondary to several causes: central processing deficit, peripheral nerve damage, cochlear injury, or disruption of the middle ear structures. Cognitive impairments that are common after TBI often interfere with the child recognizing this difficulty. It is important for clinicians to have a high index of suspicion in children and initiate screening for hearing impairment.

Vestibular impairments have already been mentioned in the discussion on balance. Vertigo secondary to vestibular impairment commonly resolves within six months of injury (112,113), but electronystagmogram abnormalities can persist for years (114).

Central auditory processing impairment occurs with damage to tracts or cortical tissue. In such individuals, pure tone audiometry is normal, but other studies, such as speech discrimination, or late wave forms of brainstem auditory evoked potentials are abnormal (115). Central auditory impairment is difficult for most families to understand. Their intuitive conclusion is that hearing is related to the ear, so they frequently anticipate that interventions such as a hearing aid may be helpful.

Hearing loss may be conductive in nature because of disruption of the ossicles or cerebrospinal fluid or blood in the middle ear. Both of these types of injuries are frequently associated with fractures of the temporal bone (116). Conductive hearing loss usually recovers spontaneously in about 3 weeks. If it persists for more than3 weeks (particularly for >30 db) a repeat audiogram and exploration of the middle ear is recommended (116). Problems related to fluid in the middle ear usually resolve spontaneously.

Sensorineural hearing loss may also be seen, but less often than conductive hearing loss (116,117). Sensorineural hearing loss is commonly noted at higher frequencies (117) and is associated with inner ear pathology (112,116). Marked variation is seen in the recovery of sensorineural hearing loss (116).

There may be trauma to the eighth cranial nerve, or injury to the labyrinthine capsule, or labyrinthine concussion, which may result in hearing loss because of the transmission of high-energy vibrations and a pattern similar to the hearing loss after prolonged noise exposure (118). Injuries to the labyrinthine capsule and the eighth cranial nerve are frequently associated with basilar skull fracture.

Visual Impairment

Because of the complexity of the visual system, a variety of visual impairments can be seen. Impairments may result from injury to cranial nerves, eyes, optic chiasm, tracts, radiations, or cortical structures (119,120). Early after injury, a child may appear to be functionally blind. Although vision is often assessed by looking at response to visual threat and visual tracking, these responses do not differentiate between peripheral and central impairments. One must assess cranial nerve function to make that differentiation.

Visual acuity reduction is the most frequently detected deficit in children, but the severity varies and is associated with severity of injury (119). Visual acuity reduction is commonly associated with frontal lobe injuries (119,120). In children with greater visual acuity impairment, optic nerve atrophy, either complete or partial, is present (119). Usually, optic atrophy is seen within 1 month after injury (120), and is correlated with the site of impact and not necessarily with the overall severity of the brain injury. Chiasmatic injury results in bitemporal visual field impairment of

varying degree and is found in 0.3% of TBI cases. It may be identified on MR imaging (121).

Homonymous hemianopsia is seen with injuries to the optic tracts and is often associated with hemorrhage and hemiparesis. Prism lenses may be of assistance, as well as learning compensatory techniques to increase scanning of the full environment (122). The presence of visual field impairments may be associated with more severe neuropsychological impairments (123).

Central visual dysfunction may be described as visual processing or visual perceptual problems. Cortical injury is responsible for this type of impairment and may not be confined to the occipital lobes. For example, involvement of temporal lobes may produce visual memory impairment, and involvement of parietal lobes may produce impairment of spatial awareness (124).

Injury of the third, fourth, and sixth cranial nerves may lead to a variety of visual problems (125). Diplopia may result from extraocular muscle imbalance most commonly due to trochlear palsy (125) and may be present at all times or just in particular gazes. Patching is commonly used to eliminate diplopia but results in monocular vision and related disadvantages (126). In children under 11 years old, it is important to patch eyes in an alternating manner to avoid difficulty with amblyopia. Visual motor impairments due to unilateral abducens nerve palsy in children usually resolve spontaneously within six months (127). Deficits that persist longer than six months are more likely to be associated with bilateral or complete abducens nerve palsy and are unlikely to resolve spontaneously (127).

Difficulties with convergence may also result in diplopia, and are believed to be due to supranuclear impairment. Anatomic correlates of diplopia have not been well described (125). Accommodation may also be impaired (128).

COMMON COGNITIVE DEFICITS

Although TBI can result in both motor and cognitive impairments, it is generally the cognitive impairments that most profoundly affect the individual's ability to function. As noted previously, the full extent of the child's cognitive impairment may not be known until a significant time after injury, as deficits may not become apparent until the child is at a developmental stage when one would anticipate that they would have a particular cognitive ability, such as abstract thinking or metacognition. In general, when children have been followed long-term after injury, those who were injured at a young age typically show more cognitive impairment than those injured later in childhood (129).

Attention and Arousal

Arousal is a precursor for attention. It has been defined as "the general state of readiness of an individual to process sensory information and/or organize a response" (130). Although there have not been systematic studies of pharmacologic interventions to improve arousal in children with TBI, a number of medications have been used and reported in case studies. One retrospective report of amantadine in children with TBI noted that compared to a group of children who had not been started on any neurostimulant medication, those on amantadine had a greater increase in their Ranchos Los Amigos level during hospitalization. The amantadine group had lower initial Ranchos scores and GCS (131). Dopaminergic medication use has also been reported, again in a retrospective review. In this report, the children's Western Neuro Sensory Stimulation Profile scores pre- and during medication were compared. Also, the rate of change in these scores before and after medication were compared. Significant differences were noted, suggesting that the medication could be contributing to the accelerated rate of improvement (132).

As noted previously, children with a prior history of ADHD are at an increased risk to sustain TBI. Likewise, attentional problems are common after TBI, affecting an additional approximately 20% (5). Severity of TBI is reported to be associated with the likelihood of developing attentional problems (133,134). The attentional problems seen after TBI in children are not the same as seen in developmental ADHD. It has been reported that skills that develop earlier in childhood are relatively spared compared to those that develop later. Therefore, sustained attention and divided attention are more significantly impaired than focused attention (135). Also, children with TBI tend to have slower response speeds than children with developmental ADHD (136). Both behavioral interventions and medications have been used as treatment for children with attentional problems after TBI. Case reports have noted improvements (137,138).

Memory Impairment

Memory impairment is another common area of concern after pediatric TBI. Typically, the memory impairment that is seen is for the formation of new memories as opposed to long-term memory. This has significant implications for a child's ability to learn new information. As observed in other areas, severity of memory impairment appears to be related to the overall severity of injury. Impairment is seen in both immediate and delayed recall in severe TBI (139–142). When

evaluating preschool children who had experienced TBI, Anderson et al. (141) found that over time, children did show developmental progress in their memory skills; however, children with more severe TBI did less well over time. They saw this trend as well for both the learning and memory measures that they evaluated. It has been reported that verbal memory is more impaired than visual memory after TBI in children and that unstructured retrieval is the most impaired aspect of memory (143). Memory impairment is a challenging deficit to attempt to address during rehabilitation. Different approaches include trying to improve recall through memory practice, using organizational strategies or mnemonics, using teaching techniques to make learning more efficient (including backward chaining), or making use of compensatory techniques such as a memory notebook or electronic device (142,144,145). Avoiding purely verbal teaching, making use of structured activities in teaching, and increased repetition have been advocated as well (143).

Behavioral Problems

Behavioral sequelae are also common after TBI in children. These can include impulsivity, personality changes, depression, anxiety, becoming easily frustrated, aggression, and sleep problems (146). These problems persist long-term and are reported in 10% to 50% of children with TBI (147,148). Some authors report an increase of emotional and behavioral symptoms over time (148). Also, a number of authors note that those who sustain TBI are more likely to have a preinjury history of behavioral or psychiatric concerns (147,149). Behavioral problems can be significantly disabling even in the absence of significant mobility or activities of daily living impairment (147). Behavioral problems appear to be more significant and more common in those injured at a younger age (149). Approaches to address behavioral concerns include providing structured environments and daily routines but allowing the individual to make choices when possible, as well as assisting in breaking down tasks to their component parts, providing cues or aids for organization, creating situations in which the individual will be successful, and helping the individual to communicate the need to escape a task or situation. Positive reinforcement of desired behaviors has also been used. Involvement of family members in the process is important (149,150). Various medication interventions have been tried in the past, but none has been shown to be ultimately superior to others in addressing this variety of behavioral symptoms. It is imperative that those working with the individual understand that the behavioral problems are neurologically based. Behavioral symptoms are strong predictors of family burden over time (148).

Communication Deficits

A variety of communication impairments can be seen after TBI in children. If there is focal injury in areas of the brain that control language, aphasia can be seen. Also, motoric impairment can contribute to dysarthria. In general, the communication impairments that are seen more commonly are due to other cognitive deficits, such as memory impairment and executive function concerns (149). Difficulties with response speed can contribute to a reduced rate of speech and, conversely, impulse control difficulties can result in a rapid rate of speech (151). Word finding and verbal learning deficits are common, potentially relating to memory impairment (149,152). Discourse, abstract language, and social interaction with language are all commonly impaired (149,153). Also, verbal working memory, which is commonly impaired, is important in acquiring language, reading, and arithmetic in children (154). Authors report that ability to use language functionally is typically more impaired than one would expect from reported results of standardized intelligence testing (155).

Executive Function

The area of executive function is one that is commonly affected, even in children who have experienced a mild TBI. It also is one in which the full effect of the injury may not be manifest until the child has matured to the point when one would expect him or her to demonstrate these particular skills. Executive function is defined as the ability to manage and direct more modular cognitive abilities in order to set, manage, and attain goals (5). This includes problem solving, organization, self-monitoring and self-regulation, self-appraisal, and self-management. It has been suggested that children are particularly susceptible to impairment in executive function if injured, as they are experiencing rapid development in this area (5). Impairments of executive function are noted to be more severe in children injured at a young age (156).

Working memory is one of the first executive function areas to develop, emerging between 7 and 12 months of age. It involves being able to temporarily store some information while concurrently processing and retrieving other data (157). It has been shown to be impaired after TBI, and the degree of impairment relates to the severity of injury (5,154,157). Other areas commonly affected by TBI include the ability to inhibit, shifting set, planning, self-monitoring and control, decision making, social cognition, and behavioral self-regulation. The Behavior Rating Inventory of Executive Function (BRIEF) is a tool that uses parent and teacher ratings to evaluate the impact of executive dysfunction on everyday life (5). Interventions

for executive dysfunction have not been rigorously studied. Some have suggested using an approach that breaks tasks into problem-solving steps. Also, the provision of a structured environment and expectations is important. Incentives for progress toward a goal can be helpful. It is imperative that parents develop an effective working relationship with their child's school program providers to have open communication around the issues of executive dysfunction and its impact on school programming (5,156,158).

Social Functioning

It is not possible to totally separate social functioning from executive function; however, separate comment on this important area will be undertaken here. A child's ability to effectively function within his or her social milieu is often significantly affected by TBI. Emotional lability is common (159). Often, children have difficulty interpreting social cues from others or recognizing the emotions being expressed (160,161). Janusz et al. (159) reported on social problem-solving skills in children with TBI. They found that although the children were able to articulate the social dilemmas, they chose less developmentally mature strategies as the best means to solve them and also used low-level reasoning to evaluate whether the strategies were effective. Social participation is also reported to be decreased in children with TBI compared to their typically developing peers. Bedell and Dumas (162) reported that 30% to 73% of the children with acquired brain injuries that they studied were restricted in at least one of the participation domains they evaluated. Family-reported institutional, social, and attitudinal barriers were more often contributing to this restriction than physical environmental barriers (162).

MEDICAL CONDITIONS ASSOCIATED WITH TBI

Medical conditions associated with TBI can vary greatly from individual to individual. Essentially, all organ systems can be affected when a child sustains a TBI.

Neuroendocrine Dysfunction

Head trauma places the pituitary gland at risk for injury due to its encasement in the sella turcica, its delicate infundibular structures, and its tenuous vascular supply. The gland may be subject to edema, ischemia, transection of the pituitary stalk, or watershed injury (163). Dysfunction of the hypothalamic pituitary axis can be categorized as either involving the anterior or the posterior pituitary. Posterior pituitary dysfunction results in syndromes including diabetes insipidus (DI) and the syndrome of inappropriate antidiuretic hormone secretion (SIADH).

DI is commonly noted early after a moderate or severe TBI and can, therefore, be considered a potential marker for global hypothalamo-pituitary injury and dysfunction (164). SIADH also is a result of posterior pituitary dysfunction and needs to be distinguished from DI. The incidence of DI in children is poorly understood and poorly researched. One study (165) demonstrated incidence around 21.6% of DI in adults with moderate or severe brain injury. The study also found DI tended to be associated with a lower GCS and with the presence of cerebral edema. The fluid and sodium imbalance of DI results in a deficiency of antidiuretic hormone and excessive water loss. As antidiuretic hormone is produced in the hypothalamus, those patients who exhibit DI are felt to be predisposed to other hypothalamo-pituitary system dysfunction. Patients with DI are hypernatremic and demonstrate polyuria and polydipsia. Although often DI is only a temporary problem for most people with TBI, it may persist. Treatment for DI is desmopressin acetate (DDAVP), which is a synthetic form of an antidiuretic hormone (166).

The syndrome of SIADH is another common fluid and electrolyte imbalance encountered in those with TBI, and needs to be distinguished from DI in order to provide appropriate treatment. In contrast to DI, these individuals exhibit decreased urine output, hyponatremia, and decreased serum osmolarity. SIADH is typically managed with fluid restriction and carefully reestablishing the serum sodium to a normal level in a cautious fashion. Rapid correction of the hyponatremia can cause pontine myelinolysis and possibly death (166).

Cerebral Salt Wasting

Cerebral salt wasting is a third cause of serum sodium imbalance in individuals with TBI. Like SIADH, cerebral salt wasting results in hyponatremia. It is essential that cerebral salt wasting be distinguished from SIADH. Unlike DI and SIADH, cerebral salt wasting does not involve the hypothalamo-pituitary system, but is believed to occur due to direct neural effects on renal tubular function. The low sodium levels seen are a direct result of abnormal renal tubular function, resulting in lost sodium along with lost fluid volume. These patients are dehydrated and, therefore, fluid restriction would cause their condition to further decline. The treatment for cerebral salt wasting involves fluid and sodium replacement (167).

Anterior Pituitary Dysfunction

Literature suggests that approximately one-third to one-half of adults who have sustained a moderate

or severe TBI have some hypothalamo-pituitary dysfunction (164). Children with TBI are at risk for hypothalamo-hypophyseal dysfunction, with one study identifying a rate of about 60% (168). Another group of 48 pediatric patients were found to have a 10% incidence of hypothalamo-hypophyseal dysfunction 6 months after their brain injuries (169). The challenge in identifying which children to screen for anterior pituitary dysfunction is that many of the symptoms of anterior pituitary dysfunction mimic the effects of TBI. For instance, low levels of growth hormone are associated with symptoms such as fatigue, cognitive dysfunction, irritability, and DI (164). An individual who has sustained a TBI would commonly complain of these symptoms and have them dismissed as sequelae of the brain injury. In 2005, a consensus statement on screening for hypopituitarism after TBI recommended systematic screening for pituitary dysfunction for individuals with moderate to severe TBI who are at risk of developing pituitary dysfunction. They recommend screening for hypopituitarism if, while the patient was hospitalized, they had DI or hyponatremia and hypotension. If they had SIADH or hypothyroidism identified, screening for anterior pituitary dysfunction would also be indicated. Reasons noted for foregoing anterior pituitary function screening include the individual being in a persistent vegetative state at a very low level of consciousness. Since little is known about the incidence of hypothalamo-pituitary dysfunction in children after TBI, the majority of the recommendations are extrapolated from adult literature. The treating physician should be knowledgeable, however, of the presenting features of hypothalamo-pituitary dysfunction in children, which may include growth failure, arrested or delayed puberty, amenorrhea, decreased libido, and short stature (170).

Precocious Puberty

Precocious puberty is defined as the onset of puberty in girls before the age of 8 years and in boys before the age of 10 years. It can occur following TBI in children, with signs developing from 2–17 months after the initial injury. There is a positive correlation between increased ventricular size secondary to cerebral atrophy and the development of precocious puberty, and girls are affected much more frequently than boys (54.5% in girls to 4.5% in boys) (171). The signs of precocious puberty include onset of secondary sexual development prior to the predicted age and accelerated linear growth. These children demonstrate advanced bone age and premature closure of the epiphyseal plates. Because precocious puberty places a social and emotional burden on the patient and family, and because of the development of short stature secondary to premature epiphyseal plate closure, it is essential that the physician have a watchful eye for precocious puberty and be prepared to evaluate for it and treat it if indicated.

Respiratory Dysfunction

Recommendations for the treatment of children with TBI include transitioning from endotracheal intubation to tracheostomy for ventilatory support around the time the patient is 7 to 10 days post-injury. The tracheostomy allows for pulmonary support, easier secretion clearance, and better long-term airway management. The tracheostomy is not without complications, though, including, the potential for vocal cord paralysis, tracheal stenosis, subglottic and glottic stenosis, and tracheomalacia (172). The ultimate goal is to move toward decanulation once controlled ventilation is no longer needed and when the patient is able to manage his own secretions. Another reason to move toward decanulation is to avoid the increased nursing and respiratory care requirements when the tracheostomy tube is in place. These increased needs can complicate discharge, as some long-term care facilities are unwilling to provide care for patients with tracheostomies and family members may be anxious and apprehensive about caring for a child who has one (173). The stepwise fashion moving toward decanulation has been described by Klingbeil (174). The process begins with downsizing the tracheostomy tube sequentially until, ultimately, an uncuffed small tube is tolerated. Then capping of the tracheostomy tube is recommended as the clinician evaluates the patient's tolerance. If the patient is able to maintain oxygen saturations with a comfortable breathing effort and demonstrate effective cough with good management of secretions, the tube is removed and an occlusive dressing is placed to allow the site to heal. If the patient is demonstrating difficulty during the process of decanulation with worsening respiratory function or distress, it is recommended that the patient undergo direct laryngoscopy prior to decanulation in order to evaluate for concerns such as tracheal granuloma.

Nutritional Management

Very early after severe TBI, it is important for the primary team to place emphasis on the child's nutritional status. Guidelines have been established for achieving adequate nutritional management in this population (175). These guidelines are mostly from the adult TBI literature, as there is quite limited pediatric research regarding nutrition after TBI. Metabolism is reported to be increased after severe TBI in children, causing increased nutritional requirement. Phillips et al. (176) studied pediatric TBI survivors who had initial GCS between 3 and 8. Overall, the energy expenditure in

those patients was 130% of their expected metabolism. Phillips also found that weight loss ranged between 2 and 26 pounds during their two-week post-injury evaluation despite aggressive nutritional support. Moore et al. (177) identified metabolic profiles of pediatric TBI survivors who had initial GCS of less than 7. They found that the increased energy expenditure in that group averaged 180%. In adult literature, hypermetabolism in TBI survivors is well established. The guidelines for the adult population include the following recommendations: (a) Full nutritional replacement should be initiated by day 7 post injury; (b) enteral nutrition should be started no later than 72 hours post injury; and (c) tight control of serum glucose is necessary to avoid hyperglycemia, which is associated with worsening ischemic injury and worse outcome. Parenteral nutrition should be started if enteral nutrition is not full and complete by day 7.

Tube Feedings

Typically, enteral support of nutrition begins with nasal jejunal or nasal gastric feedings. Jejunal tube feedings are often tolerated better due to delayed gastric emptying (172), but the goal is to move to gastric feeds with boluses of nutritional formula for more typical meal feedings to decrease the complexity of equipment needs and to more approximate the typical physiology of enteral feedings. Percutaneous gastronomy (PEG) tubes are often placed at the time tracheostomies are placed, with the presumption that the patient who requires the tracheostomy will require tube feedings for longer than an acceptable time to leave a nasal tube in place. Nasal gastric and nasal jejunal tubes are associated with an increased risk of sinusitis, and the presence of the tubes in the posterior pharynx may be a source of irritation for the restless and the agitated child with TBI. If the child's cognitive status improves and they achieve full nutrition by mouth with no risk for aspiration, the PEG tube can be discontinued as early as two to six weeks after it was placed once the cutaneous-gastric fistula has matured (178). Janik et al. (179) found that gastrostomy tubes that remained in place in the pediatric patient for greater than 8 months required surgical closure of the fistula in 92% of the patients studied.

Gastroesophageal Reflux Disease

Prior to placing a gastrostomy tube, consideration should be given to the child's likelihood of having gastroesophageal reflux disease. This can be evaluated with an upper gastrointestinal radiologic study, a pH probe study, or a milk scan. Occasionally, a Nissen fundoplication will be done in conjunction with the placement of a gastrostomy tube to avoid reflux and the risk for aspiration and its associated morbidity. Children with gastrostomy tubes in place should usually be placed on an H2 receptor blocker or proton pump inhibitor to decrease risk for acid reflux as well as gastrointestinal bleeding (180).

Transition to Oral Feedings

Evaluation of the child with TBI at bedside by speech and language pathologists and/or occupational therapists is usually the first step in determining whether to begin transitioning to oral feeding. Studies show that dysphagia, oral motor impairment, and cognitive impairment are all highly correlated in the child with TBI; therefore, evaluation at the bedside of oral motor control as well as cognitive impairment helps to determine the degree to which dysphagia is present (181). The incidence of dysphagia in this population varies by severity of the brain injury. Overall incidence is reported to be 5.3%. Children with mild brain injuries have an incidence of dysphagia of 1%, moderate brain injuries demonstrating a 15% incidence, and severe brain injuries 68% (182). The strongest factor predicting whether dysphagia will be present in a pediatric patient is the GCS. If the GCS is less than 9 (representing a severe TBI), the child is more likely to have dysphagia. These children tend to exhibit both oral and pharyngeal deficits with reduced lingual control and a delayed swallow reflex in the majority (181). Most dysphagia in this population resolves about 12 weeks post injury and a normal diet is resumed. Once a child is evaluated by modified barium swallow and no silent aspiration is identified, the rehabilitation team can begin oral feeding in a stepwise fashion. Typically, the team will begin with tastes of pureed foods and thickened liquids, with progression over time, as tolerated, to solid foods and thin liquids.

Bowel Management

It is important to maintain regular bowl movements early on in the critical care course of a child with a TBI. A bowel management program may involve stool softeners, suppositories, and/or laxatives in order to cause regular and routine bowel movements. The patient's bowel management program needs to be adjusted depending on their clinical response. Narcotic medications are constipating and antibiotic medications can cause loose stools, so close follow-up and regular adjustments are indicated. Once the child is medically stable and the routine for their bowel routine is better established, the team may choose to cause bowel movements at the same time of day with the use of a suppository. In this way, "functional continence" may be obtained, with the child's bowel movements being more predictable. The agents commonly used include

Senna, docusate sodium, polyethylene glycol, or glycerin suppositories.

Bladder Management

During acute care of the child with severe TBI, it is common for a Foley catheter to be in place for measuring urine output. After the child has been transferred from the critical care unit to the rehabilitation unit, they typically are placed in diapers for management of their incontinence. In children with cognitive impairment, the majority of their incontinence is a result of a disinhibited bladder. In these children, the bladder is emptied completely and bladder volume is reduced. If their cognitive status allows, they may be able to participate in a timed voiding program to achieve functional continence or to learn continence. Children may also have a neurogenic-type bladder with uninhibited bladder contractions, which can be treated with anticholinergic medication. This will allow for increasing bladder volume. If the clinician is faced with bladder emptying that resembles an upper motor neuron or lower motor neuron voiding pattern, it is imperative that spinal cord injury be ruled out. This possibility can usually be eliminated by demonstrating low postvoid residual volumes (183).

Central Autonomic Dysfunction

Central autonomic dysfunction (CAD) is a clinical entity that is manifest by a myriad of symptoms, including hyperthermia, hypertension, diaphoresis, generalized rigidity, tachypnea, decerebrate posturing, tachycardia, and pupillary changes. It has many names, including, diencephalic seizures (184), autonomic storming, autonomic dysfunction syndrome (185), hypothalamic midbrain disregulation syndrome (186), central seizures, central storming, central fevers (187), and posttraumatic hyperthermia (188). CAD is a result of an injury to the brain that interrupts the diencephalic–brainstem connection, leading to what is called " brainstem release phenomenon (186)." Signs and symptoms will often disappear as neurologic improvement is noted, but medical management may be necessary for six months or more after injury in a select group of patients (189).

Management of CAD is usually initiated due to concern about an elevated body temperature. It is imperative the clinician rule out an infectious etiology, as central temperature elevation is a diagnosis of exclusion. CAD responds poorly to antipyretic medication (185), such as the nonsteroidal anti-inflammatory drugs. This may be helpful when ruling out infection. Initial management at the bedside usually consists of attempting to lower the temperature by providing cooling blankets and ice packs, turning down the temperature in the room, or providing a fan in the room to cool the patient. Often, the patient's hypertension is marked enough to warrant treatment with a beta blocker such as propranolol, which will also help reduce heart rate and can be used on an as-needed basis (187). Bromocriptine is used by some clinicians to reduce the symptoms of CAD and has ultimately resulted in a decreased need for antipyretics (189). Morphine in combination with bromocriptine has been useful in one study. ITB has also been reported to effectively treat CAD associated with TBI (100,190).

CAD is associated with a poor prognosis. In a retrospective review of a series of children with acquired brain injury, CAD correlated positively with more protracted periods of unconsciousness and overall worse cognitive and motor outcomes one or more years postinjury. Follow-up computed axial tomography (CAT) scans in these children revealed ventricular enlargement and marked brain atrophy (189).

Heterotopic Ossification

Pediatric TBI survivors have about a 14% to 23% chance of developing heterotopic ossification (172). It is more common in children who are over 11 years of age and also more common in children who have two or more extremity fractures (191). Heterotopic ossification in children with TBI is most common at the hips and knees. Diagnosis is often made approximately one month after injury (191,192). Signs and symptoms of heterotopic ossification include pain, decreased range of motion, and occasionally swelling (192). Deep venous thrombosis may be present concomitantly with heterotopic ossification and warrants further evaluation (193).

Treatment of heterotopic ossification begins with regular and aggressive passive range of motion for these patients. Occasionally, splinting is necessary to prevent worsening contracture. Nonsteroidal antiinflammatory drugs such as indomethacin, ibuprofen, or aspirin are often employed in an effort to halt progression once it is identified (191,192). Although in adult rehabilitation patients, heterotopic ossification is often treated with high-dose disodium etidronate, it is avoided in pediatric patients due to concerns for development of rickets or rachitic syndrome (194). Rarely does pediatric heterotopic ossification require surgery (172,191,192).

Posttraumatic Epilepsy

In recent years, whether pediatric TBI survivors should be treated with antiepileptic drugs (AED) prophylactically has been discussed frequently in the literature. Seizures after TBI are separated into immediate, early, and late posttraumatic seizures. Immediate seizures

happen within the first 24 hours of injury, and early seizures happen within the first 7 days. Late seizures occur anytime after the first week following the brain injury and may begin many years after injury (195).

In adults who have TBI, early seizures correlate with the development of late seizures. However, this correlation is not seen in the pediatric population after brain injury (196). The incidence of posttraumatic seizures is greater in children than in adults. Although the majority of posttraumatic seizures in children are immediate seizures, the incidence of early seizures ranges from 20% to 39% (54, 196, 197, 198) and the incidence of late seizures ranges from 7% to 12% (196, 199, 202). It should also be noted that lower GCS and younger age are associated with a higher risk of early posttraumatic seizure (54, 196, 197, 198, 200). Children less than 2 years of age have a three-fold greater risk of early posttraumatic seizures compared with children who are 2–12 years of age (197). In one study of children who were 3 years of age and younger at injury, the risk of late posttraumatic seizures was greatest in the children who were under 1 year of age at the time of injury (55).

Consensus guidelines established in 2003 state that currently there is insufficient data to support a standard guideline for the prevention of pediatric posttraumatic seizures (201). The guidelines recommend that prophylactic AED not be used to prevent the development of late seizures. They did note, however, the bulk of the evidence does suggest considering AED as a treatment option to prevent early seizures in high-risk patients. The American Academy of Physical Medicine and Rehabilitation agrees that "[a]ntiepileptic drugs are not recommended after one week for seizure prophylaxis in nonpenetrating traumatic brain injuries." Young et al. (202) conducted a randomized, double-blinded, placebo-controlled study to evaluate phenytoin in 41 children with TBI who were followed for 18 months post-injury for the development of seizures. No statistically significant difference was distinguished between the groups in the development of late posttraumatic seizures.

Posttraumatic epilepsy is diagnosed when the patient has two or more seizures in the late period after TBI. For the child who transfers to the pediatric rehabilitation medicine unit on phenytoin or another AED, the process of weaning the medication is fairly simple. If serum levels of the AED are subtherapeutic, it is safe to discontinue the medication without weaning. Otherwise, the dose can be reduced by approximately 50% the first week and can be discontinued thereafter. Since early seizures in children are not correlated with the development of late seizures, one can obtain an EEG in children who had early seizures and if no epileptiform activity is identified, consideration can be given to weaning the antiepileptic drug (166).

In children who develop posttraumatic epilepsy, AED therapy should use medications that have the least effect on cognitive function. This medication should then be used at the lowest clinically effective dose in order to maximize the cognitive recovery of these patients. The consulting pediatric neurologist considers which AED to use in a given child based on factors including the clinical seizure pattern, the EEG activity, and the side effect profile of the AED.

Posttraumatic Hydrocephalus and Cerebral Atrophy

Ventriculomegaly is seen commonly after severe TBI in children (61). The enlargement of the ventricular system can be either from high-pressure hydrocephalus or from cerebral atrophy resulting in hydrocephalus ex vacuo. True hydrocephalus is a result of either an obstruction in the cerebral spinal fluid flow or impairment in the absorption of cerebral spinal fluid, ultimately resulting in an increase in cerebral spinal fluid volume and pressure. Hydrocephalus can be described, therefore, as either communicating (where there is abnormality in absorption) or noncommunicating (where there is an obstruction in the flow of the cerebral spinal fluid). The majority of hydrocephalus is caused by impaired cerebral spinal fluid absorption, secondary to inflammation or secondary to subarachnoid hemorrhage.

Hydrocephalus ex vacuo describes enlargement of the ventricular system that results after cerebral atrophy and loss of brain volume (Fig. 10.1). To distinguish between clinically significant hydrocephalus and the expected consequence of cerebral atrophy after severe TBI, one must consider the patient's clinical status as well as the amount of time that has passed since the injury. Overall, if the patient is continuing

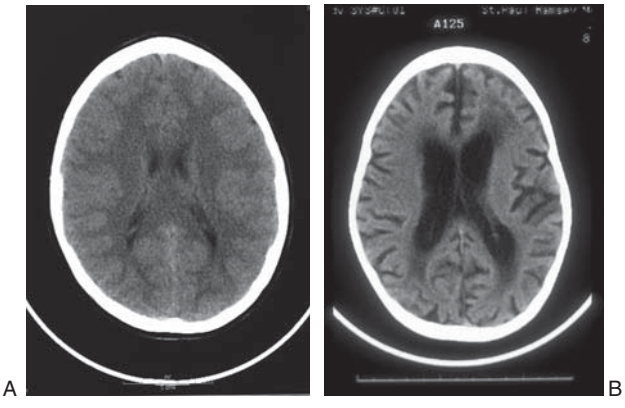

Figure 10.1 Cerebral atrophy. (A) Normal computed tomography (CT) scan. (B) CT scan showing posttraumatic brain injury cerebral atrophy with ventriculomegaly and increased sulci.

to demonstrate ongoing and regular improvements in their clinical status, ventriculomegaly is more likely to be due to cerebral atrophy. The patient who has hydrocephalus typically continues with poor clinical improvement or clinical deterioration. The CT scan findings will yield clues as well, with cerebral atrophy demonstrating areas of encephalomalacia or enlargement of sulci, while hydrocephalus demonstrates more specific changes around the ventricular system outlined in Table 10.2 (203).

Hydrocephalus should be suspected if clinical improvement is not noted in a patient status post-TBI or if the clinical picture includes functional decline, seizures, abnormal posturing, or increased tone. Consideration of hydrocephalus in these patients is paramount, as failure to identify hydrocephalus when it is present may delay recovery. CT scan of the brain allows for rapid detection of hydrocephalus. The treating team may then choose to have a ventricular peritoneal shunt placed, which may improve the clinical status of the patient when normal ventricular pressures are reestablished (204).

REHABILITATION

Rehabilitation's goals are to reduce disability and help a child achieve the maximum degree of age-appropriate functional independence in physical, cognitive, social, and emotional areas after having sustained a TBI (205). In addition to prevention of secondary impairment, facilitation of improved function, education in the use of compensatory techniques, and evaluating and potentially modifying the child's environment are also important considerations in minimizing handicap. Parent and caregiver education are important as well. It is, therefore, imperative that children with TBI be involved with rehabilitation services (206). Also, it is important that these rehabilitation services be provided by individuals knowledgeable in child development (207).

Rehabilitation efforts include attempting to restore function or, when that is not possible, to teach adaptive techniques to compensate for areas of deficit (207).

10.2 Computed Tomography Criteria for the Evaluation of Hydrocephalus

1. Increased size of the lateral ventricles at the anterior horns
2. Increased size of the temporal horns and the 3rd ventricle
3. Increased size of the basilar cisterns and 4th ventricle
4. Sulci appear normal or of decreased size
5. Periventricular hypodensity

Context-sensitive rehabilitation with integration across many domains of functioning and providers should be practiced (208). For example, when a child is returning to school, in addition to appropriate special education, social reintegration, help with activities of daily living, and comfortable positioning should all be addressed.

Early Rehabilitation

Initiating rehabilitation services early shortens the overall hospital and rehabilitation stay (209,210). Rehabilitation efforts, therefore, should begin early while the child is in the intensive care unit (ICU). Early efforts should be aimed at reducing potential complications of immobility, including ischemic ulcers, compression neuropathies, and contractures (211). Complications due to excessive pressure can be prevented by frequent repositioning, special mattresses, and padding bony prominences. Contractures can be prevented by initiating range of motion exercises and use of resting splints. Also, stimulation therapy is important during the ICU stay. Stimulation therapy involves presenting a brief structured stimulus for which one anticipates a response. It is a means of frequently assessing the child but does not cause awakening. Sometimes, rehabilitation interventions in the ICU must be limited because stimulation can increase intracranial pressure (87).

It is also helpful to have a social worker begin to meet with the family while the child is still in the ICU to begin education about brain injury and the rehabilitation process, as well as to provide support (87). Early transfer to a rehabilitation setting is indicated as soon as the patient is medically stable (212).

Inpatient Rehabilitation

Inpatient rehabilitation requires the participation of an interdisciplinary, specialized team lead by a rehabilitation physician to manage the multiple physical, cognitive, and social issues with which the child is faced (213,214). Central to this team is the injured child and their family.

Sensory Stimulation

Even before a child is following commands, rehabilitation may be initiated. In addition to providing structured stimulation and assessing responses on a frequent basis, physical and occupational therapy may work with positioning, including specialized equipment, and activities. Head and trunk control are facilitated. Also, localized responses are channeled into more purposeful activity using hand-over-hand techniques. Oral stimulation is started to help with evaluating oral motor function, and may facilitate more

control and begin the process of evaluating for attempt to transition to oral feeding (87).

Computer-assisted rehabilitation can be used at many times in the rehabilitation continuum. Even when a child is not yet consistently following commands, computer programs may be useful to elicit auditory or visual attention. As responses increase, various types of switches can be used to assess the understanding of causality. Obviously, with children who are cognitively able, a wealth of software is available to work on various cognitive areas and provide structure and immediate feedback in reference to performance (87). The use of computers in rehabilitation activities can continue after discharge from the inpatient service. Although commonly used, there is no certainty whether computer-assisted therapy is more effective than more traditional neurorehabilitation intervention. Computers are only one facet of the overall rehabilitation approach (215).

Interventions Based on the Cognitive Level

As children become more responsive and interactive, therapy can become more cognitively based, addressing specific areas of identified deficits that have been previously noted. An eclectic therapeutic approach should be used (87). Classic neurorehabilitative therapy approaches, adaptive equipment, the use of technology, and environmental modification all have the ultimate goal of increasing the child's independence and ability to function, and continue to facilitate ongoing development and acquisition of skills. Cognitively based rehabilitation should continue even after discharge from the inpatient rehabilitation setting, as improvement in this area has been noted as far as two years post-injury (211).

Speech can also be impaired after a TBI. Children therefore should be assessed by a speech pathologist that can provide them with directed therapy or communication aids as appropriate (211).

Psychosocial Services

An acquired brain injury of a child changes the entire family. Roles and responsibilities change, and the degree of disability affects the family's future activities and opportunities (87,216). Supportive services are essential not only for the injured child, but also for the entire family. It is also important to assess preinjury family functioning because this factor has been shown to have an impact on long-term outcome, especially with regard to behavioral problems (217). The injured child participates in supportive counseling in addition to cognitive rehabilitation activities. Counseling is imperative to assist in preparing for community reentry and in the recognition of the differences seen after return to the community as contrasted to the artificial environment of the inpatient rehabilitation unit.

Providing supportive counseling and education for the patient's siblings is also important. Medical play can be an effective technique for both injured children and their siblings. Siblings may also benefit from peer support (87).

Counseling and education about TBI and its consequences can be helpful to parents. Proper training enables them to become advocates for their children and to help their children deal with the challenges they face because of the injury (217). These counseling and education needs may be long-term because the parents initially may be in denial concerning the severity of injury and permanence of impairment (87,218,219). The injury results in the need to negotiate systems with which parents were previously unfamiliar. These include special education, medical and rehabilitation services, and publicly supported programs (217). Also, for families of children with severe injury and those who had difficulties before injury, stressors continue long-term, and families may need additional attention and resources to assist them in coping with the consequences of their child's injuries (220). One of the areas most severely affected after a TBI is social and peer reintegration. The inpatient rehabilitation process should also address this issue (162).

Another issue that requires attention is the potential impact of a child's TBI on family finances. Osberg and colleagues (221) found that parents of children who required transfer to a rehabilitation unit experienced difficulty with work and finances. Proactive planning, contact with employers, and the exploration of alternative funding sources can be of substantial benefit.

Discharge Planning

Rehabilitation has become a continuum of care, being provided at many different sites and intensities of service. It is important to begin discharge planning early in the rehabilitation hospitalization. The costs of caring for children with TBI are significant. The majority of those costs relate to the acute care hospitalization, but for those with significant injury, up to 47% of the hospital costs are due to inpatient rehabilitation (199).

Most children are discharged to home after TBI. Determining the appropriate services, assisting the family in obtaining them (depending on their third-party payer and network requirements), and coordination with the public school system are essential elements in this planning process. Working closely with the third-party payer case manager can be helpful in obtaining the appropriate services for optimal transition. Family or other caregiver training is imperative

in medical or nursing procedures as well as the management of behavioral problems after TBI.

After discharge from the acute care setting, rehabilitation continues, with reintegration into the community. Coordination of medically and educationally based services and effective communication among providers are essential. Accommodations to facilitate effective reintegration can be physical, environmental, or instructional (207).

COMMUNITY REINTEGRATION

School Services

Children who have experienced TBI are more likely than the general population to require special education services (222).Children with TBI have learning problems (223). Twenty-five percent to 75% of children with TBI demonstrate school failure or require special education services within the first five years of injury. Studies demonstrate that the severity of injury is correlated with cognitive functioning after brain injury. Areas of concern include intelligence, adaptation, adaptive problem solving, memory, academic performance, motor abilities, and psychomotor problem-solving (199). Other studies have demonstrated poor overall academic performance and academic promotion despite average academic achievement scores in nearly all children who have sustained TBI (222).

Most children return to school relatively soon after TBI, and many schools have an inclusive service model so that these children are in regular classrooms receiving supportive services. The wide variety of potential impairments post-TBI makes general statements about school programming challenging. It is necessary to identify the student's needs by evaluating their level of function and plan strategies to address those needs. Most likely, a student with a TBI will need a program that is unique to their individual needs, requires flexibility, frequent communication with family, and regular monitoring (151).

Individual Educational Plans

The Individuals with Disabilities Education Act (IDEA) was enacted in 1990 as Public Law 101–476 and allowed for the inclusion of TBI as a condition of eligibility for special education and educational assistance within the public school system. With this law in place, emphasis was placed on the child's global functioning rather than on academic performance alone. This resulted in increased emphasis on executive function deficits, memory and attention deficits, and slowed perceptual motor functions that tend to be characteristic of children with TBI (224). A team approach to the management of the IEP for the child with a TBI is important. The child's team should include a rehabilitation specialist, the child's school, and the child's family, at a minimum. Preparation of the initial IEP should begin while the child is still an inpatient on the rehabilitation ward. This allows for smooth transition from the inpatient rehabilitation program back to the school system (225). The involvement of the family is essential to facilitate a sense of continuity of care, and demonstrates to the parents that return to school does not represent return to the child's previous level of functioning. Ongoing difficulties will likely persist and need to be addressed accordingly. It is imperative that the team understand the dynamic and changing needs of the child with a brain injury, such that regular review and updating of the IEP occurs. The role of family involvement and family support for these children cannot be minimized, as it has been shown that there is an increased risk for maladaptive behavior in children with TBI who came from poorly functioning families. Therefore, individual and family counseling, parent training, and child behavior management is recommended to improve these children's outcomes (226).

In recent years, a push toward identifying the best approach to assisting children with TBI within the school system has been investigated. Some states have responded with programs that provide consulting services to the school systems and their educators with a TBI team model. The school system then presents on a case-by-case basis their concerns for a given pupil, and the consulting team assists in developing an IEP. The state brain injury team will then reassess the child and the IEP. It has been demonstrated that educators who receive training in childhood TBI have increased confidence in working with these pupils (227).

Too often, children with TBI remain underserved and, in some cases, forgotten. Sometimes educators are unaware the child had a previous TBI, or if their academic performance on achievement tests was within the average or acceptable range, they are deemed to be unaffected by the brain injury. Their diagnosis is forgotten until they have failed academically. This is highlighted in the research estimating that there are approximately 130,000 students in the United States with special education needs after TBI; however, the U.S. Department of Education reported only about 15,000 students receiving services under the TBI label (208).

Community Support

When the child with TBI is discharged from the hospital, it is almost certain that the child, at a minimum, will have a need for increased supervision. Ideally, the child will be transitioned back to school full-time, but the family will need to care for that child when school

is not in session. Community services become paramount in caring for these family units.

In-Home Services

There are many reasons why additional support may be needed within the family home to care for the child with TBI. If the child is dependent for all aspects of care, personal care assistants (PCAs) or skilled nursing care may be necessary for a time. Even if the child is not dependent for mobility, marked behavioral changes in the child with TBI may warrant some of these services. Furthermore, if ongoing therapy services are needed to meet active rehabilitation goals, these therapies can sometimes be provided in the home setting. Social workers and case managers may be helpful, especially when poor family functioning is present. This is especially critical to attempt to offset the development of behavioral problems in these children status post-TBI (226).

Out-of-Home Services

The majority of children with TBI are discharged to home in the care of their families. Some children transition to medical foster placement, group homes, or skilled nursing facilities as an alternative living situation. In these circumstances, the children still need to have school services identified and accessed locally, as well as potential outpatient therapy services for their ongoing rehabilitation goals and needs.

Planning for Long-Term Needs

Ultimately, the child with a TBI becomes an adult with a remote TBI, and often ongoing services as well as resources are still needed. The time may come for the child who is dependent for all cares to require transition out of the home and into a long-term care facility or medical foster care placement. Resources are often limited in this regard, so early planning with the help of a social work team and perhaps legal consultation is appropriate. Vocational rehabilitation services should also be identified for these patients if appropriate. Often, the school system can be helpful in accessing these resources. The school may collaborate with local vocational services, independent living centers, community-based advocacy agencies, and other support systems to establish and coordinate a transition plan from school to the community (225).

Returning to Sports and Recreational Activities

Since sports and other recreational activities are typically an integral part of the childhood lifestyle, return to the community for children often involves planning for return to these activities.

For the child who has sustained a TBI, counseling the family on the safety of returning them to playing sports is challenging. This is partly due to a lack of evidence or guidelines in the rehabilitation literature. In recent years, better guidelines have become available for the management of return to play within sports after a child sustains a concussion during sporting activities (see the section on concussion), but these recommendations do not necessarily translate to appropriate recommendations in the child who sustains a TBI unrelated to sport activities. For instance, the grading of non-sports traumatic brain injuries as mild, moderate, and severe is a different rating scale than grading the sports-related concussion as mild, moderate, or severe.

For the child who was injured with a moderate to severe brain injury, the guidelines remain unclear. It is known that in certain sports, such as high school football, approximately 20% of players incur a concussion each year, though other "collision" sports can result in concussions as well, including boxing, and ice hockey (228). Furthermore, sports such as basketball and soccer may result in an inadvertent concussion if players come into contact with each other, though with less force than one would expect in the collision sports. Other high-risk sports, including downhill skiing, snowboarding, and gymnastics, can be as dangerous as contact or collision sports from potential resulting blows to the body (229). For these reasons, it is challenging as a rehabilitation clinician to allow a patient who sustained a TBI to return to these activities. It is known that cognitive impairments will follow multiple mild concussions. Mildly concussed athletes demonstrate a decline in memory compared with their baseline performance (230), and athletes with a history of multiple concussions score significantly lower on memory testing (60). In the individual with a recent TBI, risking subsequent brain injury or concussion and worsening their clinical outcome is not recommended. Furthermore, the patient may sustain other traumatic injuries in attempting to return to sports as a result of poor performance due to impaired speed, response time, and information processing (30).

OUTCOMES

Measurement Tools

Several measures of function have been used to assess outcomes after TBI. They are variable and can involve neuropsychological assessment as well as motor testing.

The Coma/Near-Coma Scale is useful in evaluating small changes in patients who are at a low level

of consciousness. It can be applied to both children and adults, and is helpful in allowing for reproducible assessment of subtle changes over time (231).

The Functional Independence Measure (FIM) and the Functional Independence Measure for Children (WeeFIM) can be used to asses global functioning (232). The FIM is useful for children who are more than 7 years of age and the WeeFIM between 6 months and 7 years of age. This tool assesses transfers, locomotion, self-care, sphincter control, communication, comprehension, and social cognition (233). The WeeFIM is often used to demonstrate gains in children with TBI during their inpatient rehabilitation stays.

The Glasgow Outcome Scale is a scale for classifying patients with traumatic brain injuries into five categories: death, persistent vegetative state, severe disability, moderate disability, and good recovery (234). This scale has been modified to differentiate outcomes as they apply to children (Table 10.3). It is divided into a cognitive component and a motor component.

The Pediatric Evaluation of Disability Inventory (PEDI) is another clinical assessment tool. It describes performance in the domains of self-care, mobility, and social function. The PEDI has questions about 197 functional skills, 20 caregiver assistance questions, and 20 equipment modification questions. This scale is used in children 6 months of age to 7 years of age, and correlates well with the WeeFIM, demonstrating good validity within both of the measures (232).

Survival

In the last two decades, morbidity and mortality associated with pediatric TBI has been on the decline, with children younger than 4 years of age and adolescents greater than 15 years of age having higher mortality rates. Infants still had the highest overall mortality (235). This improved mortality rate in children and adolescents may be due to improvements in medical care and surgical treatment. Potoka et al. (236) reported that for children who sustain severe TBI, mortality was significantly lower if the child was treated at a pediatric trauma center or at an adult trauma center with qualifications to treat children. The mortality of patients who sustained a TBI was higher if the child was treated at a level 2 adult trauma center instead. More than two-thirds of deaths from brain injury occur at the scene or en route to the hospital in a population in which both adults and children were studied (237), but children with acquired brain injury who survive the initial injury generally live for many years. The pediatric literature evaluating mortality after TBI suggests that death from profound brain injury is only seen in children who remained in vegetative states longer than 90 days after anoxic or traumatic injury (238). These findings stand in contrast to adults who have sustained an acquired brain injury. The adult literature notes that approximately 50% of adults in vegetative states die within one year of their injury, whereas in the pediatric population, one-half of the children still in vegetative states one year after injury were still living seven to eight years later (238,239).

10.3 Modified Glasgow Outcome Scale

Cognitive Status
0-Normal
1-Verbal communication, needs help in academic setting
2-Limited language, can express needs and wants, significant adaptation of academic setting
3-No language, responds to voices
4-Persistent vegetative state

Motor Status
0-Normal
1-Near-normal ambulation, needs supervision for ADLs
2-Ambulates with assistive devices and/or needs adaptive equipment for ADLs
3-Needs assistance for ambulation or ADLs
4-Nonambulatory, assistance for transfers, dependent for ADLs
5-No purposeful movement

Morbidity by Injury Severity

Concussions

A concussion is the transient and immediate change in neurologic function due to a mild TBI, with or without a brief loss of consciousness (240). A concussion is often referred to as getting "dinged" or having your "bell rung." Neuroimaging is typically normal following a concussion (241), and the diagnosis is made clinically. Symptoms of concussion usually resolve within 20 minutes, but postconcussive symptoms can last for days and weeks. Common concussive symptoms include headache, memory lapses, cognitive problems, confusion, feeling dazed or "foggy," dizziness, sleep problems, behavioral changes, bizarre statements, poor attention span, photophobia, diplopia, and sadness (242).

Common causes of concussions in children are sports injuries, falls, bicycle accidents, and automobile accidents (243). Yearly in the United States, more than 300,000 TBIs, mostly concussions, occur due to youth sports (244). Female athletes have a higher rate of concussions than males, thought to be secondary to their relatively weaker neck muscles being less able to absorb head and neck trauma (245). Concussions are graded by severity (Table 10.4), and return to activities

depends on the concussion severity. Postconcussive symptoms (246) may resolve before cognitive functioning returns to normal (247). Neuropsychologic testing can detect these persistent cognitive changes. Many youth sports programs use cognitive assessment tools such as ImPACT™ (248) prior to participation and will not allow a return to activities until cognition returns to baseline (249). In general, a person should be symptom-free for one week before returning to activities.

In the days and weeks after a concussion, the injured brain cells are vulnerable to repeat injuries, which can cause extensive neuronal loss (240). For this reason, the brain should be rested following a concussion until all symptoms have resolved. Symptoms can be exacerbated and recovery slowed by strenuous physical and cognitive activities. During this "cognitive rest," physical and academic activities should be limited. Once symptoms have resolved, the patient should gradually return to activities as tolerated (250).

Repeated concussions over months or years can lead to long-term cognitive deficits (60) and increase the risk of neurodegenerative disorders such as Alzheimer's disease (251). So activities that have a higher risk of concussions, such as football, boxing, and ice hockey, should be restricted if a person has suffered several concussions. Persons who have had previous concussions may be more susceptible to recurrent concussions and slower brain healing (60). Repeat concussions over hours, days, or weeks can lead to catastrophic changes, such as second impact syndrome, previously described in the pathophysiology section.

Mild to Moderate Injury

Children who sustain minor TBI may demonstrate few, if any, consequences, or they may complain of subjective complaints such as headaches, mild memory impairment, and fatigue. This constellation of symptoms is consistent with postconcussive syndrome.

10.4 When to Return to Play

GRADES OF CONCUSSION	GRADE 1	GRADE 2	GRADE 3
Definitions	1. Transient confusion 2. No loss of consciousness 3. Concussion symptoms last <15 minutes	1. Transient confusion 2. No loss of consciousness 3. Concussion symptoms last > 15 minutes	1. Any loss of consciousness
Management recommendations	1. Remove from activity 2. Examine immediately and every 5 minutes for change in status, at rest and with exertion 3. May return to activity if symptoms clear within 15 minutes	1. Remove from activity for remainder of day 2. Examine immediately and frequently for signs of deteriorating neurologic status 3. Trained person reexamine the next day 4. Full neurologic exam by physician to OK return to activity after asymptomatic for one full week at rest and with exertion	1. Transport to nearest emergency department if still unconscious or other concerning signs 2. Thorough neurologic exam on emergent basis and appropriate neuroimaging, if indicated. 3. Hospital admission if pathology detected or mental status abnormal
When to return to play (period of time being asymptomatic with normal neurologic exam at rest and with exertion)	1. One grade 1 concussion: 15 minutes 2. Multiple grade 1 concussions: 1 week	1. One grade 2 concussion: 1 week 2. Multiple grade 2: 2 weeks	1. Grade 3 with brief loss of consciousness (seconds): 1 week 2. Grade 3 with prolonged loss of consciousness (minutes): 2 weeks 3. Multiple grade 3: 1 month or longer, as per evaluating physician

Source: Adapted from Quality Standards Committee of the American Academy of Neurology. The Management of Concussion in Sports (practice parameter). *Neurology*. 1997;48:581–585.

Although the child with a mild TBI may not require a prolonged hospital stay on the rehabilitation unit, they may still have difficulty returning to school. The challenges these children may encounter include difficulty with timed tasks, impaired attention, and impaired memory. Subtle language dysfunction and impaired prosody of speech may be notable, as well as behavioral and personality changes. For these children, neuropsychological testing to identify any deficits is imperative, lest they be allowed to fall behind in their academic progress as the effect of the injury on their cognitive function goes unnoticed (166). It is encouraging, however, to note that by one year after injury, children who sustained a minor TBI rarely have impairment that continues to challenge them academically (252). In 2004, Hawley et al. identified a group of 67 school-aged children who sustained TBI (35 mild, 13 moderate, 19 severe) and gathered 14 control subjects as well. They reported that two-thirds of the children with TBI exhibited significant behavioral problems and 76% of the children with behavioral problems also had difficulties with schoolwork (253). Another study has noted that children with mild TBI also demonstrate difficulties compared to typically developing peers in some areas of metacognition—specifically in their ability to recognize semantic anomalies in spoken sentences (254). These findings suggest that although it is encouraging that so many children do well academically after sustaining mild TBI, caution must be taken to not overlook behavioral concerns or higher executive functions that may affect academic performance.

Moderate to Severe Injury

Outcome studies regarding children who sustained significant TBI have demonstrated overall fair recovery. One such study evaluating 30 subjects noted that only 1 out of the 30 subjects failed to become ambulatory by two or more years post-injury, and 6 out of the 30 subjects ultimately attended college. The evaluators found that 13 out of 30 of those subjects returned to their previous level of functioning (255). Another study in 1980 by Brink et al. (63) noted 73% of pediatric survivors of severe TBI were able to demonstrate independence in ambulation and self-care within 1 year post-injury.

The literature regarding academic outcomes for children after severe TBI is less encouraging. These children demonstrate lower scores on standardized tests (199). Ewing-Cobbs (224) reported these children have lower reading recognition, spelling, and arithmetic scores compared with patients who sustained only a mild to moderate brain injury. Two years post-TBI, 39% of these patients had failed a grade and 73% of them needed special education assistance. Ewing-Cobbs (256) also reported that moderate to severe TBI sustained prior to the age of 6 had adverse persistent consequences for intellectual and academic development. These children were assessed five years after injury and were found to have continuing deficits with no further recovery of function, demonstrating a persistent performance gap with no "catch up" phenomenon. They also found that children with focal nonprogressive brain injury demonstrated relatively good intellectual and academic outcomes. They concluded that there appeared to be significant limits on neurologic and cognitive plasticity. An interesting note was that the older children did fairly well on achievement testing but demonstrated poor functional academic recovery by failing a grade and needing ongoing support services. It seems that contributing components to success at school are the comorbid behavioral problems that almost two-thirds of children display after TBI and approximately three-quarters of those children demonstrate difficulties with schoolwork (253).

Profound Injury

Children with profound brain injury and unconsciousness that lasted for greater than 90 days demonstrated a less favorable prognosis for recovery. In a series evaluating profoundly injured children by Kriel, only 1 of the 36 subjects had a normal motor outcome and no children demonstrated a normal cognitive outcome. Two-thirds of the patients recovered some language function, and one-quarter recovered independent ambulation with or without assistive devices (238).

Anoxic Brain Injury

Generally speaking, the children who sustain an anoxic brain injury tend to demonstrate a worse outcome than those with TBI. In a study that evaluated children who were unconscious for greater than 90 days secondary to an acquired brain injury, 75% of the subjects who had a TBI eventually regained consciousness. Only 25% with anoxic brain injury ultimately regained consciousness. One-quarter of children with TBI became ambulatory, and most of them regained some language function. Children with anoxic brain injury who were unconscious for more than 60 days did not regain language skills or become ambulatory. A greater percentage of the children who had anoxic injuries died during the years of follow-up (257).

Morbidity Related to Age at Time of Injury

Since children have a better rate of survival after TBI, it is often assumed that pediatric outcomes are more favorable than adult outcomes. This is often attributed

to the plasticity theory, suggesting that the young brain has a better opportunity to recover function. As noted in the pathophysiology section, however, injury to the developing brain may affect response to injury and the ability for future development and learning to occur. Also, the pediatric brain has had less time to learn skills and overlearn skills.

TBI during infancy has been shown to result in difficulty developing expressive and receptive language skills. Infants sustain a higher proportion of TBI that are secondary to nonaccidental trauma and their outcomes are poor. Koskiniemi (258) reported the long-term outcome of TBI in children and identified that the worst outcomes typically occurred in those children who were younger than 4 years of age. That study demonstrated similar results to a study done by Kriel (62) in which 97 pediatric patients who were unconscious for greater than 24 hours were followed, with the worst outcomes seen in children who were younger than 6 years of age and involved both cognitive and motor impairment.

Older children show fairly good recovery of language function and independent ambulation. This was evaluated in a study of 28 adolescents followed longitudinally after brain injury. Twenty-five of them recovered language function, and 21 of them recovered independent ambulation. However, they had a lower high school graduation rate and employment rate than an age-referenced population. Their social interactions are impaired, as two-thirds of these individuals reported that after their TBI, their social life declined, and in fact, only 1 of the 28 subjects was married at the time of the follow-up, compared with 61% of the reference population (61).

Prevention

Prevention campaigns against child abuse and shaken impact syndrome have largely been educational campaigns provided by perinatal hospital staff and pediatricians.

Seatbelt use has been shown to reduce fatalities by 45% in passenger cars and by 60% in light trucks. Child safety seats, like seatbelts, decrease injury and death in the pediatric population when correctly installed. Their use has been associated with a reduction in mortality by 70% for infants and by 47% to 54 % for toddlers. Seatbelt use in children decreased the need for hospitalization by 69% (2). Helmet use during motorized vehicle use has been documented to decrease the number of hospital-treated head injuries and the severity of motorcycle-related TBI (259).

Aggressive injury prevention campaigns, such as the "ThinkFirst" National Injury Prevention Foundation program, aim to educate children on the effects of brain injury related to gun accidents and sporting accidents, as well as the benefits of seat belt use and general safety (235). The use of bicycle helmets has reduced the frequency and severity of brain injuries (260–263). Greenwald (2) reported bicycle helmet use decreased the risk of serious brain injury by up to 85%. Rule changes and better equipment in football have significantly reduced severe neurological injuries (29,249). Efforts should be made to prevent mild brain injuries by avoiding risky behavior, wearing helmets when appropriate, following sports rules, and training properly. Following these guidelines can minimize the incidence and long-term consequences of concussions. Furthermore, in sporting activities, as previously discussed, guidelines for returning to play should be followed to avoid multiple concussive events and worsening cumulative effects.

Other prevention strategies to reduce TBI include lowering the height of playground equipment to no higher than 5 feet and fabricating play surfaces on the playground out of rubber, sand, or wood chips for better absorption of impact in the event of a fall (2). Finally, prevention of pediatric TBI begins with adults modeling safe behaviors within the home. Whenever adults are around children, safety-conscious behaviors should be demonstrated, including regular and routine safety belt use and helmet use during sporting activities.

Long-Term Rehabilitation Follow-up

The role of the pediatric physiatrist in caring for the child with TBI continues throughout the child's development. Cognitive deficits may not actually be evident in the very young child until higher cognitive skills are expected to develop. Follow-up should continue throughout the child's development, with their need for intervention intermittently reevaluated by the patient's physiatrist, therapists, and school team.

PEARLS AND PERILS

1. Injury at a younger age (younger than 4–6 years) typically results in poorer outcomes. This is perhaps due to increased vulnerability of the young child's brain to injury and the injury's impact on development.
2. Following a concussion, the injured brain cells are vulnerable to repeat injuries, which can cause extensive neuronal loss. For this reason, the brain should be rested following the concussion until all symptoms have resolved.
3. The long-term outcomes in motor, cognitive, and behavioral function may be better in focal injuries versus diffuse injuries, given the isolated nature of the brain damage.

4. Context-sensitive rehabilitation, with integration across many domains of functioning, and providers using the team approach should be practiced.
5. Care needs to be taken to distinguish cerebral atrophy (hydrocephalus ex vacuo) from posttraumatic hydrocephalus.
6. In children, seizures early after injury do not correlate with late seizures.
7. Long-term anticonvulsant prophylaxis has not been shown to decrease the development of late seizures.
8. Children often perform better in an evaluation setting than in their daily life.
9. It is important to be able to distinguish between diabetes insipidus, syndrome of inappropriate antidiuretic hormone, and cerebral salt wasting.

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11

Spinal Cord Injuries

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Children and adolescents with spinal cord injury (SCI) must deal with the multisystem involvement imposed by the injury that is compounded by physical and psychological growth and development, which cause complications not seen in the adult. Rehabilitation is a process that extends at least until the child is physically and psychosocially an adult. Involvement by a team that is expert in the management of children and adolescents with SCIs should continue throughout this period. This chapter discusses some of the main points to be considered by those who are involved in assisting this rehabilitation process. Advances have been made in the acute management of pediatric SCI, and there is new equipment and technology for rehabilitation, but the basics of rehabilitation in this area remain the same.

EPIDEMIOLOGY

Incidence and Prevalence

Compared to other disorders discussed in this text, SCI is a relatively rare disorder in the general population. The most comprehensive data on the epidemiology of SCI comes from the National SCI Database (NSCID), which is generated by the Model SCI Care Systems. The Model SCI Care Systems is a network of 26 centers funded by the National Institute on Disability and Rehabilitation Research, an institute in the U.S. Department of Education. Since SCI is not a reportable condition, data collection is limited. The NSCID estimates that it collects data on 13% of the new SCI cases per year. From this data, the incidence is estimated to be approximately 40 cases per million in the United States, roughly 12,000 new cases per year. This data does not include injuries that resulted in death prior to hospitalization (1). Acute SCI primarily occurs in young adults, though the average age has been rising. In the 1970s, the average age of injury was 28.7 years; since 2005, the average has increased to 39.5 years. The cause in this shift is unknown, though the incidence in SCI over the age of 60 has more than doubled since the 1980s. Other factors, such as prevention programs, advances in automobile safety, or referral patterns to Model Systems, may also be affecting the data. It was previously reported that 3% to 5% of all SCI occurred in children under age 15 and 20% of injuries occurred in those under 20. In their January 2008 report, the NSCID estimated that there were between 227,080 and 300,938 people living in the United States with SCI.

Demographics

Publications have combined data from the Shriners Hospitals for Children and the NSCID. As in adults, males are four times more likely to have SCI than females overall, with the ratio being 1.5:1 in children under age 9 years. In children under 3 years, females have outnumbered males in some studies. In younger children, there are no statistically significant racial trends. In those over age 15, there is an increased risk

in African American and Hispanic American populations. These figures are all from specialized hospital data and may not represent those with milder injuries (eg, incomplete lesions and paraplegia) who are treated in smaller hospitals or in adult settings. Since the year 2000, the racial make-up for SCI treated in the Model Systems has been 63.0% white, 22.7% black, 6% Hispanic, and 2.4% other racial groups (1).

Cause of Injury

Trauma is the primary cause of spinal cord injury, accounting for at least 93% of all SCI. Since 2000, motor vehicle crashes (MVCs) account for 42% of SCI, falls 27.1%, violence 15.3%, and sports injuries 7.4%. The remaining 8.1% are other and unknown causes (1). In those under age 20, violence and sports injuries are more common than falls. The sports most commonly associated with SCI are American football, ice hockey, wrestling, diving, skiing, snowboarding, rugby, and cheerleading (2).

Hadley and colleagues (3) reviewed 122 cases of spine injury in children 16 years and younger. Median age was 15 years in males and 14 years in females. SCIs were due to MVC in 39% overall, with MVC the cause in 17% of children under 10 years of age, 26% of those 10–14 years, and 52% in those 15–16 years. Pedestrian versus MVC were 11% overall and 33%, 16 %, and 3%, respectively, for the three age groups. Falls were the second leading cause under 10 years, with sports the second leading cause at ages 15–16 years. Fifty percent of those under the age of 10 had an occiput-C1 injury, with all levels of cervical injuries occurring in 72%, 60%, and 55% in the three age groups. Fifty percent of the subjects were neurologically intact, with bony or ligamentous injury only.

More recently, Bilston and Brown (4) have reported similar data from Australia, looking at children 16 years and younger. MVC accounted for 30% of all spine injuries and 50% of serious injuries. Sports were the next most common cause of all spine injuries, though falls resulted in a higher (20% vs 16%) risk of serious injury. Gender plays a significant role in cause of injury. Violence and sports-related injuries are more common in males, while MVC injuries are less gender-specific. The authors again demonstrate that children are at higher risk for cervical spine injury, especially under the age of 8, with higher injuries occurring in younger children.

The incidence in sports-related injuries is 8.7%. SCIs in American football have decreased markedly since the mid-1970s, when "spearing" was made illegal. This now-banned tackling technique resulted in a high degree of axial cervical loading. Since institution of the ban, SCI in football has decreased by 80% (5). Since the 1980s, the incidence of spine injuries and SCI have been increasing in ice hockey. These injuries are most often the result of a player being checked from behind, with his head down, into the boards, again resulting in high axial compression loads (6). Over the last several years, cheerleading has evolved into a competitive sport. This often involves gymnastic moves, tosses, jumps, and pyramid formations. While the incidence of SCI is low, this is a risk category where females are more likely to be injured (7).

CLASSIFICATION OF SPINAL CORD INJURY

Level of Injury-ASIA Impairment Scale

The most common method of classifying impairment from SCI is the American Spinal Injury Association (ASIA) impairment scale. The classification is based upon assessment of strength and sensation to light touch and pinprick in defined myotomes and dermatomes. Key muscle groups and sensory points are shown in Figure 11.1. The ASIA impairment scale has been modified over the years, originally based on the classification system defined by Frankel. Completing the ASIA examination on a child requires a certain level of maturity in being able to follow motor commands and respond appropriately to sensory stimulation. The examiner must take this into account when assessing children. Other factors that may limit examination (eg, long bone fractures or decreased level of consciousness) need to be taken into account as well.

The motor examination scores strength on a six-point scale: 0–5. For each strength grade, the joint being assessed must be moved through full available range of motion. A strength grade of 0 is given for total paralysis. A 1 is given for a visible or palpable contraction that cannot move the joint through its available range of motion with gravity removed. A grade 2 is given if the muscle group can move the joint through its range of motion with gravity removed. The joint is positioned parallel to the ground to limit the effect of gravity. A grade 3 is given when the patient can move the joint through full available range of motion against gravity but cannot bear any additional resistance. If a patient can bear additional resistance, they are given a grade 4, and a grade 5 is given for normal strength. Motor scores are documented on the ASIA form and summed for a total motor score. A rectal exam must be performed to assess for voluntary contraction, and is scored as yes/no. As individual muscles are almost always innervated by multiple spinal cord levels, a strength grade of 3 is considered normal for a muscle group if the level above has grade 5 strength. This implies that the grade 3 muscle group is only partially innervated and the more proximal innervation level is intact.

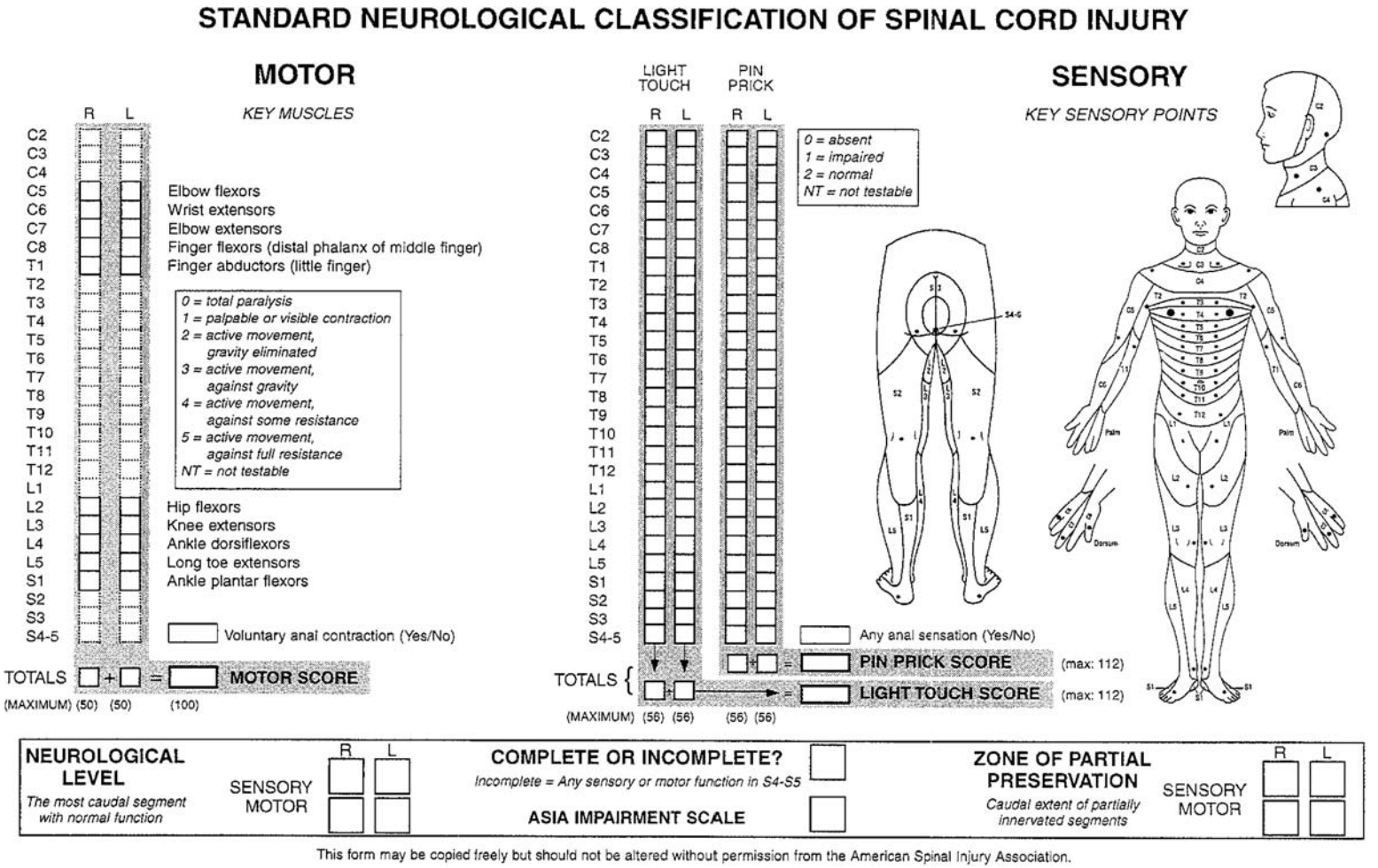

Figure 11.1 American Spinal Injury Association guide.

Sensory examination is performed using pinprick and light touch at key points, and grades as 0 for absent, 1 for impaired, and 2 for normal. These results are summed as well for total light touch and pinprick scores. Again, a rectal exam is necessary to assess anal sensation, also scored yes/no.

The ASIA neurologic level is the most caudal segment with intact motor and sensory exam. In addition to the level is whether the injury is complete or incomplete. With a complete injury, there is no motor or sensory function in the lowest sacral segment (ie, no anal sensation or voluntary anal contraction). A complete injury is classified as ASIA-A. Incomplete injuries are classified as B–E, as listed in Figure 11.1. While an "E" is described as normal sensory and motor function, this is in the context of a previously abnormal examination.

Paraplegia affects the lower extremities and, to varying degrees, the trunk. It does not affect the upper extremities; thus, T2 must be normal and any deficits are below that sensory and motor level. The preferred term from ASIA for involvement of all four extremities is tetraplegia, though quadriplegia is much more commonly used. Any injury that affects motor and/or sensation at or above the T2 level is tetraplegia. In addition, a number of syndromes have been described based upon the patterns seen after specific areas of the spinal cord have been injured.

Central Cord Syndrome

The central cord syndrome was first described in 1954 (8). As its name implies, this is damage to the central area of the spinal cord. This most commonly happens in the cervical region. Disruption of decussating spinothalamic fibers at the site of the lesion results in impaired pain and temperature sensation at those dermatomes. Dermatomes above and below the lesion may have normal sensation. As a lesion enlarges, damage may extend into the anterior horn cells and medial corticospinal tracts, causing weakness. Reflexes may be lost at the level of the lesion as well, with possible hyperreflexia at lower levels. As this is primarily a cervical syndrome, there are typically motor and sensory changes in the arms, with sparing of the legs, bowel, and bladder function.

Brown-Sequard Syndrome

Brown-Sequard syndrome results from a hemisection of the spinal cord. This is most commonly seen with low-speed penetration wounds, such as a stabbing. Corticospinal tracts and the dorsal columns cross in the brainstem, so their damage in this type of lesion leads to ipsilateral weakness and loss of vibration and position sense. The lateral spinothalamic tracts cross soon after entering the spinal cord, thus causing contralateral loss of pain and temperature sensation.

Anterior Cord Syndrome

The anterior (or ventral) cord syndrome is most commonly related to a vascular insult, causing infarction of the ventral spinal cord. This includes corticospinal, spinothalamic, and descending autonomic tracts to the bladder. This syndrome results in urinary incontinence, paralysis, and loss of pain and temperature sensation. Vibratory and position sense, whose tracts are in the dorsal columns, are spared.

Cauda Equina Syndrome

Compressive injuries in the lower lumbar and sacral vertebral levels may result in damage to the cauda equina, as the spinal cord proper has terminated at a higher level. This results in scattered symptoms, depending upon which nerve roots are damaged. The cauda equina syndrome results in damage to the axon of lower motor neurons, leading to a flaccid paralysis.

SCIWORA

Spinal cord injury without radiographic abnormality (SCIWORA) in children has been a known entity since at least the early twentieth century (9), though the acronym did not come about until 1982 in an article by Pang and Wilberger (10). The initial definition focused on children with traumatic SCI, who did not have evidence of vertebral column injury on spine x-rays, conventional and computed tomographic studies, myelograms, or dynamic flexion/extension studies. This excluded injuries caused by penetrating trauma, electrical shock, obstetric complications, and congenital spinal anomalies. In 2004, Pang published a review on what is now known about SCIWORA (11). Incidence of SCIWORA ranges between 5% and 67% of cases of pediatric SCI, with an average of 34.8%. The incidence is much higher in children 9 years and younger. Pooled data indicated an incidence of SCIWORA of 63.1% in younger children and only 19.7% in older children. Most injuries occur in the cervical cord, most commonly with C5–C8 lesions. Thirteen percent of injuries were in the thoracic cord. This injury is thought to be primarily present in children due to the unique physiology of the developing spine, being much more mobile, without resulting in bony fractures, but causing stretch injury to other tissues. This increased mobility was thought to result in damage to the soft tissue structure of the spine, including ligamentous and neural structures, which cannot be demonstrated on radiographic studies. The advent of more advanced imaging techniques has demonstrated these soft tissue injuries.

SCIWORA has been reported to cause complete and incomplete SCI, as well as central cord and Brown-Sequard syndromes. Pang classified ASIA-B/C as severe injuries and ASIA-D as mild. From this pooled data, SCIWORA results in ASIA-A 22.1%, ASIA-B/C 12.6%, ASIA-D 23.2%, central cord syndrome 29.4%, and Brown-Sequard 12.7% (11).

With the development of magnetic resonance imaging (MRI), damage to the soft tissue structures in the spinal column is readily apparent. This information was recently reviewed by Yucesoy and Yuksel (12). These authors suggest that in the MRI era, SCIWORA may be an ambiguous term and suggest that those with no lesions on neuroimaging be classed as "real SCIWORA" or spinal cord injury without neuroimaging abnormality (SCIWNA). In a strict sense, radiographs do not include MRI, yet in standard use, most practitioners would consider most imaging to fall into the category of radiograph, regardless of the nature of the physics involved in the imaging process.

PROGNOSIS FOR NEUROLOGIC RECOVERY

One of the most challenging aspects of rehabilitation medicine is talking with patients and families regarding prognosis for recovery. We must be able to present the best available information regarding prognosis (which is often not good) in a manner that people with varied levels of education and sophistication can understand and offer a reasonable and realistic degree of hope. Spinal cord injury is truly devastating, and as rehabilitation specialists, we must take the impact of that into account when communicating with patients and families. We must also be aware that during these times of stress, communication may not always be effective. We must also be clear that neurologic recovery can be markedly different from functional recovery.

A complete spinal cord injury examination is necessary for any discussion of prognosis. Examination at least 72 hours after injury has been determined to be a better prognostic indicator than earlier examinations (13–15). (This does not mean earlier examinations are not necessary, only that they are of less prognostic value, as they may be limited for a variety of reasons.)

Most recovery from spinal cord injury occurs during the first six months, with a plateau reached around nine months postinjury, though later recovery can occur. Neurologic recovery after a complete injury is poor. Ninety-six percent of those with complete paraplegia and 90% with complete tetraplegia at one month will remain ASIA-A. Muscle groups with a grade of 0 at initial examination are unlikely to regain functional strength. Muscles with grades 1–2 have a 64% chance of increasing to functional strength in paraplegia and 97% in tetraplegia. Incomplete paraplegia has an average motor score increase of 12 points at one year postinjury. Seventy-six percent of those with incomplete paraplegia became community ambulators. For incomplete tetraplegia, 46% became community ambulators at one year (16).

PREVENTION

Prevention of injury is always more effective than treatment, and this is especially true in SCI. The hallmark of prevention is safety education beginning in early childhood. Use of safe equipment is the second tenet of prevention, and nowhere has this been more effective than in the use of infant and child auto restraints and adult lap and shoulder belts. This practice has also caused lap belt injuries, however, including SCIs, which are more common in children than in adults. Other prevention relating to motor vehicles is substance abuse education and laws relating to driving while impaired. Pedestrian safety is promoted almost exclusively through parent and child education.

Prevention of sports-related SCIs has improved because of education, rules changes noted previously (such as no spearing in football, no checking from behind in ice hockey), better coaching, and better conditioning of players.

The ThinkFirst National Injury Prevention Foundation promotes safety education. "ThinkFirst programs educate young people about their personal vulnerability and the importance of making safe choices. The message is: You can have a fun-filled, exciting life, without hurting yourself if you 'ThinkFirst' " (www.thinkfirst.org). There are separate programs for teens and children, which promote injury prevention through talks and publications.

EARLY TREATMENT

Spinal Stabilization

Once it has been determined that the child has an SCI, the spine must be stabilized. The halo external skeletal fixation device was first described in 1968 for use in adults with cervical fractures by Nickel and colleagues (17). It has subsequently been adapted for use in children, with modifications required by the unique characteristics of the child's skull, which is thinner. Fixation pins must be carefully placed, with attention paid to both location and depth of insertion. For thoracolumbar and lumbar fractures, nonsurgical

management with a thoracolumbosacral orthosis (TLSO) may be used either in place of or in addition to surgical stabilization (18).

Use of Steroids

Various studies of the efficacy of the uses of methylprednisolone in acute SCI were conducted during the 1980s. The National Acute Spinal Cord Injury Study 2 (NACSIS 2) was published in 1990 (19), with the conclusion that patients with acute SCI treated with high-dose methylprednisolone in the first eight hours after injury had better neurologic outcome than did those treated with placebo or naloxone. However, this was an adult study, with only 15% of patients being under 19 years of age and the youngest being 13 years old. Data are lacking in the pediatric population.

Respiratory Function

Most children with SCIs have impairment of normal respiratory function because of their injuries, even in the absence of other trauma causing pulmonary problems. The basic muscles of respiration are the diaphragm, intercostal muscles, abdominal muscles, and neck accessory muscles. Any SCI that weakens one or more of these muscles impairs respiration. The child with weak or absent diaphragm function needs ventilatory support. If the diaphragm is functional but intercostals and abdominal muscles are weak or nonfunctional, the child will need assistance with coughing and may need ventilator support during respiratory illnesses or during sleep. If the child only has weakness of the abdominal muscles, assistance with coughing may be the only respiratory support needed.

All children with acute SCIs should have respiratory function evaluated. At the very least this evaluation should include chest radiographs and measurement of oxygen saturation and end tidal carbon dioxide or arterial blood gases. If the child is able, vital capacity and inspiratory and expiratory forces should be measured on a daily basis until the child is medically stable. Because the child with SCI has restrictive respiratory dysfunction (so-called bellows failure), not lung disease, the earliest pulmonary abnormality will be hypercarbia, not hypoxia. End tidal carbon dioxide measurement is a simple noninvasive way to follow this, and may be used for outpatients as well as for inpatients.

Urinary Function

Most children with acute SCIs have neurogenic bladders. These are initially in spinal shock or flaccid, and may subsequently become spastic or dyssynergic. Flaccid bladders need to be drained either continuously or intermittently. Because indwelling catheters are associated with infections, the child should be converted to a clean intermittent catheterization program as soon as there is no medical reason to have continuous monitoring of urine output.

Gastrointestinal Function

After acute SCI, the gastrointestinal tract usually stops functioning initially, thus requiring the use of nasogastric suctioning. Once the ileus is resolved and the child is taking enteral feeding, a bowel program should be instituted, with the ultimate goal of continence without impaction. The consistency of the stool is normalized through the use of fluids, fiber, and medications, as needed. Evacuation is assisted through the use of digital stimulation or oral or rectal medications.

Fluids and Nutrition

Careful attention must be paid to fluid balance and nutrition in the child with an acute SCI. There must be a balance between enough fluids for hydration and to prevent constipation and not so much that intermittent catheterization must occur too frequently to prevent the bladder from becoming overdistended.

To promote healing, the child must also receive adequate nutrition. A common standard is to start some form of nutrition within 24 hours of injury. Typically, this is parenteral nutrition initially, followed by either oral or tube feedings when the ileus is resolved. For some children who have the ability to eat orally, refusal to eat may be the only way they have of refusing treatment, so nutritional intake should be closely monitored during the acute and rehabilitation hospitalizations.

Rehabilitation

Rehabilitation of the child with spinal cord injury is a lifelong process that starts soon after injury. It does not start and end with admission to and discharge from a rehabilitation unit, and this must be made clear to the patient and family. Goals of rehabilitation will be dependent upon a number of factors, primarily the patient's age, level of injury, and amount of neurologic recovery. Rehabilitation of the child with SCI is comparable to rehabilitation of the child with any other acute change in function, usually with less need for cognitive rehabilitation. The entire rehabilitation process should focus on the whole child in the context of his or her family and community, and be performed by a rehabilitation team of professionals that focuses on the needs of children. It should be noted that on occasion, some older children may be more appropriate for an adult rehabilitation service, while some young

adults or people with cognitive impairment may be better served on a pediatric rehabilitation service. When older children are treated on an adult service, it is important that the appropriate pediatric, social, and education services be available.

Goals for rehabilitation should include maintenance or attainment of good health and prevention of secondary complications, while promoting maximal and age-appropriate functional independence. Focus of rehabilitation can range from primarily family education (eg, C4 or higher complete tetraplegia with ventilator dependence) to primarily complete patient functional independence (eg, T10 complete paraplegia). The goals of rehabilitation will change as the child matures. Table 11.1 lists expected functional goals for levels of spinal cord injury.

Mobility

Mobility for the child with SCI begins with the process of learning to sit again. Sitting is compromised as both a result of the lack of neurologic control of the trunk related to the SCI, impairments in the autonomic nervous system, and physiologic adaptations to deconditioning during the acute hospitalization. Hypotension and syncope can result. Support hose, wrapping of the lower extremities with elastic bandages, and abdominal binders may help maintain blood pressure. Progression to sitting is a gradual process and should be started as soon as possible to minimize deconditioning. Short periods of sitting as tolerated can be done multiple times during the day, gradually increasing the duration of the time up. Early use of a power or manual wheelchair (as appropriate) is encouraged. It is important to monitor insensate skin as the duration of time up increases to minimize the development of pressure ulcers. Patients will begin working on bed mobility, rolling in bed, and transferring from the bed to the wheelchair. As the patient improves, more advanced transfers will be worked on.

Standing and walking, either with orthoses or independently, will be done as appropriate. Table 11.2 shows mobility guidelines from recommendations by

11.1 Mobility Guidelines

LEVEL OF INJURY	AGE	GOALS	ORTHOTIC OPTIONS
C1–4	Bracing available from age 1 year–prepuberty No standing after puberty	Standing	Prone and supine standers (stationary standers)
C4–7	Encourage from ages 1–5 years Available from age 5 years- prepuberty	Static standing and mobility	As above plus parapodiums/ swivelwalkers/mobile stamders
T1–5	Encourage ages 1–10 years after rehabilitation goals are met increase upper extremity strength/endurance); if surgery is performed, intensive gait training available postoperatively	Standing and household ambulation	As above plus RGO
	Ages 11–21 years need to meet criteria: 6 parallel bar pushups; 25 wheelchair pushups; transfer level heights; <20° of hip flexion contracture; <15° of ankle plantarflexion contracture.		
T6–12 and L1	Strongly encourage in ages 1–10 years	Household and limited community ambulation	Same as above
L-4	Strongly encourage for all ages	Community ambulation	Above plus HKAFOs, KAFOs, AFOs
L-5-S1	Strongly encourage for all ages	Community ambulation	Include AFOs, GRAFOs; strongly encourage for joint protection

AFO, ankle-foot orthosis; GRAFO, ground reaction ankle-foot orthosis; HKAFO, hip-knee-ankle-foot orthosis; KAFO, knee-ankle-foot orthosis; RGO, reciprocating gait orthosis.

Source: Adapted from Betz RR, Mulcahey MJ, eds. *The Child with a Spinal Cord Injury*. Rosemont, IL: American Academy of Orthopaedic Surgeons, 1996:849.

11.2 Functional Independence After Spinal Cord Injury

	LEVEL OF INJURY				
ACTIVITIES	C1–4	C5	C6	C7	PARAPLEGIA
Feeding	N	A	Y	Y	Y
Dressing UE	N	A	Y	Y	Y
Dressing LE	N	A	A	A	A
Bathing	N	N	N	Y*	Y
Bladder	N	Y*	A	Y	Y
Bowel	N	N	A	Y	Y
Rolling in bed	N	N	Y*	Y	Y
Transfers-level	N	N	Y*	Y	Y
Manual wheelchair	N	Y*	Y	Y	Y
Power wheelchair	Y	Y	Y	X	X
Driving	N	Y*	Y	Y	Y

N, not independent; Y, independent; A, independent with assistive devices.Y*, may be independent, but not expected; X, not usually needed; UE, upper extremities; LE, lower extremities.

the American Academy of Orthopedic Surgeons and Shriner's Hospitals for Children. It should be noted that often younger children with high lumbar or thoracic paraplegia may be ambulators with appropriate bracing. As with paraplegia from myelodysplasia, it is not unusual for these children to become more wheelchair-dependent as they reach adolescence. Ambulation at these levels is quite energy-inefficient, while use of a wheelchair is very efficient. This later use of a wheelchair should be discussed with the patient and family well before it occurs so that it is seen as the expected path and not as a failure of the patient or family.

SELF-CARE AND ACTIVITIES OF DAILY LIVING

By the time children are 5 years old, they are independent in the majority of self-care activities with supervision. Regaining this independence after it has been lost because of SCI is of utmost importance, especially to adolescents and preadolescents. The first step in this process is allowing some control over the environment in the rehabilitation unit. This may be as simple as a remote control for the television and an accessible call system to alert the nursing staff. A variety of systems are available, including "sip and puff" systems, head switches, mouth switches, and large buttons. It is important that the family try to promote the child's independence as well, and they must be encouraged to give the patient a certain degree of freedom and independence.

Relearning self-care should follow an orderly pattern, but may begin with the activity in which the child is most interested, often self-feeding. Activities that must be relearned include dressing, bathing, hygiene, feeding, transfers, writing, computer skills, and leisure activities. For young children, teaching these activities may need to be incorporated into games and play activities. Children with high tetraplegia may not be expected to manage their own self-care needs, but should be taught how to direct caregivers to perform various activities.

Cognition

It is important to assess cognition during the rehabilitation of the child with SCI. Just as MVCs are the primary cause of SCI, they are also the primary cause of traumatic brain injury (TBI). Any force significant enough to cause a spinal cord injury can also cause a TBI. Any child who has had an SCI should also be at least screened for a TBI. These screenings may also be useful in assessing possible hypoxic injury in ventilator-dependent children (20).

Bladder Management

After SCI, most patients develop a neurogenic bladder. While still in spinal shock, this tends to be a hypotonic bladder, but as spinal shock resolves, the bladder often transitions to a spastic bladder. Cauda equina syndrome and damage to the conus medullaris may result in a flaccid/hypotonic bladder. In the acute period, an indwelling catheter is typically placed to drain the bladder. This protects the bladder and kidneys, and allows close management of fluid status. While easy to manage, long-term use of indwelling catheters may lead to increased risk of urinary tract infection, shrinking of the bladder, stretching of the sphincters, and breakdown of the urethra.

The goal of bladder management is to gain continence of the urinary bladder, promote independence, minimize urinary tract infections, and protect the kidneys. Voiding pressures need to be maintained less than 40 cm H_20 to minimize the risk of ureteral reflux. During the acute phase, baseline evaluations of renal and urinary function need to be performed. These include blood urea nitrogen levels, serum creatinine, urinalysis, urine culture, and renal ultrasound or intravenous pyelogram. When the patient is out of spinal shock, they should undergo urodynamic testing.

Clean intermittent catheterization or intermittent self-catheterization is the method most commonly used today for bladder management after SCI. Numerous studies have shown its efficacy and safety for long-term management of the neurogenic bladder. Self-catheterization is easier for males and more problematic for females. Mirrors are frequently used by females to better visualize the urethral opening.

For those who cannot independently manage intermittent self-catheterization, external sphincterectomy may be considered for continuously draining the bladder, but this is rarely recommended in children because it destroys any chance for urinary continence when the child is older. It is also rarely recommended in females, as there is no good external collecting device. Condom catheters are commonly used in males after sphincterectomy. Complications of external sphincterectomy may include penile erosions from the condom catheter, need for reoperation, and erectile dysfunction.

A surgical procedure such as the Mitrofanoff procedure may be used to ease self-catheterization. This creates a stoma in the abdominal wall, typically through the umbilicus, which allows easier accessibility for catheterization. This is a major surgical procedure and should be performed by an experienced pediatric urologist. It should not be performed during the initial rehabilitation period, but later, after the child has had an opportunity to live at home. Reports of outcomes of this procedure have come from Shriners Hospitals for Children (21,22) and report relatively high satisfaction with the procedure and improved level of independence.

Various medications have been used in the management of the neurogenic bladder. In addition to treating urinary tract infections, antibiotics are sometimes used for prophylaxis with a catheterization program or treating asymptomatic bacteriuria. Recently, Clarke et al (23) completed a randomized trial of prophylactic antibiotics in 85 children with neurogenic bladder. They noted a six times higher incidence of urinary tract infection (UTI) in subjects treated with antibiotic prophylaxis compared to those without antibiotics. This was thought to be a result of bacteria developing antibiotic resistance. Schlager's group (24) investigated the use of nitrofurantoin to clear asymptomatic bacteriuria. Approximately 70% of subjects had asymptomatic bacteriuria, which was not cleared by nitrofurantoin. While there was a change in type of bacteria, it resulted in the growth of resistant organisms. At this time, it is not clear that antibiotics should routinely be used for neurogenic bladder, and use may increase the risk for resistant organisms. Cranberry juice is commonly recommended to prevent urinary tract infections, though it has not been shown to be effective in children (25).

Anticholinergic agents are commonly used to relax the urinary bladder, which results in a larger bladder capacity and decreased bladder pressures. Commonly used oral agents are oxybutynin, tolterodine, imipramine, and hyoscyamine. Side effects include dry mouth, decreased sweating, blurred vision, heat intolerance, and constipation. As children with spinal cord injuries, especially cervical levels, may have impaired thermal regulation, special caution must be used regarding anticholinergics and hot environments. Oxybutynin has been used intravesically to relax the bladder directly and avoid systemic side effects. The tablet is crushed, suspended in distilled water, and instilled in the bladder after catheterization. This practice is particularly useful where environmental temperatures are high and children wish to pursue outdoor activities.

In recent years, botulinum toxin A has been used as an intravesicular injection to decrease bladder tone. This was initially evaluated in 2000 and has been increasingly used in Europe, less so in the United States (26,27). Injections seem to last, on average, 9–11 months and are effective with repeat injections (28). Botulinum toxin type A may also be used to relax the external urinary sphincter in a dyssynergic bladder (29).

Neurogenic Bowel

With the loss of neural control, the gastrointestinal tract loses voluntary control, and peristalsis slows. Stiens

and associates reviewed the anatomy, physiology, and management of the neurogenic bowel. A program to control incontinence while preventing impaction must fit into the child's daily life. Factors to consider are premorbid bowel function, timing, consistency, frequency, and volume of bowel movements. The new bowel regimen should duplicate, as closely as possible, the premorbid patterns. If possible, bowel movements should be timed shortly after a meal to take advantage of the gastrocolic reflex. It is often more practical to try to time this after the evening meal, as the child is likely to be home and have more time to manage the bowel movement. Factors to be considered in the new program are diet, physical activity, equipment, oral and rectal medications, and scheduling. The diet should contain adequate fluid and fiber to provide sufficient bulk to facilitate transit through the gastrointestinal tract. Table 11.3 summarizes commonly used medications for bowel programs in SCI. Young children may only need digital stimulation or no special program to evacuate completely. Older children may, likewise, need only digital rectal stimulation to evacuate completely, but, more commonly, one or more oral or rectal medications are necessary.

Sometimes, bowel continence cannot be attained just with medications, and surgical intervention may be necessary, especially for those prone to constipation or impaction. The Malone procedure or antegrade continence enema (ACE) creates a stoma to allow antegrade use of enemas to improve bowel evacuation. This procedure has been shown to be effective in improving continence in SCI (30).

RESPIRATORY FUNCTION

Although acute pulmonary problems may not be as frequent during rehabilitation as during the initial phase

11.3 Bowel Medications

MEDICATION	EFFECTS	NEGATIVE EFFECTS
Bulk-forming agents Psyllium (Metamucil, Fibercon, Citrucel, Perdiem)	Absorb water to keep stool formed and prevent dry, hard stool	Bloating, flatulence
Stool softeners Docusate (Colace, Surfak)	Allows water to enter stool	Diarrhea, liquid form tastes bitter and is poorly tolerated
Mineral oil	Lubricant	Interferes with absorption of fat-soluble vitamins, causes lipid pneumonia after aspiration
Stimulants Senna (Senokot) Bisacodyl (Duicolax)	Increases intestinal motility, takes 6–12 hours to work Increases intestinal motility	Diarrhea, cramping Diarrhea, cramping (less with rectal suppositories)
Saline laxatives Milk of Magnesia Magnesium citrate Saline enemas (Fleet's)	Draws water into gut to stimulate colonic motility Stimulates colonic motility, used for complete bowel evacuation Acts to evacuate distal colon	Diarrhea Large volume, tastes bad, may cause electrolyte imbalance Cramping, may cause electrolyte disturbance
Hyperosmolar Lactulose, sorbitol Polyethylene glycol (Miralax) Glycerine suppositories	Draws fluid into intestine Draws fluid into intestine, used for complete bowel emptying Irritant	Diarrhea, cramping, flatulence Cramping, diarrhea
Prokinetic agents Metaclopramide (Reglan)	Affects neurotransmitters to increase gastrointestinal motility, including gastric emptying antiemetic Promotes gastric emptying	Interacts with many drugs, cardiac arrhythmia Behavior problems
Rectal agents Therevac mini-enemas Carbon dioxide suppositories (Ceo-Two)	Triggers colonic peristalsis Causes rectal distention	

after SCI, close attention should be paid to pulmonary status, especially in children who are younger and less able to communicate and in those with tetraplegia, high paraplegia, or more complete lesions. Though children with lower cervical and thoracic lesions have full diaphragmatic innervation, complete or partial paralysis of the abdominal wall and accessory respiratory muscles will weaken the cough and clearance of pulmonary secretions. Clinical symptoms of respiratory problems often develop long before radiologic or laboratory evidence is present. The child should be carefully watched for changes in secretions or cough, shortness of breath, headache, changes in mental status, sleepiness, and snoring. Presence of morning headache should be assumed to be a sign of hypercarbia and promptly investigated. Routine monitoring of pulmonary status during rehabilitation should, at the least, include daily auscultation, measurement of end-tidal carbon dioxide tension and transcutaneous oxygen saturation, and measurement of vital capacity and maximal inspiratory and expiratory forces in all children with quadriplegia and infants and young children with high paraplegia. Consideration should be given to monitoring oxygen saturation overnight in children with complete quadriplegia because some studies have found that a high percentage of adults with complete quadriplegia have frequent nocturnal desaturations (31–33). Prevention of problems may include percussion and postural drainage, assisted cough techniques, respiratory muscle training, pneumococcal immunization and yearly influenza vaccines, adequate nutritional status, and a cardiopulmonary fitness program. An abdominal binder or thoracolumbrosacral orthosis may be beneficial by providing support to the abdominal muscles.

NUTRITION

Adequate nutrition is necessary to promote healing of injuries and provide energy to participate in the rehabilitation process. For many children, refusal to eat may be present, either because of lack of appetite or because this may be the only activity over which they have any control. Loss of the sense of smell may accompany some injuries, also contributing to anorexia. Nutrition must become a non-negotiable issue during rehabilitation. If the child is unable or unwilling to eat, short-term use of nasogastric tube feedings should be considered. If the inability to eat continues longer, the placement of a gastrostomy tube should be considered. Once a child has finally begun to eat, care must be taken that he or she not overeat and thus become overweight. No calorie guidelines are available for children with SCI, but careful monitoring of weight can assist in determining the correct level of calories necessary for growth without promoting obesity.

SKIN

Pressure ulcers are a common complication of pediatric SCI and are caused by pressure, shear, and friction, with moisture being a complicating factor. Ulcers cause a huge burden in terms of time lost from school and other activities, cost, and psychological distress. Prevention is clearly a better solution than any treatment. The basis of prevention is thorough education of the child and family about pressure relief, avoiding moisture, and treatment of ulcers in the earliest stage. Data from Model SCI Care Systems in 2006 show that 33.5% of patients developed ulcers while still hospitalized, including 53.4% of those with complete tetraplegia, 39% of those with complete paraplegia, 28.7% of those with incomplete tetraplegia, and 18.3% of those with incomplete paraplegia (34). Fifteen to twenty percent of those seen for annual examinations developed ulcers per year during the first five years after injury. Although these figures may be less in children, ulcers nonetheless are costly. Various systems of classification are used for pressure ulcers (Tables 11.4 and 11.5).

Large pressure ulcers may not heal with the relief of pressure for long periods, and surgery may be necessary. Various types of closures include linear closure and several types of flaps, which are well detailed by Apple and Murray (35).

AUTONOMIC DYSREFLEXIA

Autonomic dysreflexia (AD) is dysfunction of the autonomic nervous system after SCI at or above T6. As a result of noxious stimuli below the level of injury, there is increased sympathetic activity leading to vasoconstriction below the level of injury and hypertension. The central nervous system response is vasodilatation above

11.4 Shea Classification of Pressure Ulcers

GRADE	DESCRIPTION
1	Red area or ulcer of epidermis or into epidermis
2	Full dermis thickness to subcutaneous fat
3	Fascia and muscle exposed
4	Bone visible
5	Large cavity through a small sinus

Source: Adapted from Bergman SB, Yarkony GM, Stiens SA. Spinal cord injury rehabilitation: Medical Complications. *Arch Phys Med Rehabil.* 1997;78:553.

11.5 National Pressure Ulcer Advisory Panel Classification

GRADE	DESCRIPTION
I	Nonblanchable erythema
II	Partial skin loss of epidermis, dermis
III	Full-thickness skin loss
IV	Damage through fascia, muscle, or bone

Source: Adapted from Yarkony GM. Pressure ulcers: Classification and overview. In: Betz RR, Mulcahey MJ. eds. *The Child with a Spinal Cord Injury*. Rosemont, IL: American Academy of Orthopedic Surgeons, 1996.

the level of injury, with increased vagal tone and bradycardia. Symptoms of AD include pounding headache, sweating above the level of the lesion, red splotches on the face and neck, and nasal congestion. Bradycardia may be present. Inciting factors are bladder and bowel distention and rapid change in position from sitting to supine. Urinary tract infection, renal or bladder stones, and suppository or enema insertion may also be inciting factors. AD can present as an acute emergency, more commonly in older adults than in children, who are better able to withstand extreme hypertension.

Treatment of AD consists of relief of inciting factors. The child is immediately placed in the sitting position, and the bladder is emptied. The child should be examined for other potential noxious stimuli, such as tight clothing or pressure sores. Most episodes of AD resolve with these treatments. If a rectal examination must be done, this may exacerbate the AD and should be done with the use of local anesthetic on the glove. If AD persists, nifedipine should be administered sublingually. An older treatment is nitroglycerine paste, which can be wiped off the skin, terminating its action once the hypertension resolves. Prevention of AD consists of effective bowel and bladder management programs.

Wheelchair tetraplegic athletes have been known to induce AD ("boosting") to improve their athletic performance. Performance is improved by the increased sympathetic tone, shunting blood away from the viscera, thus improving cardiac output. AD can be induced by maintaining a full bladder or using a noxious stimulus (eg, a tack) below the level of injury. Boosting is dangerous, and thus is banned in wheelchair athletics.

HYPERCALCEMIA

As discussed previously, hypercalcemia is most likely to occur in adolescent boys in the first two to three months after SCI. Serum calcium should be routinely followed throughout the rehabilitation inpatient course, and treatment with fluids, furosemide, and calcitonin, as described previously, should be instituted.

DEEP VENOUS THROMBOSIS

Deep venous thrombosis (DVT) and pulmonary embolism are common, potentially life-threatening complications in SCI. Although DVT is somewhat less common in prepubertal children, it still does occur. The most common time of occurrence is during the first few weeks after the SCI. Recommendations for prophylaxis against DVT in pubertal children include low-dose heparin or low-molecular-weight heparin and calf compression pumps during the rehabilitation hospitalization. Late-occurring DVT most commonly occurs with increased immobilization related to illness or surgery.

Symptoms of DVT include a swollen, warm extremity, with or without fever. If the child has sensation, this may be accompanied by pain. Differential diagnoses include cellulitis, fracture, reflex sympathetic dystrophy, and heterotopic ossification. Diagnosis is confirmed by Doppler ultrasound. If the ultrasound is negative and the index of suspicion for DVT is high, a venogram or MR imaging may be necessary. Plain radiographs should be obtained, especially in prepubertal children and in those whose SCI occurred more than three months previously to rule out fractures. Once a DVT is confirmed, treatment is bed rest until adequate heparinization is achieved to maintain the partial thromboplastin time 1.5 to 2.5 times control values. Treatment should continue for three to six months. Complications of heparin and warfarin include bleeding for both and heparin-induced thrombocytopenia. Warfarin may interact with many medications, and the patient and family should be fully educated about this if warfarin is to be continued after hospital discharge.

TEMPERATURE REGULATION

Children with SCI above T6 frequently have problems with temperature regulation because of the loss of central control of sympathetic and voluntary muscles (36). They must thus dress according to the environmental temperature. Before investigating the source of hyperthermia or hypothermia, investigation should be made into the temperature of the environment where the child has been. Often, undressing the child or putting a blanket over the child is all that is necessary to treat the hyperthermia or hypothermia. For children who reside in areas with cold weather, the use of a Mylar space blanket to maintain

body heat is recommended for emergency situations. Baclofen withdrawal with resultant severe spasticity may cause extreme hyperthermia (37).

LATEX ALLERGY

Latex allergy is commonly seen in children with myelodysplasia and is now being recognized in children with SCI. A report states the incidence of latex allergy in children with SCI is 6% to 18% (38). Children and families should be educated about this potential problem and encouraged to avoid latex when possible. Latex allergy can lead to an anaphylactic reaction. Any child (and caregivers) with any type of latex allergy should be instructed on the use an EpiPen for emergency use.

SPASTICITY

Approximately 50% of children with SCI have spasticity, which tends to be more common in those with incomplete lesions (39). Management of spasticity has the goals of promoting function and preventing contractures and pain because of the spasticity. Simple measures include ranging, positioning, and the use of orthoses. Some patients and families think that spasticity is reduced with a daily passive standing program. If spasticity still interferes with function, medications may be considered. See Table 11.6 for a summary of common antispasticity medications.

Local spasticity may be treated with splinting or casting or, if severe, with the use of intramuscular botulinum toxin. If spasticity continues to be severe and generalized after physical measures are employed and medications are maximized, surgical management of spasticity should be considered. Selective dorsal rhizotomy has been used in the United States since the mid-1980s. Although usually performed in children with spasticity of cerebral origin, the same technique may be used in children with SCI who are at least six months postinjury.

A newer surgical technique is the implantation of a subcutaneous pump for continuous administration of baclofen into the intrathecal space (40). Potential complications seen with intrathecal baclofen include infection, catheter disconnection or blockage, seroma around the pump, cerebrospinal fluid leak, seizures, failure to respond to increasing doses of baclofen, and pump failure. Some deaths have been reported after implantation of baclofen pump in children with SCI (41).

Psychosocial Issues

The primary psychosocial issue during rehabilitation is funding for care, equipment, therapies, and environmental modifications after discharge from inpatient rehabilitation. While parents are dealing with these issues, they must also adjust to the new needs of their child and assist their child in adjusting. The child must adjust to the new function of his or her body and learn to reenter home, community, and school. Recreation therapy can be of great help in assisting the child learn to move about in the community, both from the physical and the psychosocial perspective.

EDUCATION AND VOCATION

While the child is relearning mobility and self care skills, he or she must also begin to resume school work.

11.6 Common Spasticity Medications

MEDICATION	SITE OF ACTION	SIDE EFFECTS
Baclofen (Lioresal)	Spinal cord-GABA receptor agonist	Sedation, nausea, seizures (especially with rapid withdrawal)
Diazepam (Valium)	Brain	Sedation, potential for substance abuse
Dantrolene Dantrium)	Muscle	Liver dysfunction, weakness
Tizanidine (Zanaflex)	Spinal cord	Sedation, nausea
Clonidine (Catapres)	Spinal cord	Hypotension (less with transdermal than oral), dry mouth, constipation
Gabapentin (Neurontin)	Central	Gastrointestinal
Botulinum toxin (Botox)	Local muscle	Weakness

Source: GABA, γ-aminobutyric acid.

Adaptations necessary in the school environment need to be addressed, including architectural barriers, attitudinal barriers, and how to function with different physical skills. The child may need new ways to access computers for school or something as simple as two sets of schoolbooks—one for home and one in each classroom—to ease the physical challenges of returning to school. School staff and students need to be educated about SCIs to the extent the child and family wish this to be done. Often, it is helpful for several members of the rehabilitation team to visit the school to discuss spinal cord injury and present a video of the child engaged in some common activities. If this can be a question-and-answer session for the other students and school staff, many misconceptions can be eliminated and school reentry eased.

EQUIPMENT AND ENVIRONMENT

Wheelchairs

Children with SCI are affected in many ways, and equipment and environment can lessen the impact of their disabilities and enable them to participate in age-appropriate activities. Age and level of injury will dictate the extent of changes needed in the environment and the type of equipment needed (Table 11.7). Infants and toddlers may be well served by usual infant/toddler equipment, although those who require mechanical ventilation may be well served by a twin stroller to accommodate all of the necessary equipment. As children approach 2 to 3 years of age, they need to be provided with a mobility device to allow them to explore their environment. This may be a riding toy, such as a hand tricycle or powered riding toy, or an appropriately sized wheelchair. Transportation in a vehicle will still require the use of an appropriate toddler car seat. For those children with tetraplegia or with medical problems that preclude the use of a manually propelled wheelchair, a power wheelchair may be necessary. Prior to prescribing such a device, various control systems should be tried to see if the child can learn to drive a wheelchair and which system best suits their needs. Prerequisites to learning to drive a power wheelchair include:

1. At least one repeatable motor movement to drive the chair (eg, hand, head movement)
2. Understanding of cause and effect (knowing that an action causes something to happen)
3. Understanding of directionality
4. Ability to follow simple commands (42)

Children as young as 18 months have been shown to have the ability to drive power wheelchairs (43). However, if they require complex controllers, such as chin control rather than hand controllers, they may need to be closer to 4 or 5 years of age. But you do not know if a child can use any specific controller until you have tried it.

School-age children and adolescents need increasingly more independence and typically travel greater distances, so they may need power mobility to allow for this independence. All children who will be transported in their wheelchairs in vehicles should have transit-ready wheelchairs that meet WC19 standards (44).

Orthotics

Orthotic management of the child with an SCI must consider the child's age, developmental status, and functional status as well as the physical features of their home and school environments. Orthotic options include orthoses for positioning as well as orthoses to enhance function in standing or ambulation. See Chapter 6 for a detailed discussion of orthotics. A recent study looked at ambulation in 169 children and youth with SCI. After a mean follow-up of 9 years, 56 of these patients were nonambulators, 17 were community ambulators, 42 were household ambulators, and

11.7 Mobility Equipment Options

AGE	LEVEL	MANUAL OR POWER	CONTROLLER	SPECIAL FEATURES
0–3 years	Paraplegia Tetraplegia	Stroller or riding toy Stroller	Hand	
3–10 years	Paraplegia Tetraplegia	Manual Manual and power	Hand, chin	Tilt, recline, vent tray, standing
>10 yrs	Paraplegia Tetraplegia	Manual Manual and power	Hand, chin	Tilt, recline, vent tray, standing

54 were therapeutic ambulators. Young age at injury and lower neurologic levels were positively associated with greater likelihood of ambulation (45).

Special Considerations in High Tetraplegia

Children with high tetraplegia (C1–C4 levels) all have some type of partial or complete respiratory dysfunction. Whether they require full- or part-time mechanical ventilation depends on their level and the completeness of their lesion. Some may be ventilated only at night via face mask, while others require tracheostomies and full-time ventilation. Issues unique to this population include increased risk of pulmonary infection, the developmental impact of being assisted by a machine for life support, and the impact of a tracheostomy on swallowing and communication. This group of patients also has more problems with maintaining blood pressure in the upright position and in accessing their environment. Wheelchairs should be "self-contained," with all necessary equipment carried.

Long-Term Follow-up

Children with SCI can expect to live a relatively long time (46) and thus will most likely be affected by complications related to growth that adults do not experience. These complications include contractures, which are most likely to occur during periods of rapid growth, hip subluxation, and scoliosis. A study at Shriners Hospital for Children in Philadelphia found that 93% of their patients who sustained SCI under the age of 10 years had hip subluxation as compared to 9% of those over 10 years old at the time of injury (47). While the sample size was small (only 62 patients total), this echos the impression of clinicians. Researchers at the same institution also looked at prevention of scoliosis in children with SCI. They found that bracing with a thoracolumbosacral orthosis before the scoliotic curve reached 20 degrees delayed the time to surgical correction of the deformity.

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12

Neuromuscular Diseases

Craig M. McDonald

Progressive acquired or hereditary neuromuscular diseases are disorders caused by an abnormality of any component of the lower motor neuron—anterior horn cell, peripheral nerve, neuromuscular junction (presynaptic or postsynaptic region), or muscle. While some neuromuscular diseases have pathologic abnormalities isolated to one anatomic region of the lower motor neuron, with primary or secondary changes in muscle, other neuromuscular diseases have been recognized as multisystem disorders. For example, myotonic muscular dystrophy may affect skeletal muscle, smooth muscle, myocardium, brain, and ocular structures; Duchenne muscular dystrophy gives rise to abnormalities of skeletal and cardiac muscle, the cardiac conduction system, and brain; Fukuyama congenital muscular dystrophy affects skeletal muscle and brain; mitochondrial encephalomyopathies may affect the mitochondria of multiple tissues.

Neuromuscular diseases may be acquired (eg, poliomyelitis, Guillain-Barré syndrome, myasthenia gravis, or polymyositis), but the most common etiology is genetic (eg, spinal muscular atrophy [SMA], Charcot-Marie-Tooth [CMT], congenital myasthenic syndrome, or Duchenne muscular dystrophy).

Tremendous advances have occurred in the past two decades in our understanding of the molecular genetic basis and pathophysiology of neuromuscular diseases affecting children and adults. Traditional approaches to the classification of neuromuscular disorders utilized clinical history, family history, clinical examination findings, electrodiagnostic findings, and histopathologic analysis of muscle and/or nerve biopsy specimens to provide clinical diagnosis. Molecular genetic advances have led to the discovery of specific genes for hundreds of neuromuscular disorders and have provided pathophysiologic explanations for phenotypically divergent disorders.

Appropriate rehabilitation management of neuromuscular diseases requires an accurate diagnosis. The clinician must be able to obtain a relevant patient and family history and perform focused general, musculoskeletal, neurologic, and functional physical examinations to direct further diagnostic evaluations. Laboratory studies include relevant molecular genetic studies in certain instances; however, specific genetic entities need to be strong diagnostic considerations, because these studies may be expensive and have limited sensitivity and specificity. Electrodiagnostic studies may help guide the acquisition of further diagnostic studies such as genetic studies and muscle and nerve biopsies. There has definitely been a trend away from the utilization of electrodiagnostic studies in the diagnostic evaluation of pediatric neuromuscular diseases. All diagnostic information needs to be interpreted, not in isolation, but within the context of relevant historical information, family history, physical examination findings, laboratory data, molecular diagnostic studies, electrophysiologic findings, and pathologic information, if obtained.

A skilled synthesis of all available information may provide the patient and family with a precise diagnosis or as accurate a diagnosis as is medically possible, prognostic information (if available for a specific entity), and anticipatory guidance for the near future. Knowledge of the natural history of specific neuromuscular disease conditions helps in the ongoing rehabilitative management of progressive impairments, disabilities, and handicap.

This chapter summarizes the diagnostic evaluation, natural history, and impairment profiles and rehabilitation management of childhood neuromuscular diseases.

DIAGNOSTIC EVALUATION IN NEUROMUSCULAR DISEASES

Neuromuscular Disease History

The common presenting chief complaints from parents or children with suspected neuromuscular disorders may include infantile floppiness or hypotonia, delay in motor milestones, feeding and respiratory difficulties, abnormal gait characteristics, frequent falls, difficulty ascending stairs or arising from the floor, and muscle cramps or stiffness. Teenagers with later-onset disorders may present with chief complaints of strength loss or decreasing endurance, falls, difficulty ascending stairs, exercise intolerance, episodic weakness, muscle cramps, focal wasting of muscle groups, breathing difficulties, or bulbar symptoms such as speech and swallowing difficulties.

Information should be obtained about the recent course of the chief complaint, specifically whether the process is getting worse, staying the same, or getting better. If strength is deteriorating, it is important to ascertain the rate of progression (ie, is weakness increasing over days, weeks, months, or years?). It is critical to determine whether the distribution weakness is predominantly proximal, distal, or generalized. It is also useful to identify factors that worsen or help primary symptoms. A history of twitching of muscles may reflect fasciculations. Tremor or balance problems may be due to distal weakness or superimposed cerebellar involvement.

Bulbar involvement may be identified if the individual has difficulty with chewing, swallowing, or speech articulation. Visual complaints (blurriness or diplopia) may indicate the presence of cataracts or possible involvement of extraocular musculature. Distal stocking glove or focal sensory complaints may be consistent with a peripheral neuropathy or focal nerve entrapment. A comprehensive past medical history and surgical history should be obtained. A history of recent illnesses should be carefully elucidated, including respiratory difficulties, aspiration pneumonias or recurrent pulmonary infections. In addition, such cardiac symptoms as dizziness, syncope, chest pain, orthopnea, or exertional complaints may indicate superimposed involvement of the myocardium. A review of pulmonary symptoms should be obtained. A history of weight loss may be due to recurrent illnesses, nutritional compromise, swallowing difficulty, or progressive lean tissue atrophy.

A detailed history regarding pregnancy (eg, quality of fetal movement or pregnancy complications) and perinatal problems (evidence of fetal distress, respiratory difficulties in the recovery room, need for resuscitation or ventilation problems in early infancy, ongoing respiratory difficulties, swallowing/feeding difficulties, and persistent hypotonia) should be obtained. Perinatal respiratory distress in the delivery room may be seen in acute infantile type I SMA, myotubular myopathy, congenital myotonic muscular dystrophy, congenital hypomyelinating neuropathy, infantile congenital myasthenic syndrome, transitory neonatal myasthenia, and severe neurogenic arthrogryposis.

History regarding the child's acquisition of developmental milestones should be ascertained relating to head control, independent sitting, crawling, standing with and without support, walking with and without support, fine motor prehension, bimanual skill acquisition (bringing objects to midline, transfer of objects), and language acquisition. Information regarding gait characteristics (toe walking, excessive lordosis, etc.), running ability, transitions from floor to standing, stair climbing, falls, recreational/athletic performance, pain or muscle cramps and easy fatigue, or lack of endurance may be important clues to the presence of a neuromuscular disorder. History regarding mental development, type of school, and school performance may be important indicators of superimposed central nervous system (CNS) involvement. For the older child, a detailed history regarding the age of onset of symptoms, paraprogression, distribution of weakness, presence of muscle cramps, fatigue, episodic weakness, presence of atrophy of fasciculations, performance in physical education, current and past ambulatory distances, ability to move from floor to standing, problems climbing stairs, and problems reaching overhead or dressing may all be important functional information.

A history of muscle cramps at rest or with exertion may be associated with a muscular dystrophy, metabolic myopathy, toxic myoglobinuria, inflammatory myositis, or other lower motor neuron disorders.

A thorough anesthetic history should be obtained. Malignant hyperthermia is associated with primary familial malignant hyperthermia, central core congenital myopathy, Duchenne muscular dystrophy (DMD), and Becker muscular dystrophy (BMD). Other neuromuscular disease (NMD) conditions occasionally

associated with malignant hyperthermia include Fukuyama congenital muscular dystrophy, limb girdle muscular dystrophy (LGMD), fascioscapulohumeral muscular dystrophy (FSHD), periodic paralysis, myotonia congenita, mitochondrial myopathy, and Schwartz-Jampel syndrome.

Family History

Suspicion of a neuromuscular disease warrants the ascertainment of a detailed family history and pedigree chart. Autosomal-dominant conditions may have pedigrees with multiple generations affected, with equal predilection to males and females. Typically, one-half of offspring within a pedigree are affected. In autosomal-recessive conditions, only one generation may be affected, with equal proportions of males and females. Proportionally, one-fourth of offspring are clinically affected. Parents in earlier generations may be normal, and the parents of affected children are presumptive heterozygote carriers of the condition. In many instances of autosomal-recessive inheritance, no other family members within the nuclear family unit are affected, making the confirmation of inheritance pattern difficult without a molecular genetic marker present or protein abnormality confirmed by immunohistochemistry techniques. In X-linked recessive conditions, males on the maternal side of the family are affected in approximately 50% of instances and females are carriers in 50% of instances.

Often, it is valuable to examine affected relatives who may be either earlier or later in the course of their neuromuscular disease relative to the affected child. In addition, medical records and diagnostic evaluations of affected family members should be reviewed and the diagnosis confirmed if possible. In some instances, the examination of a parent can help establish the diagnosis in an affected infant or child, as is frequently the case in myotonic muscular dystrophy 1 (MD1). In this disorder, genetic anticipation with abnormal CTG trinucleotide expansion of unstable DNA results in progressively earlier onset of the disease in successive generations with increasing severity.

In the case of dystrophic myopathies, a definitive molecular genetic or pathologic diagnosis, established in a sibling or close relative, may allow the clinician to establish the diagnosis in a child or adult based on clinical examination, easily obtained laboratory data such as creatine kinase, or molecular genetic testing, thus allowing the avoidance of further invasive testing such as muscle biopsy.

Physical Examination

Physical examination findings help focus further diagnostic evaluation, utilizing such tools as electrodiagnosis, molecular genetic testing and histopathologic analysis of biopsy specimens. All diagnostic information must be interpreted within the context of relevant clinical information. In many instances, a precise molecular genetic diagnosis is not medically possible. However, the accurate characterization of an individual patient within the most appropriate NMD clinical syndrome still allows the clinician to provide the patient and family with accurate prognostic information and anticipatory guidance for the future.

Specific aspects of the physical examination, relevant to the neuromuscular disease population, includes simple inspection for the presence of focal or diffuse muscle wasting or focal enlargement of muscles, as with the "pseudohypertrophy" seen in such dystrophic myopathies as Duchenne and Becker muscular dystrophy (Fig. 12.1). The increase in calf circumference in DMD is caused by an increase in fat and connective tissue rather than true muscle fiber hypertrophy in the gastrocnemius. Over time, the reduced bulk of musculature may be caused by more severe fiber loss in a more "active" dystrophic process affecting proximal musculature. Other neuromuscular disorders may show calf pseudohypertrophy, such as childhood-type acid maltase deficiency.

Focal atrophy of particular muscle groups may provide diagnostic clues to specific neuromuscular disorders, such as spinal muscular atrophy, Emery-Dreifuss muscular dystrophy, FSHD, and LGMD. Those with CMT, particularly those with type II axonal forms, demonstrate distal atrophy or "stork leg appearance" relatively early in the disease course. Palpable nerves in the cubital tunnel, posterior auricular region, or around the fibular head may be indicative of "onion bulbs" seen in hereditary demyelinating neuropathies such as CMT I subtypes or Dejerine-Sottas disease (CMT III).

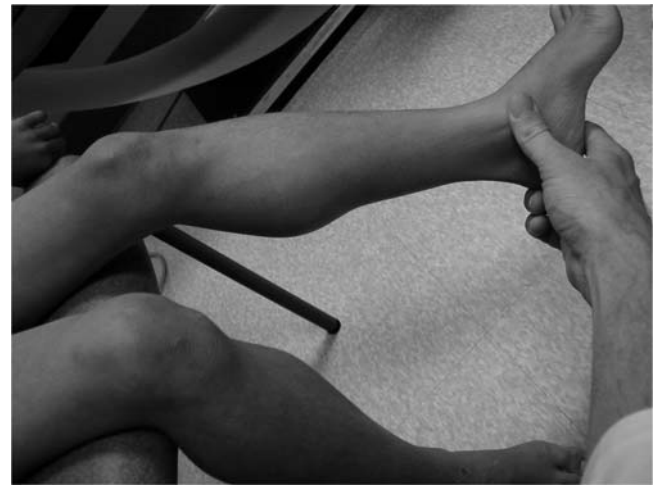


Figure 12.1 Calf pseudohypertrophy in a male with Duchenne muscular dystrophy.

Muscle fasciculations may be seen as nonspecific findings of a variety of lower motor neuron disorders. Fasciculations are particularly common in such lower motor neuron disorders as SMA. Distal fine tremor may be seen in a large proportion of CMT patients (30–50%), and in other patients with weakness such as SMA. "Polyminimyoclonus," another variant of muscle fasciculations characterized by a fine tremor of the fingers and hands, may be evident in SMA I and II.

Infants with NMD often show infantile hypotonia (Fig. 12.2), the differential for which is large (see chapter on pediatric electrodiagnosis).

A thorough general physical examination of cardiac, pulmonary, and gastrointestinal systems should be performed on all patients suspected of having a neuromuscular disease. Hepatomegaly may be seen in such metabolic myopathies as acid maltase deficiency (type II glycogenosis or "Pompe disease") and type III and IV glycogenosis. The skin should be evaluated for characteristic skin rashes and nail bed capillary changes if an inflammatory myopathy such as dermatomyositis is suspected. Keratosis pilaris is a characteristic skin rash seen in congenital muscular dystrophy with collagen VI deficiency (Fig. 12.3). Craniofacial changes and dental malocclusion are common in congenital myotonic muscular dystrophy, congenital myopathies, congenital muscular dystrophy, and type II SMA. A neurological examination should include a thorough evaluation of cranial nerve function, muscle tone, muscle strength, sensory and cerebellar function, and deep tendon reflexes. An assessment for the presence of percussion and grip myotonia (Fig. 12.4) should be performed in situations where a myotonic syndrome is suspected. Musculoskeletal examination will reveal the presence of limb contractures, deformities, and spinal deformity.

A

B

Figure 12.2 (A, B) Hypotonia in an 18-month-old child with spinal muscular atrophy.

Some neuromuscular disorders, such as congenital myotonic muscular dystrophy (congenital DM1), Fukuyama congenital muscular dystrophy, selected cases with mitochondrial encephalomyopathies, and a small proportion of Duchenne muscular dystrophy cases, may have significant intellectual impairment.

A thorough functional examination is essential in the diagnostic evaluation of a patient with suspected neuromuscular disease. This includes an evaluation of head control, bed/mat mobility, transitions from supine-to-sit and sit-to-stand, sitting ability without hand support, standing balance, gait, stair climbing, and overhead reach.

An evaluation of overhead reach is performed while examining the patient from the front and from behind in order to evaluate shoulder girdle weakness. Careful assessment of scapular winging, scapular stabilization, and scapular rotation is helpful in the assessment of patients with FSHD or other limb girdle syndrome. The scapula is stabilized for overhead abduction by the trapezius, rhomboids, and serratus anterior. Abduction to 180 degrees requires strong supraspinatus and deltoid muscles in addition to strong scapular stabilizers.

Patients with proximal weakness involving the pelvic girdle muscles may rise off the floor using the classic "Gower's sign," where the patient usually assumes a fou- point stance on knees and hands, brings the knees into extension while leaning forward the upper extremities, substitutes for hip extension weakness by pushing off the knees with the upper extremities, and sequentially moves the upper extremities up the thigh until an upright stance with full hip extension is achieved (Fig. 12.5). A Gower's sign is not specific to any neuromuscular condition, but may be seen in a variety of neuromuscular diseases, including DMD, LGMD, SMA type III, severe childhood autosomal recessive muscular dystrophy (LGMD II subtypes), congenital muscular dystrophy, congenital myopathy,

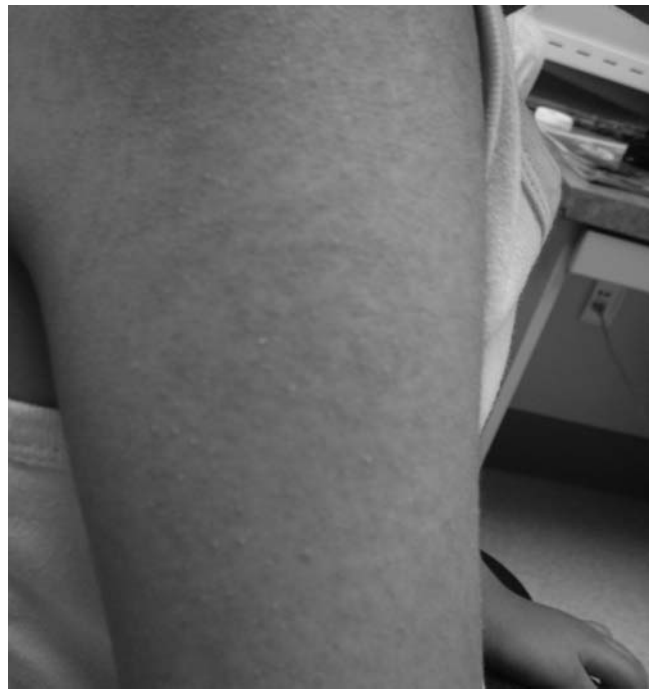
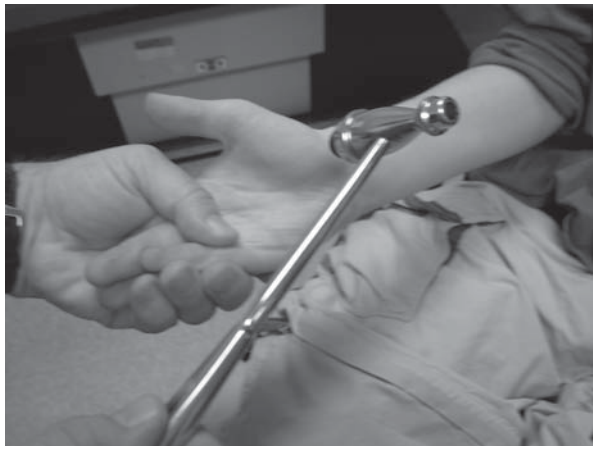
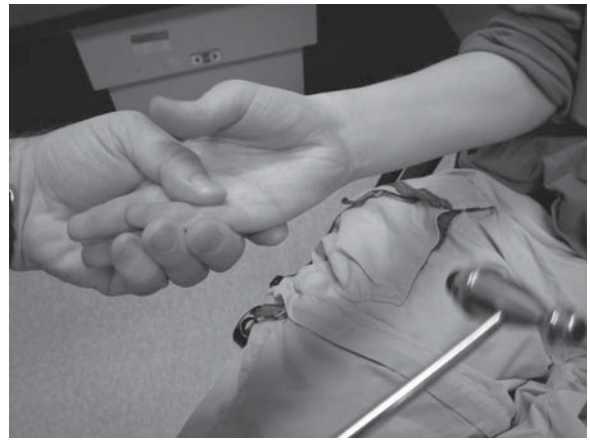


Figure 12.3 Keratosis pilaris skin rash on the extensor surface of the arm in Ullrich congenital muscular dystrophy with collagen type VI abnormality.

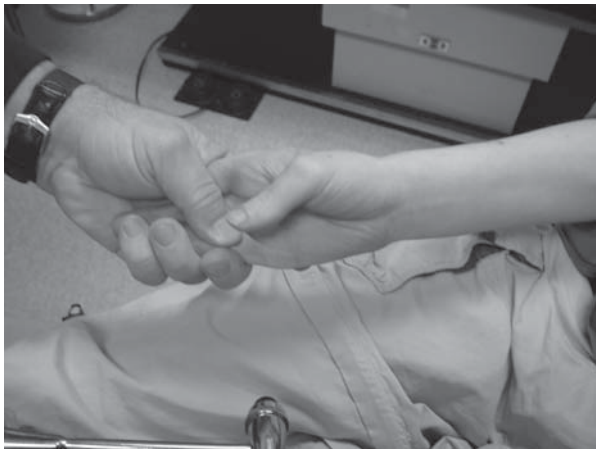


A



B

Figure 12.4 (A–C) Percussion myotonia of the thenar eminence.



C

myasthenic syndromes, severe forms of CMT (eg, CMT III and CMT IV), and in other neuromuscular disease conditions producing proximal weakness. Patients with proximal lower extremity weakness often exhibit a classic myopathic gait pattern (Fig. 12.6). Initially, weakness of the hip extensors produces anterior pelvic tilt and a tendency for the trunk to be positioned anterior to the hip joint. Patients compensate for this by maintaining lumbar lordosis, which positions their center of gravity/weight line posterior to the hip joints, thus stabilizing the hip in extension on the anterior capsule of the hip joint. Subsequently, weakness of the knee extensors produces a tendency for patients to experience knee instability and knee buckling

with falls. Patients compensate for this by decreasing stance-phase knee flexion, and they posture the ankle increasingly over time into plantar flexion. This produces a knee extension moment at foot contact and plantarflexion of the ankle during mid- to late-stance phase of gait, which helps position the weight line/center of gravity anterior to the knee joint (thus producing a stabilizing knee extension moment). Patients with DMD will progressively demonstrate toe walking with initial floor contact, with the foot contact increasingly moving forward onto the midfoot and finally the forefoot as they reach the transitional phase of ambulation before wheelchair reliance (see Fig. 12.7). Finally, weakness of the hip abductors produces a tendency toward lateral pelvic tilt and pelvic drop of the swing phase side. Patients with proximal weakness compensate for this by bending or lurching the trunk laterally over the stance-phase hip joint (Fig. 12.8). This produces the so-called "gluteus medius lurch" or Trendelenburg's gait pattern.

Patients with distal weakness affecting the ankle dorsiflexors and ankle everters and less severe proximal

A

B

C

D

Figure 12.5 (A–F) Gower's sign in a seven-year-old boy with Duchenne muscular dystrophy due to hip extension weakness.

E

F

Figure 12.5 ***Continued***

weakness (eg, CMT, Emery-Dreifuss muscular dystrophy, myotonic muscular dystrophy, FSHD, and other conditions) often exhibit a foot slap at floor contact with a steppage gait pattern to facilitate swing-phase clearance of the plantar-flexed ankle. Alternatively, these patients may clear the plantar-flexed ankle using some degree of circumduction at the hip or vaulting on the stance-phase side. Milder distal lower extremity weakness may become clinically evident by testing heel walking and toe walking.

Serum Laboratory Studies

Those neuromuscular diseases with inherent sarcolemmal muscle membrane injury often show significant elevations in transaminases, aldolase, and creatine kinase (CK). The CK enzyme catalyzes the release of high-energy phosphates from creatine phosphate. It occurs mainly in muscle and leaks into the serum in large amounts in any disorder involving muscle fiber injury. The MM fraction is specific to skeletal muscle. The CK value may be significantly elevated in early stages of DMD and BMD, with values up to 50–100 times normal. A normal CK value may help exclude DMD and BMD. Overlap in CK values occurs between DMD and BMD. Other forms of muscular dystrophy, such as Emery-Dreifuss muscular dystrophy (EMD), LGMD, FSHD, and congenital muscular dystrophy, may show moderate elevations in CK. However, in congenital muscular dystrophy, the CK value may be extremely variable, ranging from normal values to a fairly marked elevation. There is no close association between disease severity and CK values. In all dystrophic myopathies, the CK values tend to decrease over time, with increasing severity of the disease owing to progressive loss of muscle fiber and irreversible cell death. Thus, a 3-year-old with DMD may have a CK value of 25,000, while a 10-year-old with DMD may show a CK value of 2,000. Other conditions with significant elevations in CK may include polymyositis, dermatomyositis, acute rhabdomyolysis, and malignant hyperthermia. In many of the congenital structural myopathies, such as central core disease, nemaline rod myopathy, and fiber-type disproportion syndrome, a serum CK is likely to be normal or only mildly elevated.

CK levels have ranged from normal to elevated two to four times in SMA I and II. SMA III patients have also been found to have normal to slightly elevated CK values, with elevations generally to two to five times normal. A serum CK level greater than 10 times the upper limit of normal generally is exclusionary criteria for SMA and, in this setting, workup for other disorders such as inflammatory or dystrophic myopathies should be pursued. In a child with muscle weakness, a normal CK does not exclude a myopathy or other NMD conditions; however, a severely elevated CK is suggestive of but not diagnostic of a dystrophic myopathy or inflammatory myopathy.

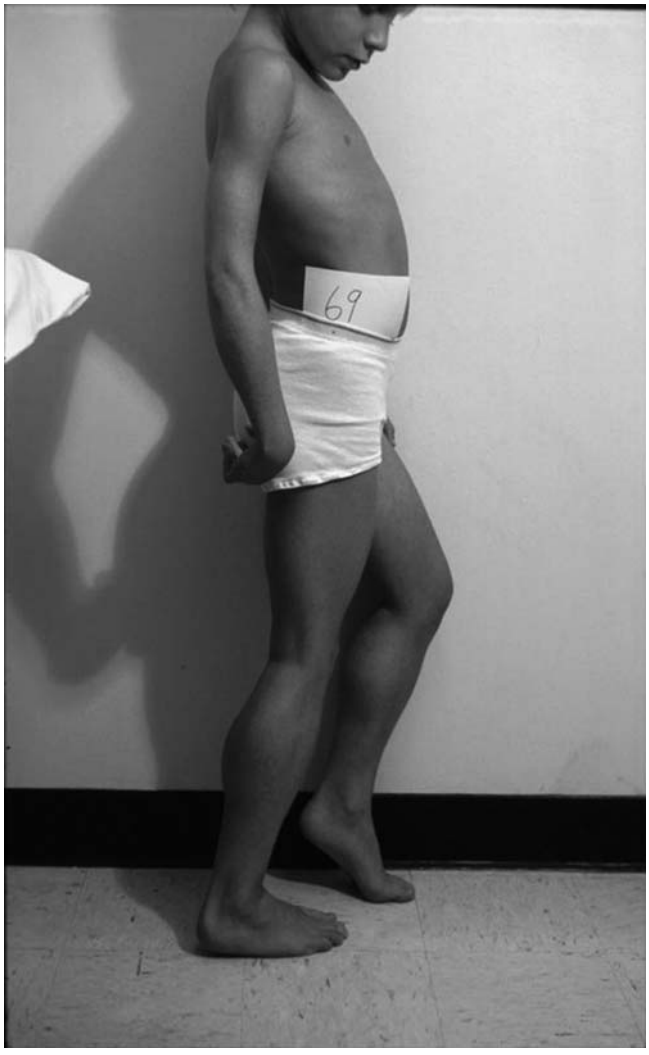


Figure 12.6 "Myopathic" stance in an eight-year-old male with Duchenne muscular dystrophy. Notice the lumbar lordosis to compensate for hip extensor weakness and primarily forefoot contact to compensate for knee extensor weakness.

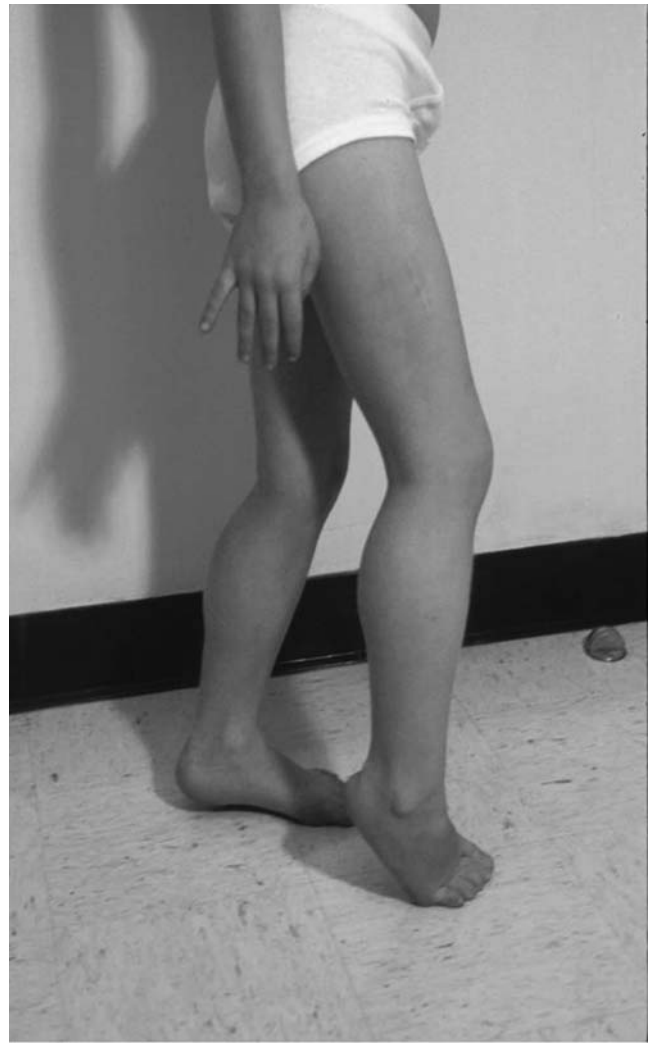


Figure 12.7 Toe walking with initial floor contact in the transitional phase of ambulation in Duchenne muscular dystrophy is, in part, a compensatory mechanism to maintain knee stability.

Lactate and pyruvate levels are useful in the evaluation of possible metabolic myopathy. The presence of a lactic acidosis may be seen in such mitochondrial encephalomyopathies as Kearns-Sayre syndrome, myoclonic epilepsy and ragged-red fibers (MERRF) and mitochondrial encephalomyopathy with lactic acidosis and strokelike episodes (MELAS). Whenever clinical evidence suggests a disorder of oxidative metabolism, blood lactate and pyruvate levels should be obtained. Arterial lactate values are a more reliable guide. Stable or reduced levels of lactate and pyruvate, with concomitany increases in ammonia with ischemic or nonischemic forearm exercise testing, suggests a mitochondrial dysfunction. In a setting of lactic acidemia, the lactate/pyruvate ratio may aid in the differential diagnosis. Children with suspected mitochondrial encephalomyopathy should be evaluated with cerebrospinal fluid (CSF) lactate and pyruvate levels, because these values are less subject to flux than are either venous or arterial values. The ischemic forearm test, initially utilized by McArdle, and the nonischemic forearm exercise test are widely used means of assessing muscle and aerobic metabolism in older, more cooperative patients (1,2).

Electrodiagnostic Studies

Nerve conduction and electromyography are an extension of the clinician's physical examination and a useful tool for the localization of lesions within the lower motor neuron. In addition, electromyogramy (EMG) and nerve conduction studies help guide further studies, such as muscle biopsy, by providing information about the most appropriate muscle site for biopsy. With spinal

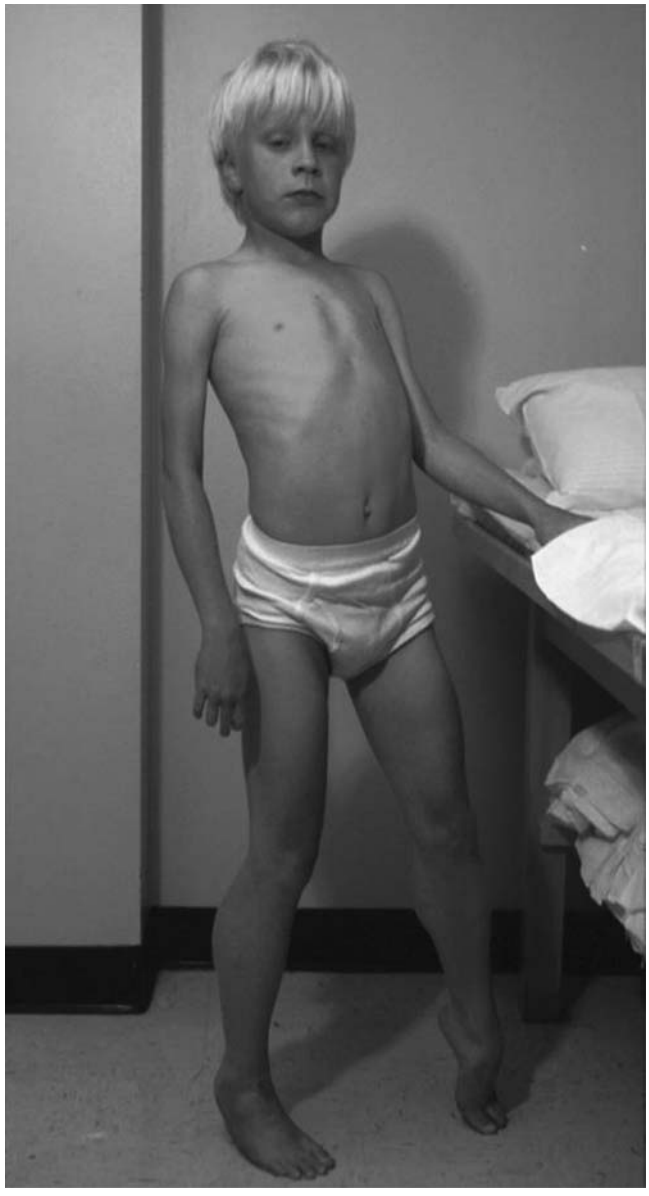


Figure 12.8 Trendelenburg or "gluteus medius" gait pattern in a male with Duchenne muscular dystrophy. Note the lateral lean over the stance side due to hip abductor weakness; ankle dorsiflexion weakness necessitates swing phase in circumduction for clearance.

muscular atrophy, an electrodiagnostic evaluation can allow the clinician to defer muscle biopsy and proceed with molecular genetic studies of the survival motor neuron gene. Electrodiagnostic studies in patients with CMT help to categorize the neuropathy as either primarily demyelinating or axonal, and such information may help focus subsequent molecular genetic analyses for a more cost-effective approach. In patients with suspected CMT and positive family histories with genetically confirmed diagnoses, the diagnosis of CMT may be confirmed in the clinic by a simple, reliable, and relatively inexpensive nerve conduction study.

A thorough discussion of the role of electrodiagnosis and neuromuscular disease is provided in the chapter on pediatric electrodiagnosis.

Molecular Genetic Studies

The application of molecular genetic techniques has resulted in enormous gains in our understanding of the molecular and pathophysiologic basis of many neuromuscular diseases. In addition, molecular genetic studies now aid in the diagnostic evaluation of the dystrophin-deficient muscular dystrophies (DMD, BMD), myotonic muscular dystrophy, predominantly proximal autosomal-recessive spinal muscular atrophy, Charcot-Marie-Tooth neuropathy (hereditary motor and sensory neuropathy), and a host of other neuromuscular disease conditions. The clinical application of molecular genetic studies is described in the following sections on specific neuromuscular disease conditions.

Muscle Biopsy Evaluation

Muscle Biopsy Technique

The two techniques for obtaining a muscle biopsy specimen include the traditional open biopsy and the needle biopsy (3,4). Either technique can be performed under local anesthesia; however, most clinicians in the United States use general anesthesia for open biopsies and local anesthesia for needle biopsies. There may be some disruption in the architecture of the tissue with needle biopsy technique, which can affect the evaluation of histologic examination and electron microscopy. Immunocytochemistry analyses, such as Western blot, and metabolic studies do not require strict maintenance of the muscle cellular architecture.

Muscle Biopsy Site Selection

Selection of the muscle is based on distribution of muscle weakness found clinically in addition to electrodiagnostic findings, if obtained. In a dystrophic myopathy, the muscle biopsy should be clinically affected, but not so severely affected that it is largely replaced by fat and connective tissue with minimal residual muscle fiber present for evaluation. The insertional activity on EMG or muscle imaging studies can be helpful in this respect. Sufficient normative information about proportional fiber type and fiber diameter should be available with age-appropriate norms. A diagnosis of congenital myopathy with fiber-type disproportion cannot be made without careful consideration of the normal fiber-type predominance in a given muscle. For example, the vastus lateralis is two-thirds type II fibers (with equal proportions of

type IIa and type IIb fibers) and one-third type I fibers. The anterior tibialis, on the other hand, contains a predominance of type I fibers, and the anconeus is mostly type I fibers. In addition, some muscles, such as the quadriceps and biceps, have longitudinally running fibers that facilitate orientation of the specimen for preparation of cross-sectional slices. The gastrocnemius and middle deltoid muscles, on the other hand, may be difficult to orient because fibers run in different planes. The posterior deltoid is preferred over the middle deltoid. Muscles that have undergone recent needle electromyographic evaluation should be avoided as muscle biopsy sites because of the possibility of cellular changes in the muscle fiber secondary to the needle study.

For routine diagnostic studies, the vastus lateralis muscle in the lower extremity and the triceps, biceps, or posterior deltoid in the upper extremity are often preferred. When proximal muscles are severely affected or only distal muscles are involved, the extensor carpi radialis or anterior tibialis muscles are often biopsied.

Histology/Histochemistry

The histopathologic study is likely to provide information on whether the basic disease process is primarily a myopathy or a neurogenic process. In some instances, the diagnosis of specific disorders (such as dystrophic myopathy or inflammatory myopathy) is delineated. When analyzing paraffin sections, basic pathologic reactions of muscle may include fiber necrosis, central nuclei indicative of regeneration, abnormalities of muscle fiber diameter (atrophy, hypertrophy, abnormal variation, and fiber size), fiber splitting, vacuolar change, inflammatory infiltrates, and proliferation of connective tissue/fibrosis. A dystrophic myopathy frequently is characterized by the presence of normal fibers, hypertrophied fibers, degenerating fibers, atrophic fibers, regenerating fibers, and connective tissue and fatty infiltration. Neurogenic changes may be characterized by small or large groups of atrophic fibers, with or without target fibers, and frequently by hypertrophy of the nonatrophic fibers. Pyknotic nuclear clumps, target fibers, and darkly staining angulated fibers are consistent with a neurogenic process. Red-rimmed vacuoles suggest inclusion-body myositis. Ragged red fibers are consistent with a mitochondrial myopathy. Perifascicular atrophy is consistent with dermatomyositis.

Frozen sections can be assessed with standard H & E NADH, ATPase, and trichrome stains. Other stains include include PAS (for glycogen), Oil Red-O (for lipid), congo red (for amyloid), acetylcholinesterase (for motor end plates), myophosphorylase (for McArdle's), acid phosphatase (for type II glycogenosis), cytochrome oxidase C, succinic dehydrogenase (mitochondrial enzymes), and specific immunostains for dystrophin and sarcolemmal membrane associated proteins.

A variety of histochemical stains, including NADH stains and ATPase stains, at different pH values can be used to differentiate fiber types (types I, IIa, IIb, IIc). Based on the histochemical analyses, information is obtained about pattern of fiber types (eg, normal predominance, fiber type predominance, selective fiber type involvement, or reinnervation evidenced by fiber type grouping), analysis of muscle fiber diameters (eg, fiber hypertrophy or atrophy, increased variability in fiber diameter, or denervation atrophy with narrow range of diameters among atrophic and nonatrophic fibers), or alterations in the muscle fiber (eg, central nuclei, necrosis, splitting or branching, regeneration or the presence of a variety of other accumulations and fiber alterations, both specific to certain conditions and nonspecific).

Congenital myopathies are a group of structural myopathies whose diagnosis is based on classic histologic characteristics seen on muscle biopsy (eg, centronuclear or myotubular, central core, nemaline rod, and fiber type disproportion myopathies).

Immunoblotting and Immunostaining

Imunoblotting of a muscle sample provides information about amounts of specific muscle protein, such as dystrophin or other structural proteins important in maintaining structural integrity of the muscle membrane. Immunoblotting can be performed with as little as 10 mg of frozen tissue. Quantitative dystrophin analysis using Western blot technique can differentiate DMD from BMD and thus help determine the prognosis in a young symptomatic patient—information not precisely determined by standard molecular genetic analysis of the dystrophin gene. Dystrophin quantity 0% to 5% is consistent with DMD, 5% to 20% dystrophin is seen in some with less severe "outlier" DMD or severe BMD, and either 20% to 80% dystrophin or normal quantity and reduced or increased molecular-weight dystrophin is consistent with BMD. A normal dystrophin level in a patient with histologic evidence of a dystrophic myopathy is suggestive of LGMD. Immunofluorescent staining of muscle biopsy sections for dystrophin helps identify symptomatic female DMD carriers and some female BMD carriers.

The progressive loss of muscle fibers evident in muscular dystrophy is now thought to be caused by primary muscle sarcolemmal membrane abnormalities due to inherited structural abnormalities (abnormal molecular weight, deficiency, or absence) of dystrophin or dystrophin-associated transmembrane glycoproteins. Membrane instability leads to membrane injury from mechanical stresses, transient breaches

of the membrane, and membrane leakage. Ultimately, after multiple cycles of degeneration and regeneration, irreversible muscle cell death occurs. The muscle fiber is then replaced by connective tissue and fat, and this fibrotic replacement of the muscle may be exceedingly aggressive. This has given rise to the concept of diseases of the *dystrophin-glycoprotein complex*. Primary genetic abnormalities lead to abnormalities of intracellular dystrophin, transmembrane sarcoglycans, or transmembrane dystroglycans. An abnormality in the muscle protein merosin, located in the extracellular matrix, gives rise to one of the forms of congenital muscular dystrophy. Immunoblotting and/or immunofluorescent staining of the proteins of the dystrophin-glycoprotein complex allows many LGMD patients to be subtyped.

Electron Microscopy

Electron microscopy (EM) is used to evaluate ultrastructural changes of muscle fiber organelles/internal components, as well as changes in the muscle fiber. At times, this may provide additional complimentary information to the histologic and histochemical assessment of muscle fibers that may be diagnostically relevant. For example, ultrastructural alterations of the mitochondria may provide important information and direct additional metabolic studies in the workup of mitochondrial myopathy. In a congenital structural myasthenic syndrome, ultrastructural alterations at the neuromuscular junction may be present by EM, either presynaptically or postsynaptically.

Metabolic Studies

Depending on clinical suspicion and histologic and ultrastructural changes on muscle biopsy, additional metabolic studies may be obtained to evaluate for the presence of metabolic myopathies, including glycogenoses, lipid disorders, or mitochondrial myopathies.

Nerve Biopsy Evaluation

Nerve biopsies are occasionally useful in the characterization of more severe hereditary motor sensory neuropathies, congenital hypomyelinating neuropathy, and neuroaxonal dystrophy. In addition, perineural immune complex deposition seen in some autoimmune neuropathies and changes consistent with vasculitis also may be useful diagnostically. Otherwise, nerve biopsies rarely add specific information to the diagnostic evaluation of the NMD patient beyond that information obtained from nerve conduction studies and EMG.

Generally, the sural nerve is utilized. Since this is a pure sensory nerve, the usefulness of this specimen is limited to those disorders giving rise to demyelinating or axonal changes in sensory fibers. Occasionally, a small portion of the motor nerve can be obtained simultaneously with an anconeus motor point biopsy, where the motor branch is excised along with the entire muscle from origin to insertion.

SPECIFIC NEUROMUSCULAR DISEASE CONDITIONS

Dystrophic Myopathies

Muscular dystrophies are debilitating myopathic disorders that present with muscle wasting and diffuse muscle weakness. They are caused by genetic mutations, which produce muscle fiber necrosis and regeneration, ultimately resulting in muscle fiber loss. Traditionally, patients with the muscular dystrophies were grouped together because they had similar pathologies, and they were subdivided into categories based upon their modes of inheritance, ages of onset, and distributions of affected muscles. Most types of muscular dystrophy are not purely muscle disorders, but multisystem disorders with disease manifestations in a variety of body systems, which may include the musculoskeletal, cardiovascular, pulmonary, and gastrointestinal systems, as well as endocrine system, skin, eyes, brain, and other organ systems. Muscular dystrophies are caused by mutations of the genes encoding for proteins important for the stability of the sarcolemmal membrane and the maintenance of muscle fiber intracellular homeostasis. They are genetically, biochemically, and clinically diverse diseases.

Dystrophinopathies

Duchenne Muscular Dystrophy

Duchenne muscular dystrophy (DMD) is an X-linked disorder caused by an abnormality at the Xp21 gene loci. The DMD/BMD gene occupies 2.5 million base pairs of DNA on the X chromosome and is about 10 times larger than the next largest gene identified to date. The gene coding sequence contains 79 exons. The primary protein product, *dystrophin*, is localized to the intracellular side of the plasma membrane of all myogenic cells, certain types of neurons, and in small amounts of other cell types (5). Dystrophin deficiency at the plasma membrane of muscle fibers disrupts the membrane cytoskeleton and leads to the secondary loss of other components of the muscle cytoskeleton. The primary consequence of the cytoskeleton abnormalities is membrane instability, leading to membrane injury from mechanical stresses, transient breaches of the membrane, and membrane leakage. Chronic

dystrophic myopathy is characterized by aggressive fibrotic replacement of the muscle and eventual failure of regeneration with muscle fiber death and fiber loss. Generally, loss of the reading frame causes complete absence of dystrophin and a Duchenne phenotype. For cases with a deletion mutation, the "reading frame" hypothesis predicts that BMD patients with in-frame deletions produce a semifunctional, internally deleted dystrophin protein, whereas DMD patients with frameshift or "out of frame" deletions produce a severely truncated protein that would be unstable (6). Characteristics of DMD and BMD are shown in Table 12.1.

Diagnostic Evaluation. Serum creatine kinase is a useful screening test. Gene abnormalities may be identified by full gene sequencing of a blood specimen in 99% of all patients with a dystrophinopathy. Full gene sequencing in addition to evaluation for large deletions to identify point mutations, deletions, duplications,

12.1 Characteristics of Dystrophinopathies

	DUCHENNE MUSCULAR DYSTROPHY	BECKER MUSCULAR DYSTROPHY
U.S. Prevalence (est.)	15,000	2,200
Incidence rate	1/3,500 male births	unknown
Inheritance	X-linked	X-linked
Gene location	Xp21 (reading frame shifted)	Xp21 (reading frame maintained)
Protein	Dystrophin	Dystrophin
Onset	2 to 6 years	4–12 years (severe BMD) Late teenage to adulthood (mild BMD)
Severity and course	Relentlessly progressive Reduced motor function by 2–3 yrs Steady decline in strength Life span <35 years	Slowly progressive Severity and onset correlate with muscle dystrophin levels
Ambulation status	Loss of ambulation: 7 to 13 yrs (no corticosteroids) Loss of ambulation 9 to 15 years (corticosteroids)	Loss of ambulation: >16 years
Weakness	Proximal > distal Symmetric Legs and arms	Proximal > distal Symmetric Legs and arms
Cardiac	Dilated cardiomyopathy first to second decade Onset of signs second decade	Cardiomyopathy (may occur before weakness); third to fourth decade frequent
Respiratory	Profoundly reduced vital capacity in second decade Ventilatory dependency in second decade	Respiratory involvement in subset of patients Ventilatory dependency in severe patients
Muscle size	Calf hypertrophy	Calf hypertrophy
Musculoskeletal	Contractures: ankles, hips, and knees Scoliosis: onset after loss of ambulation	Contractures: ankles and others in adulthood
CNS	Reduced cognitive ability oReduced verbal ability	Some patients have reduced cognitive ability
Muscle pathology	Endomysial fibrosis and fatty infiltration Variable fiber size and myopathic grouping Fiber degeneration/regeneration Dystrophin: absent Sarcoglycans: secondary reduction	Variable fiber size Endomysial connective tissue and fatty infiltration Fiber degeneration Fiber regeneration Dystrophin: reduced (usually 10%–60% of normal)
Blood chemistry and hematology	CK: Very high (10,000–50,000) High AST and ALT (normal GGT) High aldolase	CK: 5,000 to 20,000 Lower levels with increasing age

inversions, etc. rather than simple deletion screening with polymerase chain reaction (PCR) is now standard of care in all patients at risk of a dystrophinopathy. This is both for diagnostic purposes and to identify candidates for future molecular-based therapies such as exon skipping with oligonucleotides, nonsense-mediated suppression therapy for the 10% to 15% of patients with DMD and BMD with stop codon mutations, and specific gene therapy strategies that will require knowledge of specific gene sequence alterations. In patients with no family history and molecular genetics that do not clearly differentiate a DMD and BMD phenotype, a muscle biopsy with immunostaining and quantitative dystrophin analysis with Western blot is critical to allow patients to be eligible for future clinical trials with rigid inclusionary criteria.

Epidemiology. The incidence of Duchenne muscular dystrophy, based on a number of population studies as well as neonatal screening, has been estimated to be around 1:3,500 male births (7). As many as one-third of isolated cases may be due to new mutations, which is considerably higher than observed in other X-linked conditions. This high mutation rate may relate to the large size of the gene.

Onset and Early Signs. While the history of hypotonia and delayed motor milestones are often reported in retrospect, the parents are often unaware of any abnormality until the child starts walking. Variability has been reported in the age of onset (8,9). In 74% to 80% of instances, the onset has been noted before the age of 4 years (8–10). The vast majority of cases are identified by 5 to 6 years of age. The most frequent presenting symptoms have been abnormal gait, frequent falls, and difficulty climbing steps. Parents frequently note the toe walking, which is a compensatory adaptation to knee extensor weakness, and a lordotic posture to the lumbar spine, which is a compensatory change due to hip extensor weakness (Fig. 12.7).

Occasionally, Duchenne muscular dystrophy is identified presymptomatically in situations where a CK value is obtained with a markedly elevated value, malignant hyperthermia occurs during general anesthesia for an unrelated surgical indication, or a diagnosis is pursued in a male with an affected older sibling.

Difficulty negotiating steps is an early feature, as is a tendency to fall due to the child tripping or stumbling on a plantar-flexed ankle or the knee buckling or giving way due to knee extensor weakness. There is progressive difficulty getting up from the floor with presence of a Gower's sign (see Fig. 12.5).

Pain in the muscles, especially the calves, is a common symptom. Enlargement of muscles, particularly the calves (see Fig. 12.1), is commonly noted. The deltoid may also be hypertrophied. With the patients arms abducted to 90 degrees and externally rotated, the hypertrophy of the posterior deltoid and infraspinatus frequently leaves a depression between these two muscles referred to as the "posterior axillary depression sign" in DMD (Fig. 12.9). The tongue is also frequently enlarged. There is also commonly an associated wide arch to the mandible and maxilla with separation of the teeth, presumably secondary to the macroglossia.

Pattern and Progression of Weakness. Earliest weakness is seen in the neck flexors during preschool years (Fig. 12.10). Weakness is generalized, but predominantly proximal early in the disease course. Pelvic girdle weakness predates shoulder girdle weakness by several years. Ankle dorsiflexors are weaker than ankle plantar flexors; ankle everters are weaker than ankle inverters; knee extensors are weaker than knee flexors; hip extensors are weaker than hip flexors; and hip abductors are weaker than hip adductors (9).

The weakness progresses steadily, but the rate may be variable during the disease course. Quantitative strength testing shows greater than 40% to 50% loss of strength by 6 years of age (9). With manual muscle testing, DMD subjects exhibit loss of strength in a fairly linear fashion from ages 5 to 13, and measurements obtained several years apart will show fairly steady disease progression. A variable course may be noted when analyzing individuals over a shorter time course (9). Quantitative strength measures have been shown to be more sensitive for demonstrating strength loss than manual muscle testing when strength is grades 4–5 (9).

Loss of Ambulation. Average age to wheelchair in a DMD population not treated with corticosteroids has

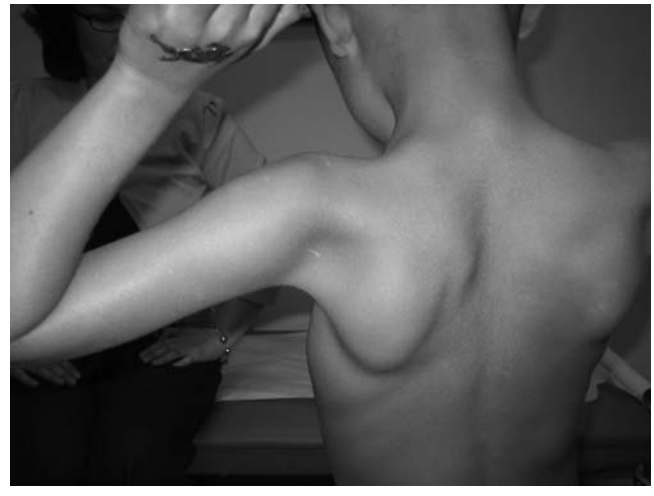


Figure 12.9 Posterior axillary depression sign in Duchenne muscular dystrophy. Note the prominent deltoid superolaterally and infraspinatus inferomedially.

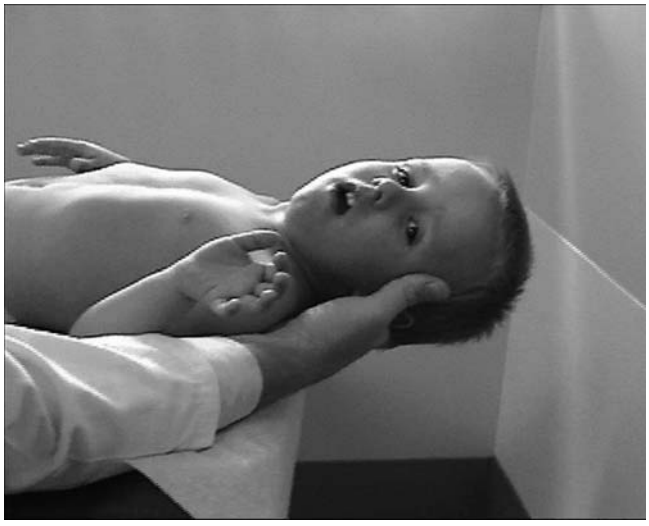


Figure 12.10 Weakness of neck flexors in an eight-year-old child with Duchenne muscular dystrophy makes it difficult for him to bring his chin to the chest when supine and to hold his head up when placed at the end of the examination table.

been age 10, with a range of 7–13 years. Treatment with prednisone or deflazacort helps maintain strength and prolongs ambulation by two years (11,12). There does not appear to be an advantage of deflazacort over daily prednisone. The optimal dose of prednisone is 0.75 mg/kg/day up to a maximum of 40 mg/day (11,12,13). The optimal dose of deflazacort appears to be 0.90 mg/kg/day. With both corticosteroid regimens, patients need to be monitored for cataracts, hypertension, weight gain, osteoporosis, growth retardation, diabetes, and behavioral side effects.

Timed motor performance is useful for the prediction of time when ambulation will be lost without provision of long-leg braces. One large natural history study showed that all DMD subjects who took nine seconds or longer to ambulate 30 feet lost ambulation within two years. All DMD subjects who took 12 seconds or longer to ambulate 30 feet lost ambulation within one year (9). Ambulation past the age of 14 in a noncorticosteroid-treated patient should raise the suspicion of a milder form of muscular dystrophy such as BMD or limb girdle muscular dystrophy. Ambulation beyond 16 years has been previously used as an exclusionary criteria for Duchenne muscular dystrophy in studies of BMD. Immobilization for any reason can lead to a marked and often precipitous decline in muscle power, rapid development of contractures, and loss of ambulatory ability. A fall with resultant fracture leading to immobilization and loss of ambulatory ability is not an uncommon occurrence.

Contractures. Significant joint contractures have been found in nearly all children with Duchenne muscular dystrophy older than age 13 (9,14,15). The most common contractures include ankle plantar flexion, knee flexion, hip flexion, iliotibial band, elbow flexion, and wrist flexion contractures (9). Significant contractures have been shown to be rare in DMD before age 9 for all joints. There is no association between muscle imbalance around a specific joint (defined as grade 1 or greater difference in flexor and extensor strength) and the frequency or severity of contractures involving the hip, knee, ankle, wrist, and elbow in DMD (9). Flexion contractures have been shown to be rare in those with ≥grade 3 extensor strength about a joint, an expected finding because of the definition of a grade 3 muscle on manual muscle testing (MMT). For those DMD subjects with less than antigravity strength about a joint, there is low correlation between the MMT strength of these specific muscle groups and the severity of joint contracture (9). The presence of lower extremity contractures in DMD has been shown to be strongly related to onset of wheelchair reliance (9). Lower extremity contractures were rare while DMD subjects were still upright, but developed soon after they developed a sitting position in a wheelchair for most of the day. The occurrence of elbow flexion contractures also appears to be directly related to prolonged static positioning of the limb, and these contractures develop soon after wheelchair reliance. The relationship between wheelchair reliance and hip and knee flexion contractures has been noted (9). Mild contractures of the iliotibial bands, hip flexor muscles, and heel cords occurs in most DMD patients by 6 years of age (16). Limitations of knee, elbow, and wrist extension occurs about two years later (9,16); however, these early observed contractures were relatively mild. Given the tremendous replacement of muscle by fibrotic tissue in DMD subjects, it is not surprising that a muscle of less than antigravity extension strength, statically positioned in flexion, would develop a flexion contracture (subsequent to wheelchair reliance). The lack of lower extremity weight bearing likely contributes to the rapid acceleration in the severity of these contractures after transition to wheelchair. Ankle plantar flexion contractures are not likely a significant cause of wheelchair reliance, as few subjects exhibit plantar flexion contractures of ≥15 degrees before their transition to a wheelchair (9). Natural history data suggests that weakness is the major cause of loss of ambulation in DMD, not contracture formation.

Spine Deformity. Reported ultimate prevalence of scoliosis in DMD subjects not treated with corticosteroids varies from 33% to 100% (17). This marked variability is primarily because of retrospective selection for scoliosis, the inclusion or exclusion of functional curves, and dissimilar age groups. The prevalence of scoliosis is strongly related to age. Fifty percent of DMD

patients acquire scoliosis between ages 12 and 15, corresponding to the adolescent growth spurt. Ten percent of older DMD subjects with no treatment of scoliosis show no clinical spinal deformity. This is consistent with Oda's report (18) that 15% of older DMD patients show mild nonprogressive curves (usually 10 degrees to 30 degrees). The rate of progression of the primary or single untreated lateral curve has been reported to range from 11 degrees to 42 degrees per year, depending on the age span studied. Johnson and Yarnell (19) reported an association between side of curvature, convexity, and hand dominance, an association recently confirmed (20). Oda and colleagues (18) reported that the likelihood of severe progressive spinal deformity could be predicted by type of curve and early pulmonary function measurements. Those with spines lacking significant kyphosis or hyperlordosis and a peak obtained absolute forced vital capacity (FVC) greater than 2,000 mL tended not to show severe progressive scoliosis.

No cause-and-effect relationship has been established between onset of wheelchair reliance and occurrence of scoliosis (9,21). Wheelchair reliance and scoliosis has been found to be an age-related phenomenon. The causal relationship between loss of ambulatory status and scoliosis is doubtful, given the substantial time interval between the two variables in most subjects (scoliosis usually develops after three to four years in a wheelchair). Both wheelchair reliance and spinal deformity may be significantly related to other factors (eg, age, adolescent growth spurt, increase in weakness of trunk musculature, and other unidentified factors) and thus represent coincidental signs of disease progression.

In retrospective series, treatment of DMD with deflazacort and prednisone have been shown to reduce the occurrence of significant scoliosis (22,23,24). It remains to be seen whether the apparent arrest in the development of scoliosis with corticosteroids will continue past the age of skeletal maturity.

Pulmonary Function. In DMD, absolute forced vital capacity volumes increase during the first decade and plateau during the early part of the second decade (9). A linear decline in percent predicted FVC is apparent between 10 and 20 years of age in DMD (9). Rideau and colleagues (25) reported forced vital capacity to be predictive of the risk of rapid scoliosis progression. In the most severe DMD cases, maximal forced vital capacity reached a plateau of less than 1,200 mL. This was associated with loss of ability to walk before age 10 and severe progressive scoliosis. Moderately severe DMD cases with respiratory compromise reached maximum forced vital capacities between 1,200 mL and 1,700 mL. Spinal deformity was present consistently in these cases, but varied in severity. The least severe DMD cases reached plateaus in FVC of greater than 1,700 mL. Similarly, McDonald and colleagues (9) found that those patients with higher peak FVC (>2,500 mL) had a milder disease progression, losing 4% predicted FVC per year. Those with peak predicted FVC less than 1,700 mL lost 9.6% predicted FVC per year. Thus, the peak obtained absolute values of forced vital capacity usually occurring in the early part of the second decade is an important prognostic indicator for severity of spinal deformity, as well as ultimate severity of restrictive pulmonary compromise due to muscular weakness. Prednisone and deflazacort both appear to reduce the loss of pulmonary function over time during the second decade in DMD (22,23,24,26).

Maximal static airway pressures (both maximal inspiratory pressure and maximal expiratory pressure) are the earliest indicators of restrictive pulmonary compromise in DMD with impaired values noted between 5 and 10 years of age. Vital capacity typically increases concomitant with growth between 5 and 10 years of age, with percent predicted FVC remaining relatively stable and close to 100% predicted. DMD patients typically show a linear decline in percent predicted FVC between 10 and 20 years of age. An FVC falling below 35% is associated with increased perioperative morbidity in DMD (27) and optimally, surgery should ideally be performed with % predicted FVC greater than 40%. Recent evidence suggests that spinal arthrodesis may be safely performed in a population of DMD with % predicted vital capacity less than 30% (28).

Ultimately, respiratory failure in DMD is insidious in its onset and results from a number of factors, including respiratory muscle weakness and fatigue, alteration in respiratory system mechanics, and impairment of the central control of respiration. Noninvasive forms of both positive and negative pressure ventilatory support are increasingly being offered to DMD patients nocturnally and continuously with acceptable quality of life. Airway clearance strategies, such as the cough assist/inexsufflator, TheraVest, or intrapulmonary percussion ventilation (IPV) are also important pulmonary management strategies (29).

Cardiomyopathy. The dystrophin protein is present in both the myocardium and the cardiac Purkinje fibers. Abnormalities of the heart may be detected by clinical examination, electrocardiogram (ECG), echocardiography, and Holter monitoring. Cardiac examination is notable for the point of maximal impulse palpable at the left sternal border due to the marked reduction in anteroposterior chest dimension common in DMD. A loud pulmonic component of the second heart sound suggests pulmonary hypertension in patients with restrictive pulmonary compromise. Nearly all patients over the age of 13 demonstrate abnormalities of the ECG (9). Q-waves in the lateral leads are the first

abnormalities to appear, followed by elevated ST segments and poor R-wave progression, increased R/S ratio, and finally resting tachycardia and conduction defects. ECG abnormalities have been demonstrated to be predictive for death from the cardiomyopathy with the major determinants including R-wave in lead V1 less than 0.6 mV; R-wave in lead V5 less than 1.1 mV; R-wave in lead V6 less than 1.0 mV; abnormal T-waves in leads II, III, AVF, V5, and V6; cardiac conduction disturbances; premature ventricular contraction; and sinus tachycardia (30). Sinus tachycardia may be due to low stroke volume from the progressive cardiomyopathy, or in some cases, may be sudden in onset and labile, suggesting autonomic disturbance or direct involvement of the sinus node by the dystrophic process (31).

Autopsy studies and thallium 201 single-photon emission computed tomography (SPECT) imaging have demonstrated left ventricular lateral and posterior wall defects that may explain the lateral Q-waves and the increased R/S ratio in V1 seen on ECG. Localized posterior wall fibrosis was found to be peculiar to DMD and was not found in other types of muscular dystrophy (32). Pulmonary hypertension leading to right ventricular enlargement also is known to affect prominent R-waves in V1 and has been demonstrated in patients with DMD (33).

Ventricular ectopy and sudden death are known complications of the cardiomyopathy in DMD, and this association likely explains observed cases of sudden death. Severe ventricular ectopy in DMD has been associated with left ventricular dysfunction and sudden death. Yanagisawa and colleagues (34) reported an age-related increase in the prevalence of cardiac arrhythmias detected by ambulatory 24-hour electrocardiographic recordings. They also noted an association between ventricular arrhythmias and sudden death in DMD. Clinically evident cardiomyopathy is usually first noted after age 10 and is apparent in nearly all patients over age 18 (35). Development of cardiomyopathy is a predictor of poor prognosis. Echocardiography has been used extensively to follow the development of cardiomyopathy and predict prognosis in patients with DMD. The onset of systolic dysfunction noted by echocardiography is associated with a poor short-term prognosis (35). The myocardial impairment remains clinically silent until late in the course of the disease, possibly caused by the absence of exertional dyspnea, secondary to lack of physical activity. Death has been attributed to congestive heart failure in as many as 40% to 50% of patients with DMD by some investigators (35). Regular cardiac evaluations with an ECG, echocardiography, and Holter monitor should be employed in teenagers with preclinical cardiomyopathy.

Recent studies suggest that early presymptomatic treatment to achieve afterload reduction with angiotensin-converting enzyme inhibitors (ACE inhibitors) such as perindopril or enalapril delayed the onset and progression of prominent left ventricle dysfunction in children with DMD (36). In another series, 43% with impaired left ventricular (LV) systolic dysfunction responded to enalapril with the normalization of function (37). Alternatively, angiotensin II type 1 receptor blockers (ARBs) such as losartan may be considered for afterload reduction in DMD. Animal studies show that the angiotensin II type 1 receptor blocker losartan attenuates TGF-beta–induced failure of muscle regeneration in dystrophinopathy, presenting an additional potential for therapeutic benefit vis-a-vis skeletal muscle in DMD (38).

Cognition and Behavioral Phenotype. There is a dystrophin isoform present in the brain. Previous studies on intellectual function on children with DMD have generally revealed decreased IQ scores when these children are compared with both control and normative groups (9). A mean score for the DMD population of 1.0 to 1.5 standard deviation (SD) below population norms has been reported. There has generally been a considerable consistency in the degree of impairment across measures reflecting a rather mild global deficit. Some studies (39) have demonstrated relative deficits in verbal IQ. In a longitudinal assessment of cognitive function, McDonald and colleagues (9) found IQ measure in DMD to be stable over time. On neuropsychological testing, a large proportion of DMD subjects fell within the "mildly impaired" or "impaired" range according to normative data (9). Again, no particular areas of strength or weakness were identified. These findings likely reflect a mild global deficit rather than focal nervous system impairment (9). Hinton found that DMD is associated with poor attention to complex verbal information (more so than verbal or memory measures), and they exhibit decreased verbal span capacity, but not impaired recall (40,41). An increased incidence of autism spectrum disorder has been found in DMD (42). In one large series of DMD subjects, 11.7% were reported to have a comorbid diagnosis of attention-deficit hyperactivity disorder (ADHD), 3.1% had autism spectrum disorder, and 4.8% had obsessive-compulsive disorder (43). In addition, impaired facial affect recognition has found to be a part of the phenotype associated with DMD (44).

Anthropometric Changes. Substantial anthropometric alterations have been described in DMD. Short stature and slow linear growth with onset shortly after birth has been reported (45). Accurate measurement of linear height is extremely difficult in this population. Arm span measurements may be an alternative measure of linear growth; however, this measurement

might also be difficult, as elbow flexion contractures of greater than 30 degrees are frequently present in patients older than age 13. Forearm segment has been proposed as an alternative linear measurement in DMD patients with proximal upper extremity contractures, and radius length may be followed for those with wrist and finger contractures. Obesity is a substantial problem in DMD, subsequent to the loss of independent ambulation (9,46). Weight control during early adolescence has its primary rationale in ease of care, in particular, ease of transfers during later adolescence.

Immediately following spine fusion, there has been a documented tendency for DMD patients to lose significant weight. Those who lost weight were unable to self-feed. The weight loss after surgery was associated with loss of self-feeding (47). There was no association with weight loss and loss of biceps strength. A correction of the kyphosis may actually make self-feeding problematic in DMD. A feeding evaluation and incorporation of kyphosis into the spinal instrumentation construct may help preserve self-feeding and prevent weight loss subsequent to spine fusion.

Longitudinal weight measurements in DMD confirm significant rates of weight loss in subjects ages 17–21 (9,48). This is likely caused by relative nutritional compromise during the later stages when boys with DMD have higher protein and energy intake requirements because of hypercatabolic protein metabolism. Protein and calorie requirements may often be 160% of that predicted for able-bodied populations during the later stages of DMD (49,50). Restrictive lung disease becomes more problematic during this time, and this may also influence caloric intake and requirements. Self-feeding often becomes impossible during this period because of significant biceps weakness. In addition, boys with DMD may develop signs and symptoms of upper gastrointestinal dysfunction (51).

Becker Muscular Dystrophy

Existence of a form of muscular dystrophy with a similar pattern of muscle weakness seen in Duchenne muscular dystrophy, X-linked inheritance, but with later onset and a much slower rate of progression, was first described by Becker and Kiener in 1955 (52). The disorder has the same gene location as the DMD gene (Xp21) and is thus allelic. On immunostaining of muscle biopsy specimens, the presence of patchy abundance of dystrophin suggests a Becker muscular dystrophy phenotype. On Western blot for quantitative dystrophin analysis, either 20% to 80% dystrophin levels or normal quantity and reduced or increased molecular-weight dystrophin is consistent with BMD. Studies show that 5% to 20% dystrophin quantity is consistent with an outlier or intermediate phenotype (5).

Epidemiology. Becker muscular dystrophy has a lower incidence than DMD, with prevalence rates for BMD ranging from 12–27 per million and a recent estimated overall prevalence of 24 per million (7,53).

Molecular Genetics and Diagnostic Evaluation. Full gene sequencing of the dystrophin gene, which demonstrates large deletions, duplications, and point mutations, identifies 99% of patients with dystrophinopathy and is now the standard of care. This is essential for identification of patients with stop codons and specific gene alterations that will be targeted for molecular-based therapies. Not all DMD and BMD patients have deletion mutations: Many have point mutations that cannot be detected by screening deletion testing. Thus, full sequence analysis is necessary. About 55% of DMD patients and 70% of BMD patients show large deletion mutations of the gene. A positive DNA test result (presence of a point mutation, duplication, or deletion) is diagnostic of a dystrophinopathy (Duchenne or Becker dystrophy)—there are no false-positives if the test is done appropriately. While genetic testing is improving with regard to the differentiation of DMD and BMD, there remains some overlap and variability. Differential diagnosis between DMD and BMD is best done by a consideration of clinical findings, family history of clinical phenotype, and muscle biopsy with quantitative dystrophin analysis. If the patient is still ambulating at 16–20 years of age and has a deletion mutation, the correct diagnosis is BMD. Mutations at the Xp21 locus, which maintain the translational reading frame (in-frame mutations), result in an abnormal but partially functional dystrophin protein, whereas in Duchenne muscular dystrophy, the mutations shift the reading frame (out-of-frame mutations) so that virtually no dystrophin is produced. The reading frame interpretation is most accurate for deletions in the center of the gene (exons 40–60) and is least accurate for deletions in the beginning of the gene (exons 1–20).

Absent dystrophin or levels less than 5% of normal generally are considered diagnostic of Duchenne muscular dystrophy; however, 5% of such patients have BMD phenotypes. In BMD, dystrophin typically has abnormally small molecular weight (<427 kDa). A minority of patients have dystrophin of larger-than-normal molecular weight (>427 kDa) or normal molecular weight. Most BMD patients with larger or smaller molecular-weight dystrophin also have decreased quantities of the protein. All BMD patients with normal molecular-weight dystrophin have decreased quantities, usually less than 30% normal. Smaller-size dystrophin typically is caused by deletion mutations, and larger-size dystrophin by duplication mutations. A further refinement is the use of antibodies specific to the carboxy-terminal (C-terminal) region of dystrophin. Using such antibodies, immunohistochemistry

reveals that the C-terminal region is almost always absent in DMD but invariably present in BMD. Thus, when this region of the molecule is missing, a more severe phenotype is likely.

Age of Onset and Presenting Signs. Studies have shown significant overlap in the observed age of onset between DMD and BMD (10). Although determination of the quantity and molecular weight of dystrophin has substantially improved the early differentiation among BMD, "outlier" DMD, and the more common and rapidly progressive DMD phenotype, Bushby and colleagues (54) found no clear correlation between abundance of dystrophin and clinical course within the BMD group.

A series of Bushby and Gardner-Medwin (54), which included 67 BMD subjects, supported the presence of two major patterns of progression in BMD: a "typical" slowly progressive course and a more "severe" and rapidly progressive course. All of the "severe" BMD cases showed difficulty climbing stairs by age 20, whereas none of the "typical" BMD cases had difficulty climbing stairs before age 20. Abnormal ECGs were seen in 27% of typical BMD subjects and 88% of severe subjects. Bushby and Gardner-Medwin (54) found BMD subjects to have a mean age of onset of 12 years in the typical group and 7.7 years in the severe group. Some patients with BMD present with major muscle cramps as an isolated symptom (54). As in DMD, preclinical cases are often identified by the finding of a grossly elevated CK value. There is also considerable overlap in CK values between DMD and BMD cases at the time of presentation. Thus, CK values cannot be used to differentiate DMD from BMD.

Calf enlargement is a nonspecific finding in BMD, as is the presence of a Gower's sign. The gait over time is similar to other neuromuscular disease conditions with proximal weakness. Patients often ambulate with a lumbar lordosis, forefoot floor contact, decreased stance-phase knee flexion, and a Trendelenburg's or gluteus medius lurch, often described as a waddle.

Other atypical clinical presentations include a sole complaint of cramps on exercise in individuals with no muscle weakness (54). In addition, patients with focal wasting of the quadriceps, previously diagnosed with quadriceps myopathy, have been diagnosed with BMD, based on molecular genetic testing and/or dystrophin analysis on muscle biopsy (10).

Age of Transition to Wheelchair. The most useful clinical criterion to distinguish BMD from DMD is the continued ability of the patient to walk into late teenage years. Those with BMD will typically remain ambulatory beyond 16 years. Some patients may become wheelchair uses in their late teens or 20s, whereas others may continue walking into their 40s, 50s, or later. DMD cases usually stop ambulating by 13 years unless treated with corticosteroids. Outlier DMD or intermediate dystrophinopathy cases generally stop ambulating between 13 and 16 years of age.

Pattern and Progression of Weakness. BMD patients have distribution of weakness similar to those with DMD (10). Proximal lower limb muscles are involved earlier in the disease course. Gradual involvement of the pectoral girdle and upper limb musculature occurs 10–20 years from onset of disease. Extensors have been noted to be weaker than flexors (10). The muscle groups that are most severely involved earlier in the course of disease include the hip extensors, knee extensors, and neck flexors (10).

Contractures. Early development of contractures does not appear to be a feature of BMD (10,54). As with BMD, nonambulatory BMD subjects may develop equinus contractures, knee flexion contractures, and hip flexion contractures. Because of the tremendous replacement of muscle in BMD subjects by fibrotic tissue, it is likely that, as in DMD, a muscle with less-than-antigravity extension strength, which is statically positioned in flexion, is more likely to develop a flexion contracture subsequent to wheelchair reliance.

Spine Deformity. Spinal deformity is not nearly as common or severe in BMD, as compared with DMD. Spinal instrumentation is rarely required by DMD patients (10,54).

Pulmonary Function. Compromised pulmonary function is much less problematic in BMD as opposed to DMD (10,25,54). The percent predicted forced vital capacity does not appear substantially reduced until the third to fourth decade. The percent predicted maximal expiratory pressure appears relatively more reduced at younger ages than the percent predicted maximal inspiratory pressure, a finding seen in DMD and other neuromuscular diseases (9,55,56,57). This may be caused by more relative involvement of the intercostals and abdominal musculature with relative sparing of contractile function in the diaphragm of BMD. As in DMD and other neuromuscular disease, it appears that predicted maximal expiratory pressure (MEP) may be a useful quantitative measure of impairment and perhaps disease progression early in the course of BMD.

Cardiomyopathy. The pattern of occasional life-threatening cardiac involvement in otherwise mild and slowly progressive BMD has been reported by many (54,58). A significant percentage of BMD cases develop cardiac abnormalities, and the rate of progression of cardiac failure may on occasion be more rapid than the progression of skeletal myopathy (58). In fact,

successful cardiac transplantation has been successfully performed in BMD subjects with cardiac failure. Approximately 75% of BMD patients have been found to exhibit ECG abnormalities (10,59). The abnormal findings most typically reported include abnormal Q-waves, right ventricular hypertrophy, left ventricular hypertrophy, right bundle branch block, and nonspecific T-wave abnormalities. Unlike DMD, resting sinus tachycardia has not been a frequent finding. Echocardiography has shown left ventricular dilation in 37%, whereas 63% have subnormal systolic function because of global hypokinesia (59). Thus, the cardiac compromise may be disproportionately severe, relative to the degree of restrictive lung disease in some BMD subjects. The evidence for significant myocardial involvement in BMD is sufficient to warrant screening of all of these patients at regular intervals using ECG and echocardiography. The slowly progressive nature of this dystrophic myopathy, which is compatible with many years of functional mobility and longevity, makes these patients suitable candidates for cardiac transplantation if end-stage cardiac failure occurs.

Some cases with BMD may present with an isolated cardiomyopathy with no clinical manifestation of skeletal muscle involvement. The diagnosis can be established by demonstration of a deletion in the Xp21 gene or by muscle biopsy. Isolated cases of cardiomyopathy in children, particularly those with family histories indicative of X-linked inheritance, should be screened for BMD with an initial serum CK estimation and molecular genetic studies of the Xp21 gene.

Cognition. Cognitive testing in BMD subjects have shown large variability in IQ scores and neuropsychological test measures. Mildly reduced intellectual performance has been noted in a subset of BMD patients; however, the degree of impairment is not as severe as noted in DMD (10).

Limb Girdle Muscular Dystrophy (LGMD)

Before the advent of genetic testing, a group of patients commonly sharing a progressive pattern of proximal greater than distal muscular weakness with either autosomal-recessive (LGMD2) or dominant (LGMD1) inheritance were termed *limb-girdle muscular dystrophies.* Recent advances in molecular and genetic analyses have now identified a number of distinct genetic mutations in these patients. LGMD1 subtypes usually have later onset in adulthood. LGMD2 usually present during childhood or adolescence, although some may present in early adulthood. Many of the LGMD2 subtypes have been linked to gene defects causing abnormalities of the sarcolemmal-associated proteins, including sargoglycans (alpha-SG, gamma-SG, beta-SG, and delta-SG), dystroglycans, calpain-3, dysferlin, fukutin-related protein (FKRP), telethonin, and titin. The most common LGMD2 subtypes include sarcoglycanopathies, dysferlinopathies, calpainopathies, and FKRP deficiencies. The distribution and pattern of weakness at onset most often affects the pelvic or shoulder girdle musculature or both. The rate of progression is slower than DMD (60,61,62). Clinical features of the most common forms of LGMD2 are shown in Table 12.2.

Sarcoglycanopathies (LGMD 2C-2F)

Disruption of the sarcolemmal membrane cytoskeleton is a common feature of the sarcoglycanopathies. Most of the primary sargoglycan abnormalities lead to secondary deficiencies of alpha-sarcoglycan. Diagnosis of sarcoglycanopathies may be made with molecular genetic studies and immunohistochemical analysis of muscle biopsies. The age of onset of sarcoglycanopathies ranges from 2 to 15 years. Progression is variable with both more severe and milder phenotypes. Loss of ambulation may vary from 10 years to young adulthood. Weakness involves proximal greater than distal musculature. Calf pseudohypertrophy scapular winging, progressive contractures, and scoliosis often occur (61). A dilated cardiomypathy may occur, particularly in alpha-SG and delta-SG. Intelligence is often normal.

Dysferlinopathies (LGMD 2B)

Dysferlin is a skeletal muscle protein localized in the muscle cell membrane (63). It is involved in muscle contraction and contains C2 domains that play a role in calcium-mediated membrane fusion events, suggesting that it may be involved in membrane regeneration and repair. Specific mutations in this gene have been shown to cause autosomal-recessive limb girdle muscular dystrophy type 2B (LGMD2B) with proximal muscle involvement as well as Miyoshi myopathy, which presents with distal weakness involving the distal legs, including the gastrocnemius and soleus muscles (61). In LGMD 2B, no specific genotype-phenotype correlations have been established. LGMD 2B presents from 12 to 39 years, with early weakness of gastrocnemius, quadriceps and psoas muscles, and atrophy of the pelvic and shoulder girdle muscles. There is no scapular winging. Patients have difficulty tip-toeing and running. Weakness occurs in a distal lower extremity and/or pelvifemoral distribution. Progression is slow, with loss of ambulation 10 to 30 years after onset. Equinus contractures are common, and toe-walking may be a presenting sign. Respiratory and cardiac muscles are spared. Intelligence is normal.

Calpainopathies (LGMD 2A)

Heterogeneous dystrophies due to mutation of the calpain-3 gene are termed calpainopathy (62). Calpain-3 is

12.2 Characteristics of Autosomal Recessive Limb Girdle Muscular Dystrophies (LGMD)

	LGMD 2A	LGMD 2B	LGMD 2C	LGMD 2D	LGMD 2E	LGMD 2F	LGMD 2I
U.S. prevalence	4,200	2,850	675	1,260	675	105	450
Inheritance	AR	AR	XR	XR	AR	AR	XR
Gene location	4p21	2p12–14	13q12	17q21	4q12	5q33	19q13.3
Protein	Calpain-3	Dysferlin	γ-sarcoglycan	γ-sarcoglycan (Adhalin)	γ -sarcoglycan	γ-sarcoglycan	Fukutin-related protein
Onset	Early: <12 years Leyden-Mobius type: 13–29 years Late: >30 years	12–39 years Mean 19 ± 3 years	Mean 5 to 6 yrs C283Y mutation: <2 years	2 – 15 years	3 years–teens Intrafamilial variability	2–10 years	0.5–27 years; 61% less than 5 years
Severity course	Variable Mild phenotype in majority Early onset has more severe progression	Slow progression Mild weakness	Variable progression (some like DMD; others like BMD) Death common in second decade	Variable Absent adhalin: rapid progression Reduced adhalin: Later onset and milder weakness	Moderate progression and severity	Rapid progression Death in 2nd decade	Variable Early onset: non ambulant by teens Later onset: slowly progressive,
Ambulation status	Loss of ambulation 10–30 years after onset	Loss of ambulation: 10–30 years after onset; Most walk until their fourth decade	Loss of ambulation: 10–37 years (mean 16 years)	Early onset: loss of adhalin Later onset: reduced adhalin	Often in wheelchair by 10–15 years; usually by 25 years	Loss of ambulation: 9–16 years	30% non-ambulant by fourth to sixth decade
Weakness	Scapula pelvic girdle and trunk weakness Proximal legs > arms	Weakness in gastrocnemius, quadriceps, and psoas Weakness in biceps after legs	Proximal > Distal Patchy distribution with some mutations Quadriceps: spared	Proximal > distal Symmetric quadriceps weakness	Proximal	Proximal Symmetric	Proximal > Distal Legs: Proximal Arms: proximal Face: mild weakness in older patients
Cardiac	No involvement	No involvement	Occasional; especially late in disease course	Dilated cardiomyopathy	Occasional cardiomyopathy	Dilated cardiomyopathy described; May occur without myopathy	Dilated cardiomyopathy in 30%–50% of patients

Respiratory	Rarely involved: PFTs rarely < 80% of normal	Rarely involved:	Functional vital capacity ranges from normal to severe	Functional vital capacity ranges from normal to severe	Variable respiratory involvement:	Variable respiratory involvement:	Variable respiratory involvement; some severe
Quality of life	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown
Muscle size	Limbs, pelvic and shoulder: Atrophy of posterior compartments	Hypertrophy: uncommon	Hypertrophy of calf and tongue in some patients	Calf hypertrophy in some patients	Prominent muscle hypertrophy	Calf hypertrophy Cramps	Calf, tongue and thigh hypertrophy Wasting in regions of weakness
Musculoskeletal	Contractures: calf (toe walking may be presenting sign)	Contractures: calf (toe walking may be presenting sign)	Lumbar hyperlordosis Scapular Winging	Scapular winging	Shoulders: scapular winging and muscle wasting	Scapular winging	Contractures in ankles (especially in non-ambulant) Scoliosis
CNS	Intelligence: Normal to mild mental retardation	No intellectual defect reported	No intellectual defect reported Hearing loss	No intellectual defect reported	No intellectual defect reported	No intellectual defect reported	No intellectual defect reported
Muscle pathology	Myopathic Necrosis and regeneration with fiber size variability Endomysial fibrosis Type I predominance with increasing weakness Normal Dystrophin and Sarcoglycan	Myopathic Necrosis and degeneration with variable fiber size □Endomysial connective tissue Absent or ↓ dysferlin staining Normal Dystrophin and Sarcoglycan	Myopathic Inflammation: occasional Severe disease: absent γ-sarcoglycan Slowly progressive: Reduced γ-sarcoglycan Dystrophin: Normal or reduced	Myopathic Degeneration and regeneration Variable fiber size Endomysial connective tissue Myopathic grouping of fibers Absent or reduced adhalin γ-sarcoglycan	Myopathic Sarcoglycans: usually absent Dystrophin: often reduced, but not absent	Myopathic Fiber degeneration Fiber regeneration □-Sarcoglycan absent Other sarcoglycans absent or reduced	Myopathic Necrosis and degeneration Variable fiber size connective tissue Type 1 fiber predominance ↓ staining for adhalin
Blood chemistry and hematology	CK: 7 to 80 times normal	CK: 10 to 72 times normal	CK: Very high	CK: Very high (often >5,000)	CK: Very high (often >5,000)	CK: 10 to 50 times normal	CK : Very high 1,000–8,000)

AR-LGMD, autosomal recessive limb girdle muscular dystrophy.

a nonlysosomal calcium-dependent proteinase specifically expressed in muscle. Muscle biopsies reveal that calpainopathy patients have normal dystrophin and sarcoglycan labeling but lack calpain-3. An early-onset form occurs before 12 years of age and has the most severe progression. The "Leyden Mobius" subtype has an onset between 13 and 30 years. Others with later onset have been reported. Pelvic girdle weakness is present and symptomatic from the onset, but often with striking sparing of the hip abductors, even relatively late into the course of the disease. Scapular winging is usually present from the early stages. The rate of deterioration varies between families. Wheelchair dependency typically occurs at 10–30 years after the onset of symptoms. The disease is predominantly symmetrical and atrophic, with prominent calves seen in only a minority of cases. Achilles tendon contractures may be an early sign, and spine deformity may also develop. Respiratory, but not cardiac, complications have been reported.

Fukutin-Related Protein

This dystrophy is caused by pathogenic mutations in the gene for fukutin-related protein (FKRP), which is involved in the glycosylation of cell surface molecules in muscle fibers (63). The majority of the LGMD 2I patients carry a common C826A missense mutation in the FKRP gene. In the LGMD 2I patients, different mutations in the FKRP gene are associated with several secondary muscle protein reductions, and the deficiencies of α2-laminin and α-DG on sections are prevalent, independent of the mutation type or the clinical severity. LGMD 2I has a relatively mild and variable course, with the age at onset varying from the first to the fourth or fifth decade of life. Progression is slow. Serum CK is elevated and intelligence is preserved, although structural brain changes have been reported.

Congenital Muscular Dystrophy

The term congenital muscular dystrophy (CMD) has been widely used for a group of infants presenting with hypotonia, muscle weakness at birth or within the first few months of life, congenital contractures, and immunohistochemical finding of dystrophic changes on muscle biopsy (muscle fiber necrosis and regeneration, increased endomysial connective tissue, and replacement of muscle with fat tissue). The early contractures may include equinovarus deformities, knee flexion contractures, hip flexion contractures, and tightness of the wrist flexors and long finger flexors. The contractures can become more severe over time with prolonged static positioning and lack of adequate passive range of motion and splinting/positioning. Classical CMDs are clinically confined to the musculoskeletal system, but other CMDs are characterized by significant cerebral neuronal migration defects and eye abnormalities. Classical CMDs are further subdivided according to the presence or absence of merosin (laminin-2) (64). An additional subgroup with collagen VI abnormalities has been identified and referred to as *Ullrich's congenital muscular dystrophy.*

Merosin-Deficient CMD

This condition (CMD 1A) has been linked to chromosome 6q22 and accounts for around half of classical CMD (64). These children show a consistently severe phenotype with multiple contractures and joint deformities (arthrogryposis) at birth. Weakness correlates with level of residual merosin (laminin α2) protein. If there is absent laminin α2 protein, weakness is severe, symmetric, proximal greater than distal, and involves the facial muscles. Contractures are present at multiple joints. CK is mildly to moderately elevated. Infants may present with respiratory failure, but if adequately supported, they can be weaned off ventilatory support. A proportion will achieve independent sitting, but independent standing or walking is almost never achieved if laminin α2 is severely reduced. Progressive spine deformity is common. The condition tends to remain relatively static, but some subjects may show slow progression. Mental development is usually normal, although minor learning disabilities and seizures do occur. Brain MRI commonly shows diffuse white matter signal changes. Nerve conduction velocities are frequently slowed, reflecting the ubiquitous expression of merosin in basement membranes. Merosin (laminin α2) is an extracellular glycoprotein that interacts with surface receptors on the sarcolemmal membrane of the muscle cell. The diagnosis of merosin-deficient CMD is dependent on the demonstration of absent merosin staining on muscle immunohistochemistry.

Merosin-Positive CMD

This is generally a milder disorder than merosin-deficient CMD and the clinical phenotype is more heterogeneous. Intellectual function is normal and the brain magnetic resonance imaging (MRI) is normal. Most of these children present with weakness and hypotonia, and they achieve the ability to stand and walk independently by age 4. The course is static, with little or no progression; however, contractures and scoliosis may develop. Respiratory failure is uncommon, as is cardiomyopathy.

Fukuyama CMD

These patients present in infancy with severe hypotonia, weakness, and wasting of the face and limbs,

occasional spasticity, large cheeks, contractures, kyphoscoliosis, microcephaly, seizures (50%), severe mental retardation (IQ 30 to 50), and occasionally progressive hydrocephalus. Muscle biopsy shows dystrophic changes. While rare in North America, the condition is common in Japan, with an incidence approaching 40% of Duchenne muscular dystrophy (65). Brain malformations are frequently seen on MRI, including polymicrogyria, pachygyria, and agyria. Frontal white matter lucencies are also evident on MR or computed tomography (CT) imaging. The gene loci has been identified to be at 9q31–33.

Muscle-Eye-Brain Disease

This is a syndrome comprising congenital muscular dystrophy, marked mental retardation due to neuronal migration defects, and ocular abnormality. Infants present with congenital hypotonia, muscle weakness, elevated CK, myopathic EMG, and a dystrophic changes on muscle biopsy. Children with muscle-eye-brain disease are usually able to stand and ambulate. Severe visual impairment is present, caused by severe myopia, retinal dysplasia, cataracts, and optic atrophy. Patients often deteriorate around 5 years of age with progressive occurrence of spasticity. CT scans have shown ventricular dilatation and low density of the white matter. Death is usually in the first or second decade, but some individuals survive well into adulthood.

Walker-Warburg Syndrome (WWS)

This is a severe condition leading to blindness at birth and early death. Infants present with congenital muscular dystrophy, mental retardation, and consistent central nervous system abnormalities on imaging (type II lissencephaly, abnormally thick cortex, decreased interdigitations between white matter and cortex, and cerebellar malformation). Ocular abnormalities and cleft lip or palate may also be present. Muscle involvement is less prominent in Walker-Warburg syndrome (WWS) than other CMDs. Several gene abnormalities with autosomal-recessive inheritance have been linked to WWS, including O-mannosyltransferase 1 (POMT1) linked to chromosome 9q34.1 and O-mannosyltransferase 2 (POMT2) linked to chromosome 14q24.3.

Ullrich Congenital Muscular Dystrophy

An emerging common group of CMD patients have a unique combination of dystrophic changes on muscle biopsy in association with weakness, low tone, selected early joint contractures, and other joints and skin demonstrating clinical laxity caused by a primary collagen VI abnormality (64). The term *collagen myopathy* is increasingly being utilized to describe these conditions. Three subunits of collagen VI have been found to be abnormal in these patients: collagen type VI, subunit α1 (COL6A1) linked to chromosome 21q22.3; collagen type VI, subunit α2 (COL6A2), also linked to chromosome 21q22.3; and collagen type VI, subunit α3 (COL6A3) linked to chromosome 2q37. Inheritance for all three groups may be recessive or dominant. Clinical features are variable, as some patients show severe weakness and some families with COL6A3 mutations have milder disease. Onset is often at birth, with congenital proximal contractures and arthrogryposis caused by reduced fetal movements, hypotonia, and early hyperlaxity of distal joints (Fig. 12.11). Knee contractures may limit walking in some. Spine rigidity and kyphoscoliosis has been noted. Torticollis may improve with increasing age. Weakness is diffuse and affects distal muscles greater than proximal and neck flexors. A minority of patients walk by age 1 to 2 years, but the majority never walk. Respiratory insufficiency and hypoventilation may begin in the first decade, and respiratory failure is not correlated with degree of weakness. The course is slowly progressive. Death has been reported in the first or second decade due to respiratory failure, but many patients live to adulthood. The skin is soft, lax, and a classic rash can often be found described as "keratosis pilaris." Patients may also show keloids, atrophic scars, striae, and petechiae. There is no associated cardiomyopathy, and intelligence is usually normal. Serum CK is normal to 10 times elevated, and EMG is usually myopathic. Muscle biopsy and skin biopsy should be obtained to make the diagnosis. Muscle biopsy shows varied muscle fiber size, some very small muscle fibers, and an increase in endomysial connective tissue. Rare or occasional necrotic muscle fibers may be found. Collagen

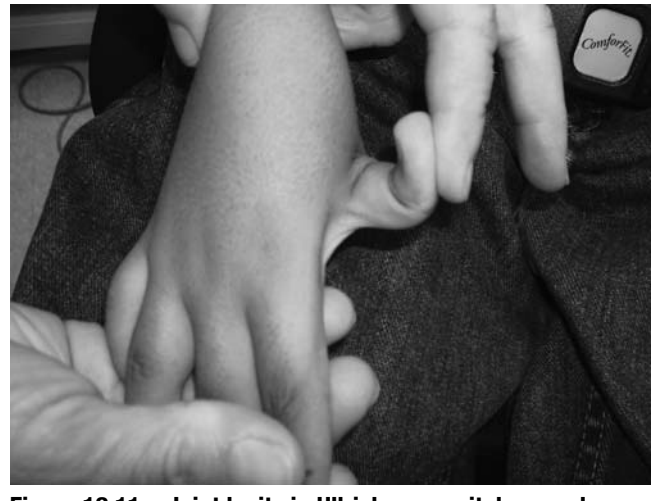


Figure 12.11 Joint laxity in Ullrich congenital muscular dystrophy with collagen VI abnormality.

VI expression may be absent in skeletal muscle and capillaries or absent on surface of muscle fibers but present in connective tissue. There has been no correlation between pattern of pathology and clinical phenotype.

Congenital Muscular Dystrophy With Early Spine Rigidity

This is a recessive condition caused by a defect in selenoprotein N, 1 (SEPN1) and linked to chromosome 1p35-p36. Clinical severity is variable, with early-onset cases in infancy and later-onset cases in the later first decade. Patients present with hypotonia and poor head control. The weakness is symmetric and involves the neck, face, and proximal and distal musculature.

Respiratory function is compromised with vital capacity below 55% by the end of the first decade. Patients often show signs of nocturnal hypoventilation and central apnea. Respiratory failure may develop. Some patients never develop walking. Muscle size is small, especially in the inner thighs and calves. Many children show early improvement, with development followed by nonprogressive or slow decline. The rigid spine develops by 3 to 7 years and is manifested by limited flexion of the neck and spine. Progressive scoliosis occurs with onset 4 to 12 years. Contractures of the elbow flexors, hip extensors, ankles, and knees are common. The rate of insulin resistance is increased, and intelligence is normal. Serum CK is usually normal. The muscle to biopsy can be best identified by MR imaging, with involved muscles often being the vastus lateralis and biceps femoris. Clinically, there is overlap with minicore congenital myopathy syndromes, and mutations in this SEPN1 gene also cause minicore congenital myopathy, congenital myopathy with desmin inclusions, and congenital fiber type size disproportion (small type I fibers).

Fascioscapulohumeral Muscular Dystrophy (FSHD)

Facioscapulohumeral muscular dystrophy (FSHD) is a slowly progressive dystrophic myopathy with predominant involvement of facial and shoulder girdle musculature (66). The condition has autosomal-dominant inheritance, with linkage to the chromosome 4q35 locus. Approximately 10% to 30% of cases are caused by sporadic mutations. FSHD is the third most common of the dystrophies, behind Duchenne and myotonic dystrophies, with an incidence of between 10 and 20 per million live births (7). Age of presentation is generally before age 20. Changes on muscle biopsy are relatively slight, with the most consistent finding being the presence of isolated small atrophic fibers. Other fibers may be hypertrophied. Serum creatine kinase levels are normal or slightly elevated in the majority of patients. Diagnosis is confirmed in more than 90% of cases by molecular genetic testing.

Facial weakness is an important clinical feature of FSHD muscular dystrophy. The initial weakness affects the facial muscles, especially the orbicularis oculi, zygomaticus, and orbicularis oris. These patients often have difficulty with eye closure but not ptosis. An individual may assume an expressionless appearance and exhibit difficulty whistling, pursing the lips, drinking through a straw, or smiling (Fig. 12.12). Even in the very early stages, forced closure of the eyelids can be easily overcome by the examiner. Masseter, temporalis, extraocular, and pharyngeal muscles characteristically are spared in FSHD.

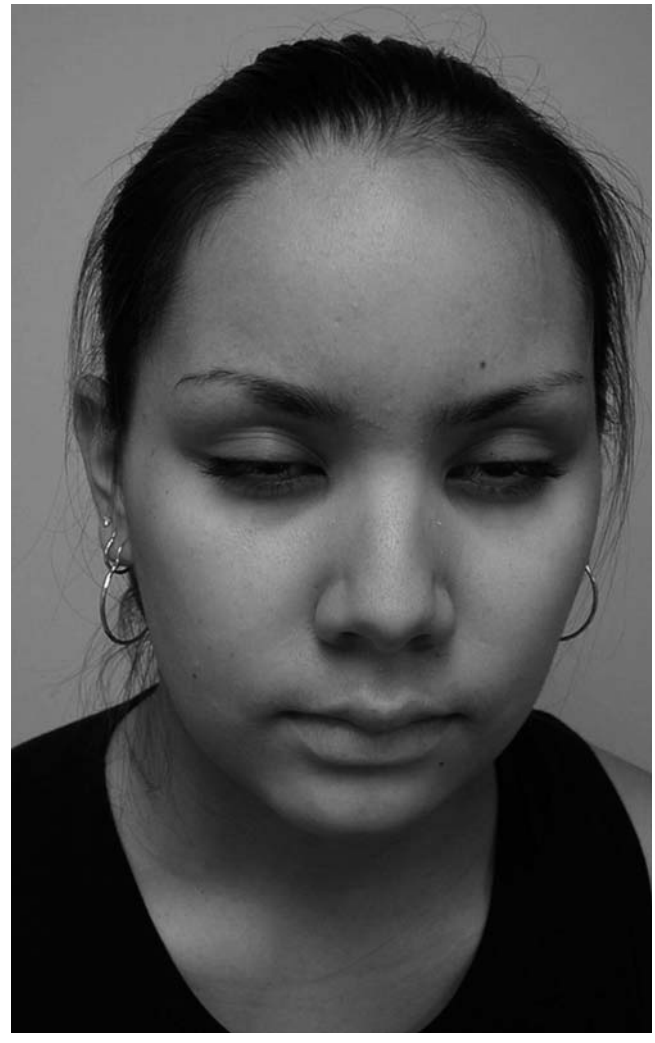


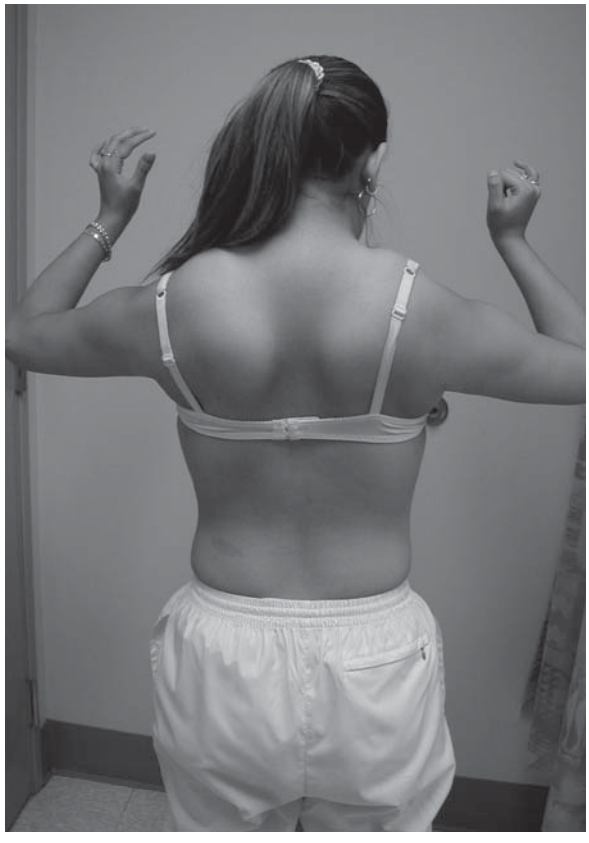
Figure 12.12 Facial weakness and expressionless facies in fascioscapulohumeral muscular dystrophy. Both father and daughter demonstrate difficulty whistling and pursing their lips.

Scapular stabilizers, shoulder abductors, and shoulder external rotators may be significantly affected, but at times the deltoids are surprisingly spared if tested with the scapulae stabilized. Both the biceps and triceps may be more affected than the deltoids. Patients with FSHD show characteristic patterns of muscle atrophy and scapular displacement. Involvement of the latissimus dorsi, lower trapezius, rhomboids, and serratus anterior results in a characteristic appearance of the shoulders, with the scapula positioned more laterally and superiorly, giving the shoulders a forward-sloped appearance (Fig. 12.13). The upper border of the scapula rises into the trapezius, falsely giving it a hypertrophied appearance. From the posterior view, the medial border of the scapula may exhibit profound posterior and lateral winging. The involvement of shoulder girdle musculature may be quite asymmetric. Some authors have found asymmetric weakness in the dominant upper extremity (67).

A sensory neural hearing deficit was originally observed in Coates syndrome (early-onset FSHD). These individuals have a myopathy that presents in infancy. The disease progression is fairly rapid, with most individuals becoming wheelchair-reliant by the late second or third decade. These individuals also have a progressive exudative telangiectasia of the retina. Early recognition and photocoagulation of the abnormal retinal vessels may prevent loss of vision. Several audiometry studies have demonstrated hearing deficits in many later-onset FSHD patients in addition to those with Coates syndrome, suggesting that impaired hearing function is more common than expected in FSHD muscular dystrophy (68). Thus, all patients with FSHD should have screening audiometry and ophthalmologic evaluation. Contractures are relatively uncommon in FSHD muscular dystrophy. FSHD patients with scoliosis have mild and nonprogressive curves. Rarely, severe and progressive hyperlordosis is associated with FSHD. The patient with severe hyperlordosis may utilize their lordotic posturing to compensate for hip extensor weakness.

Mild restrictive lung disease has been reported in nearly one-half of FSHD patients (66). The expiratory muscles involved in respiration appear to be more affected than inspiratory muscles in FSHD (67). Patients rarely require nocturnal ventilatory support.

The presence of cardiac abnormalities in FSHD muscular dystrophy is debated. While diverse ECG abnormalities have been noted, one study showed no abnormalities on ECG, chest radiography, Holter monitoring, and echocardiography (69). Nuclear scanning with thallium-201 has demonstrated diffuse defects consistent with diffuse fibrosis (32). Abnormalities in systolic time intervals on echocardiography and elevations in atrial natriuretic peptide are consistent with subclinical cardiomyopathy. Cardiac complications in FSHD muscular dystrophy are rare, and patients in general have normal longevity. There is usually no associated intellectual involvement in this dystrophic myopathy.



A

B

Figure 12.13 (A) Posterior and lateral scapular winging, high-riding scapula, and (B) hyperlordosis in fascioscapulohumeral muscular dystrophy.

Emery-Dreifuss Muscular Dystrophy (EMD)

Emery-Dreifuss muscular dystrophy (EMD) refers to a group of muscular dystrophies with weakness, contractures and cardiac conduction abnormalities. Inheritance pattern is variable among subtypes.

Emery-Dreifuss 1

Emery-Dreifuss 1 (EMD1) is an X-linked recessive progressive dystrophic myopathy due to an abnormality of the protein "Emerin" with a gene locus identified at Xq28 (70,71). The protein is associated with the subcellular nucleus and cytoplasm membranes, and is found in muscle, nerve, mucosal epithelium, skin, and cardiac tissue. Patients usually present in teenage years, but age of presentation may vary from the neonatal period with hypotonia to the third decade. Early elbow flexion contractures are a hallmark of the disease. Severe contractures, including elbow flexion, ankle equinus, rigid spine, and neck extension contractures, are often more limiting than weakness, which begins in a sapulohumeral peroneal distribution. The biceps and triceps show wasting and weakness, and the deltoids and forearms are often more spared. The calf frequently shows wasting. Ankle dorsiflexors often are weaker than ankle plantar flexors leading to the equinus contractures. Scapular winging is frequent. Tightness of the cervical and lumbar spinal extensor muscles, resulting in limitation of neck and trunk flexion, with inability to flex the chin to the sternum and to touch the toes, also has been reported in EMD. The face is either spared or affected late. Functional difficulties are experienced walking or climbing stairs. Progression is slow and loss of ambulation is rare. Some cases with EMD1 may show evidence of nocturnal hypoventilation, as a result of restrictive expansion of the chest in association with the rigid spine, and partly due to involvement of the diaphragm.

Progressive cardiac disese is almost invariably present, with onset in the early second decade to the fourth decade. Arrhythmia may lead to emboli or sudden death in early adult life. The cardiomyopathy may progress to left ventricular myocardial dysfunction or four-chamber dilated cardiomyopathy due to fibrosis with complete heart block and ventricular arrhythmias (72). Initially, atrial arrhythmia usually appears prior to complete heart block. Reported features include first-degree heart block, followed by Wenckebach phenomenon, and then complete atrial ventricular dissociation and atrial fibrillation or flutter with progressive slowing of the rate (72). Frank syncope may develop in the late second and early third decade, and patients often require a cardiac pacemaker by age 30 with an indication being bradycardia with heart rate below 50. EKG changes include slow heart rate, absent or small P waves, AV block, and atrial fibrillation/flutter. Evidence of cardiac arrhythmia, sometimes only present at night, may be detected on 24-hour Holter monitoring. A significant percentage of female carriers have conduction defects and arrhythmias, so they warrant monitoring with annual EKGs.

Laboratory evaluation is usually with molecular genetic studies and/or muscle biopsy. Serum CK is mildly elevated to <10 times normal, and levels decrease with age. MRI of posterior calf shows the soleus to be involved and the gastrocnemius to be relatively spared. Muscle biopsy shows variable muscle fiber size, endomysial fibrosis, inflammation, type I fiber atrophy, type I or II predominance, and nuclear membrane pathology on electron microscopy. Other muscle changes in some patients include rimmed vacuoles ("IBM-like"). Immunohistochemistry reveals Emerin loss in muscle in >95% of patients.

Emery Dreifuss Muscular Dystrophy 2

EMD2 is due to a lamin A/C protein abnormality, and it has been linked to chromosome 1q21.2. Inheritance may be dominant or recessive, and lamin A/C mutations may be either frameshift or missense (70). Those with missense mutations have childhood onset, with a mean age of onset of 2.4 years. Weakness is in a scapuloperoneal and facial distribution. Patients demonstrate paravertebral weakness or rigidity, and tendon contractures are common. Those with frameshift mutations producing a truncated protein have adult onset, with mean age of 30.5 years, and cardiomyopathy is more frequent than weakness (70). Contractures are rare, and weakness is in a limb girdle distribution. The disorder is allelic with autosomal-dominant LGMD 1B.

CONGENITAL MYOPATHIES

The term *congenital myopathy* is used to describe a group of heterogenous disorders usually presenting with infantile hypotonia due to genetic defects, causing primary myopathies with the absence of any structural abnormality of the central nervous system or peripheral nerves. A specific diagnosis of each entity is made on the basis of specific histologic and electron microscopic changes found on muscle biopsy. While patients may be hypotonic during early infancy, they later develop muscle weakness that is generally nonprogressive and static. The weakness is predominantly proximal, symmetric, and in a limb girdle distribution.

The serum creatine kinase values are frequently normal, and the EMG may be normal or may show mild, nonspecific changes, usually of a myopathic

character (small-amplitude polyphasic potentials). The only congenital myopathy consistently associated with spontaneous activity is myotubular (centronuclear) myopathy. In this disorder, the EMG reveals myopathic motor unit action potentials with frequent complex repetitive discharges and diffuse fibrillation potentials. These myopathies may be considered primarily structural in nature and thus, patients do not actively lose muscle fibers, as is the case in dystrophic myopathies.

Central Core Myopathy

This is an autosomal-dominant disorder with gene locus at 19q13.1, the same gene locus as the malignant hyperthermia gene (ryanodine receptor gene, RYR1). Indeed, these patients have a high incidence of malignant hyperthermia with inhalational anesthetic agents. Histologically, the muscle fibers have amorphus-looking central areas within the muscle that may be devoid of enzyme activity. There are densely packed disorganized myofibrils ("cores") in the center of the majority of type 1 fibers. Electron microscopy shows the virtual absence of mitochondria and sarcoplasmic reticulum in the core region, reduced muscle enzymes (cytochrome oxidase, NADH), a marked reduction in the interfibrillary space, and an irregular zig-zag pattern (streaming) of the Z-lines. This gives rise to the characteristic central pallor. There is a predominance of high oxidative, low glycolytic type I fibers and a relative paucity of type II fibers, resulting in a relative deficiency of glycolytic enzymes.

Clinically, patients generally demonstrate mild and relatively nonprogressive muscle weakness, either proximal or generalized, and arreflexia, which presents in either early infancy or later. There may be mild facial weakness but normal extraocular movements. Patients often achieve gross motor milestones, such as walking, rather late, and they continue to have difficulty going upstairs. Proximal limb weakness is typical, and patients may show a Gower's sign. The disorder remains fairly static over the years. There may be a frequent occurrence of congenital dislocation of the hip, kyphoscoliosis, and pes cavus. The condition is largely nonprogressive, with affected children remaining ambulatory into adult life. One-third show anesthesia-related malignant hyperthermia. Central core myopathy and familial malignant hyperthermia appear to be allelic, as the ryanodine receptor chain implicated in malignant hyperthermia has the same locus. Individuals within the same family can exhibit one or both phenotypes.

Nemaline Myopathy

Nemaline myopathy, also referred to as rod body myopathy, represents a varied group of disorders with different modes of inheritance, but the most typical form is autosomal recessive. While the rods may be easily overlooked on routine hematoxylin-and-eosin (H&E) staining, they can be rarely demonstrated with the Gomori trichrome stain. The rods are readily demonstrated on electron microscopy. They are thought to be an abnormal deposition of Z-band material of a protein nature and possibly alpha-actinin. The disease has been linked to at least seven distinct genes. The severe congenital form has been linked to α-Actin, nebulin, and troponin T1 mutations. A milder childhood form has been linked to α-actin, nebulin, α-tropomyosin 3 (TPM3), and β-tropomyosin (TPM2) mutations.

A severe form of the disease may present in the neonatal period with severe weakness, respiratory insufficiency, and often a fatal outcome. Most cases present with a mild, nonprogressive myopathy with hypotonia and proximal weakness. In more severe cases, swallowing difficulty may be present in the neonatal period. Skeletal abnormalities, such as kyphoscoliosis, pigeon chest, pes cavus feet, high arched palate, tent-shaped mouth, and an unusually long face has been noted. Cardiomyopathy has been described in both severe neonatal and milder forms of the disease. Autosomal-dominant inheritance has been described in a few instances.

Centronuclear (Myotubular) Myopathy (Non-X–linked)

Patients with non-X–linked myotubular myopathy have muscle biopsies that show a striking resemblance to the myotubes of fetal muscle. Patients typically present with early hypotonia, delay in motor milestones, generalized weakness of both proximal and distal musculature, and ptosis with weakness of the external ocular muscles, as well as weakness of the axial musculature. The author has seen severe cardiomyopathy in an adult female with documented autosomal-dominant inheritance. Nocturnal hypoventilation has been described.

Several gene loci with autosomal-dominant inheritance have been identified in centronuclear congenital myopathy, including dynamin 2 (DNM2) linked to chromosome 19p13.2, and MYF6 linked to chromosome 12q21.

Severe X-linked Centronuclear (Myotubular) Myopathy

Cases with neonatal onset and severe respiratory insufficiency have been identified with an X-linked recessive mode of inheritance. The gene for this disorder codes for myotubularin (MTM1), and has been linked to chromosome Xq27.3-q28. Muscle biopsy shows

characteristic fetal-appearing myotubes with rows of centrally placed internal nuclei.

Patients present with severe generalized hypotonia, associated muscle weakness, swallowing difficulty, and respiratory insufficiency. They often become ventilator-dependent at birth. If they are able to be weaned from the ventilator, subsequent death due to pulmonary complications is not uncommon. Mean age of death is 5 months, but some children survive for many years with mechanical ventilation. Aspiration pneumonias are common. Additional clinical features include congenital contractures, facial weakness with an elongated expressionless face, tent-shaped mouth, high arched palate, weakness of the external ocular muscles, and long digits. Progressive kyphoscoliosis is common. Systemic features in some survivors >1 year of age include pyloric stenosis, spherocytosis, gallstones, renal stones or calcinosis, a vitamin K-responsive bleeding diathesis, rapid linear skeletal growth, advanced bone age, and hepatic dysfunction. Electromyography shows many fibrillations and positive sharp waves.

Minicore Disease (Multicore Disease)

This is a relatively rare congenital myopathy with muscle biopsies showing multiple small randomly distributed areas in the muscle, with focal decrease in mitochondrial oxidative enzyme activity and focal myofibrillar degenerative change. Characteristic changes are present on electron microscopy. There is a predominance of type I fiber involvement.

Clinically, patients present with hypotonia, delays in gross motor development, and nonprogressive symmetric weakness of the trunk and proximal limb musculature. There may be mild facial weakness, ptosis, and ophthalmoplegia. There is also associated diaphragmatic weakness, placing patients at risk for nocturnal hypoventilation. Subtle ultrastructural changes allow this condition to be distinguished from central core disease. The cores are smaller in size (minicores) and not confined to the center of the fiber. Inheritance is usually autosomal-recessive, and two genes—the ryanodine receptor gene (RYR1), linked to chromosome 19q13.1, and the selenoprotein N, 1 (SEPN1) gene, linked chromosome 1p35-p36, account for 50% of cases.

Congenital Fiber-Type Size Disproportion

Congenital fiber-type size disproportion represents a heterogenous group of conditions most likely with varied genetic defects. The condition was initially delineated by Brooke (73) on the basis of the muscle biopsy picture demonstrating type I fibers that are smaller than type II fibers by a margin of more than 12% of the diameter of the type II fibers. The mean reduction in fiber diameter is 41% and ranges up to 78%. A number of disorders, such as congenital myopathies (nemaline rod, centronuclear, and multi-minicore), Emery-Dreifuss MD and myotonic dystrophy 1, rigid spine syndromes, congenital muscular dystrophy (SEPN1), LGMD 2A, and severe spinal muscular atrophy, all may show small type I fibers and should be excluded. The diagnosis of congenital fiber-type disproportion should be made only in the presence of normal-sized or enlarged type II fibers and not in cases where both type I and type II fibers are small. Serum CK has been normal to <3 times the upper limit of normal.

Patients typically present with infantile hypotonia and delay in gross motor milestones. The severity has been noted to be quite variable, but it is generally nonprogressive or improves with time. Limb weakness of variable severity may be diffuse or affect proximal muscles. Deep tendon reflexes are reduced. Ophthalmoplegia, facial weakness, and bulbar weakness are rare findings but associated with more severe cases. Intelligence is normal. There is generally short stature and low weight. Patients may exhibit a long, narrow face; high-arched palate; and deformities of the feet, including either flat feet or occasionally high-arched feet. Kyphoscoliosis has been reported. Lenard and Goebel (74) documented a case with fairly severe weakness and associated respiratory deficit, necessitating tracheostomy. The author has managed two cases (a mother and son with presumed autosomal-dominant inheritance), who both developed nocturnal hypoventilation requiring bilevel positive airway pressure.

Patients with muscle biopsies indicative of congenital fiber-type disproportion and ptosis should be evaluated for a congenital myasthenic syndrome, as the author has seen a number of cases in recent years of congenital structural neuromuscular junction disorders that have associated nonspecific changes on muscle biopsy, interpreted to be congenital fiber-type disproportion. This is an important distinction, as some of these patients with congenital myasthenia respond to pharmacologic intervention.

The mode of inheritance for congenital fiber-type disproportion is varied, with both autosomal-recessive and autosomal-dominant patterns of inheritance reported.

MYOTONIC DISORDERS

Myotonic Muscular Dystrophy 1 (DM1)

Myotonic muscular dystrophy 1 (DM1) is an autosomal-dominant multisystem muscular dystrophy

with an incidence of 1 per 8,000 (7). It represents the most common inherited neuromuscular disease of adults. The disorder affects skeletal muscle, smooth muscle, myocardium, brain, and ocular structures. Associated findings include baldness and gonadal atrophy (in males), cataracts, and cardiac dysrhythmias. Insulin insensitivity may be present. The gene has been localized to the region of the myotonin protein kinase (DMPK) gene at 19q13.3. Patients demonstrate expansion of an unstable CTG trinucleotide repeat within the region. Molecular genetic testing is available for diagnosis. Normal individuals generally have <37 repeats, which are transmitted from generation to generation. DM1 patients may have 50 to several thousand CTG repeats with remarkable instability. The age of onset is inversely correlated by the repeat links (75). Mild, late-onset DM1 usually is associated with 50–150 repeats, classic adolescent or young adult-onset DM1 shows 100–1,000 repeats, and congenital DM1 patients show greater than 1,000 repeats. The expanded CTG repeat further expands as it is transmitted to successive generations, providing a molecular basis for genetic anticipation. Both maternal-to-child and paternal-to-child transmission occurs. Repeat size in offspring exceeding 1,000 CTG repeats is generally seen in maternal rather than paternal transmission. Affected fathers seldom transmit alleles larger than 1,000 copies to offspring owing to a lack of sperm containing such alleles.

Several characteristic facial features of DM1 may be noted on inspection (Fig. 12.14A). The adult with long-standing DM1 often has characteristic facial features. The long, thin face with temporal and masseter wasting is drawn and has been described by some as "lugubrious." Adult males often exhibit frontal balding. Children with congenital myotonic muscular dystrophy often exhibit a triangular or so-called tent-shaped mouth (Fig. 14B).

Myotonia, a state of delayed relaxation, or sustained contraction of skeletal muscle is easily identified in school-age children, adolescents, and adults with DM1. Grip myotonia may be demonstrated by delayed opening of the hand with difficult extension of the fingers following tight grip. Percussion myotonia may be elicited by percussion of thenar eminence with a reflex hammer, giving an adduction and flexion of the thumb with slow return (see Fig. 12.4). Symptomatic myotonia may be treated with agents such as mexiletine or membrane stabilizers such as carbamazepine or dilantin, which have been shown to affect the symptoms; however, patients treated have shown little functional gain (76).

DM1 is one of the few dystrophic myopathies with greater distal weakness than proximal weakness, although neck flexors, shoulder girdle musculature, and pelvic girdle musculature may become significantly involved over decades. Weakness initially is often most predominant in the ankle dorsiflexors, ankle everters and inverters, and hand muscles (77). As with other dystrophic myopathies, significant muscle wasting may occur over time. In DM1 patients with infantile onset, a congenital club foot or talipes equinovarus are fairly common deformities (Fig. 12.15). In patients with noncongenital DM1, contractures at the wrist, ankle, and elbows are relatively uncommon and

A

B

Figure 12.14 (A) Typical facial characteristics in myotonic muscular dystrophy 1 (DM1) and congenital DM1. The symptomatic mother has 660 trinucleotide CTG repeats at the DM protein kinase (DMPK) gene loci in chromosome 19q13.3, while the child has 1,560 repeats. (B) Siblings with DM1.

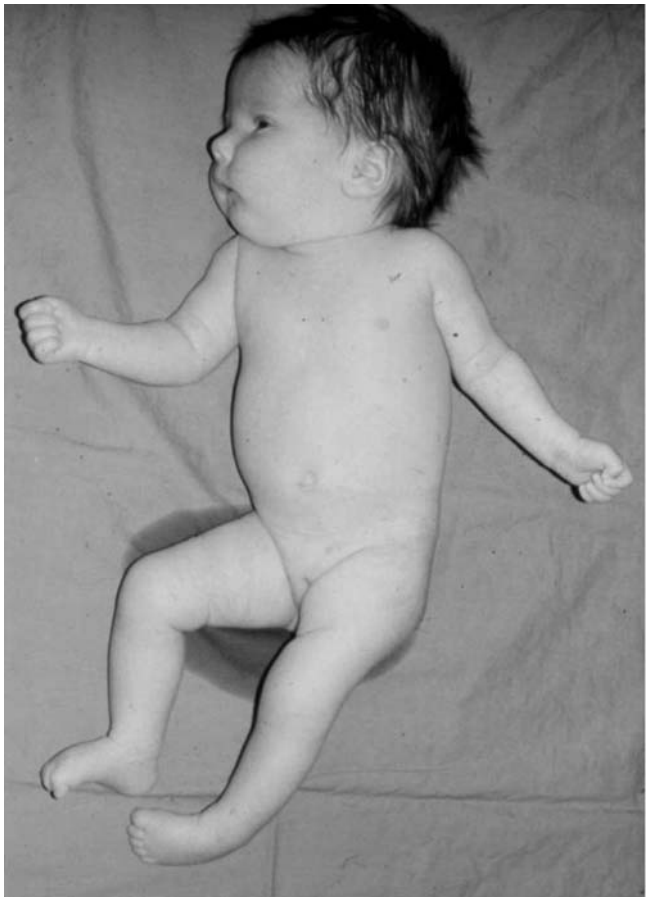


Figure 12.15 Talipes equinus in congenital myotonic muscular dystrophy 1 (DM1).

mild (77). Patients with congenital-onset DM1 may develop spinal deformity requiring surgical spinal arthrodesis (77).

Excessive daytime sleepiness is commonly seen in DM1, and it is thought to be related to the loss of serotonergic neurons in the dorsal raphe and superior central nucleus of the brainstem. Treatment of the hypersomnolence with modafinil has been helpful.

Endocrinopathies are frequently found in DM1. These include hypothyroidism, increased insulin resistance and type 2 diabetes, reduced insulinlike growth factor-1 (IGF-1) levels, hypogonadism in males with reduced testosterone levels and oilgospermia, pituitary deficiency with reduced growth hormone release, and increased follicle-stimulatin hormone (FSH) levels. DM1 patients should also be screened for diabetes mellitus, as insulin insensitivity is not uncommon.

Cardiac involvement is common in DM1. Abnormalities on ECG and echocardiography are demonstrated in approximately 70% to 75% of patients (77). Prolongation of the PR interval, abnormal axis, and infranodal conduction abnormalities are all suggestive of conduction system disease, which may explain the occurrence of sudden death, which occurs in less than 5% of DM1 patients (78). Ventricular tachycardia may also contribute to the syncope and sudden death associated with DM1. Some patients have required implantation of cardiac pacemakers. Q-waves have been reported on screening ECGs in DM1 patients, and this abnormality may reflect myocardial fibrosis (77,78). Occassionally, teenagers may present with atrial arrythmias. Any DM1 patient with dyspnea, chest pain, syncope, or other cardiac symptoms should receive thorough cardiac evaluation.

Individuals with congenital and noncongenital DM1 have a very high incidence of restrictive lung disease (77). Involvement of respiratory muscles is a major cause of respiratory distress and mortality in affected infants with DM1. Swallowing difficulties that produce aspiration of material into the trachea and bronchial tree, along with weakened respiratory muscles and a weak cough, have been reported as factors that may result in pulmonary complications in DM1 patients. Constipation is a fairly common complaint in congenital DM1, owing to smooth muscle involvement. Care should be taken during general anesthesia in DM1 due to risk of cardiac arrhythmias and malignant hyperthermia.

Twenty-five percent of infants born to myotonic mothers have congenital DM1 and 10% to 15% of all DM1 patients have congenital presentations. CTG repeats in these cases may range from 1,000 to more than 4,000 repeats. Obstetric problems are inversely related to age of presentation of the mother with DM1, and they include polyhydramnios, decreased fetal movements, breech presentation, and preterm labor. Infants show hypotonia, failure to thrive due to an inability to suck, bilateral facial and jaw muscle weakness, craniofacial changes (including a tented upper lip and high-arched palate), neonatal respiratory distress (50%), delayed motor milestones, and delayed speech. Equinovarus deformaties are common. Most children are weaned from the ventilator and walk independently. Clinically, children with congenital DM1 usually show no myotonia over the first five years of life. Those with congenital DM1 usually show significantly reduced IQ, often in the mentally retarded range (77,79). The cognitive impairment is nonprogressive. Behavioral abnormalities include hyperactivity attention-deficit and autistic behavior. Hydrocephalus may be seen in nearly half of patients with congenital DM1. MRI may show hypoplasia of corpus callosum and cerebral white matter changes and diffuse cerebral atrophy. Diagnosis of congenital DM1 is made by molecular genetic studies, as EMG shows no myotonia and CK is usually normal. Muscle biopsy is normal or nonspecific.

In noncongenital DM1, there is evidence for a generally lower intelligence of a mild degree (full-scale IQs have been reported in the 86–92 range) (77). There is a wide range of IQ values found in this population, with many subjects scoring in the above-average range. Cognitive functioning also appears to be related to the size of the CTG expansion at the DM1 gene locus.

Proximal Myotonic Myopathy (PROMM; DM2)

Proximal myotonic myopathy, also referred to as myotonic muscular dystrophy 2 (DM2), is a disorder with clinical similarities to DM1 (80). The abnormal protein in this autosomal-dominant disorder is the zinc finger protein 9 (ZNF9) with genetic loci at chromosome 3q21. Clinical severity is unrelated to variable-size CCTG repeats. The prognosis is more benign than DM1, and there is not a severe congenital onset form. Onset is 8 to 60 years, and there is intrafamilial variability. Patients present with muscle stiffness and pain. Weakness involves the proximal legs (hip flexors and extensors) greater than the proximal legs as well as thumb and finger flexors. Facial weakness is seen in a minority of patients. Distal legs and respiratory muscles are not clinically affected. A hallmark is the enlargement of calf muscles. Muscle pain may be exercise-related, or at rest and increases with cold. The myotonia is severe, asymmetric, and intermittent from day to day. The myotonia actually inceases with warmth and decreases with cold. There is both grip and percussion myotonia. Cataracts are noted in all patients over 20 years with slit lamp examinations. Cardiac conduction defects are present in 20%, diabetes mellitus in 20%, and hearing loss in 20%. MRI shows white matter hyperintensity on T2-weighted images. CK is normal to less than 10 times elevated. EMG shows profound myotonia and compound muscle action potential (CMAP) amplitudes increment by 60% with exercise and reduce by 40% with rest. There is no decrement on short exercise or slow or rapid repetitive stimulation. Myopathic motor units are seen proximally. MRI shows selective muscle involvement of the erector spinae and gluteus maximus. Diagnosis is confirmed by molecular genetic studies.

Myotonia Congenita

Myotonia congenita (Thomsen's disease) presents in infancy and is inherited as an autosomal-dominant condition. There is an abnormality of the muscle chloride channel, and the disease is linked to the 7q35 loci. There is variable penetrance. Symptoms may be present from birth, but usually develop later. The myotonia is relatively mild and may manifest as difficulty in releasing objects or difficulty walking or climbing stairs. Most patients do not show overt weakness. Functional difficulties in climbing stairs may be present. The myotonia is exacerbated by prolonged rest or inactivity. There is a "warm-up" phenomenon with reduced myotonia after repeated activity. Myotonia may be aggravated by cold, hunger, fatigue, and emotional upset. Patients may demonstrate grip myotonia or lid lag following upward gaze or squint and diplopia following sustained conjugate movement of the eyes in one direction. Nearly all have electrical myotonia by EMG, but there is a warm-up phenomenon with the myotonia reduced after a period of maximal contraction. Half of individuals have percussion myotonia. Patients may be symptom-free for weeks to months. The other common feature of myotonia congenita is muscle hypertrophy. Patients may exhibit a "Herculean" appearance. Patients have shown some benefit from treatment with quinine, mexiletine, dilantin, procainamide, carbamazepine, and acetazolamide.

A recessive form of myotonia congenita (Becker form) also exists with later onset (ages 4 to 12), more marked myotonia, more striking hypertrophy of muscles, and associated weakness of muscles, particularly with short exercise. EMG shows myotonia in distal muscles and less myotonia after maximal contraction. On repetitive stimulation, there is a decremental CMAP response at high stimulation frequency (30 Hz) and following exercise. The dominant form seems more prone to aggravation of the myotonia by cold. Diagnosis is suspected based on clinical information and the presence of classical myotonic discharges on EMG. Diagnosis is confirmed with molecular genetic testing. Muscle biopsy is essentially normal, apart from the presence of hypertrophy of fibers and an absence of type II-B fibers.

Paramyotonia Congenita

Paramyotonia congenita is an autosomal-dominant myotonic condition with at least two distinct genetic etiologies involving the sodium channel: α subunit (SCN4A) located at chromosome 17q35 and a muscle chloride channel (CLCN1) located at chromosome 7q35. The worsening of the myotonia with exercise is referred to as paradoxical myotonia. Weakness or stiffness may occur together or separately, there is cold and exercise aggravation, hypertrophy of musculature, and more severe involvement of hands and muscles of the face and neck. Myotonic episodes usually subside within a matter of hours, but may last days. Some patients become worse with a potassium load. On electrodiagnostic studies, there is a drop in CMAP amplitude with cooling. Dense fibrillations disappear below 28 degrees Celsius, myotonic bursts disappear below 20 degrees Celsius, and electrical silence

may occur below 20 degress Celsius. Treatment has involved mexiletine or tocainide.

Schwartz-Jampel Syndrome (Chondrodystrophic Myotonia)

Schwartz-Jampel syndrome is an autosomal-recessive disorder with myotonia, dwarfism, diffuse bone disease, narrow palpebral fissures, blepharospasm, micrognathia, and flattened facies (see Fig. 12.10). Onset is usually before age 3. Patients have respiratory and feeding difficulties with impaired swallowing. Limitation of joint movement may be present along with skeletal abnormalities, including short neck and kyphoscoliosis. Muscles are typically hypertrophic and clinically stiff. There is a characteristic facies with pursed lips, micrognathia, and small mouth. Patients may be difficult to intubate. Ocular changes include myopia and cataracts. There may be hirsutism and small testes. The symptoms are not progressive. The protein perlecan with gene loci at chromosome 1p34-p36 has been implicated.

Electrodiagnostic studies show continuous electrical activity, with electrical silence being difficult to obtain. There is relatively little waxing and waning in either amplitude or frequency of complex repetitive discharges. Abnormal sodium channel kinetics in the sarcolemma of muscle has been demonstrated. Some therapeutic benefit has been reported with procainamide and carbomezapine.

METABOLIC MYOPATHIES

Inborn errors of glycogen metabolism and fatty acid metabolism may result in neuromuscular disorders. The major clinical presentations include fixed and progressive weakness or exercise intolerance, cramps and myalgias, and myoglobinuria.

Fixed and progressive weakness may be caused by glycogenoses (acid maltase deficiency or "Pompe disease," debrancher deficiency, brancher deficiency, and aldolase A deficiency), or disorders of lipid metabolism (primary systemic carnitine deficiency, primary myopathic carnitine deficiency, secondary carnitine deficiency, short-chain acyocoenzyme A synthetase defiency [SCAD], medium-chain acylocoenzyme A synthetase dehydrogenase deficiency [MCAD], etc.).

Exercise intolerance, cramps/myalgias, and myoglobinuria may be caused by glycogenoses (myophosphorlase deficiency or "McArdle's disease," phosphorylase kinase defiency, phosphofructokinase [PFK] deficiency, phosphoglycerate mutase deficiency [PGAM], etc.); disorders of lipid metabolism (carnitine palmitoyltranferase II deficiency [CPT II], VLCAD deficiency, and TP deficiency, etc.); and respiratory chain defects (coenzyme Q10 deficiency, complex I deficiency, complex III deficiency, and complex IV deficiency). While an exhaustive review of metabolic myopathies is not presented here, two prototypical metabolic myopathies—McArdle's disease and Pompe's disease—deserve mention.

Myophosphorylase Deficiency

The most common glycogen storage disease is myophosphorylas deficiency, also known as McArdle's disease or glycogenosis type 5. The autosomal-recessive disorder has been linked to chromosome 11q13, and more than 65 different disease-causing mutations have been identified. Initial onset of symptoms often occurs during childhood and consists of poor endurance, fatigue, and exercise-induced cramps and myalgia that mainly affects active muscle groups. Myoglobinuria may also be absent during childhood, with prevalence of fixed muscle weakness increasing as patient ages. Symptoms can be precipitated by activities such as lifting heavy weights or climbing long flights of stairs. The "second wind phenomenon" is characteristic of this disorder. With the onset of myalgia, patients who rest briefly are then able to continue their physical activity with few or no symptoms. The normal function of muscle myophosphorylase is to catalyze the removal of 1,4-glycosyl residues from glycogen to produce glucose-1-phosphate. Its absence leads to decreased metabolic substrate for glycolysis to produce adenosine triphosphate. CK is persistently elevated between episodes of myoglobinuria. EMG is normal when patients are asymptomatic, but can show myotonic discharges and fibrillation potentials during an acute attack. Nonischemic forearm exercise testing shows only an increase in ammonia and stable levels of lactic acid and pyruvate. Diagnosisis made by demonstrating absence of myophosphorylase on muscle biopsy or by genetic mutation analysis. Possible treatments include high protein diet, pyridoxine, and creatine monohydrate.

Acid Maltase Deficiency

Acid maltase deficiency, also referred to as glycogenosis type 2 or Pompe's disease, is caused by a deficiency of acid α-1,4-glucosidase (GAA). Inheritance is autosomal recessive, with linkage to chromosome 17q23. Disease incidence is 1 in 40,000 to 50,000 live births. The level of residual enzyme activity correlates with the severity of disease. Those with infantile onset (birth to 1 year) show <1% GAA activity, childhood and juvenile onset (1 year to teens) show 2% to 6% GAA activity, and those with adult onset (third decade or later) show 1% to 29% GAA activity. There is no clear correlation with residual activity within adult

population. There is glycogen accumulation in tissues. Clinically in those with infantile onset, symptoms and signs usually present within the first six months, with hypotonia, weakness, cardiomegaly, congestive heart failure, and arrhythmia. There is liver involvement and pulmonary involvement. Anesthesia risks with succinylcholine include arrhythmia, hyperkalemia, and rhabdomyolysis. Propofol also produces risks. Safer anesthetics include ketamine and etomidate. Death occurs within the first year of life in 80% to 95% of untreated patients. In childhood onset, there is mildly enlarged tongue, symmetric proximal weakness, and calf hypertrophy. Death occurs between 3 to 24 years due to respiratory failure. There is glycogen accumulation mainly in muscle. In adult-onset Pompe's, patients present with lower extremity weakness, restrictive lung disease from diaphragm involvement, headache, somnolence, and increased dyspnea when supine. Sleep-disordered breathing is common. Expiration is more involved than inspiration due to chest wall muscle involvement. Nocturnal noninvasive ventilation is occasionally necessary. There is atrophy of paraspinous muscles and scapular winging. The disease course is one of slow progression over years. Pain, fatigue, and cramps are common complaints. There may be mild calf hypertrophy and diffuse muscle atrophy more proximally. Progressive disability is related to disease duration rather than age of onset. Eventual respiratory involvement is common, and many need wheelchairs or walking devices. Death is most often due to respiratory failure.

Diagnosis of Pompe's disease is confirmed with either molecular genetic studies or biochemical analysis of acid maltase activity with muscle biopsy. However, new methods using blood samples to measure GAA activity are rapidly becoming adopted because of their speed and convenience (81,82). Typically, serum CK is elevated (less than 10 times) in infants and less elevated in adults. EMG shows an irritative myopathy with fibrillations, complex repetitive discharges, and myotonic discharges. Treatment now involves enzyme replacement with intravenous administration of recombinant α-glucosidase (Genezyme). Better outcomes are seen with earlier initiation of therapy. Genzyme has been shown to benefit infantile disease and possibly late-onset disease with improved strength of distal and proximal muscles, improved pulmonary function, improved cardiomyopathy, and improved survival (83,84).

MITOCHONDRIAL DISORDERS

Mitochondrial encephalomyopathies, also referred to as mitochondrial cytopathy, represent a complex group of disorders that affect multiple organ systems. Mitochondria are essential cellular organelles that convert carbohydrates, lipids, and proteins into usable energy in the form of adenosine triphosphate (ATP) via aerobic metabolism. Although the human mitochondrial genome is only 16.5 Kb and encodes 13 proteins, many different clinical syndromes can result from mutations of these genes. Mutant mitochondrial DNA can be present in different proportions in various cell populations in a phenomenon known as heteroplasmy. The pathogenic effect of the mutation will only be manifested when a critical level of mutation is reached. Mutant and normal mitochondrial DNA segregate randomly during cell division, thus changing the proportion of mutant DNA in different cells over time. All mitochondria and mitochondrial DNA are derived from the mother's oocyte. Thus, a family history compatible with maternal inheritance is strong evidence for a primary mitochondrial DNA mutation. Different family members in the maternal lineage may be asymptomatic or oligospermatic.

Of the many clinical features of mitochondrial disorders that involve multiple organ systems, some are frequently present together and should alert the clinician to a mitochondrial etiology. Ptosis, progressive external ophthalmoplegia (PEO), or both are hallmarks of Kearns-Sayre syndrome, which produces diplopia and blurred vision. Myopathy is common among patients with mitochondrial disorders. Neck flexors may be affected earlier and more severely than neck extensors. Progressive fixed proximal weakness is more common, and patients may develop decreased muscle bulk. Premature fatigue, exercise intolerance, myalgia, and recurrent myoglobinuria can be symptoms of mitochondrial disorders. Serum lactate and pyruvate often are elevated at rest, and these levels may increase significantly after moderate exercise. Sensorineural hearing loss is frequently associated with mitochondrial encephalomyopathies. The hearing loss may be asymmetric and fluctuating in severity. Maternally inherited deafness and diabetes (DAD) is another phenotypic combination in patients with mitochondrial DNA mutations. Dementia can be a prominent feature in mitochondrial cytopathy.

The diagnostic workup of a mitochondrial disorder often includes a complete blood count, serum electrolytes (including calcium and phosphate), liver function tests, blood urea nitrogen, creatinine, blood lactate and pyruvate, ECG, lumbar puncture for CSF protein, glucose, lactate, pyruvate, EMG and nerve conduction study, brain imaging with MRI, and muscle biopsy for histology and electron microscopy. Histochemical stains for mitochondrial enzymes (SDH, NADH-TR, and COX) may be obtained, and the activities of mitochondrial respiratory chain enzymes can be measured in muscle tissue. The identification of numerous mitochondrial DNA (mtDNA) mutations, including

duplications, deletions, multiple deletions, and more than 100 pathgenic point mutations, provides specific genetic diagnoses.

Treatment is symptomatic for seizures (with avoidance of valproic acid, which is contraindicated because of depletion of carnitine and direct inhibitory effects on the mitochondrial respiratory chain). Electrolyte disturbances related to hypoparathyroidism and diabetes mellitus are corrected. Thyroid replacement alleviates hypothyroidism, Cardiac pacemaker placement prolongs life in Kearns-Sayre syndrome (KSS) with conduction defects. Impairments in the oxidative phosphorylation pathway may generate increased amounts of free radicals; therefore, antioxidants are prescribed, which include β-carotene, vitamin C, vitamin E, and CoQ_{10}. CoQ_{10} shuttles electrons from complex I and II to complex III and may stabilize the oxidative phophorylation enzyme complexes within the inner mitochondrial memebrane. The dose for CoQ_{10} in adults is 50 to 100 mg, three times per day. L-xarnitine is also recommended. Dicholroacetate increases the pyruvate dehydrogenase complex and reduces lactate. Aerobic training is recommended for some conditions.

More common mitochondrial disorders presenting in childhood are discussed in the following sections.

Kearns-Sayre Syndrome

These patients show progressive external ophthalmoplegia, retinitis pigmentosa on fundoscopic examination, and complete heart block. Onset is usually before 20 years of age. Cerebellar findings may be present on physical examination, and patients may show limb weakness, hearing loss, diabetes mellitus, hypoparathyroidism, irregular menses, and growth hormone deficiency. Dementia may be progressive. CSF protein is frequently greater than 100 mg/dL.

Myoclonus Epilepsy With Ragged-Red Fibers

This clinical syndrome is defined by the presence of myoclonus, generalized seizures, ataxia, and ragged red fibers on muscle biopsy. Symptoms usually begin in childhood. Other common clinical manifestations include hearing loss, dementia, exercise intolerance, and lactic acidosis. Multiple lipomatosis is common. Multiple members of a pedigree usually show the full syndrome.

Mitochondrial Encephalopathy, Lactic Acidosis, and Strokelike Episodes

This clinical syndrome is characterized by strokelike epidodes at a young age and typically before 40 years; encephalopathy evident as seizures, dementia, or both; lactic acidosis, ragged-red fibers on biopsy, or both as manifestations of the respiratory chain defects. Other frequent clinical features include normal early development, myogenic limb weakness, ataxia, myoclonus, migrainelike headaches, recurrent nausea and vomiting, and hearing loss. The abrupt-onset strokes often affect the occipital cortex, but may involve other regions of the brain. These patients often describe an antecedent history of migraine headaches that often occur prior to the strokelike event. Patients may experience improvement over weeks to months, but events virtually always recur. The lesions do not conform to territories of large vessels, a finding that favors the term *strokelike episodes.* Dementia may occur and be progressive. There is infrequent occurrence of the full syndrome in more than one member of a pedigree. Based on the hypothesis that MELAS is caused by impaired vasodilation in an intracerebral artery, investigators have evaluated the effects of administering L-arginine, a nitric oxide precursor to patients acutely with the first signs of strokelike episodes. Oral L-arginine administration within 30 minutes of a stroke was shown to significantly decrease frequency and severity of strokelike episodes (85).

Neuropathy Ataxia and Retinis Pigmentos

This disorder consists of the variable combinations of proximal neurogenic limb weakness, sensory neuropathy, ataxia, pigmentary retinopathy, developmental delay, dementia, and seizures. The onset occurs in teens and young adults, and the course is gradually progressive.

Mitochondrial Neurogastrointestinal Encephalomyopathy

This syndrome is clinically recognized by the unusual combination of six features: PEO, severe gastrointestinal dysmotility, cachexia, peripheral neuropathy, diffuse leukoencephalopathy on MR imaging, and evidence of mitochondrial dysfunction (histologic, biochemical, or genetic). The peripheral neuropathy and the prominent gastrointestinal dysmotility are defining features. Lactic acidosis at rest is present in two-thirds of patients. Both axonal and demyelination polyneuropathy is frequent. Muscle biopsy reveals ragged red fibers (RRFs) and neurogenic changes.

NEUROMUSCULAR JUNCTION DISORDERS

Transient Neonatal Myasthenia

Transient neonatal myasthenia occurs in about 10% to 15% of infants born to myasthenic mothers and is

due to transplacental transfer of circulating acetylcholine receptor (AChR) antibodies from the myasthenic mother to the fetus. Symptoms appear within the first few hours of birth; however, occasionally onset may be delayed for three to four days. Typical clinical characteristics include feeding difficulty, generalized weakness and hypotonia, respiratory difficulties, fetal cry, facial weakness, and, less frequently, ptosis.

The author prefers diagnostic confirmation by evaluating the response to edrophonium or neostigmine, with repetitive nerve stimulation studies performed at baseline and subsequent to infusion of the anticholinesterase agent. A response decrement with slow rates of stimulation (2–5 Hz) over a train of four to five stimuli may be repaired by the edrophonium (Tensilon) or neostigmine.

Treatment is largely supportive and the condition itself limiting, with resolution generally occurring within two to three weeks, although occasional cases may persist longer.

Congenital Myasthenic Syndromes

Congenital myasthenia syndrome (CMS) is a term used for a heterogenous group of disorders that are genetically determined rather than autoimmune-mediated. Patients may present in the neonatal period, later in childhood, or even in adult life. Patients often exhibit ptosis, external ophthalmoparesis, facial weakness, general hypotonia, proximal greater than distal muscle weakness, and variable degrees of functional impairment. Patients show absence of anti-AChR antibodies. More than 20 subtypes have been described, and congenital myasthenia may be classified according to the following: 1) presynaptic defects (eg, choline acetyltransferese [CHAT] deficiency causing CMS with episodic apnea, paucity of synaptic vesicles and reduced quantal release, or congenital Lambert-Eatonlike syndrome), 2) synaptic basal lamina defects (eg, endplate acetylcholinesterase [AChE] deficiency at neuromuscular junctions [NMJs]), and 3) postsynaptic defects (eg, AChR disorders involving α, β, δ, e subunits; kinetic abnormalities in AChR function caused by AChR deficiency; slow AChR channel syndromes; fast-channel syndromes; endplate rapsyn deficiency, etc.).

Several congenital myasthenic syndromes have been associated with arthrogryposis syndromes. For example, "multiple pterygium syndrome" (Escobar's syndrome) has been associated with AChR gamma, alpha 1, and delta subunit mutations.

For diagnostic workup, standard EMG with repetitive nerve stimulation is utilized initially, and subsequently stimulated single-fiber EMG may be useful. Ultrastructural evaluation of the neuromuscular junction with electron microscopy usually is performed on a biopsy of the deltoid or biceps, including the muscle region containing the neuromuscular junction (NMJ) or the "motor point." For in vitro electrophysiologic and immunocytochemical studies of the neuromuscular junction, a short muscle usually is removed from origin to insertion along with its motor branch and NMJ (a "motor point biopsy"). Muscles obtained have included the anconeus muscle near the elbow, the external intercostal muscle in the fifth or sixth intercostal space near the anterior axillary line, or the peroneus tertius muscle in the lower extremity. Such in vitro electrophysiologic studies allow specific delineation of the congenital myasthenic syndrome into one of the numerous specific subtypes. More recently, the diagnostic evaluation of CMS has increasingly relied upon molecular genetic studies.

For treatment of a CMS subtype, a definitive diagnosis is important because some CMS syndromes deteriorate with empiric treatment with AChE inhibitors such as pyridostigmine (Mestinon). For example, slow channel syndromes may deteriorate on pyridostigmine and endplate acetylcholinesterase deficiency may deteriorate or show no response. Some presynaptic syndromes may show response to 3,4-diaminopyridine, which increases release of acetylcholine at the presynaptic terminal. This drug has been used in Lambert-Eaton syndrome and in presynaptic CMS on a compassionate-use basis.

Autoimmune Myasthenia Gravis

This disorder is similar to the autoimmune myasthenia gravis observed in adults. The onset is often insidious, but at times, patients may present with acute respiratory difficulties. Patients usually present with variable degrees of ophthalmoparesis and ptosis. In addition, patients may exhibit facial weakness, swallowing difficulties, speech problems, and weakness of the neck, trunk, and limbs. Proximal muscles are more affected than distal, and the upper limits are more affected than the lower.

Fluctuation in the disease course with relapse and remission is common. Patients often complain of fatigue and diplopia, as well as progressive difficulty with chewing or swallowing. Patients are often worse with fatigue towards the end of the day. Thymoma, which occurs in about 10% of adult cases, is not a feature of the childhood-onset disease.

Serum AChR antibodies are an important diagnostic screening tool. Anti-AChR antibodies can be detected in the serum in about 85% to 90% of patients with generalized myasthenia gravis and greater than 50% of those with ocular myasthenia. The most common antibodies detected are AChR binding, followed by AChR modulating and then striational AChR antibodies. Muscle-specific kinase (MUSK) antibodies are an additional marker present in some seronegative patients and many patients with ocular myasthenia.

Diagnosis may also be confirmed by clinical response to an anticholinesterase drug such as edrophonium (Tensilon) Alternatively, neostigmine, a longer-acting

agent, can be used. Repetitive nerve stimulation studies show a characteristic decrement in the compound muscle action potential with slow stimulation rates (2–5 Hz) over a train of four to five stimuli. A decrement greater than 12% to 15% is often noted. Electrophysiologic studies may be more sensitive with proximal muscle groups such as the accessory nerve to the trapezius or study of the facial nerve. Abnormal repetitive nerve stimulation studies may also be seen in Lambert-Eaton syndrome, botulism, and congenital myasthenic syndromes. Single-fiber EMG is usually impractical in children; however, stimulated single-fiber EMG may be performed under anesthesia. Management may include treatment with anticholinesterase drugs, such as pyridostigmine, corticosteroids (prednisone), intravenous (IV) immunoglobulin, immunosuppressants (azathioprine, cyclosporine, mycophenolate mofetil, or cyclophosphamide), plasma exchange, or thymectomy.

Infantile Botulism

Infants with botulism usually present between 10 days to 6 months, with an acute onset of hypotonia, dysphagia, constipation, weak cry, and respiratory insufficiency. The neurologic examination shows diffuse hypotonia and weakness, ptosis, ophthalmoplegia with pupillary dilation, reduced gag reflex, and relative preservation of deep tendon reflexes. The diagnosis may be made by electrodiagnostic studies (see Chapter 7) or by measuring *Clostridium botulinum* toxin in a rectal aspirate containing stool.

Noninfantile Acquired Botulism

Older children and adults acquire botulism through poorly cooked, contaminated food with the toxin or through a cutaneous wound that becomes contaminated with soil-containing *Clostridium botulinum*. The toxin can often be identified in the serum and the food source. Clinical findings include acute onset of constipation, ptosis, diplopia, bulbar weakness, respiratory difficulties, ophthalmoparesis, pupillary dilation, and diminished deep tendon reflexes. Recovery may take months. The diagnosis is generally made from electrodiagnostic studies.

PERIPHERAL NERVE DISORDERS

Acute Inflammatory Demyelinating Polyradiculoneuropathy (Guillain-Barré Syndrome)

Acute inflammatory demyelinating polyradiculoneuropathy (AIDP) is a primarily demyelinating neuropathy with autoimmune etiology. Motor axons are affected more than sensory axons. Incidence in children is similar to that seen in adults. Children often have a prodromal respiratory or gastrointestinal infection occurring within one month of onset. Common precipitating infections include *Mycoplasma*, cytomegalovirus, Epstein-Barr virus, *Campylobacter jejuni*, and various vaccinations. Weakness generally begins distally in the lower extremity, with a progressive ascending paralysis ultimately involving the upper limbs. Pain and sensory symptoms are not uncommon. The most common cranial nerve abnormality is an ipsilateral or bilateral lower motor neuron facial paralysis. Objective sensory loss has been documented in the minority of children (85). In one series, only 15% required mechanical ventilation (86). The maximal degree of weakness generally reaches a peak within two weeks of onset, and time to maximum recovery was 7 months ± 5 months in one series (87). Complete recovery occurs in most children. Classic criteria for poor recovery in adults (low-median CMAPs and fibrillation potentials) may not apply to children (87).

Disturbances of the autonomic nervous system are common in children, including transient disturbances of bowel and bladder, excessive sweating or vasoconstriction, mild hypertension or hypotension, and occasionally cardiac arrhythmias.

The acute motor axonal neuropathy (AMAN) involves predominantly motor nerve fibers with a physiologic pattern suggesting axonal damage, whereas the acute inflammatory demyelinating polyneuropathy (AIDP) involves both motor and sensory nerve fibers with a physiologic pattern suggesting demyelination. Another clinical variant is the Miller-Fisher syndrome characterized by acute onset ataxia, ophthalmoparesis, and areflexia.

Diagnosis is generally confirmed by electrodiagnostic studies (see Chapter 7), and the CSF protein is characteristically elevated in a majority of children. Serum autoantibodies that may be elevated include IgM and IgG versus beta-tubulin and heparin sulfate. AMAN patients may show increased IgG antibodies to GM1 ganglioside. The Miller-Fisher syndrome is associated with a high frequency of the IgG GQ1b antibodies. The major considerations in differential diagnosis of AIDP or AMAN include transverse myelitis, toxic neuropathies, tick paralysis, infantile botulism, myasthenia gravis, and dermatomyositis.

Treatment has typically included corticosteroids, plasma exchange, or, more recently, intravenous immune globulin (88,89,90,91). AIDP patients respond to both plasma exchange and intravenous immunoglobulin (IVIG). Patients with AMAN respond preferentially to IVIG over plasma exchange. Recovery is often quite good in children without treatment. After standard intravenous immunoglobulin therapy, children

with axonal forms of Guillain-Barré syndrome (GBS) recover more slowly than those with the demyelinating form, but outcome at 12 months appears to be equally favorable in two groups (92).

Chronic Inflammatory Demyelinating Polyradiculoneuropathy

Children with chronic inflammatory demyelinating polyradiculoneuropathy (CIDP) often have a presentation similar to AIDP; however, the disorder continues with a chronic or relapsing course. The disorder may begin as early as infancy, but is seen in children and adults. Electrophysiologic studies show focal conduction block, temporal dispersion of CMAPs, prolongation of distal motor latencies, markedly slow conduction velocities, and absent or prolonged H-wave and F-wave latencies. CIDP cases often demonstrate axonal loss on EMG. The CSF protein is elevated in most cases.

The differential diagnosis usually includes CMT types I and III. The presence of acute relapsing episodes point towards CIDP. Due to the more severe involvement of proximal nerves and nerve roots, a distal sural nerve biopsy may not always show inflammatory changes and demyelination.

Treatment may include corticosteroids (prednisone) and IVIG as first-line approaches and subsequently plasma exchange.

Charcot-Marie-Tooth (Hereditary Motor Sensory) Neuropathy

Charcot-Marie-Tooth (CMT) neuropathy (also called hereditary motor sensory neuropathy or HMSN) is a heterogenous group of inherited disease of peripheral nerve that affects both children and adults and causes significant progressive neuromuscular impairment (93,94). It has been estimated that 1 per 2,500 to 3,000 persons has a form of CMT. CMT 1 denotes individuals with a hypertrophic demyelinating neuropathy ("onion bulbs") and reduced nerve conduction velocities, whereas CMT 2 refers to individuals with an axonal neuropathy and normal or slightly reduced nerve conduction velocities. Individuals with CMT 3 (Dejerine-Sotttas disease) have a primarily demyelinating peripheral neuropathy with a more severe phenotype presenting in infancy. Historically, types 1, 2, and 3 were felt to be autosomal-dominant conditions, with type 3 CMT patients exhibiting point mutations with frameshift and either dominant or recessive inheritance. CMT 4 refers to autosomal-recessive CMT. However, recently, axonal forms of CMT have been identified with autosomal recessive inheritance (deemed AR-CMT 2A, 2B, etc.)

In general, in most CMT subtypes, onset is usually during the first or second decade of life. Both motor and sensory nerve function are affected. The clinical features include distal muscle weakness, impaired sensation, and absent or diminished deep tendon reflexes. Weakness usually is greatest initially present in the foot and hand intrinsics and distal lower extremities, and subsequently in the distal upper extremities. Slow progressive weakness, more proximally in the knees, elbows, and pelvic and shoulder girdles may occur over decades (56). There is variable penetrance in most subtypes. The various gene locations and known protein abnormalities associated with various forms of CMT (HMSN) are given in Table 12.3.

The majority of *CMT 1* pedigrees (70%) demonstrate linkage to chromosome 17p11.2–12 and are designated CMT 1A (95). CMT 1A duplication results in increased expression of peripheral myolin protein-22 (PMP-22). Conduction velocities are uniformly slow in all nerves, with a mean of 17–20 M/s and a range of 5–34 M/s. Onset is typically in the first decade, with leg arreflexia, gait disorder (toe-walking or steppage gait), foot muscle atrophy or pes cavus, occasionally short Achilles tendons, and enlarged nerves owing to onion bulb formation in half of patients. Distal weakness develops initially in intrinsic muscles of the feet and hands with development of wasting of musculature occurring slowly over time (Fig. 12.16). Ankle dorsiflexion, ankle eversion, and extensor hallucis longus weakness develops with more normal strength proximally. Progressive cavus foot deformities with clawing of the toes often develop (Fig. 12.17). Orthopedic procedures are limited to soft tissue procedures and correcting wedge osteotomies, and joint fusion should be avoided if possible to avoid late pain. Late in the disease, diaphragm or bulbar weakness may develop in rare cases. Progression is slow over many decades. Defects in the human myelin zero gene (P_0) on chromosome 1q22-q23 leads to CMT 1B. P_0 is the major protein structural component of peripheral nervous system myelin. The clinical presentation is similar to CMT1A; however, onset may lag into the second to third decade in a minority of patients and there is more variability in severity. Nerve conduction velocities are usually less than 20 m/s. P0 mutations may lead to other clinical variants, referred to as CMT 1E (demyelinating CMT with deafness), and predominantly axonal neuropathy with late adult-onset (eg, CMT 2I, and CMT 2J with hearing loss and pupillary abnormalities).

CMT 2 is a less common disorder than CMT 1. Generally, CMT 2 patients demonstrate later age of onset, less involvement of the small muscles of the hands, and no palpably enlarged nerves. Wasting in the calf and anterior compartment of the leg may give rise to an "inverted champagne bottle" or "stork-leg" appearance. Conduction velocities are mildly reduced,

12.3 Hereditary Motor Sensory Neuropathy (HMSN) Types: Comparison of Clinical Features

DISORDER	GENE	LOCATION	USUAL ONSET	EARLY OR DISTINCT SYMPTOMS	TENDON REFLEXES	AVERAGE NCVs
CMT1: Dominant; Demyelinating						
CMT 1A	PMP-22	17p11	1st decade	Distal weakness	Absent	15 to 20 M/s
CMT 1B	P0	1q22	1st decade	Distal weakness	Absent	<20 M/s
CMT 1C	LITAF	16p13	2nd decade	Distal weakness	Reduced	16 to 25 M/s
CMT 1D	EGR2	10q21	2nd decade	Distal weakness	Absent	26 to 42 M/s
CMT X (S-D*)	Connexin-32	Xq13	2nd decade	Distal weakness	Absent distal	25 to 40 M/s
HNPP	PMP-22	17p11	3rd decade	Focal episodic weakness	Normal	Entrapments
Dejerine-Sottas (HMSN 3)	PMP-22 8q23 EGR2	17p11 8q23 10q21	2 years	Severe weakness	Absent	<10 m/s
CMT Intermediate NCV	DNM2 0q24 1p34 P0 CMT-X	19p12 10q24 1p34 1q22 Xq13	1st or 2nd decade	Distal weakness		25 to 50 M/s
CMT2: Dominant; Axonal						
CMT 2A	KIF1Bβ Mitofusin 2	1p36 1	10 yrs	Distal weakness	Absent distal	> 38 M/s
CMT 2B	RAB7	3q13	2nd decade	Distal weakness Sensory loss Acromutilation	Absent distal	Axon loss
CMT 2C		12q23-q24	1st decade	Vocal cord and Distal weakness	Absent	> 50 M/s
CMT 2D	GARS	7p15	16 to 30 yrs	Distal weakness Arms > legs	Reduced	Axon loss
CMT 2E	NF-68	8p21	1 to 40 yrs	Distal weakness	Reduced	Axon loss
CMT 2F/ Distal HMN	HSPB1 (HSP 27)	7q11	6 to 54 years	Difficulty walking	Reduced ankle	Axon loss
CMT 2G		12q12	15 to 25 years	Distal weakness	Reduced	42 to 58 M/s
CMT 2L	HSPB8	12q24	15 to 33 years	Distal weakness	Reduced	Axon loss
HMSN-P		3q13	17 to 50 yrs	Proximal weakness, cramps	Absent	Axon loss
HSMN + Ataxia		7q22	13 to 27 yrs	Gait ataxia	Absent	Axon loss
CMT 2 P0	P0	1q22	37 to 61 years	Leg weakness Pupil or Hearing	Reduced	< 38 M/s to Normal
AR-CMT2: Recessive; Axonal						
AR-CMT2A	Lamin A/C	1q21	2nd decade	Distal weakness	Reduced	Axon loss
AR-CMT2B		19q13	3rd and 4th decade	Distal weakness	Absent distal	Axon loss
AR-CMT2 Ouvrier		Autosomal	1st decade	Distal weakness	Reduced	Axon loss

Continued

12.3 *Continued*

DISORDER	GENE	LOCATION	USUAL ONSET	EARLY OR DISTINCT SYMPTOMS	TENDON REFLEXES	AVERAGE NCVs
HMSN 3: Infantile						
Dejerine-Sottas (HMSN 3)	P0 PMP-22 Periaxin	Autosomal Dominant/ recessive	2 years	Severe weakness	Absent	<10 m/s
Congenital Hypomyelinating Neuropathy	P0 EGR2 PMP-22	Autosomal Recessive	Birth	Severe weakness	Absent	<10 m/s
CMT4: Recessive; Demyelinating						
CMT 4A	GDAP1	8q13	Childhood	Distal weakness	Reduced	Slow
CMT 4B	MTMR2	11q22	2 to 4 yrs	Distal and Proximal weakness	Absent	Slow
CMT 4B2	SBF2	11p15	1st 2 decades	Distal weakness Sensory loss	Absent	15 to 30 m/s
CMT 4C	KIAA1985	5q23	5 to 15 yrs	Delayed walking	Reduced	14 to 32 M/s
CMT 4D (Lom)	NDRG1	8q24	1 to 10 yrs	Gait disorder	Absent	10 to 20 M/s
CMT 4E	EGR2	10q21	Birth	Infant hypotonia	Absent	9 to 20 M/s
CMT 4F	Periaxin	19q13	1 to 3 yrs	Motor delay	Absent	Absent
CMT 4H	FGD4	12q12	10 to 24 mo	Walking delay	Absent	< 15 M/s
CCFDN	CTDP1	18q23	1st or 2nd decade	Distal leg weakness	Reduced	20 to 34 m/s

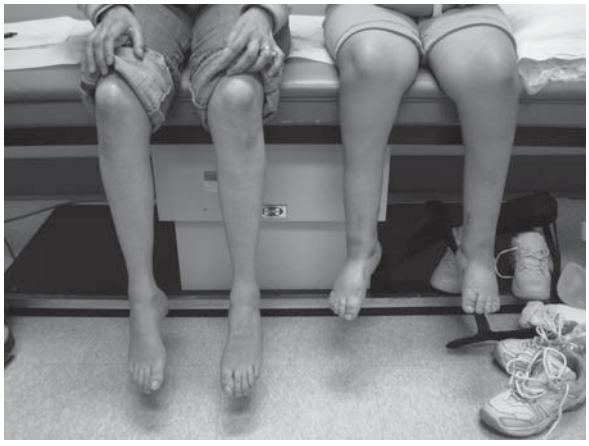
and CMAP amplitudes and sensory nerve action potential (SNAP) amplitudes are usually reduced. CMT 2A2 with mitofusin abnormality accounts for approximately 20% of CMT 2 probands. CMT 2C linked to chromosome 12q23-q24 has interesting features of early onset in the first decade and diaphragm and intercostal weakness producing shortness of breath. Vocal cord paralysis may alter the voice of these patients. The disease may progress to proximal and facial muscles. Arthrogryposis is present in some patients. Phrenic nerve CMAPs are often reduced. CMT 2E with abnormality in neurofilament light chain (NFL) linked to chromosome 8p21 may have associated hearing loss in 30% of cases. While most axonal CMT is autosomal-dominant, emerging pedigrees are being identified with recessive inheritance.

Dejerine-Sottas disease (CMT 3) is a severe hypertrophic demyelinating polyneuropathy with onset in infancy or early childhood. Most patients achieve ambulation, but some may subsequently progress to wheelchair reliance. Nerve conduction velocities are greatly slowed (often below 10 m/s), and elevations in cerebrospinal fluid protein may be present. Dejerine-Sottas disease may be associated with point mutations in either the PMP-22, P_0 or EGR2 gene (95). While this disorder was previously felt to be autosomal-recessive, many cases are due to denovo point mutations and actually have dominant inheritance.

Congenital hypomyelinating neuropathy is a severe and often fatal newborn disorder that often presents with respiratory distress in the delivery room. These infants often have severe generalized hypotonia and associated arthrogryposis. Diagnostically, these infants have absent sensory nerve action potentials (SNAPS) or low-amplitude SNAPS with prolonged distal latencies. Compound muscle action potentials are either absent or low-amplitude, with motor conduction velocities ranging from 3–10 meters per second. The disorder has been linked to PMP-22, P0, and EGR2 genes. Sural nerve biopsy may be useful. Inheritance is usually autosomal-recessive, with some dominant inheritance linked to EGR2.

Autosomal recessive CMT 4 is relatively rare. Most are demelinating with more severe phenotypes, and onset is often in childhood. CMT 4C linked to 5q23 is a relatively more common form of CMT 4.

Hereditary neuropathy with liability to pressure palsies (HNPP) is an autosomal-dominant disorder



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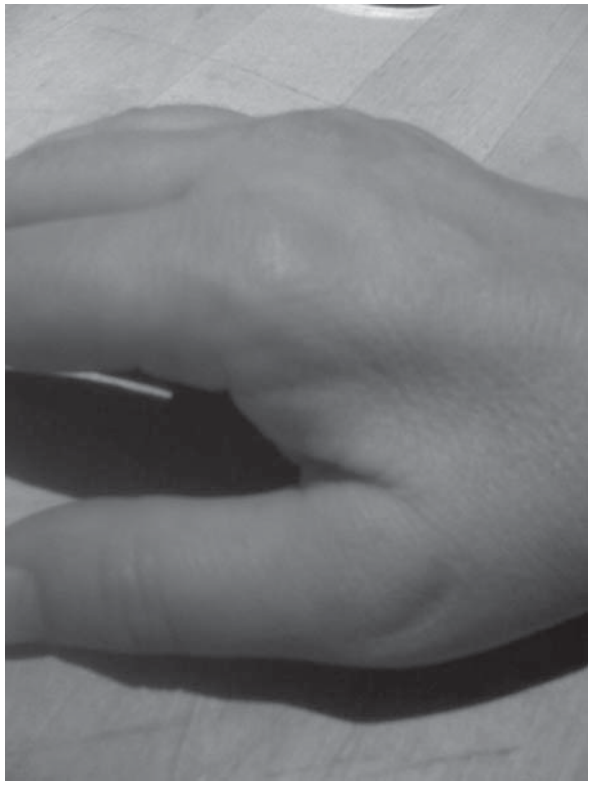

B

Figure 12.16 Distal weakness of intrinsic muscles of the feet (A) and hands (B) with wasting in Charcot-Marie-Tooth syndrome.

that produces episodic recurrent nerve entrapments with focal demyelination. Patients may present with peroneal palsies, carpal tunnel syndrome, and other entrapment neuropathies. A positive family history of entrapments often exists. Peripheral nerve biopsies may demonstrate segmental demyelination and tomaculous or "sausagelike" formations. A deletion at the PMP-22 gene locus (chromosome 17p11.2–12) causes this autosomal-dominant condition, in contrast to a duplication of this gene, which causes CMT 1A.

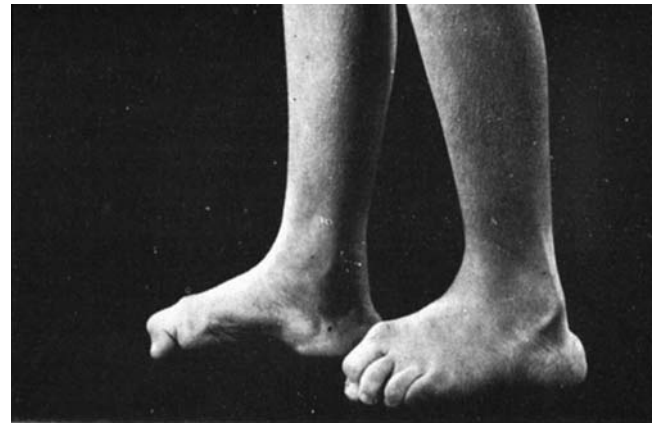

Figure 12.17 Progressive cavus foot deformities with clawing of the toes on Charcot-Marie-Tooth syndrome.

Patients with an X-linked dominant form of CMT (CMT-X) have been described. Male-to-male transmission is not observed, and the disorder generally shows earlier onset and faster rate of progression. The gene locus code for connexon 32 protein is Xq13, which encodes a major component of gap junctions, which provides a pathway for the transfer of ions and nutrients around and across the myelin sheath.

DNA testing for many of the CMT subtypes (particularly CMT 1) is available, but the ordering of extensive CMT batteries is expensive unless guided by nerve conduction study findings. Nerve conduction studies may be more expeditiously carried out on an affected parent to guide the molecular genetic workup of an affected child. Given the overlap of some gene abnormalities with several CMT clinical subtypes, it is often difficult to make a definitive diagnosis on genetic study results without the additional information provided by nerve conduction testing. Hereditary motor sensory neuropathy remains one clinical entity that continues to warrant electrodiagnostic evaluation.

Toxic Neuropathies

Toxic polyneuropathies are rare occurrences in children in North America. Toxic exposure to heavy metals and environmental toxins may be more common in other regions of the world. Expeditious diagnosis is critical to identify and remove the source of the toxicity and to establish treatment with agents such as penicillamine. Arsenic polyneuropathy is a sensorimotor neuropathy that may be axonal or, at times, predominantly demyelinating, simulating Guillain-Barré syndrome or CIDP. Gastrointestinal (GI) symptoms are common, as well as tachycardia and hypotension. Mee's lines may be seen in nails, along with other skin changes and allopecia. The diagnosis is established by

obtaining levels of arsenic in blood, urine, hair, and nail samples.

Lead polyneuropathy is most commonly observed in children who have ingested old lead-based paint. Acute exposures cause lead encephalopathy more commonly. Clinical findings may include anorexia, nausea and vomiting, gastrointestinal disturbance, fatigue, clumsiness and ataxia, and occasionally cognitive impairment, seizures, mental status changes, papilledema, and coma. The weakness is predominantly in the lower limbs, but the upper limbs may be involved. Electrophysiologic studies show a primarily axonal degeneration affecting motor greater than sensory axons. A microcytic hypochromic anemia with basophilic stippling of red blood cells establishes the diagnosis. Lead lines may be evident in long bone films. Lead levels may or may not be elevated in urine and blood, but levels of delta aminolevulinic acid are usually elevated in the urine.

Mercury poisoning may occur from the ingestion of mercuric salts, exposure to mercury vapor, or use of topical ammonia mercury ointments. Patients present with a generalized encephalopathy, fatigue, and occasionally a skin rash. A predominantly distal motor axonal neuropathy occurs. Deep tendon reflexes may be absent, and the gait is often ataxic. Sensory examination is often normal, although patients may complain of distal paresthesias. Electrophysiologic studies show motor axonal degeneration with normal sensory conduction studies.

Organophosphate poisoning may be due to exposure to insecticides or high-temperature lubricants or softeners used in the plastic industry. Patients present with an encephalopathy manifested by confusion and coma. In acute-exposure cholinergic crisis, manifested by sweating, abdominal cramps, diarrhea, and constricted pupils, may be present. A predominantly motor polyneuropathy is a late effect. However, the disorder may present as a rapidly progressive polyneuropathy mimicking Guillain-Barré syndrome. Severe paralysis with respiratory failure requiring ventilatory support may occur, and in this situation there may be a superimposed postsynaptic defect in neuromuscular transmission.

Glue-sniffing (N-hexane) neuropathy may be seen in teenage recreational glue sniffers. Repeated use may cause symptoms and signs of a predominantly distal motor and sensory polyneuropathy, which is predominantly demyelinating. Motor and sensory nerve conduction studies demonstrate moderate slowing.

Chemotherapeutic agents, in particular, vincristine, often produce a relatively pure motor axonal polyneuropathy. Severity is dose-dependent. Clinical findings include distal weakness, absent deep tendon reflexes, and at times foot drop. The disorder is often readily apparent by clinical examination, and electrophysiologic studies or nerve biopsy are usually not necessary. The neuropathy usually improves with discontinuation of the medication, although significant electrophysiologic abnormalities (reduced CMAP amplitudes and neuropathic recruitment) may persist. Vincristine may be particularly troublesome for children with hereditary motor sensory neuropathy.

Metabolic Neuropathies

Uremic neuropathy often occurs in children with end-stage renal disease. If clinical manifestations are present, they consist of a predominantly distal motor and sensory polyneuropathy with glove and stocking loss of sensation, loss of vibratory sense, and distal weakness, particularly involving peroneal innervated musculature. With successful renal transplantation, clinical findings and electrophysiologic abnormalities normalize (143). Diabetic polyneuropathy usually is a mixed motor and sensory polyneuropathy with both axonal changes and mild demyelination. The polyneuropathy is less common in children with diabetes mellitus, as compared with adults. The severity of the neuropathy may be related to the degree of glucose control (96).

MOTOR NEURON DISORDERS

Predominantly Proximal Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a term used to describe a varied group of inherited disorders characterized by weakness and muscle wasting, secondary to degeneration of both anterior horn cells of the spinal cord and brainstem motor nuclei without pyramidal tract involvement. Three subtypes of autosomal-recessive predominantly proximal SMA have been described, all linked to chromosome 5q. A common nomenclature subdivides SMA into types I, II, and III, based on age of onset and age of death, whereas the other approach classifies cases as severe, intermediate, and mild, based on ability to achieve independent sitting, independent standing, and walking. The International Consortium on SMA attempted to standardize the classification of childhood SMA to provide a rational basis for linkage studies and therapeutic trials (Table 12.4) (97).

SMA type I (Werdnig-Hoffman, severe form) was defined by the International Consortium on SMA as follows: onset from birth to 6 months, no achievement of sitting without support, and death usually prior to age 2 years. In SMA type II (intermediate form), onset is before 18 months, sitting is usually obtained, but standing and ambulation are never obtained and death

12.4 Childhood Onset Proximal Spinal Muscular Atrophy (SMA)

	SMA I (WERNIG HOFFMAN)	SMA II (INTERMEDIATE SMA)	SMA III (KUGELBERG WELANDER)
Onset	<6 months	6 to 18 months	>8 months IIIa <3 years IIIb >3 years
Genetics	SMN1: AR homozygous SMN2: <3 copies	SMN1: AR homozygous SMN2: three copies	SMN1: AR homozygous SMN2: 4–8 copies
Phenotype	Severe hypotonia, weak suck, weak cry, proximal weakness, absent refexes, respiratory failure common	Hypotonia, proximal weakness, muscle wasting, contractures, scoliosis, absent reflexes, tongue fasiculations	Proximal symmetric weakness, lordotic gait, Gowers's sign, decreased reflexes, tremor, tongue fasiculations
Milestones	Poor head control; Never sit independently	Sit with head control; never stand unassisted; may require ventilatory support	Stand and walk unassisted; may lose standing or continue to walk IIIa: onset 18 mo to <3 years (80% not walking at age 40) IIIb: onset > 3 years (40% not walking at age 40)
Life Expectancy	1 to 2 years 10% living at age 20	Most live to 3rd decade; many live to 4th to 5th decade	Normal life expectancy

occurs above the age of 2 years, usually much later in adulthood. In SMA type III (Kugelberg-Welander, mild form), the onset is after the age of 18 months, patients develop the ability to stand and walk, and death is in adulthood.

There is considerable variability and severity within each of the three groups, and occasionally some overlap exists. For example, patients with onset prior to 6 months may exhibit prolonged survival well past 4 years of age. Patients with onset between 6–18 months may ultimately achieve standing and independent ambulation. An adult-onset type of SMA with mild disease phenotype presenting usually in the second or third decade has been recognized. These patients usually are able to ambulate with minor motor impairments. Although the adult-onset SMA was not classified formally by criteria set forth by the consortium, among clinicians, SMA type IV has been used widely to classify later-onset patients with mild disease features. A modified classification has been proposed by Zerres and Rudnik-Schoneborn, which defines adult SMA as type IV (98).

The carrier frequency for SMA in the general population is estimated at about 1 in 40 to 50 individuals. Autosomal-recessive inheritance has long been documented in proximal SMA with childhood onset. In 1990, all three forms of SMA were mapped to chromosomal region 5q13, indicating that allelic variance of the same disease locus accounts for the clinical heterogeneity (99,100). During the past two decades, tremendous advances have been made in our understanding of the genetic basis for SMA (100–105). A detailed analysis of the 5q13 region revealed that this chromosomal region in humans contained a large inverted duplication, with at least two genes present in telomeric and centromeric copies.

Further studies have identified the SMA causative gene as the survival motor neuron (SMN) 1 gene (SMN1, telomeric copy), along with a disease-modifying gene (SMN2, centromeric copy) (100–102). Briefly, the two SMN genes are nearly identical, except for a difference of only five nucleotides in their 3' regions, without any alteration of the amino acid sequence of the protein. However, the critical difference between the SMN1 and SMN2 genes is a C-T transition located within the exon-splicing region of the SMN2 that affects the splicing of exon 7. This change results in frequent exon 7 skipping during the splicing of SMN2 transcripts (107,108). It is thought that the resulting truncated SMN protein, without its exon 7 contribution, is a less stable form of SMN protein, and, therefore, rapidly degraded. In about 95% of SMA patients, both copies of SMN1 exon 7 are absent because of mutations. In the remaining SMA-affected patients, other small or subtle mutations have been identified (101).

Genetic studies have now established that SMA is caused by mutations in the telomeric SMN1 gene, with all patients having at least one copy of the centromeric SMN2 gene. At least one copy of the SMN2 gene must be present in the setting of homozygous SMN1 mutations; otherwise, embryonic lethality occurs. The copy number of SMN2 varies in the population, and

this variation appears to have some important modifying effects on SMA disease severity (109–111). All SMA patients have >2 SMN2 genes. It appears that a higher number of SMN2 copies in the setting of SMN1 mutations is associated with a less severe clinical SMA phenotype: SMA I (severe): two or three gene copies of SMN2; SMA II: three copies of SMN2; SMA III: four to eight copies of SMN2. However, substantial variations in SMA phenotype and disease severity can exist with a given SMN2 copy number, so it is not recommended that disease severity be predicted based soley on SMN2 copy numbers. Although we now know that SMN protein is expressed widely in many tissues throughout the body, its function is still not completely understood at this time (112).

Spinal Muscular Atrophy I (Werdnig-Hoffman Disease)

The majority of cases of SMA I present within the first two months, with generalized hypotonia and symmetrical weakness. The age of onset of symptoms is less than 4 months in the vast majority of cases. Weak sucking, dysphagia, labored breathing during feeding, frequent aspiration of food or secretions, and weak cry are frequently noted by history.

Examination shows generalized hypotonia and symmetric weakness involving the lower extremities earlier and, to a greater extent, in the upper extremities. Proximal muscles are weaker than distal extremities. In the supine position, the lower extremities may be abducted and externally rotated in a "frog-leg" position. The upper extremities tend to be adducted and externally rotated at the shoulders with a semiflexed elbow. Volitional movements of fingers and hands persist well past the time when the shoulders and elbows cannot be flexed against gravity. The thorax is flattened anteroposteriorly and bell-shaped as a result of intercostal weakness. Pectus excavatum may be variably present. The diaphragm is usually preserved, relative to the intercostal and abdominal musculature. This results in a diaphragmatic breathing pattern during respiration with abdominal protrusion, paradoxical thoracic depression, and intercostal retraction. Neck flexor weakness may result in persistent posterior head lag when the trunk is lifted forward from the supine position. Neck extensor weakness may result in forward head lag when the infant is positioned in the horizontal prone position. With advanced disease, the mouth may remain open as a result of masticatory muscle weakness. Facial weakness may be noted in up to half of patients. The diagnostic criteria for SMA outlined by the International SMA Consortium (97) lists marked facial weakness as an exclusionary criterion for SMA, but this is not an absolute criterion. Tongue fasciculations have been reported in 56% to 61% of patients (113), so the absence of this finding does not necessarily exclude the disease. In one series (113), deep tendon reflexes (DTRs) were absent in all four extremities in 74% of cases. Thus, the preservation of DTRs does not exclude the diagnosis of SMA. Appendicular muscle fasciculations and distal tremor are also associated examination findings. Extraocular muscles are spared, as is the myocardium. Mild to moderate hip flexion, knee flexion, and elbow flexion contractures may be observed in some patients, along with wrist contractures and ulnar drift of the fingers. Severe arthrogryposis is not typically observed.

Diagnosis is confirmed by a consideration of clinical findings, molecular genetic studies, and, occasionally, electrodiagnostic studies. Muscle biopsy is generally not required to confirm the diagnosis.

In a large series from Germany (98), 197 patients classified as type I (never sits alone) had the following survival probabilities: 32% at age 2; 18% at age 4; 8% at age 10; and 0% at age 20.

Spinal Muscular Atrophy II

Spinal muscular atrophy II disease onset is usually more insidious than that of SMA I. The findings of generalized hypotonia, symmetrical weakness, and delayed motor milestones are hallmarks of SMA II. Weakness also involves proximal muscles more than distal muscles and lower extremity more than upper extremity. A fine tremor of the fingers and hands occurs in a minority of patients. This "polyminimyoclonus" may be attributed to spontaneous, repetitive rhythmical discharges by the motor neurons that innervate a large territory of muscle. Wasting tends to be more conspicuous in SMA II versus SMA I. DTRs are depressed and usually absent in the lower extremities. Appendicular or thoracic muscle wall fasciculations may be observed. Tongue fasciculations have been observed in 30% to 70% of SMA II patients (97,113,114) Progressive kyphoscoliosis and neuromuscular restrictive lung disease is almost invariably seen in the late first decade. Contractures of the hip flexors, tensor fasciae latae, hamstrings, triceps surae, and elbow and finger flexors are quite common. Hypotonic hip dislocations have been noted commonly in SMA II patients. Sensory examination is completely normal, and extraocular muscles and the myocardium are spared. In a large series from Germany (98), of 104 cases classified as SMA II (sits alone, never walks), 98% survived to the age of 10 and 77% to the age of 20. Thus, a longer lifespan is possible with adequate supportive care.

SMA II is a slowly progressive condition affecting proximal musculature more than distal. The calculated grade of progression for SMA may be less than one-half manual muscle testing units decline per decade (69).

Longitudinal series of 12–39 months' duration have shown essentially stable strength measurements but slow loss of function (115,116).

Pathologic changes on muscle biopsy have been consistent with hypotrophic change in fetal muscle development. Other changes are consistent with a more active denervating process. Thus, SMA includes a component of myofiber atrophy comparable to that seen in other denervating diseases and is not a pure hypotrophic process occurring during early fetal development.

Spinal Muscular Atrophy III (Kugelberg-Weilander Syndrome)

In more chronic SMA III, also referred to as Kugelberg-Welander syndrome, weakness usually initially occurs between the ages of 18 months and late teens. Motor milestones may be delayed in infancy. Proximal weakness is observed, with the pelvic girdle being more affected than the shoulder girdle (57). There is an exaggerated lumbar lordosis and anterior pelvic tilt owing to hip extensor weakness. There is also a waddling gait pattern with pelvic drop and lateral trunk lean over the stance-phase side, secondary to hip abductor weakness. If ankle plantar flexion strength is sufficient, the patients may show primarily forefoot or toe contact and no heel strike similar to patients with Duchenne dystrophy. This is a compensatory measure for knee extensor weakness to maintain a stabilizing knee extension moment at the knee. The patient may exhibit a Gower's sign when arising from the floor; stair climbing is difficult due to hip flexor weakness. Facial weakness is sometimes noted. Fasciculations are noted in about half of the patients (97) and are more common later in the disease course. Fasciculations in the limb muscles and thoracic wall muscles are common. Calf pseudohypertrophy has been occasionally noted, but wasting of affected musculature is more prominent. Deep tendon reflexes are diminished and often become absent over time. Contractures are generally mild as long as patients remain ambulatory. Scoliosis may be observed in SMA III, but it occurs less frequently and is less severe than scoliosis and SMA II. While no survival data exist for patients with SMA III, cases have been followed into the eighth decade without mechanical ventilation (57,98). Ventilatory failure due to neuromuscular restrictive lung disease is a rare event in SMA III, occurring only in adulthood (57,117).

Zerres and Rudnik-Schoneborn (98) have proposed further subtypes, including SMA IIIa (walks without support; age of onset less than 3 years) and SMA IIIb (walks without support; age of onset 3–30 years). In their series, only 44% of SMA "IIIa" patients remained ambulatory 20 years after onset of weakness, whereas 89% of "IIIb" patients remained ambulatory after a similar 20-year duration.

Distal Spinal Muscular Atrophy

Distal spinal muscular atrophy is an increasingly recognized group of rare diseases with varied genetic etiologies. More than 20 distinct genetic subtypes have been identified. The patients may be clinically misdiagnosed as having CMT due to the distal weakness of the foot and hand intrinsics. Some subtypes of distal SMA have predominant upper extremity involvement. Other variants of distal SMA may present initially with distal lower extremity weakness. Sensory function is always normal clinically and electrodiagnostically. The course is usually slowly progressive, although some patients may experience a prolonged period of stability. Other associated features in some subtypes include vocal cord paralysis and diaphragm weakness. Some subtypes have associated pyramidal signs.

Juvenile Segmental SMA (Benign Focal Amyotrophy; Hirayama Disease)

This disease was originally described by Hirayama as a slowly progressive focal motor neuron disease affecting the upper extremities. Most cases occur on a sporadic basis. The onset of this syndrome is typically between 15 and 25 years, with a range of 2 to 30. Wasting and weakness develop segmentally in C8–T1 hand and forearm muscles and unilaterally and often but not always in the dominant extremity. Sensation is completely normal. The disease progresses to more proximal upper extremity muscles. The lower extremities are never affected, and typically the disease progression plateaus after two to six years. Symptoms worsen in the cold ("cold paresis"). Tremor may occur due to distal weakness. Hyperhidrosis of the involved limb is a common complaint. Reflexes are typically spared but not brisk. EMG studies are consistent with an anterior horn cell disorder. MR imaging abnormalities of the cervical spinal cord (segmental atrophy, stenosis, or foraminal narrowing) have been described in a proportion of patients. The disease is more common in Asian populations.

Progressive Bulbar Paralysis of Childhood (Fazio-Londe Disease)

Fazio-Londe disease, or progressive bulbar paralysis of childhood, is a progressive bulbar paralysis that is probably genetically transmitted. This is a disorder of bulbar motor neurons. Patients present with cranial nerve findings, including ptosis, facial weakness,

dysphagia, normal hearing, and respiratory stridor. They may show hyperreflexia. Dominant transmission is rare. One group with recessive inheritance had early onset in infancy and rapid progression, with death from respiratory failure less than two years from the age of onset. Another group with recessive inheritance shows later onset (3 to 12 years), less respiratory involvement but slowly progressive dysarthria, dysphagia, and facial weakness. These patients may have progressive motor neuron disease with primary involvement of the anterior horn cells in the cervical and upper thoracic core segments. In addition, there may be widespread degenerative changes in the brainstem. Cranial nerve VII is almost always affected. These patients develop dysphagia secondary to cranial nerve XII involvement. The nuclei of cranial nerves III, IV, VI, and X may also be involved; however, clinical impairment of extraocular movement is rare.

SPINOCEREBELLAR DEGENERATION DISEASES

Friedreich's Ataxia

Friedreich's ataxia is a spinocerebellar degeneration syndrome with the onset of symptoms before age 20 years. This autosomal-recessive condition has been linked in one subtype to chromosome 9q13–21.1 (FRDA), with the protein implicated being termed "frataxin." A second subtype referred to as FRDA2 is linked to chromosome 9p23-p11.

The incidence of Friedreich's ataxia is 1 in 25,000 to 50,000. Carrier frequency is 1 in 60 to 110. Age of onset is usually <20 years, typically around puberty, with a range from 2 to 25 years. Obligate signs and symptoms include progressive ataxic gait, cerebellar dysfunction with tremor and dysmetria, dysarthria, decreased proprioception or vibratory sense (or both), muscle weakness, and absent deep tendon reflexes. Other common signs include cavus foot deformity, cardiomyopathy, scoliosis, and upper motor neuron signs such as a Babinski's sign and spasticity. Weakness is progressive, affecting lower extremities and small muscles in the hands and feet. Sensory loss is typical and especially affects vibration and joint position sensation. Tendon reflexes are often absent. An occasional patient may have chorea without ataxia. With electrodiagnostic studies, sensory nerve potentials may be absent or reduced. Progression is slow, with mean time to wheelchair 15 years of age and death from cardiomyopathy ranges from the third to seventh decade.

The prevalence of scoliosis approaches 100%, but some cases have more severe progressive spinal deformity than others. Those Friedreich's ataxia cases with onset of disease before the age of 10 years generally have more severe progressive scoliosis. Those with the onset of disease during or after puberty have later onset spinal deformity, which may not require surgical intervention.

Frataxin is a mitochondrial protein located on the inner mitochondrial membrane. It is likely required for maintenance of mitochondrial genome, and it is involved in iron homeostasis and iron transport into mitochondria. Idebenone is a powerful antioxidant and a synthetic analogue of coenzyme Q. It may improve iron homeostasis and mitochondrial function in Friedreich's ataxia. In randomized clinical trials, longer-term idebenone treatment has been shown to prevent progression of cardiomyopathy and cardiac hypertrophy in both pediatric and adult patients with Friedreich's ataxia. Its stabilizing effect on neurological dysfunction has been shown to be present only in the pediatric population, mainly before puberty. This suggests that the age at which idebenone treatment is initiated may be an important factor in the effectiveness of the therapy (118).

Other Hereditary Ataxias

The hereditary ataxias are a group of genetic disorders characterized by slowly progressive incoordination of gait and often associated with poor coordination of hands, speech, and eye movements. Frequently, atrophy of the cerebellum occurs. The hereditary ataxias are categorized by mode of inheritance and causative gene or chromosomal locus. The genetic forms of ataxia are diagnosed by family history, physical examination, and neuroimaging. Molecular genetic tests are available for the diagnosis of many but not all spinocerebellar ataxias (SCAs). At least 28 genetically distinct autosomal-dominant SCA subtypes and four other autosomal-dominant hereditary ataxias have been identified. Childhood onset has been found commonly in SCA7, SCA13, SCA17, and SCA21 and more rarely in SCA1, SCA2, SCA3, SCA5, SCA 21, and SCA 22. Other pedigrees of SCA with childhood onset have been identified in only single families. Autosomal-dominant episodic ataxia 1 (EA1) with episodic attacks of myokymia and ataxia and linkage to chromosome 12p13.3 has onset in the first decade. At least seven autosomal-recessive ataxias in addition to Friedreich's ataxia have been identified, most of which have childhood onset. The most common of these is ataxia-telangiectasia, with linkage to chromosome 11q22.3, which presents in the first decade with ataxia, dysarthria, ocular telangiectasias, immune deficiency, and risk of cancers.

MANAGEMENT OF CHILDHOOD NEUROMUSCULAR DISEASES

Diseases affecting the lower motor neuron, including those primarily affecting anterior horn cell, peripheral nerve, neuromuscular junction (presynaptic or postsynaptic) or muscle, ultimately lead to progressive loss of functional muscle fiber over time. This loss of functional muscle fiber may lead to progressive weakness, decreased endurance, limb contractures, spine deformity, body composition changes, decrease in mobility, decreased pulmonary function, and occasionally cardiac impairment if the myocardium is affected. Genetic defects causing CNS structural protein alterations may lead to intellectual impairment. Rehabilitation approaches directed at improving impairment and/or resultant disability may substantially improve the quality of life and community integration of children with neuromuscular diseases. The following discussion emphasizes general principles in the rehabilitation management of childhood neuromuscular disease, with several specific conditions used to illustrate key concepts.

Exercise in Neuromuscular Disease

Exercise prescriptions and recommendations in childhood neuromuscular disease need to consider the specific disease condition as well as the developmental and maturational status of the child.

Strengthening Exercise in Rapidly Progressive Disorders

The more rapidly progressive neuromuscular disorders of childhood generally include the dystrophic myopathies. The inherent instability of the sarcolemmal membrane predisposes to membrane injury due to mechanical loads. Theoretically, eccentric or lengthening contractions produce more mechanical stress on muscle fiber than concentric or shortening contractions. Indeed, many of the muscle groups that show the greatest weakness early in the course of Duchenne muscular dystrophy are muscle groups that perform a great deal of eccentric activity, such as the hip extensors, knee extensors, and ankle dorsiflexors. In addition, lower extremity muscles in this population experience more mechanical loads than upper extremity muscle groups, and weakness in the lower extremities generally predates weakness in the upper extremities. Edwards and colleagues (119) proposed that routine eccentric contractions occurring during gait are a likely source of the pattern of weakness typically seen in myopathies.

There may be increased weakness following strengthening exercise in DMD (120). There are other instances that have raised concerns regarding overwork weakness in dystrophic myopathies. The dominant upper limb has been found to be weaker in persons with FSHD muscular dystrophy than the nondominant, providing circumstantial evidence for overwork weakness (67,121). A single subject with scapuloperoneal muscular dystrophy had a reversal of rapid strength decline after reducing daily physical activity. Other studies evaluating strengthening intervention in DMD subjects have shown maintenance of strength or even mild improvement in strength over the period of the investigation. However, these studies are limited by use of primarily nonquantitative measures (122), lack of a control group (123), and use of the opposite limb as a control without considering the effects of cross training (124). Animal work utilizing dystrophic dogs has shown significant increases in creatine kinase values immediately following exercise.

No systemic studies using the DMD population have shown any deleterious effects of resistance exercise. Based on the theoretic susceptibility of the dystrophin-deficient sarcolemmal membrane to mechanical injury and the relative paucity of investigations, it is prudent to recommend a submaximal strengthening program in DMD and other rapidly progressive dystrophic disorders. A great concern is how to incorporate these activities effectively into the daily routine of the child, avoiding use of mundane and tedious regimens that employ progressive resistive exercises. Incorporation of the activity into recreational pursuits and aquatic-based therapy are probably the most reasonable approaches for the preadolescent child.

Strengthening Exercise in Slowly Progressive Neuromuscular Diseases

Only supervised strengthening programs in this population have been advocated. Recently, a moderate resistance home exercise program (using a less supervised approach) was devised that demonstrated similar strength gains in both neuromuscular disease patients and normal control subjects without evidence of overwork weakness (125). Based on this encouraging result, the home program was advanced to high resistance training in similar subjects without apparent additive beneficial effects; in fact, eccentrically measured elbow flexor strength actually decreased significantly (126).

Based on these investigations, the author believes that there is adequate evidence to generally advocate a submaximal strengthening program for persons with slowly progressive NMD. There seems to be no additional benefit to high-resistance, low-repetition training sets, and the risk of actually increasing weakness becomes greater. Improvement in strength will hopefully translate to more functional issues such as improved endurance and mobility.

Aerobic Exercise in Neuromuscular Disease

Aerobic exercise refers to rhythmic, prolonged activity of the level sufficient to provide a beneficial training stimulus to the cardiopulmonary and muscular systems but below the threshold where anaerobic metabolism of fuels is the primary source of energy. The response of normal skeletal muscle to this type of training includes increased capillary density in the muscle to improve substrate transfer, increased skeletal muscle mitochondrial size and density, higher concentrations of skeletal muscle oxidative enzymes, and improvement in utilization of fat as an energy source for muscular activity. Patients with neuromuscular disease have a diminished capacity for exercise. Children with Duchenne muscular dystrophy have been demonstrated to have low cardiovascular capacity and peripheral oxygen utilization with higher resting heart rate compared with controls (127). Physical ability and exercise capacity is more likely to be limited by muscle strength than by deterioration of cardiorespiratory function. In a recent study using a home-based aerobic walking program, slowly progressive neuromuscular disease subjects showed modest improvement in aerobic capacity without evidence of overwork weakness or excessive fatigue (128). It is likely that alternative exercise approaches, such as aquatic-based therapy, will need to be utilized in children with more severe neuromuscular diseases who are nonambulatory and have less-than-antigravity muscle strength.

One group recently studied the effect of endurance training on conditioning and strength in adult Becker muscular dystrophy (BMD). Eleven patients with BMD and seven matched, healthy subjects cycled 50 30-minute sessions at 65% of their maximal oxygen uptake (VO_2max) over 12 weeks, and six patients continued cycling for 1 year. Endurance training for 12 weeks significantly improved VO_2max by 47 ± 11% and maximal workload by 80 ± 19% in patients. This was significantly higher than in healthy subjects (16 ± 2% and 17 ± 2%). CK levels did not increase with training. Strength in muscles involved in the cycle exercise (knee extension, and dorsi- and plantarflexion) increased significantly by 13% to 40%. Cardiac pump function, measured by echocardiography, did not change with training. All improvements and safety markers were maintained after one year of training. Endurance training was demonstrated to be a safe method to increase exercise performance and daily function in patients with BMD, and the findings support an active approach to rehabilitation of patients with BMD (129).

Management of Limb Contractures and Deformity

The management of limb contractures in progressive neuromuscular disease and the role of stretching, orthotics, and surgery has recently been comprehensively reviewed (130). Contracture is defined as the lack of full active of passive range of motion (ROM) due to joint, muscle, or soft tissue limitation. Contractures may be arthrogenic, soft tissue, or myogenic in nature, and a combination of intrinsic structural changes of muscle and extrinsic factors leads to myogenic contractures in selected neuromuscular disease conditions. These factors include the following: degree of fibrosis and fatty tissue infiltration; static positioning and lack of full active and passive range of motion; imbalance of agonist and antagonist muscle strength across the joint; lack of upright weight bearing and static positioning in sitting; compensatory postural changes used to biomechanically stabilize joints for upright standing; and functional anatomy of muscles and joints (multijoint muscle groups in which the origin and insertion crosses multiple joints). In general, dystrophic myopathies have a high degree of fibrosis and fatty infiltration, placing these patients at higher risk for contractures. Significant contractures have been most commonly identified in Duchenne muscular dystrophy, Becker muscular dystrophy, Emery-Dreifuss muscular dystrophy, congenital muscular dystrophy, autosomal recessive LGMD, FSHD muscular dystrophy, myotonic muscular dystrophy, hereditary motor sensory neuropathy, and spinal muscular atrophy.

Contractures and progressive NMD conditions should be managed with the following concepts in mind:

1. Prevention of contractures requires early diagnosis and initiation of physical medicine approaches, such as passive ROM and splinting wall contractures, are still mild.
2. Contractures are inevitable in some NMD conditions, such as DMD.
3. Advanced contractures become fixed and show little response to stretching programs.
4. A major rationale for controlling contractures of the lower extremity is to minimize the adverse effect of contractures on independent ambulation. However, the major cause of wheelchair reliance in NMD is generally weakness, not contracture formation.
5. Static positioning of both upper and lower extremity joints in patients with weak musculature is the most important cause of contracture formation.
6. Passive stretching for control of lower limb contractures is most successful in ambulatory patients with early mild joint contractures.
7. Upper extremity contractures may not negatively affect the function if they are mild.
8. Joint range of motion should be monitored regularly by physical therapists and occupational therapists using objective goniometric measurement.

Principle therapy modalities must be regularly carried out to prevent or delay the development of lower extremity contractures for those at risk for musculoskeletal deformity. These include: regularly prescribed periods of daily standing and walking if the patient is functionally capable of being upright; passive stretching of muscles and joints with a daily home program; positioning of the leg to promote extension and oppose joint flexion when the patient is non-weight bearing through the lower extremities; and splinting, which is a useful measure for the prevention or delay of ankle contracture.

In the upper extremity, elbow flexion contractures in dystrophic myopathies may occur soon after transition to the wheelchair, secondary to static positioning of the arms and elbow flexion on the armrests of the wheelchair (9). Other associated deformities in DMD and other dystrophic myopathies include forearm pronator tightness and wrist flexion-ulnar deviation in the later stages of the disease. The regular palm-down position of the hand increases the occurrence of forearm pronator contracture. Mild elbow flexion contractures of ≤ to 15 degrees are of no functional consequence to the patient using crutches or a wheelchair. Contractures of the elbows over 30 degrees can interfere with the use of crutches in ambulatory patients with NMD. Severe elbow flexion contractures of >60° are associated with decreased distal upper extremity function and produce difficulty when dressing.

Passive stretching of the elbow flexors may be combined with passive stretching into forearm supination to help prevent contractures. Prophylactic occupational therapy management of the wrist and hand is recommended in NMD to slow the development of contractures and to maintain fine motor skills. Daily passive stretching of the wrist flexors and intrinsic and extrinsic muscles of the hand and wrist are recommended, as are active range-of-motion exercises for the wrist and long finger flexors. Nighttime resting splints, which promote wrist extension, metacarpophalangeal extension, and proximal interphalangeal flexion, are recommended. Daytime positioning should emphasize wrist and finger extension, but any splinting should not compromise sensation or function.

Shoulder contractures are less problematic in patients with profound proximal muscle weakness. Combined shoulder internal rotation, adduction contracture, and elbow flexion deformity may interfere with self-feeding. Severe shoulder internal rotation deformities may complicate dressing, produce pain on passive range of motion, and cause pain during sleep.

Bracing/Orthotic Management and Orthopaedic Surgical Management of Limb Deformity

Management in Neuromuscular Diseases With Proximal Weakness

The prototypical disorder in which bracing and surgical management of contractures for prolonged ambulation has been applied is Duchenne muscular dystrophy. In this population, wheelchair reliance is imminent when knee extension strength becomes less than antigravity and time to ambulate 30 feet is greater than 12 seconds (9). A number of principles should be emphasized for these populations. First, with an appropriate and aggressive home-based therapy program, equinovarus contractures generally are absent or very mild in DMD at the time walking ability ceases (9). In addition, hip and knee flexion contractures are also absent or extremely mild in ambulatory DMD patients at the time of transition to wheelchair. The wide-based Trendelenburg's gait exhibited by these patients with gluteus medius weakness places the hip in an abducted position, leading to iliotibial band contractures. The late phase of ambulation often is associated with more marked joint contractures involving the iliotibial bands and heel cords because DMD patients spend more time sitting and less time standing. The release of contractures at both the heel cord and iliotibial band generally is necessary to obtain successful knee ankle foot orthotic (KAFO) bracing (131–134). Other authors have reported bracing of DMD patients without surgical release of the iliotibial bands (135,136). Hip and knee flexion contractures generally are not severe enough to interfere with bracing at the time of transition to wheelchair (9). The iliotibial band contractures may be released with a low Young fasciotomy and a high Ober fasciotomy.

The ankle deformity may be corrected by either a tendo-Achilles lengthening (TAL) or a TAL combined with a surgical transfer of the posterior tibialis muscle tendon to the dorsum of the foot. The posterior tibialis tendon transfer corrects the equinovarus deformity but prolongs the time in a cast and recovery time, and it increases the risks of prolonged sitting.

Orthopedic surgical release of these contractures allows the DMD patient to be braced in lightweight polypropylene KAFOs with the sole and ankle set at 90 degrees, drop-lock knee joints, and ischial weight-bearing polypropylene upper thigh component. DMD patients who are braced may or may not require a walker for additional support. At times, DMD patients who have had excellent home stretching programs can be placed immediately into KAFO bracing without surgical tenotomies.

While DMD subjects are still ambulating independently without orthotics, they often use their ankle equinus posturing from the gastrocnemius-soleus group to create a knee extension moment at foot contact, thus stabilizing the knee when the quadriceps muscle is weak. Several authors have cautioned against isolated heel cord tenotomies while DMD patients are still ambulating independently. Overcorrection of the heel cord contracture in a DMD patient may result in immediate loss of the ability to walk without bracing unless the quadriceps are grade 4 or better (131).

The duration of ambulation in DMD has been successfully prolonged by prompt surgery and bracing, immediately implemented following loss of independent ambulation. Generally, the gains in additional walking time have been variable, but generally reported between two and three years.

Long-term benefits of prolonged walking include decreased severity of heel cord and knee flexion contractures at age 16 (137). This may ultimately improve shoe wear tolerance and foot positioning on the wheelchair leg rests. Prolonged ambulation by lower extremity bracing in DMD has never been documented to be an independent factor in the prevention of scoliosis. Disadvantages of braced ambulation center around the excessive energy cost of braced ambulation and safety concerns in the event of falls. DMD subjects with KAFO bracing usually need gait training by physical therapy, and they need to be taught fall techniques.

Weakness is the major cause of loss of ambulation in DMD, not contracture formation. Thus, the primary indication of orthopedic surgical tenotomies and posterior tibialis tendon transfers likely is the provision of optimal alignment for KAFO bracing. Little evidence supports the efficacy of early prophylactic lower extremity surgery in DMD for independently producing prolonged ambulation (9,131,138).

In general, with the increased utilization of corticosteroids in DMD, there has been a trend over the past two decades towards reduced use of lower extremity surgery and long leg bracing to prolong ambulation.

Management of NMD Patients With Distal Lower Extremity Weakness

Ankle dorsiflexors are often clinically weaker than ankle plantar flexors in neuromuscular disease because of selective involvement of the peroneal nerve in many neopathies and isolated anterior and lateral compartment weakness in several myopathic conditions such as FSHD, scapuloperoneal distribution LGMD, DMD, and Emery-Dreifuss muscular dystrophy. Ankle foot orthotics (AFOs) often are used for patients with distal weakness. AFOs are generally contraindicated in situations where NMD patients utilize equinus posturing with forefoot initial contact to maintain a knee extension moment in the setting of quadriceps weakness. Heel cord contractures may need to be surgically lengthened to allow for AFO or KAFO bracing. Cavus feet are common in peripheral neuropathies. Intrinsic muscle weakness of the foot results in hyperextension at the metatarsophalangeal joints and flexion at the interphalangeal joints with resultant claw toe deformities. This constellation of deformities may cause difficulty in walking, lack of balance and painful callosities. Treatment of the cavus foot depends on the patient's age, flexibility of the foot, bony deformity, and muscle imbalance. A supple foot can be managed nonoperatively by serial casting in a walking cast, followed by an AFO with a solid ankle in neutral position and a lateral heel wedge if significant hindfoot varus exists. Fixed soft tissue or bony deformity may require orthopedic surgery to produce a plantigrade foot. In skeletally immature children, triple arthrodesis is contraindicated. Triple arthrodesis should only be considered as a salvage procedure for severe heel varus and severe midfoot deformity, with the goal being achievement of hindfoot stability in a skeletally mature patient.

Management of Spinal Deformity

Severe spinal deformity and progressive NMD lead to multiple problems, including poor sitting balance, difficulty with upright seating and positioning, pain, difficulty in parental or attendant care, and potential exacerbation of underlying restrictive respiratory compromise (Fig. 12.18). Severe scoliosis and pelvic obliquity can, in some instances, completely preclude upright sitting in a wheelchair. The management of spinal deformity and progressive neuromuscular disease has recently been reviewed (17). Populations at risk for scoliosis include DMD, autosomal-recessive LGMD, congenital muscular dystrophy, FSHD muscular dystrophy, congenital myotonic muscular dystrophy, spinal muscular atrophy II and III, and Friedreich's ataxia. While previous estimates of incidence of severe scoliosis in DMD approached 80% to 90%, recent evidence suggests that corticosterorids (specifically deflazacort) may significantly decrease the incidence of severe progressive scoliosis in DMD (22).

Close clinical monitoring is essential for children with NMD at risk for scoliosis. Curves may progress rapidly during the adolescent growth spurt, and children need to be monitored every three to four months during this time, with clinical assessment and spine radiographs if indicated. In addition, patients who are likely to require surgical arthrodesis at some point should be monitored with pulmonary function tests every six months. A forced vital capacity falling below

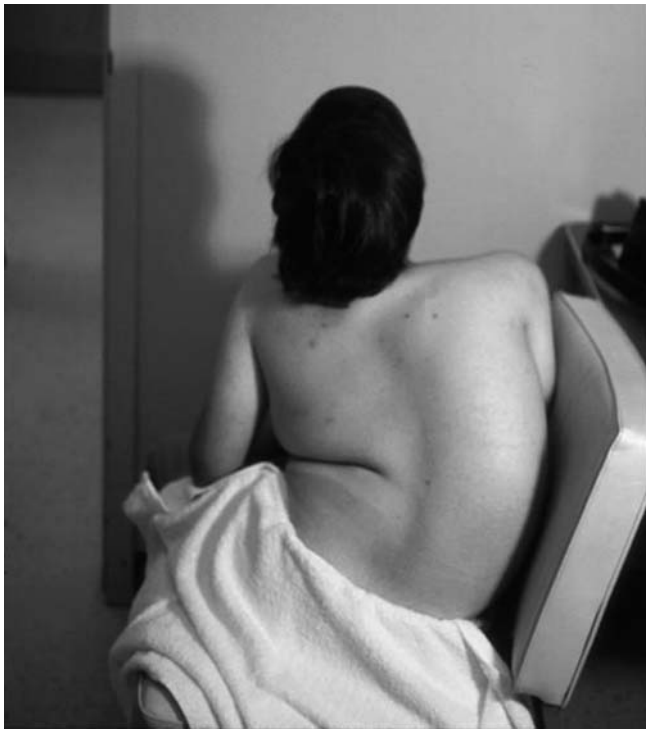

Figure 12.18 Scoliosis in Duchenne muscular dystrophy compromising long-term comfortable supported sitting in a power wheelchair.

30% to 40% of predicted does not contraindicate surgery (28), but is associated with increased perioperative morbidity and likely the need for prolonged noninvasive ventilatory support during the postoperative recovery period (27). Thus, there is often a critical window of time where the spinal deformity is evident and likely to continue to progress and the restrictive lung disease is not of a severity that would contraindicate surgery or be associated with perioperative complications.

The management of spinal deformity with orthotics is ineffective in DMD and does not change the natural history of the curve. Spinal orthoses are often reported to be uncomfortable and poorly tolerated by DMD patients. Furthermore, vital capacity potentially can be lowered with constrictive orthoses. On the other hand, in neuromuscular diseases with spinal deformity beginning in the first decade of life, such as SMA, congenital muscular dystrophy, congenital myotonic muscular dystrophy, some congenital myopathies, and congenital myasthenic syndromes, spinal bracing is generally used to improve sitting balance in patients who are unable to walk. In addition, spinal orthotics are employed in these younger patients in an attempt to halt curve progression until children are 10 to 11 years of age, when a single posterior spinal arthrodesis procedure is sufficient. Children younger than the age of 10 generally require both anterior and posterior spinal arthrodesis because of continued spinal growth, which decreases in rate after age 11 to 12. If a younger child has a severe progressive curve and severely compromised pulmonary function, a posterior fusion may be considered, with acceptance of the fact that some rotational "crank shaft deformity" will ensue.

Spinal arthrodesis is the only effective treatment for scoliosis in DMD, autosomal-recessive LGMD, congenital muscular dystrophy, congenital myotonic muscular dystrophy SMA, and Friedreich's ataxia. The decision to pursue posterior spinal instrumentation involves a consideration of the severity of the restrictive lung disease, severity of the cardiomyopathy, severity and flexibility of the spinal deformity, and likelihood that the spinal deformity will continue to progress. Surgical spinal arthrodesis should be deferred to a later date in marginally ambulatory patients with LGMD, congenital muscular dystrophy, FSHD, and spinal muscular atrophy type III, as these individuals may use significant lumbar lordosis during gait to compensate for hip extensor weakness.

Provision of Functional Mobility

Generally, antigravity quadriceps are required for community ambulation in childhood neuromuscular disease. Short-distance ambulation may be achieved by some patients with more severe weakness using KAFO bracing, with or without a walker. Such orthotic intervention is often provided to children with SMA type III, severe childhood autosomal recessive muscular dystrophy (SCARMD), congenital muscular dystrophy, DMD, and Becker muscular dystrophy during adulthood. Children with DMD SMA type II, congenital muscular dystrophy, congenital myopathies, some myasthenic syndromes, and more severe hereditary motor sensory neuropathies utilize power mobility devices for functional mobility. Generally, children can be taught to safely operate a power wheelchair when they are at the developmental age of approximately 2 years (139,140). The initial power wheelchair prescription needs to consider the natural history of the neuromuscular disease condition over the following five years, as some children will subsequently develop the need for a power recline system and the chair needs to be able to accommodate such a recline or be retrofit. In more severe disability, the power wheelchair electronics should be sufficiently sophisticated to incorporate alternative drive control systems, environmental control adaptations, and possibly communication systems in patients who are unable to vocalize.

Pulmonary Management

Pulmonary complications are recognized as the leading cause of mortality in childhood neuromuscular

disease. Respiratory insufficiency in neuromuscular disease results from a number of factors, including: respiratory muscle weakness and fatigue, alteration of respiratory system mechanics, and impairment of a central control of respiration. Progressive muscle weakness and fatigue lead to restrictive lung disease and ultimately to hypoventilation, hypercarbia, and respiratory failure. Increased inelastic load on respiratory muscles occurs because of chest wall stiffness, airway secretions, and ineffective cough mechanism. This may result in atelectasis and increased airway resistance, and kyphoscoliosis can further alter respiratory mechanics. Defects in central control of respiration may be secondary to hypoxemia and hypercarbia, associated with severe restrictive lung disease. Significant nocturnal decreases in partial pressure of oxygen, as well as elevations in arterial partial pressure of carbon dioxide, occur in more severe restrictive lung disease. Hypercapnia or hypoxemia occurring at night may have a role in reducing daytime central respiratory drive. A chronic increase in the bicarbonate pool may blunt the stimulus to breathe, generated by respiratory acidosis and perpetuating the hypercapnic state. Expiratory muscle weakness may produce ineffective cough, problems with clearance of secretions, and predisposition to pulmonary infections.

Respiratory failure may present acutely or insidiously. Respiratory difficulties in the delivery room or early infancy may be seen in acute infantile type I SMA, myotubular myopathy, congenital hypomyelinating neuropathy, congenital infantile myasthenia, congenital myotonic muscular dystrophy, transitory neonatal myasthenia, and severe neurogenic arthrogryposis. In most other childhood neuromuscular diseases, the respiratory insufficiency develops more insidiously unless an acute decompensation occurs from an event such as an aspiration episode or acute onset of weakness, as seen in Guillain-Barré syndrome, botulism, and myasthenic syndromes. Signs and symptoms of significant respiratory difficulties may include subcostal retractions, accessory respiratory muscle recruitment, nasal flaring, exertional dyspnea or dyspnea at rest, orthopnea, generalized fatigue, and paradoxic breathing patterns. A history of nightmares, morning headaches, and daytime drowsiness may indicate nocturnal hypoventilation with sleep-disordered breathing. Pulmonary function tests have been used to help in the decision-making process regarding the institution of mechanical ventilation. In a study of 53 patients with proximal myopathy, hypercapnia occurred when the maximal inspiratory pressure was less than 30% of predicted and when vital capacity was less than 55% of predicted (141). Other authors (142,143) have noted lower values for vital capacity measurements in their patients with DMD at the time they require institution of mechanical ventilatory support. Hahn and colleagues (144) have reported the predicted value of maximal static airway pressures in predicting impending respiratory failure. Splaingard (145) reviewed a series of 40 patients with a diverse group of neuromuscular disease conditions. They noted that all their patients who required mechanical ventilation had a vital capacity of ≤25%, with at least one of the following associated findings: $PaCO_2$ >than 55 mmHg, recurrent atelectasis or pneumonia, moderate dyspnea at rest, or congestive heart failure.

Noninvasive forms of both positive and negative pressure ventilation are being increasingly applied to children with neuromuscular diseases. Initially, patients may require ventilatory support for only part of the day. Noninvasive nocturnal ventilation has become a widely accepted clinical practice, providing ventilatory assistance for patients while sleeping and allowing them to breathe on their own during the day. Intermittent ventilation may ameliorate symptoms of respiratory failure, reduce hypercarbia, increase oxygenation (even during periods off the ventilator), and prolong survival in patients with neuromuscular disease. The long-term use of noninvasive ventilation (Fig. 12.19) may be associated with fewer complications than ventilation via a tracheostomy; however, bulbar muscle function should be adequate for safe swallowing (117). Ventilatory support has allowed prolonged survival and acceptable quality of life in SMA I, SMA II, and DMD (143,146,147,148).

Improved pulmonary toilet and clearance of secretions can be achieved with assisted cough; deep breathing; and setup spirometry, percussion, and postural

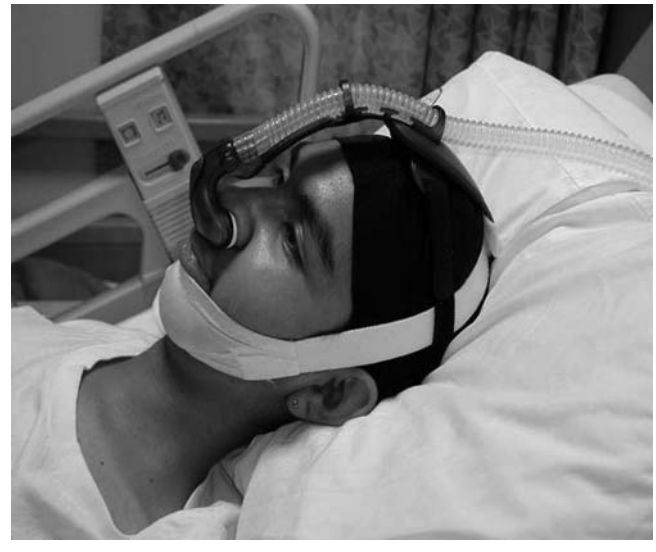

Figure 12.19 Noninvasive ventilatory support using bilevel positive airway pressure and nasal pillows mask interface in young adult with Duchenne muscular dystrophy.

drainage, and, in more severe cases, the additional use of interpulmonary percussive ventilation (IPV), given two to three times daily.

Nutritional Management

Management of Swallowing Problems

Involvement of palatal and pharyngeal muscles may produce dysphagia. Patients at particular risk include those of SMA, myasthenia gravis, congenital myasthenic syndromes, and congenital myopathies, such as myotubular myopathy, oculopharyngeal muscular dystrophy, late-stage Duchenne muscular dystrophy, and late-stage SCARMD. The presence of dysphagia in patients with neuromuscular disease has been documented by others (51,149). The function of the swallowing mechanism is best evaluated with a fluoroscopic video dynamic swallowing evaluation. DMD patients have a high prevalence of dysphagia during the late stages of the disease (51). DMD patients may also rarely develop acute gastric dilatation secondary to gastric paresis (150). Bulbar dysfunction and/or respiratory distress may affect feeding in SMA patients. In SMA I, therapeutic modifications may include use of a premature baby nipple with a large opening, use of proper head and jaw position, along with a semireclined trunk position and use of frequent small feedings to minimize fatigue. These larger bolus feeds may distend the stomach and encroach on the diaphragm, thus affecting respiratory status. Improved nourishment in SMA leads to a feeling of well-being and therefore a better quality of life. Poor nutritional status, labored feeding, and/or symptoms of dysphagia are indications for initiation of supplemental enteral feedings via nasogastric tube or gastrostomy. Gastroesophageal reflux with risk of aspiration may be an indication for placement of a gastrojejunostomy tube.

Energy and Protein Supplementation

Severe deficits in energy and protein intake have been documented in DMD (49,50) during the second decade. Substantial weight loss has been documented in DMD to occur between the ages of 17 and 21 (Fig. 12.20). Protein and calorie needs in DMD may be approximately 160% of that required for able-bodied adolescents. Beneficial effects in weight gain, anthropometric measurements, and nitrogen balance were documented for DMD patients aged 10–20 years, subsequent to a three-month nutritional supplementation, which consisted of an additional 1,000 kcals and 37.2 grams of protein (151). The positive effects on metabolism observed in this study warrant further investigation.

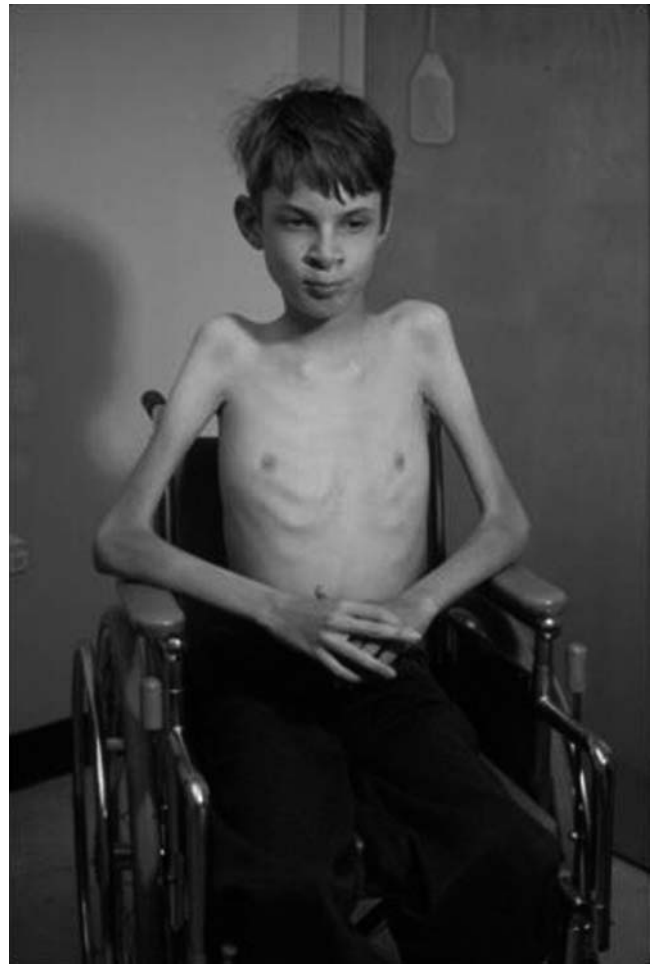

Figure 12.20 Severe weight loss in young adult with Duchenne muscular dystrophy.

Branched-Chain Ketoacid Supplementation

Based on the observations that muscle protein degradation is accelerated in DMD and administration of branched-chain ketoacids reduces protein breakdown in fasting obese subjects, Stewart and colleagues (152) conducted a trial of branched-chain ketoacid supplementation. The ketoacids of the branched-chain amino acids leucine, valine, and isoleucine were administered orally as ornithine salts at a dosage of 0.45 gm/kg body weight/day for four days in nine boys with DMD, aged 5–9 years. An equivalent amount of protein was removed from the diet during this time. A small but significant reduction in muscle protein degradation was observed as a result of the treatment, and no negative effects were noted. The results warrant further investigation regarding the effects of longer-term branched-chain ketoacid supplementation on muscle protein degradation.

Weight Reduction

DMD patients typically gain excessive weight between 9–13 years of age, subsequent to the onset of wheelchair

reliance. This is likely due to a reduction in total daily energy expenditure with increased sedentary existence. Edwards and colleagues (153) demonstrated that weight reduction through a medically supervised decrease in energy intake could be achieved successfully in DMD without compromising skeletal muscle mass. Obesity has also been observed in SMA III patients and has been attributed to a relatively sedentary lifestyle. Increased adiposity has been documented in adults with slowly progressive neuromuscular diseases (154). Approaches to weight reduction in slowly progressive neuromuscular disease patients has been previously reviewed (155).

Management of Cardiac Complications

Early treatment with ACE inhibitors is probably warranted in DMD when the measured ejection fraction falls below 55% (36,37). The benefits of earlier protective treatment with either ACE inhibitors or ARBs is under investigation. Digitalis has been demonstrated to be effective in decreasing morbidity from heart failure, but not mortality, and probably is also indicated for the treatment of heart failure observed in DMD patients with cardiomyopathy. Beta blockers may also have a role in DMD. Treatment with coenzyme Q10 remains controversial. Cor pulmonale, confirmed on echocardiography, may benefit from continuous supplemental oxygen. Patients with known arrhythmias who are at risk for fatal tachyarrhythmias may benefit from antiarrhythmic medication. DMD patients with mitral valve prolapse and mitral regurgitation should be given antibiotic prophylaxis for dental and surgical procedures in accordance with current guidelines.

The management of the cardiomyopathy, seen in Becker muscular dystrophy, is similar to that seen in DMD; however, in cases of severe end-stage cardiomyopathy, cardiac transplantation should be considered.

Cardiac conduction abnormalities observed in myotonic muscular dystrophy may ultimately require implantation of cardiac pacemakers. In rare instances with cardiomyopathy, treatment may consist of ACE inhibitors, digitalis, and diuretics, based on proven efficacy in cardiomyopathies of other etiologies.

Emery-Dreifuss muscular dystrophy patients with symptomatic bradycardia or heart block should undergo implantation of a permanent cardiac pacemaker. Atrial standstill, atrial fibrillation, and atrial flutter are all disorders in which blood can pool in the atria, leading to thrombus formation and possible embolic events, including stroke. Anticoagulation with warfarin to an international normalized ratio (INR) of 2–3 has demonstrated a reduction in the incidence of stroke in patients with atrial fibrillation. Prompt referral to a cardiologist should be made for children with cardiac signs or symptoms nr screening ECG, echocardiography, or for those with Holter recording abnormalities suggestive of cardiac disease. Late-stage DMD, BMD, and Emery-Dreifuss muscular dystrophy patients should be followed by a cardiologist on a regular basis. Appropriate management of cardiac complications in childhood neuromuscular disease will hopefully increase life expectancy.

Pharmacologic Intervention

The rehabilitation specialist may become involved in the prescription of pharmacologic agents, which affect the pathophysiology of various neuromuscular diseases. Evaluation of therapeutic efficacy for pharmacologic agents requires careful objective measurement of strength, with quantitative measurements, functional status using timed motor testing and the six-minute walk test, pulmonary function parameters, cardiac parameters, and patient-reported quality of life measures.

Corticosteroids such as prednisone and deflazacort may have an effect on the inflammatory component of the dystrophic myopathy and other disease pathways in DMD, slow the progression of the strength loss, prolong ambulation by two years, reduce the occurrence of scoliosis, and slow the loss of pulmonary function (11–13,22–24,156). Alternative pulsed-dosing regimens, such as high-dose weekend administration (5 mg/kg/day on both Saturday and Sunday) may decrease the side effects of weight gain and growth retardation with similar clinical efficacy. Deflazacort is an alternate corticosteroid with a potentially better side effect profile and equal efficacy to prednisone in DMD.

The identification of specific genes and the protein products implicated in the pathogenesis of various neuromuscular diseases provides hope that meaningful therapeutic interventions that target the defective genes will alter the natural history of many of these neuromuscular diseases. It is likely that traditional rehabilitation approaches will need to be used adjunctively with newer pharmacologic interventions, molecular genetic-based therapies, and possibly gene therapy in the management of these conditions.

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13

Pediatric Limb Deficiencies

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CONGENITAL DEFICIENCIES

Incidence

Epidemiologic surveys have determined limb deficiencies to occur ranging from 5 to 9.7 per 10,000 live births in a ratio of 3:1 upper to lower extremity (1). The United States does not have a formal complete registry of birth defects, so the precise number is unknown. The National Birth Defects Prevention Study has reported that 6% of all types of birth defects are limb deficiencies (2).

Early identification of limb anomalies occurs with routine ultrasound. A detailed level 3 ultrasound, as well as echo 3-D, amniocentesis, and cordocentesis to anticipate syndromes, is recommended if limb deficiencies are detected (3). Prenatal counseling will provide resources and psychological support for parents who undergo the loss of the idealized child (4,5).

Etiology

The first trimester is crucial for the genesis of limb production. Congenital limb deficiency occurs as a result of failure of formation of part or all of the limb bud. The mesodermal formation of the limb occurs at 26 days gestation and continues with differentiation until 8 weeks gestation. The various limb segments develop in a proximal-to-distal order so that the arm and forearm appear before the hand, and the thigh and leg before the foot (6). Limb development is a complex process that involves orchestration of a number of genes; some are well known and studied, and account for various syndromes and abnormalities (7). A relatively small set of genes and gene families appear to control the early stages of limb development. More than 80% of heritable limb deficiencies are associated with anomalies outside the musculoskeletal system (7).

Upper limb deficiencies are more commonly associated with other anomalies, particularly craniofacial, cardiac, and hematological disorders; this is due to the chronology of development in the first trimester (8). Bilateral deficiencies are more common with craniofacial abnormalities, whereas left-right asymmetry of organogenesis is more commonly associated with unilateral and left axial deficiency (9,10). Vascular pathology is not inherited, so the risk of recurrence is small (7). Conditions with implied vascular disruption include Adams-Oliver syndrome, gastroschisis, Klippel-Feil syndrome, Moebius syndrome, Poland syndrome/sequence, and terminal transverse limb deficiency (11–13). Two key features of Moebius are demonstrated in Figure 13.1: craniofacial anomalies and upper limb deformity.

Other factors that put a child at risk for limb deficiency include maternal diabetes, including gestational diabetes (14,15). Although alcohol, heroin, and cocaine have not been found to be related to limb deficiency, all maternal ingestions

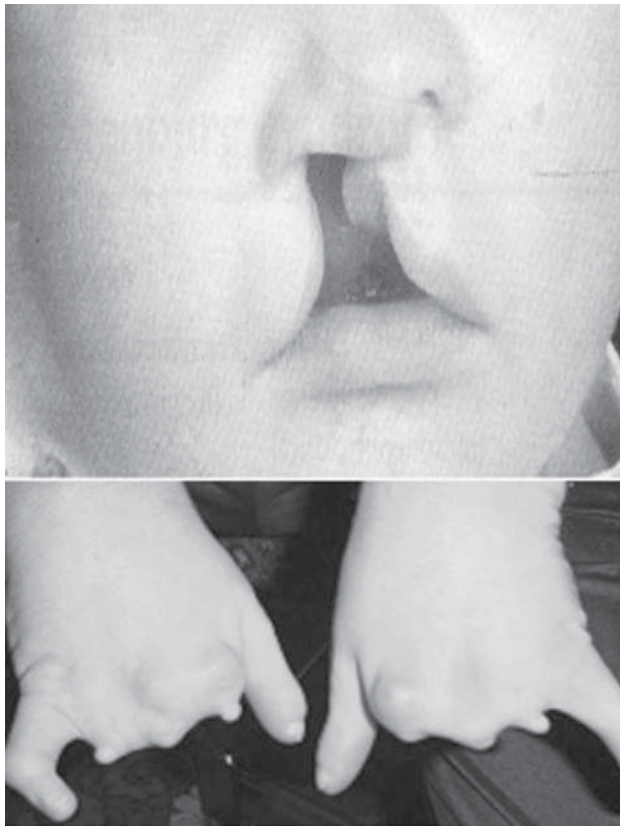


Figure 13.1 Two key components of clinical presentation in Moebius syndrome. Craniofacial (A) and upper-limb (B) deformities.

and first-trimester abnormalities should be documented (16,17). Smoking increases the risk of digit anomalies (18). Thalidomide historically presents a clear association with limb reductions (19). Recent case reports implicate valproic acid and calcium channel blockers (20,21). Maternal occupation may play a role, with exposure to chemicals, as in the agricultural setting (22). Uterine abnormalities have been reported in several cases of limb deficiencies, theoretically due to compression of the fetus (6). In addition, disturbances to the uterine environment, such as chorionic villi sampling, are implicated in deficiencies (23). Amniotic band syndrome is associated with fibrous bands that may constrict the limbs (24,25). Radiological findings of amniotic band are illustrated in Figure 13.2. Prenatal vitamins reduce the risk of limb deficiencies (26).

Postnatal problems, such as gangrene from vascular emboli and neonatal injury from vascular compromise secondary to umbilical catheters, may necessitate immediate amputation (27–29). Although the causes may be different from congenital disorders, the clinical issues for the child and the rehabilitation team are more similar to congenital disorders than acquired disorders.

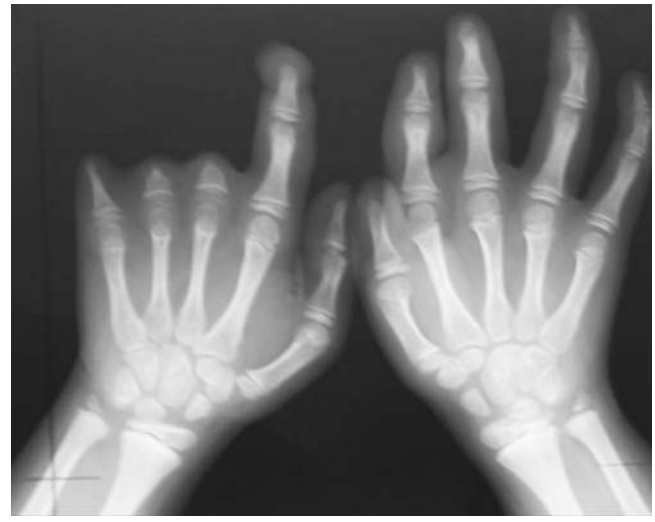


Figure 13.2 X-ray of hands affected by amniotic band syndrome.

Classification

The International Society for Prosthetics and Orthotics (ISPO) has adopted a definitive system for congenital deficiencies. No longer is it necessary to learn ancient language roots to describe the limb deficiency (30–32). However, like an old language and culture, the terminology once used in clinics is difficult to change. Clinical teams often use a fusion of terms.

Many clinics still describe deficiencies by the Frantz classification system. In this system, deficiencies are either terminal, representing the complete loss of the distal extremity, or intercalary, denoting the absence of intermediate parts with preserved proximal and distal parts of the limb. Those deficits are then divided into horizontal and longitudinal deficits.

The ISPO classification system is used in research and academic endeavors because this system facilitates communication and creates a logical, accurate approach. The ISPO classification divides all deformities into transverse or longitudinal. A transverse deficiency has no distal remaining portions, whereas the longitudinal deficiency has distal portions. The transverse level is named after the segment beyond which there is no skeletal portion. Longitudinal deficiencies name the bones that are affected, beginning with the most proximal long bone. Any bone not named is presumed present and of normal form. The affected bone is designated as total or partially absent. The approximate fraction of the limb in a transverse deficiency is estimated in *thirds*, while the longitudinal deficiencies describe a *partial* or *complete* bone absence. Involved digits are then identified. Digit numbering proceeds from the radial or tibial side of the limb. *Ray* refers to the metacarpal or metatarsal and corresponding phalanges (33). Tables 13.1 and 13.2 describe transverse and longitudinal deficiencies.

13.1 Transverse Limb Deficiencies

UPPER LIMB	"DESCRIBED AS"	LOWER LIMB	"DESCRIBED AS"
Arm	Complete Upper one-third Middle one-third Lower one-third	Thigh	Complete Upper one-third Middle one-third Lower one-third
Forearm	Complete Upper one-third Middle one-third Lower one-third	Leg	Complete Upper one-third Middle one-third Lower one-third
Carpals	Complete Partial	Tarsals	Complete Partial
Metacarpals	Complete Partial	Metatarsals	Complete Partial
Phalanges	Complete Partial	Phalanges	Complete Partial

13.2 Longitudinal Limb Deficiencies

UPPER LIMB	"DESCRIBED AS"	LOWER LIMB	"DESCRIBED AS"
Humerus	Complete Partial	Femur	Complete Partial
Radius	Complete Partial	Leg	Complete Partial
Carpals	Complete Partial	Tarsals	Complete Partial
Metacarpals	Complete (1, 2, 3, 4, 5) Partial	Metatarsals	Complete (1, 2, 3, 4, 5) Partial
Phalanges	Complete (1, 2, 3, 4, 5) Partial	Phalanges	Complete (1, 2, 3, 4, 5) Partial

ACQUIRED AMPUTATIONS

Terminology

The terminology utilized for acquired amputations follows the convention for adult limb loss. Upper-extremity amputations include shoulder disarticulation, transhumeral (above-elbow amputation), elbow disarticulation, transradial (below-elbow amputation), wrist disarticulation, and partial hand amputations. The types of lower-extremity amputations are translumbar (hemicorpectomy), transpelvic (hemipelvectomy), hip disarticulation, transfemoral (above-knee amputation), knee disarticulation (through-knee), transtibial (below-knee amputation), ankle disarticulations (ie, Syme, Boyd, and Pirigoff), and partial foot (ie, Chopart and LisFranc) (34). Figure 13.3 illustrates present classifications of acquired amputations.

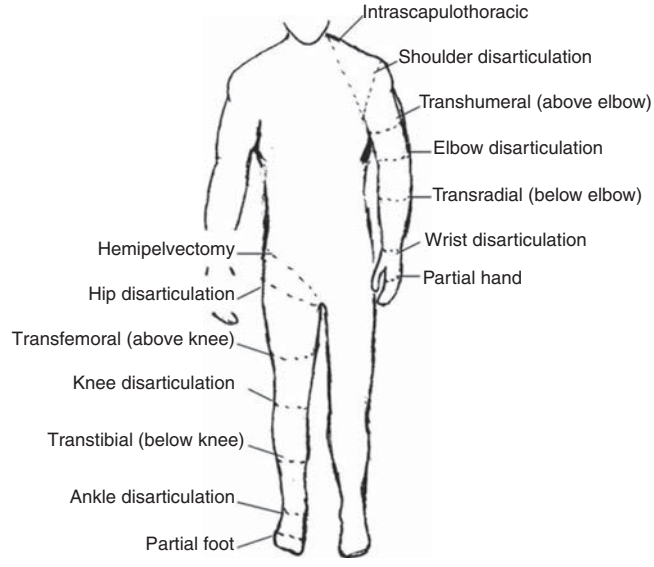


Figure 13.3 Classifications of acquired amputations.

Traumatic

In the pediatric age group, the most common causes of acquired amputations are trauma and disease (35). Trauma causes limb loss twice as often as disease (36). The most common traumatic injuries result from automobile and motorcycle collisions and train accidents. Causes for traumatic injuries vary by region. In rural areas, farm accidents, lawnmower accidents, and high-tension wire injuries occur more frequently (37–40). For the older child, vehicular accidents, burns, gunshot wounds, and power tools are the most frequent causes of limb loss. Boating accidents can produce amputations by propeller injury. Sadly, in the 1–4-year-old age range, power tools such as lawn mowers and household accidents are frequent mechanisms of amputation (41,42).

A single limb is involved in more than 90% of acquired amputations, of which 60% involves the leg. The male-to-female ratio of acquired amputation is 3:1.

Tumors

Tumors are the most frequent cause of amputations due to disease. Tumors represent the most common cause of amputations in the European Surveillance of

Congenital Anomalies (EUROCAT) data system (1). The highest incidence of malignancy is in the 12–21-year-old age group. Osteogenic sarcoma, Ewing's sarcoma, and the rare rhabdomyosarcoma are responsible for the majority of tumors resulting in amputation (43,44). Unprecedented improvement in survival has occurred with earlier detection and combined therapy (45). Definitive surgery for osteosarcoma depends upon the site of the primary tumor and the extent of invasion or metastasis (46). Surgical removal of the affected bone and the surrounding soft tissue remains the treatment of choice, whether by amputation or limb salvage procedure. Limb salvage with an endoprosthesis can be offered to 90% of children with osteosarcoma (45–47). This procedure, which involves replacing the affected bone with a metal endoprosthesis, is accompanied by orders to prohibit contact sports. Compliance with these orders is often questionable. With the advent of extendable endoprostheses, it has been suggested that children who have undergone this treatment have results that are superior to those who have undergone amputation surgery (48–50).

The surgical procedure of choice attempts to obtain a tumor-free margin of 5–8 cm above the proximal limit of the medullary tumor. The decision to proceed with limb salvage or amputation is dependent on the aggressiveness of the tumor, the stage, the responsiveness to neoadjunct therapy, and the likelihood of obtaining tumor-free margins (51–53). The knee poses a challenge for soft tissue sarcomas. Despite complications, the knee may be reconstructed with allografts (54,55).

Chemotherapy has now proven an effective adjunct to surgery. Prior to 1972, only 15% of the children were disease-free and survived with surgery, compared to the 60% to 70% who now survive with surgery and the addition of chemotherapy (56,57). Rehabilitation may be confounded by factors of fatigue and the psychological aspects of combined treatments. Physical therapy emphasizing range of motion, strengthening, and functional activities is important for children with lower-extremity sarcoma after limb salvage surgery (58). Outcomes were similar for ambulation, stair climbing, employment, and psychological adjustment when comparing amputation to limb salvage for surgical management of sarcomas (134, 147).

Amputation of a limb during adolescence, when body image is particularly important, may complicate the completion of tasks required during adolescence (59). Psychological reactions are varied. However, emotional distress was less when pain and functional loss preceded the diagnosis (60,61).

Infections

Infectious emboli from meningococcemia may autoamputate limbs or digits (62). The process frequently involves all four limbs. Growth plates may be affected, resulting in angular deformity and the need for surgical epiphysiodesis (63). Frequently, the skin is affected as well as the limb (64). Multiple surgical skin grafts limit the prosthetic fitting; a coordinated burn team is often best prepared to handle initial management (65). Over the past few decades, the incidence of invasive meningococcal disease in the United States has remained relatively stable (66–68). Pneumococcal septicemia also can produce purpura fulminans, characterized by acute onset of rapidly progressive hemorrhagic necrosis of the skin and thrombosis (28). An example of the distal and multiple amputations caused by emboli from infections are seen in Figure 13.4.

Surgical Approach: General Principles

Adherence to the general principles of childhood amputation surgery guides one to optimal function. The principles are: (a) preserve length, (b) preserve growth plates, (c) perform disarticulation rather than transosseous amputation, (d) preserve the knee joint whenever possible, and (e) stabilize and normalize proximal portions of the limb (126).

The cardinal surgical dictum to conserve all limb length if possible is true for children as well as adults. In growing children who require amputation, disarticulation rather than a transdiaphyseal amputation may

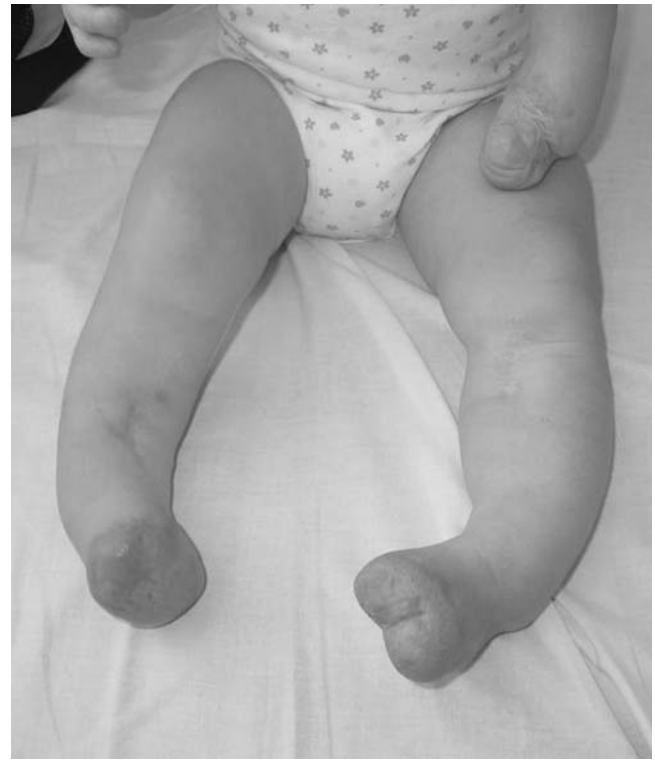


Figure 13.4 Amputations as a result of meningococcemia and subsequent purpura fulminans.

be preferred (69). Disarticulation preserves the epiphyseal growth plates and ensures longitudinal growth (70). Disarticulation also avoids the development of terminal or appositional overgrowth of new bone.

Terminal overgrowth, often referred to as *spiking*, at the transected end of a long bone is the most common complication following amputation in the immature child (148,149). Diaphyseal overgrowth may also occur in children with congenital anomalies, such as amniotic band syndrome, in which the epiphysis is no longer present. It occurs most frequently in the humerus, fibula, tibia, and femur, respectively. During appositional growth, the distal bone begins to form in the shape of an icicle. As the pointed segment creates insult to the soft tissue, a bursal formation often occurs to protect the distal residuum. During this time, the child may experience significant pain and be unable to tolerate wearing prosthesis. Frequent socket modifications are necessary to accommodate these anatomical changes. Treatments such as aspiration, steroid injections, and stump wrapping are usually ineffective. Unfortunately, the rate of growth may be so vigorous that the bone pierces the skin; at this stage, the treatment of choice is surgical revision. Distal resection and stump capping with the use of autografts or plastic polymers are surgical options (97). Once surgery becomes necessary, the problem is likely to recur until skeletal maturity. Each time that bone is resected, the overall length of the bone is reduced, thereby affecting its mechanical advantage and potential control of the prosthesis. Bone spurs may form at the periphery of the transected bone, and resection may be necessary. The resulting stump scarring, which interferes with weight bearing, requires prosthetic modifications. Plastic surgeons are involved with reconstruction of skin flaps or with complicated repairs of residual limbs (47,71). In Figure 13.5 an example of complicated residual scarring is shown.

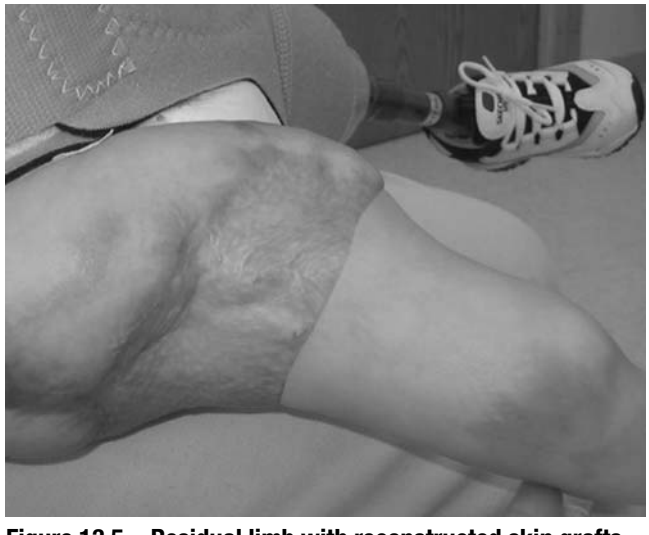


Figure 13.5 Residual limb with reconstructed skin grafts and custom liner.

Phantom Sensation

Phantom sensation is an individual's awareness of the missing limb. It is rarely unpleasant. Since phantom sensation is not painful, no treatment is necessary. Children with congenital deficiencies do experience phantom sensation, though it is not painful (72). Phantom sensations in children with limb deficiency is explainable if we recognize the brain as a generator of sensory information (73). Phantom limb pain rarely occurs in children under 10 or during growth, but is reported in teenagers. In addition, children with congenital limb deficiencies are less likely to experience phantom sensations than those with acquired amputations (74,75).

UPPER LIMB

There are differences in the approach, acceptance, and management of the upper limb amputee versus the lower-extremity amputee. The upper limb prosthesis does not replace the sensory function of the hand and is best used as a mechanical tool (76). The hand is used to explore the environment and to manipulate objects within it. The hand needs to reach the body and precisely approach an object, grasp, and then release it. Acceptance of the prosthesis is variable (77). Frequently, the exposed skin of a deficient limb is preferable to an encased limb. Stump sensation may even be enhanced to compensate for the loss of prehensile area (78).

Common Upper Limb Deficiencies

Digital Deficiencies

Digital deficiencies are common but rarely present in isolation. Removal of additional digits or intervention with Z-plasty procedures produce acceptable results for the children with polydactyly and syndactyly, respectively. Amniotic band syndrome or Streeter's dysplasia commonly presents with digital constriction banding. In addition, other anomalies are often present, such as lower limb amputations that have occurred in utero. While the hand impairments can be attended to, they may affect the child's ability to perform activities of daily living (ADLs) or don and doff a lower-extremity prosthesis (79).

Etiologies such as Moebius syndrome and Poland syndrome (sequence) result in digital deformities associated with a more serious underlying condition. Moebius syndrome often affects the sixth and seventh cranial nerves, which compromises the child's ability to visually follow objects, swallow, and communicate. In addition to hand anomalies, Poland syndrome involves a partial absence of the ipsilateral pectoralis muscle and hypoplastic chest.

Absence of individual digits creates a multitude of surgical and nonsurgical options. These include no intervention, therapy to enhance hand function, pollicization, or toe transfers. Due to the physiological function of the normal thumb, hand impairments can vary widely, depending upon which digit(s) is/are absent. There is often greater consideration for surgery if the thumb is absent. Pollicization can occur to the most radial digit in order to provide oppositional grasp (80). Toe transfers can now be transplanted from the second or third ray and minimize effects on gait mechanics (81,82).

Partial Hand and Wrist Disarticulation Deficiencies

Partial hand deficiencies are quite common and are often treated as wrist disarticulation–level limbs. Nubbins (very small underdeveloped vestigial digits) are present in a majority of these cases, as are shortening of the ipsilateral radius and ulna. Nubbins are rarely problematic or surgically removed. The child can be quite functional with no intervention. The major functional drawback of this particular limb length is the inability to perform prehensile tasks with the involved limb. Plastic surgeons are often consulted for digit- and hand-level deformity.

Transverse Deficiencies of the Forearm

Transverse deficiency of the upper third of the forearm is the most common (major) upper limb deficiency (83). The clinical presentation of these children is similar to that of children with longer, transradial residual limbs. Ipsilateral humeral shortening and the presence of smaller nubbins are common to this level. The proximal radius in these shorter residua is often unstable, subluxing anteriorly during full extension. This creates a challenge to prosthetic fitting. The longer residual limbs, in the middle third of the forearm, tend to be more easily fit with prostheses, as they have more surface area over which to distribute the forces of the socket interface. They also have longer lever arms with which the patient can control the prosthesis.

Rarely will there be any surgical intervention to this level of limb deficiency (84). If prosthetic intervention is not attempted or accepted, bimanual tasks will be performed via grasping of objects in the cubital fold, between one's legs, in the axilla region, or under the chin.

Elbow Disarticulation and Transhumeral Deficiencies

The more articulations that are involved, the greater is the functional deficit. When the elbow joint is compromised or absent, the child has fewer options to assist in prepositioning his or her distal limb in space. The child relies solely on the muscles and range of motion of the shoulder complex. The true elbow disarticulation limb has the distal epiphysis present, which is important to overall growth of the residuum. A drawback of any disarticulation is the lack of room to fit prosthetic components and maintain humeral length equality.

Transverse deficiencies of the humerus are analogous to acquired transhumeral amputations in children. The residual limbs are often medium to short in length compared to their contralateral limb. This level of deficiency has been previously noted as the most common to experience diaphyseal overgrowth. This leads to a short, nonfunctional residuum when multiple surgeries have been completed.

Shoulder Disarticulation and Intrascapulothoracic Deficiencies

It becomes increasingly difficult to restore the functions of the anatomical arm as the level of deficiency reaches the shoulder and higher. Children with remnant humeri have the ability to use these segments to assist in their activities. Often, the axilla will be used to assist these individuals to grasp and manipulate objects. If the child has unilateral limb deficiencies, the contralateral noninvolved limb will be the dominant side for grasping, with holding for manipulation taking place between the knees, in the mouth, or trapped between chin and chest or chin and shoulder. When the child has bilateral deficiencies at the shoulder level, the latter method is all that is possible to grasp objects. In these cases, the child will be strongly encouraged to use his or her feet to grasp and manipulate objects.

Many designs of upper-extremity prostheses require a degree of body movement (excursion) to operate the mechanical components. Most of this excursion is not present in the shoulder disarticulation level, as glenohumeral flexion no longer exists as a source of control input. This is further magnified when the child has an intrascapulothoracic (forequarter) level of involvement, as they only have uniscapular motion to capture for prosthetic limb control. These two issues will be discussed at length in the following sections.

Uncommon Upper-Limb Deficiencies

Longitudinal Deficiencies of the Forearm

Radial deficiencies are approximately three times as common than ulnar deficiencies, occurring in 1 in 30,000 and 1 in 100,000 live births, respectively (85). Fanconi anemia; thrombocytopenia and absent radius (TAR); Holt-Oram syndrome; vertebral defects, imperforate anus, tracheoesophageal fistula, and

renal defects (VATER); and Robert's syndrome are just a few examples of etiologies with associate radial involvement (86,87). Figure 13.6 illustrates the complex issues with Robert's syndrome. The clinical presentation of radial deficiencies usually involves the radial-side digits of the hand as well. Depending upon the classification of the radial deficiency, prehensile capabilities may be compromised by a hypoplastic or absent thumb. In these situations, pollicization or toe-transfer procedures are often discussed. Treatment for radial deficiencies is focused on reconstructing the thumb and, in both the radial and ulnar deficiencies, is directed at centralization of the hand (88).

Ulnar deficiencies are associated more with musculoskeletal conditions than systemic conditions, and isolated genetic predispositions have been discovered (89). Cornelia de Lange syndrome, ulnar-mammary syndrome, and ulnar fibula dysplasia are examples of syndromes that involve ulnar deficiencies. With ulnar-side involvement, the thumb and another digit are usually present.

Central ray syndrome, a form of ectrodactyly, had been described previously as having genetic predisposition. This is commonly referred to as "lobster claw," as the central component of the hand and/or feet are absent. This can present as a mild condition, with the more ulnar and radial digits still present, or it can present as two longer and thicker digits. Functional abilities with this condition will vary, depending upon the degree at which the syndrome affects the deformity. Many of these individuals will not need prosthetic restoration, as the limbs are at full length and have prehensile and tactile capabilities. Surgical reconstruction may be recommended if the child lacks the oppositional capabilities that the thumb usually offers.

Figure 13.6 Child with Robert's syndrome. Note flexion contractures of all limbs.

Longitudinal Deficiencies of the Humerus

When a longitudinal deficiency of the humerus is present, it is often associated with deficiencies in the radius and ulna and with phocomelic digits. The length of the arm is compromised, which leads to the inability to work in a larger envelope of space when attempting to perform bimanual tasks. For this reason, prosthetic fitting is more likely a consideration instead of longitudinal deficiencies of the forearm. The shoulder complex is quite often compromised as well. Therefore, if the child were to be fit with a prosthesis, he or she would most likely receive some externally powered components. Frequently, the phocomelic digits will be used to provide input to these components.

Intervention, Prosthetic Treatment, and Adaptive Equipment

Although prosthetic treatment may seem indisputable for an individual with a limb absence or acquired amputation, it is not as straightforward as one might imagine. The inability to provide or restore the function of the human arm and hand poses great challenges to individuals with partial or complete limb loss (77). These fittings are generally limb-level–dependent as well and vary between passive, body-powered, and externally powered options. Acceptance of prosthesis is a complex issue; factors that influence acceptance include level of limb loss, presence of other complicating medical conditions, comfort and usefulness of the prosthesis, and acceptance of the limb deficiency by the family. In general, the higher the limb absence, the less likely it is that a child will find a prosthesis useful enough to wear it regularly. For example, transradial patients will tend to wear their prostheses more than transhumeral patients, and transhumeral patients will tend to wear their limb more than shoulder disarticulation patients (90).

Goals of early intervention and training revolve around achieving age-appropriate milestones. Children with upper limb differences frequently achieve developmental milestones at or around the same age as children without limb anomalies. Prostheses are generally considered around 3 to 6 months of age (91). Until recently, 6 months of age used to be the time at which fitting was initiated (92,93). This was the age chosen because it was the time the child was expected to have achieved sitting balance. Clinical experience versus evidence-based study guides fitting timetables (94). Although there are general guidelines for fittings, the initial fitting is something that is discussed in the clinic between the team members and family. Many children will be fitted with prostheses prior to 1 year of age. Early prosthetic fitting is designed to encourage bimanual tasks, establish a wearing pattern, increase overall

independence, provide for symmetrical crawling, and reduce "stump dependence"—sensory dependence on the end of the residual limb (95). Early fitting does not guarantee acceptance (96). The prostheses needs to fit comfortably, which can be challenging to assess in an infant, be relatively easily donned, equalize lengths with the noninvolved limb, allow for growth, and provide restoration acceptable to family (97).

Several different terminal devices may be considered for the first prosthesis. Age-appropriate prostheses are fitted to children; passive prostheses are generally the first design utilized. Options include hands, hooks of various shapes, mitts, and other non-hand designs. The vast majority of parents prefer a terminal device that looks like a hand. For this reason, it is recommended that a passive hand be provided rather than a hook or other non-hand device. The two basic passive hand options for infants are the closed, "crawling hand" design and the open hand design. The parents should be involved in the decision-making process—this involves providing information about the pros and cons of each style and, more importantly, letting the parents decide which design is most acceptable in their eyes. Once the parents accept the child's limb deficiency, they will be more inclined to evaluate prosthetic components based on functional qualities in addition to appearance. If parents are involved in the decision and accept the device, they are more likely to encourage the youngster to wear the prosthesis. Figure 13.7 shows an infant passive hand.

It is questionable whether it is appropriate to fit children with partial hand deficiencies and wrist disarticulations at a very young age. They have long residua and can use them for bimanual tasks. Figure 13.8 illustrates a transcarpal limb deficiency with adequate length for function. The prosthesis would serve the purpose of providing a wearing pattern and also reducing dependence on the sensation of the limb. The latter can arguably be considered as a positive rather than a negative. The passive terminal device options can be similar, although there are limitations due to the length available distal to the residuum. Opposition posts are sometimes considered for the child with carpals and wrist motion. These devices can be rigidly fixed or placed in several different positions to accommodate for grasping different-sized objects.

For the child with a limb that extends distal to the elbow, the initial prosthesis is usually self-suspending, using a supracondylar design, with or without a suspension sleeve. If this is not achievable, a narrow Dacron harness may be designed in a figure-eight configuration. This harness should be easy to put on the child, have elastic as part of the straps for increased shoulder motion, and have snaps or fasteners that make it easy to put on and take off.

The same passive terminal device options are appropriate for the child with a limb deficiency proximal to the elbow. The major difference between these levels is that the absence of the elbow joint makes it more difficult to preposition the terminal device for

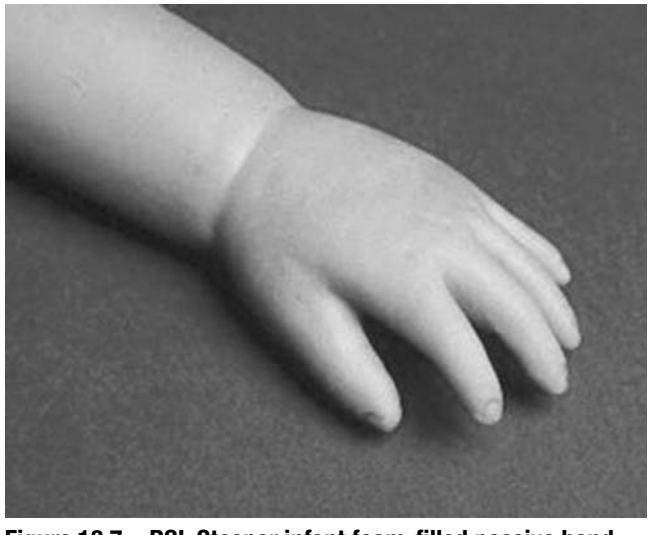


Figure 13.7 RSL Steeper infant foam-filled passive hand.

Figure 13.8 Transcarpal limb deficiency with adequate length for bimanual tasks.

bimanual tasks. The child is not cognitively ready for an articulating elbow; therefore, a curved shaped "banana" arm is often provided in order for this child to engage the prosthesis with the contralateral hand as well as reach levels that are closer to the midline and face (98). Figure 13.9 displays the passive "banana arm" prosthesis.

The next developmental milestone is walking, which usually occurs at 11 to 13 months of age. This will indicate that the child is ready for a more sophisticated upper-extremity prosthesis. At this time, the child is ready to perform simple grasp-and-release activities using the prosthesis. It is imperative that the family be involved in the clinical decision-making about their child's prosthesis. The prosthetists should have designed the prosthesis in a manner to accommodate growth. It is best to keep the control system as simple as possible at this early age in order to ensure early success. Other developmental factors to be considered are understanding of holding function, attention span longer than five minutes, and willingness to be handled by an occupational therapist to go through terminal device opening motion.

When the child is developmentally ready for terminal device activation, options include body-powered hooks or hands as well as myoelectrically controlled hands. The majority of parents prefer hands; the hands that provide optimal function at this age are myoelectrically controlled. At this age, the simplicity of control is of paramount importance. An electric hand that is controlled by one electrode in a voluntary opening control scheme has proven effective and natural. This electronic scheme permits the child to activate the hand opening with a contraction (usually on the side of the wrist extensors) and relaxation that enables the hand to automatically close. This electromechanical design is analogous to a split hook, voluntary-opening prosthesis. Designing such an electronic control scheme eliminates the need for the child to maintain muscle contraction in order to continue grasping the object. As the child grows older, another electrode can be added to the flexor side of the forearm, enabling the child to have volitional control opening and closing the myoelectric hand (99).

Myoelectric hands of the past were too large and difficult for a 1- or 2-year-old child to use successfully. Therefore, it was recommended that these hands not be fitted on children until 4 to 5 years of age. Today, it is common for these hands to be fitted successfully on 1-year-old children. Prosthetic technology has improved dramatically as a result of miniaturization and simpler control to better meet the needs of very young children. Figure 13.10 shows a transradial myoelectric prosthesis with myoelectric hand terminal device.

Body-powered devices may not work well for this age group because they lack the requisite force and excursion, as well as the cognitive ability, to relate shoulder motions to terminal device operation. The voluntary-opening–style terminal devices permit the user to grasp an object and allow the force of the elastic bands or springs to keep the object in the terminal device. This may be ineffective if the child cannot overcome the force required to activate the terminal device. The designs of voluntary-opening terminal devices for children are not very aesthetically pleasing, with the exception of the mechanical hands. The hands, however, have the drawback of providing minimal efficiency. Once a cosmetic glove is applied to the mechanical hand, it can lose up to 40% of its efficiency, compared to the function of the hand without the glove voluntary closing terminal devices have gained in popularity, although the child must maintain force and excursion through the harness to maintain grasp on an object. The amount of grasping force is directly proportional to the force that the child puts into the

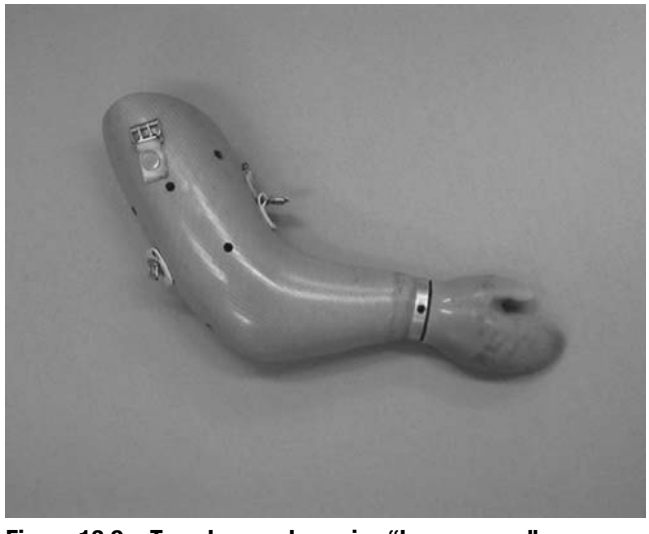


Figure 13.9 Transhumeral passive "banana arm" prosthesis.

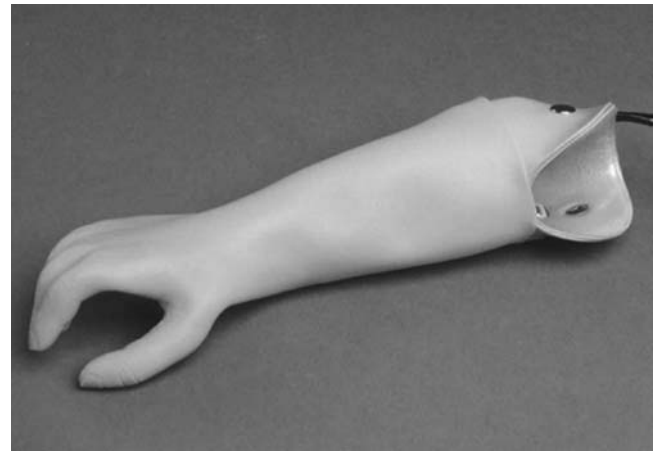


Figure 13.10 Transradial myoelectric prosthesis.

harness (100). The figure-of-eight harness is pictured in Figure 13.11 with straps shown. The prosthetic team can predict the ability of the child to control the myoelectric components when using evaluative tools such as the capacity of myoelectric control (101,102).

By the time children are 4 or 5 years old, they are able to operate virtually all types of prosthetic components and control schemes presently available (100).

The developmental milestones described previously should guide the fitting schedule of the transhumeral limb-deficient child. Because of the nature of a transhumeral prosthesis, it can be more of an encumbrance than the transradial design. This can cause difficulty in rolling over, and may impede the child's development if fitted too early. The terminal device should be activated shortly after the child begins to walk. Terminal devices for the transhumeral level are the same as the transradial. The addition of a prosthetic elbow is the key difference. The first prehensile prosthesis will employ a friction elbow to allow positioning of the terminal device. It is useful to limit the range of motion at the elbow by producing flexion and extension stops to prevent the elbow from flexing excessively during weight bearing activities (eg, crawling). The initial prosthesis may be suspended by a harness or by silicone suction suspension. The silicone suction socket (3S) has proven effective because it allows free range of motion at the shoulder and provides excellent suspension. The child with a transhumeral deficit should be fitted with an activated terminal device once he or she begins to walk.

Considerations for terminal device selection include appearance, weight, ease of operation, and cost. The myoelectric hand offers reasonable appearance and ease of operation when controlled by a single-site voluntary opening circuit; however, it is a heavier and more expensive prosthesis compared to body-powered. Either voluntary opening or voluntary closing designs can be used successfully by the transhumeral limb-deficient child once the child has sufficient strength and the cognitive ability to understand how to operate the device. This usually is possible at 2 to 3 years of age. When the child is strong enough to operate an active elbow, usually at age 4 to 5, a conventional body-powered elbow may be provided; however, locking of the elbow by conventional methods may prove challenging. If the child has insufficient strength/excursion to operate the body-powered elbow, an electric elbow may be considered, although the increased weight may preclude this option. The terminal device illustrated in Figure 13.12 is a voluntary opening split hook and can be utilized on both transhumeral and transradial deficiencies.

The shoulder disarticulation level is treated differently due to the challenge in positioning the shoulder, elbow, and terminal device. The child may be fitted with passive endoskeletal shoulders and elbows with an active terminal device. Current fittings have been utilizing externally powered hands controlled by either electromechanical rocker switches or force-sensing resistors. The child is encouraged to maintain good ranges of shoulder elevation/depression and protraction/retraction in order to make contact with these input devices. The return or enhancement of function using these devices is quite limited. Therefore, there are no "right" philosophies for the fitting of these complicated cases. The team should recognize that prostheses need to be useful to the child in order for him or her to choose to wear them. A useful sport's prosthetic terminal device is illustrated in Figure 13.13.

The phocomelic or bilateral total upper-extremity transverse deficiency patient rarely requires amputation revision; indeed, the terminal digits can activate

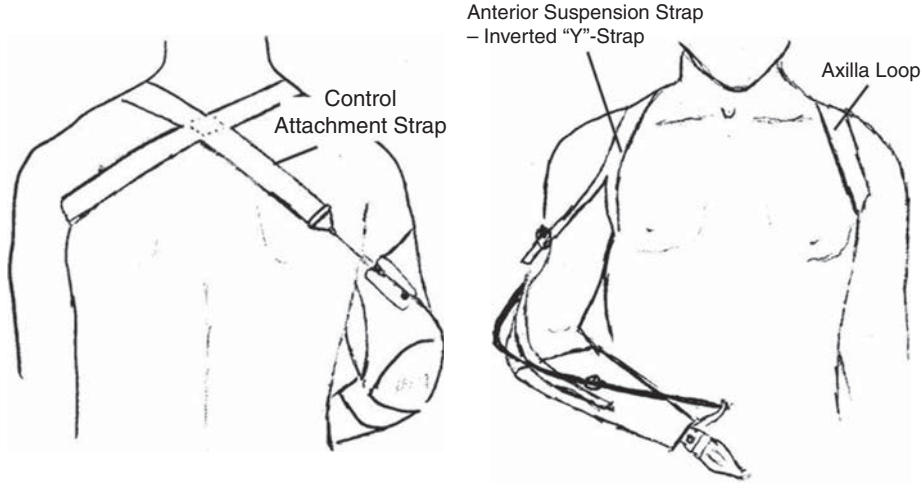


Figure 13.11 Figure-eight harness.

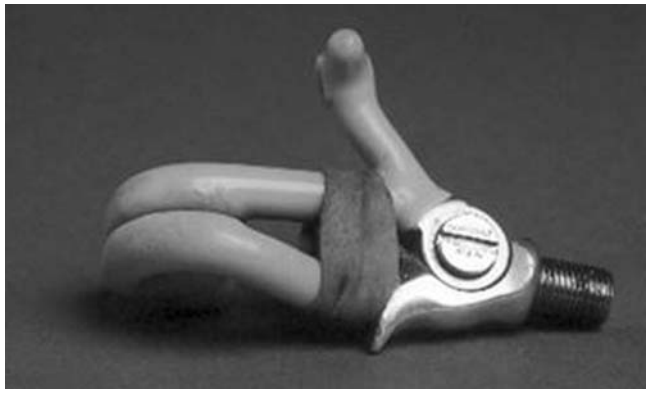


Figure 13.12 Hosmer voluntary opening, split hook terminal device.

Figure 13.13 Child with TRS High-Fly Fielder terminal device.

switches or myoelectric sensors (90). In the case of higher-level bilateral deficiencies, it is wise to start as simple as possible, recognizing that each child has a certain level of tolerance for "gadgets." With the vast array of prosthetic components now available, it would be easy for the well-intentioned clinic team to recommend components that would overwhelm the user. Critical factors in the success of the high-level bilateral are prosthetic weight, complexity of control, proprioceptive feedback, wearing comfort components, and motivation and attitude of child and family.

Therapy and Training

The preprosthetic period is mainly focused on the needs of the parents (103). The family level of distress or stress related to the child's limb deficiency will vary (104). It is important during this initial contact for the clinic team to present an honest forecast of the prosthetic plan. The team members must walk a fine line between presenting the prosthetic options in an honest manner without sounding negative or disheartening. After all, prosthetic technology with all of its sophistication is still far from the ideal of replacing a physiological arm.

The parents should be encouraged to treat the child as they would a child with normal limbs. Many parents benefit from being introduced to other parents and children with similar limb deficiencies (106).

Typically, children younger than 3 years of age have therapy provided in the home. Prone positioning is important for encouraging trunk extension and mobility. Gross motor milestones are generally not delayed, but may be affected by asymmetry imposed by unilateral upper limb deficiencies. Children compensate and substitute for the missing action of limbs. Therapists bridge the delivery of the prosthesis to the initiation of function and create a comfortable environment for children to explore options, with or without the prosthesis. The goal is to increase the child's awareness of the affected side, including the prosthetic device. The child should also be encouraged to use the prosthesis for transitional movements, such as sitting to crawling, and leaning on the prosthesis for weight bearing while reaching with the dominant hand. The parent is encouraged to maintain telephone contact with the therapist to answer questions regarding follow-through with prosthetic usage. A recheck through the clinic should be scheduled within a month after delivery of the prosthesis and then every three to four months.

When the terminal device is activated, the therapist will again provide initial instruction to the parents and child. A structured approach to use of the terminal device assists parents, child, and therapist in gaining confidence and competence (106). The therapist will work with the child and parents using toys that encourage bimanual use, such as Lego bricks, pop beads, and stringing beads. Initially, it is useful to concentrate on activities that require the prosthetic side to hold while the dominant hand manipulates. When training a child in the use of a myoelectric hand using this control scheme, the therapist should encourage activities that cause the hand to open. Because of the placement of the electrode over the forearm extensors, activities that elicit an extensor activity are appropriate. Once the hand is open, the therapist can quickly place a toy in the hand and

encourage the child to release it. The child will learn through repetition.

It is unrealistic and inappropriate to teach the child to use the prosthesis for dominant hand activities. Children with high-level bilateral upper-extremity limb deficiencies will utilize their feet in a natural manner. Assistance is necessary for donning and doffing prostheses. The child with an isolated limb deficiency or amputation is capable of achieving age-level academic skills. Few studies have been done to define achievement academically. Good social adjustment is reported for children with myoelectric prosthetic users (107). School placement is almost entirely within the regular school system, with an Individual Education Program (IEP) to address educationally related function. Occupational therapists will assist with issues of grasp and fine motor control for paper, computer, and ADL tasks needed in school. Informational pamphlets have been developed for the teacher to prepare the able-bodied students for integration of children with physical disabilities into the regular education classroom.

Adapted physical education may be necessary, but regular physical education is often sufficient. The philosophy promoted for children with physical disability is that of "participation, not observation." Participation in athletic endeavors such as skiing, tennis, and other more mundane exercise improves the self-concept of the child or adult with limb deficiency. Specialized, adaptive prosthetic components that enable unilateral or bilateral handless persons to participate in sports such as golf, shooting, and ball sports have escalated since the 1980s.

Functional assessments recently developed to determine the use of upper-extremity prosthetics and function have included Assisting Hand Assessment, the Prosthetic Upper Limb Functional Index, The University New Brunswick Test of Prosthetic Function, Child Amputee Prosthetic Project-Functional Status Index, Child Amputee Prosthetic Project-Functional Status Index Preschool, Shriners Hospital Upper Extremity Evaluation (SHUEE), Capacity for Myoelectric Control (101), and Unilateral Below Elbow Test (108–114). It is typical for children to perform activities of daily living with their prosthesis, but often choose not to utilize them (115). In addition, prosthetics are often utilized for specific tasks versus everyday tasks. Children typically utilize nonprosthetic options ages 3.5 through 13 years of age (103).

Outcomes related to patient satisfaction are increasingly important to evaluate for prosthetics (116,117).

Recent studies have indicated that children with unilateral, below-elbow deficiencies who do not wear prostheses perform as well or better than their counterparts who wear prostheses (77).

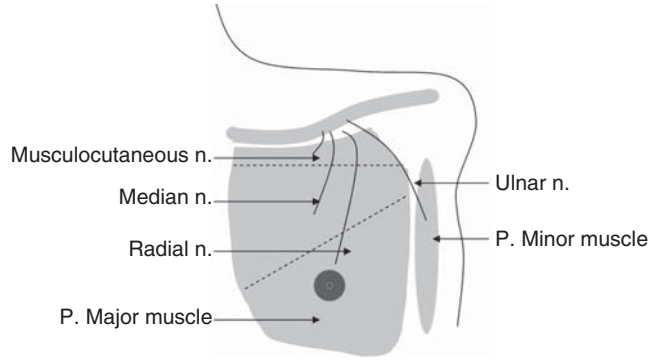


Figure 13.14 Proposed nerve transfer for targeted muscle reinnervation of an individual with shoulder disarticulation amputation.

Advancements in Upper-Extremity Prosthetics

There have been many additional components offered for children with upper limb involvement; however, one of the most exciting advancements comes in the form of a new application to nerve transfers. Although only performed in adults to date, targeted muscle reinnervation (TMR) has proven an effective means of creating additional, physiologically appropriate myoelectric sites for individuals with high-level, upper-extremity amputations (118–120).

Following amputation, the remaining peripheral nerves (ie, median and distal radial nerves) are grafted to denervated muscle sites in order to create additional, distinct myoelectric sites for the user that are physiologically appropriate, as illustrated in Figure 13.14. For example, on the transhumeral limb, the medial head of the biceps and the lateral head of the triceps are dennervated and reinnervated by the median and distal radial nerves, respectively. When the reinnervation is complete (after approximately four to six months) these two additional myoelectric sites are available for physiological control of closing and opening of a myoelectric terminal device. The prosthetic socket then incorporates four independent myoelectric sites for control of elbow flexion and extension, via native lateral biceps and medial triceps, and control of the terminal device by the aforementioned reinnervated muscles. Increased efficiency and ease of use have been positive outcomes from this surgical intervention.

LOWER LIMB

Deficiencies of the lower limb are less frequent than deficiencies of the upper limb, but surgical and rehabilitation management may be more involved. Most of the common lower limb anomalies are longitudinal

deficiencies. Despite the complexity of the early intervention, lower limb prostheses generally have high acceptance rates. Most individuals wish to ambulate independently, and these prostheses afford the children the opportunity to do so. In addition, mobility demands less precision than the positioning and fine motor skills of the upper limb.

Surgical intervention is often required to correct the deformity or provide a functional lower limb. This is the most challenging aspect of the early management of these children. Parents are often faced with difficult decisions of choosing among such surgeries as foot ablation, angulation osteotomies, epiphysiodeses, limb lengthening, and rotationplasty. In addition to the usual risks of surgery and uncertain outcome, ethnic and religious barriers are important in family decisions. Parents may benefit by meeting other families who have faced similar situations. This may ease the discomfort of the decision making for the parents and child (105).

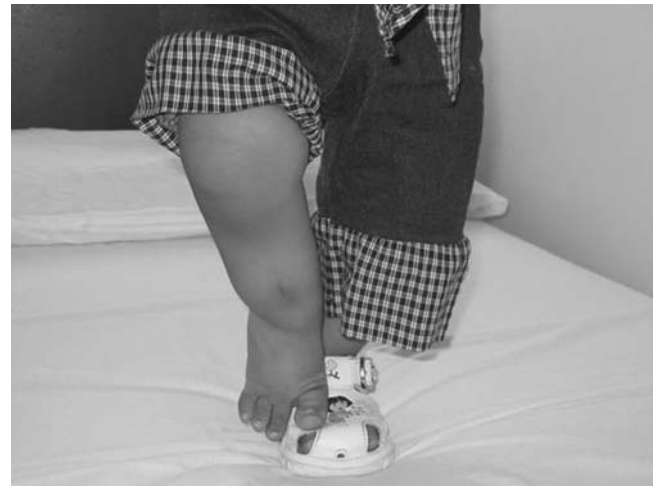


Figure 13.15 Child with fibular deficiency. Note subcutaneous dimple, leg length discrepancy, and four-rayed foot.

Common Lower-Limb Deficiencies

Longitudinal Deficiency of the Fibula

The most common, and possibly the most controversial, deficiency is the longitudinal deficiency of the fibula. Many classifications and levels of involvement exist. With partial deficiencies of the fibula, outcomes will vary.

There is no evidence that this anomaly, in isolation, is genetically transmitted (12). It has been suggested that the fibula is undergoing "regressive evolution" and that may be the reason for the prevalence of deficiency of the fibula and susceptibility to congenital absence (122).

The clinical presentation of longitudinal deficiency of the fibula, which is a completely absent fibula, generally has a foreshortened tibial section, and frequently has ipsilateral femoral shortening. This tibial section appears shorter than it is as a result of kyphoscoliotic bowing. This anterior bowing of the tibia shortens the segment longitudinally and creates an anterior prominence of the tibia. This anterior prominence is indicated by a subcutaneous dimple, which can range from superficial to invaginated. Figure 13.15 shows a child with a fibular deficiency. Proximally, the limb is often in genu valgum or drifts into genu valgum as the child grows. The distal involvement is usually an equinus position and a valgus posture during weight bearing due to lack of lateral support. Lateral tarsal and ray absences are often associated with this lateral long bone absence. As the child grows, the popliteal area becomes convex, with the medial hamstrings descending much lower than the lateral hamstrings. On physical examination, the degree of internal hip rotation is often less than that of external hip rotation.

Many surgical options are available for treatment of longitudinal deficiencies of the lower limb. Historically, the most common treatment of a complete fibular absence has been with a Syme amputation, which is successful in providing an end-bearing surface for ambulation, with or without a prosthesis (123,124). Amputation takes place when the child is beginning to pull to stand and cruise with the assist of furniture or toys. Migration of the heel pad posterolaterally has been noted in the follow-up of many Syme amputations, as shown in Figure 13.16. This migration may be due to the use of the posterior calf musculature during active ambulation in the prosthesis. The Boyd amputation serves to centralize the heel pad more effectively and is the surgery of choice in many clinics. In addition to the ankle disarticulation procedures, it may be necessary for the child to undergo unilateral epiphysiodesis or angulation osteotomies if the genu valgum becomes a prosthetic challenge to fit. Outcomes from Syme amputations have shown that these children are able to perform very well in their communities, have a good self-image, and are rarely limited in activities (125).

External fixator applications and advancements have provided options which challenge the team and orthopedists to reconsider amputation. Saving the foot would be the first choice if the procedure were proven to be as successful as the Syme amputation (125). Considerations for these procedures include level of involvement or "grade," risks and psychological effects of multiple surgeries, potential (and probable) infections around pin sites, and physical effects of "down time" the child will experience during and

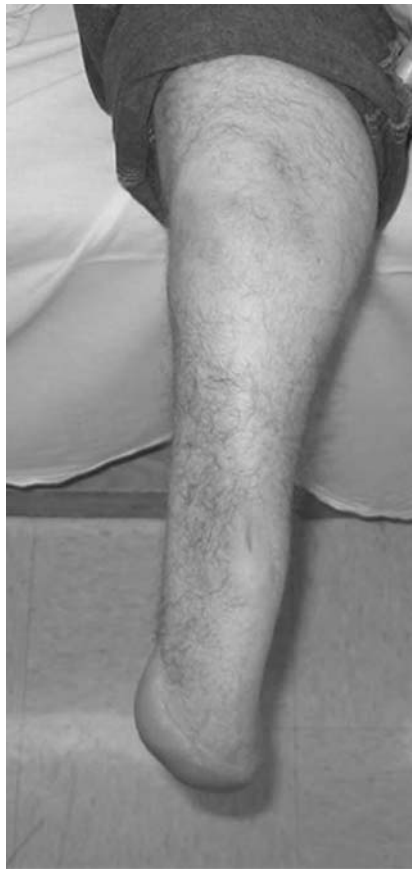


Figure 13.16 Limb with longitudinal deficiency of the fibula following Syme amputation. Note posterior lateral migration of heel pad.

after wearing the external fixator (7). In the event that the foot ablation is not imminent, orthotic fitting combined with shoe modifications will be necessary for the child to ambulate successfully.

Femoral Abnormalities

The term that has been used to define the most common deficiency of the femur has been *proximal femoral focal deficiency* (PFFD). With varying levels described first by Aitken in the late 1960s, PFFD has been the acronym of choice for many femoral anomalies. Congenital short femur differs from PFFD by having the proximal aspect of the femur and intact ipsilateral acetabulum.

Although the skeletal structures are quite variable, the clinical presentation for these limbs is similar. The femoral section is shorter, with a larger mass of soft tissue, which includes musculature, between the pelvis and the involved knee. A typical appearance is shown in Figure 13.17. These muscles are highly ineffective as a result of being slack and not stretched to their full potential. The hip posture and stability is quite variable. All of the limbs present with some degree

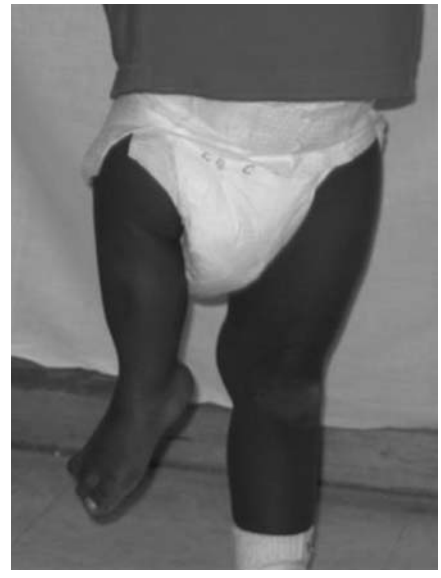
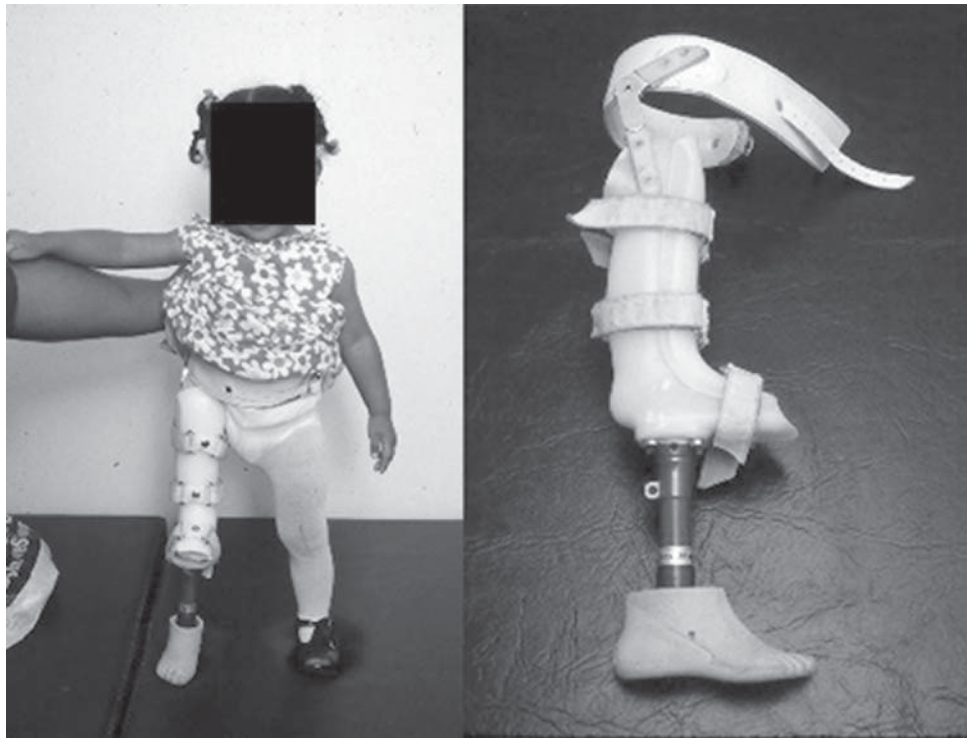


Figure 13.17 Child with femoral abnormality. Note clinical presentation of the hip flexion, abduction, and external rotation.

of hip flexion, abduction, and external rotation. For the congenitally short femur and lesser involved PFFD, the labrum is present, resisting proximal subluxation. More involved presentations have progressive suluxation to dislocation of the femur. Often, there is an associated fibular absence.

Surgical options for the congenitally short femur are numerous. The first option is "no surgery." In some cases, some clinics and families feel that no surgical intervention is the best option for treatment of this disorder. In these cases, various lengthening devices such as shoe lifts and extension prostheses are necessary. These are sometimes referred to as "prosthoses," because they often combine a proximal orthosis with distal prosthetic components. An example of the "prosthosis" is seen in Figure 13.18. Children with bilateral femoral involvement often have no surgeries and no prostheses. For those who have an intact short femur with both proximal and distal growth plates, this option may be a consideration; the addition of an external fixator to lengthen the foreshortened femur may also be considered. If the amount of lengthening necessary is unattainable, the limb lengthening may be performed in conjunction with appropriately timed epiphysiodesis of the contralateral leg to equalize leg lengths at full maturity.

Ankle disarticulation along with knee arthrodesis is another option. It is performed when the proximal femur is affected and when the length discrepancy is such that an external fixator could not achieve the desired lengthening. The ankle disarticulation amputation is either a Syme or Boyd. Figure 13.19 is an example of a Syme amputation. Fusion of the knee may be delayed, as arrest of the proximal tibial and



A B

Figure 13.18 (A) Child with femoral abnormality wearing "prosthosis." Prosthosis is a combination Bi-valve KAFO with pelvic band and hip joint (B), extended with a prosthetic pylon and foot.

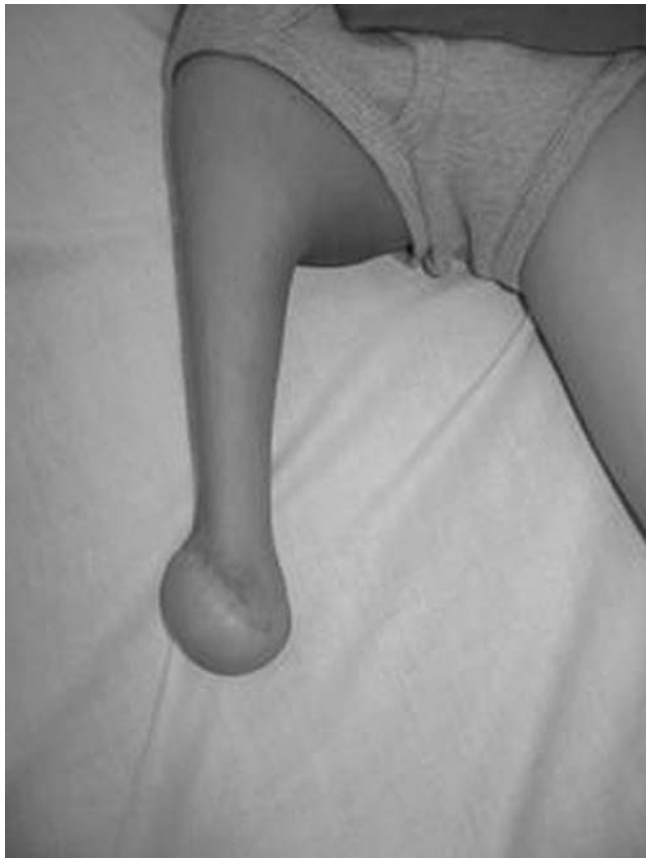


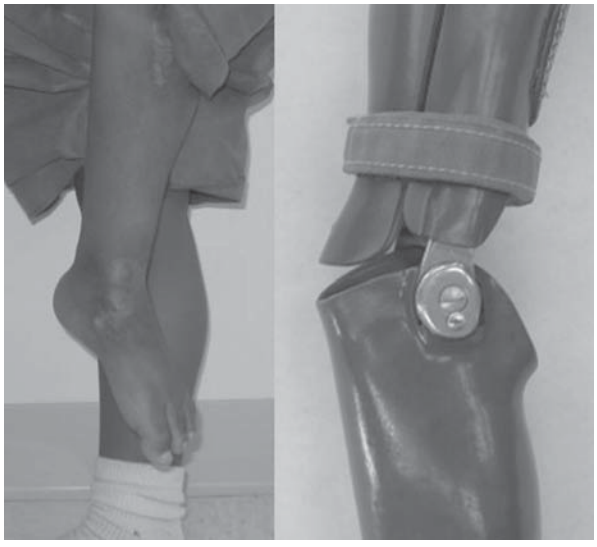
Figure 13.19 Child with femoral abnormality following Syme amputation. Note bulbous distal end of limb and proximal thigh musculature.

distal femoral growth plates will occur at that time, leaving a shorter overall limb length to control a prosthesis. This delay may be unnecessary if the overall limb length and the attempt to provide adequate space for congruency of a prosthetic knee with the noninvolved knee may be achieved during one surgery.

Another surgical option is that of a rotationplasty procedure. An intact fibula is preferred for this surgical procedure. The procedure involves rotation of the foot 180 degrees through removal of the distal femoral and proximal tibial epiphyses, and rotation of the distal segment prior to internal fixation (126). The rotated foot can now act as a knee, utilizing ankle dorsiflexion as knee flexion and ankle plantar flexion as knee extension, as in Figure 13.20. This procedure has demonstrated effective outcomes; however, the aesthetic appearance of the limb following surgery has limited its popularity (127,128). It is crucial that the therapist and family aggressively work on maintaining the full range of motion of the ankle, especially in the sagittal plane. If this does not occur, all that has been accomplished is turning a foot "backwards" on the leg. Derotation of the foot has occurred on occasion, requiring additional surgery to again position the forefoot posteriorly.

The Van Ness procedure has been utilized in other disorders such as burns and osteosarcomas (129,130).

In order to address the proximal subluxation of the femur and provide for a single articulation within the prosthesis, an iliofemoral fusion may be performed.



A B

Figure 13.20 Rotationplasty limb (A) and close-up of prosthetic socket (B). Note knee center discrepancy and use of external knee hinges.

This may be in conjunction with a rotationplasty procedure or in isolation (131).

Longitudinal Deficiency of the Tibia

Longitudinal deficiency of the tibia occurs in 1 in 1 million births. Genetic transmission has been associated with these anomalies, particularly when a bifurcated distal femur exists; 30% of partial tibial deficiency occurs as an autosomal-dominant inherited pattern. The treatment is straightforward since the tibia is the major weight bearing bone. Differences in treatment occur between complete versus partial tibial absence. Figure 13.21 shows examples of tibial deficiency by radiograph.

The clinical presentation of a longitudinal tibial deficiency may include a varus foot and lower leg, a short leg, and an unstable knee and ankle (or both). The foot may have medial tarsals, metatarsals, and rays missing as well. On radiograph, a distal femoral bifurcation may add to the challenge of prosthetic fitting as well as being an indicator for genetic influence (132).

When there is a complete absence of the tibia, the treatment of choice is disarticulation at the knee. The fibula cannot sustain weight bearing of the individual at full maturity, and the instability of the knee and ankle is too great for corrective measures. For the child with a partial tibial deficiency, the segment length is important. If the tibial segment is short, the surgeon creates a synostosis with the intact fibula in conjunction with amputation of the foot. If the heel pad is retained, this procedure will create a walking surface for the child, providing stability without a prosthesis. However, this limb length is shorter than

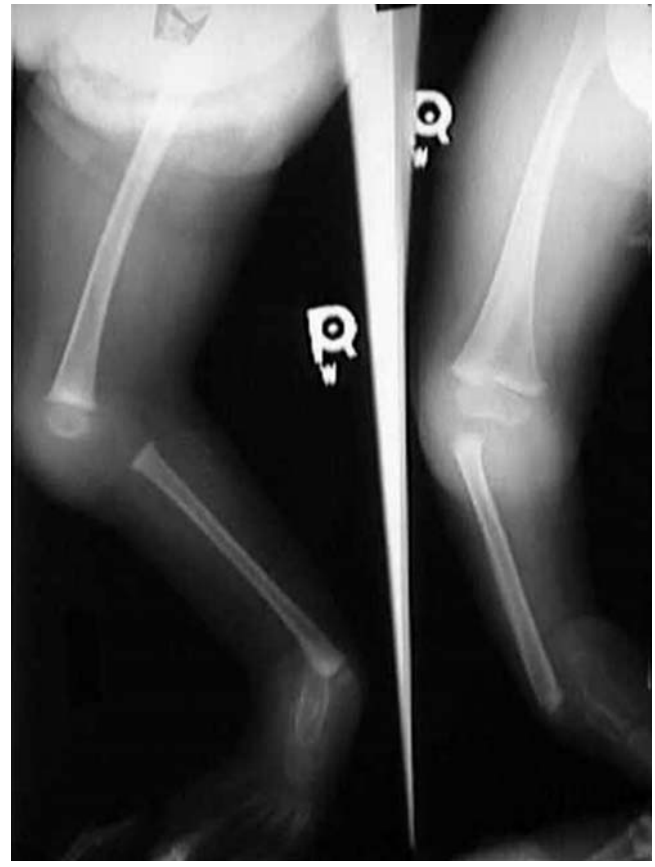


Figure 13.21 Radiographs of child with tibial deficiency.

the aforementioned Syme amputation and may prove difficult for the child to walk without his or her prosthesis. Although treated as a "transtibial amputation," many of these residua grow in quite a different manner. Often, the distal tibia and fibula fuse, while the fibula continues to grow at a faster rate than the tibia. The resulting deformity is a laterally bowing lower leg (the distal end is pushed medially), a fibular head that becomes more prominent and continues to grow proximal-laterally, or both. These additional deformities may not be able to be accommodated by the prosthetists in the socket fitting.

Uncommon Lower-Limb Deficiencies

A few of the less common lower-limb deficiencies that may be seen in the clinic include those resulting from amniotic band syndrome, central ray syndrome, Robert's syndrome, and sacral agenesis. The amniotic banding can occur at any level, but has been frequently seen to cause auto-amputation at the transtibial level (24). The critical factor in working with and fitting these children is keeping in mind that the likelihood of bony overgrowth is great as a result of the banding occurring at the diaphyseal level. Central ray syndrome and Robert's syndrome

have autosomal-dominant inheritance. They can present with a wide array of lower limb anomalies. The result of these presentations and subsequent surgeries will vary. It should be noted that in the case of Robert's syndrome, maintenance of range of motion should be stressed to prevent severe limb contractures; these contractures can affect both fit and function in prostheses. Sacral agenesis is a frequent cause of hip disarticulation or hemipelvectomy; they are difficult to address from the standpoint of a functional prosthetic fitting.

Commonly Acquired Lower-Limb Amputations in Children

Acquired lower limb amputations are the result of trauma, tumor, or infection. Traumatic lower limb amputations occur more frequently than traumatic amputations of the upper limb. These amputations occur secondary to lawn mower, train, motor vehicle, and farming accidents (38–40). Lawn mower accidents often result in partial foot amputations. Train accidents are generally a result of teenagers attempting to board slow-moving trains. The amputations are often high and/or bilateral in nature as a result of the current that draws the youngster under the wheels. Motor vehicle and farming accidents present with varying amputation levels.

Meningococcemia and staphylococcal infections with the onset of purpura fulminans can be destructive to the child. If the child is fortunate enough to survive, there are often multiple limb amputations. Lower limb amputations can range from partial foot to transfemoral levels; the most frequent are transtibial levels. Complications due to growth plate arrest, bony overgrowth, and fragile skin may necessitate revision to the knee disarticulation level (63,67).

Both osteogenic and Ewing's sarcoma are more prevalent in the lower limb than in the upper limb. Osteogenic sarcoma tends to have a better survival rate, in that it involves more skeletal than soft tissue structures. Limb salvage techniques and endoprostheses in conjunction with chemical and radiation therapies have often averted the necessity of amputation (45,46,48–50,54,55,129,133–137).

Many lower limb amputations that occur resulting from osteogenic sarcoma are at the transfemoral level, while Ewing's sarcoma tends to migrate more proximally to the region of the upper thigh and pelvis.

Intervention, Prosthetic Treatment, and Adaptive Equipment

Prosthetic fittings for the child with a fibular deficiency and subsequent ankle disarticulation should be successful. At the time of the child's first prosthetic fitting, there is still significant soft tissue surrounding the lower leg and ankle region. As the child grows, the definition around their ankle will become greater, resulting in a "bulbous-shaped" distal residuum. This is not as large as the typical Syme amputation because the lateral malleolus is not present. Therefore, using the ankle as a sole means of suspension is inappropriate (124). Often, a waist belt and fork strap will be fitted to the child for suspension of his or her prosthesis. This will permit unencumbered range of motion and provide the parents a means of keeping the prosthesis with the child. The child may still be crawling when he or she begins to wear the prosthesis. Because the foot cannot actively plantar flex during these activities, the prosthesis may drag on the floor and pull off the child's limb. As the toddler becomes more active, the waist belt and fork strap may be replaced by a suspension sleeve. This sleeve will serve the purpose of keeping the prosthesis on the child's limb, but should be pliable enough not to restrict range of motion during ambulation (48). As the child and limb matures, anatomical suspension can be utilized. Children with the Boyd amputation may be able to take advantage of this sooner, as the distal residuum becomes more bulbous sooner. The prosthetists should be able to take advantage of the distal residuum and eliminate the need for auxiliary suspension. Many methods have been used to accommodate donning of bulbous residuum into prostheses. The major challenge is permitting the larger, distal end to pass through a narrower portion of the socket that should provide total contact with the limb when it is fully seated.

Intervention for children with femoral abnormalities varies from shoe inserts to transfemoral (PFFD-style) prostheses. The child with a small femoral length discrepancy may need nothing more than a shoe insert or external shoe lift in order to equalize the length of the legs. This is only possible if hip flexion, abduction, and external rotation are addressed. Hips contracted in a flexed position often lead to a compensatory flexed knee. This posture is unstable in early stance phase of gait and may, therefore, require further intervention. Ankle foot orthoses (AFOs) that accommodate for the attitude of the leg and foot (usually equinus) can be used with shoe modifications. These orthoses may need to provide an external extension moment (ie, floor-reaction AFO design) if contractures have not been resolved.

As leg length discrepancy increases, the orthoses begin to morph into prostheses. The components used are no longer shoe lifts, but prosthetic feet inside regular shoes. The gait of the child can be asymmetrical because of different knee center heights. Transtibial prostheses have been used for some of these children with stable knees and knee centers that are higher

than the contralateral side. Benefits of this are better gait mechanics and control of external knee flexion movement in early stance by quadriceps versus hip extensors. A drawback of this type of fitting is that when this individual sits, the top of the affected knee will be much higher than the nonaffected knee because the tibia is longer than the femur. This is generally acceptable to the user and preferred to lengthening or amputation. In the event that the entire leg length, including foot or ankle disarticulation limb, is equal to or more proximal than the contralateral knee, a PFFD-style, transfemoral prosthesis is indicated. Capturing the proximal contours of the limb, with or without a foot present, and determining the appropriate height of the prosthesis are just two problems for the prosthetists. Attempts are made to fit ischial containment sockets to block the motion of the pelvis with respect to the femur, thus preventing subluxation of the femoral head. The difficulty with this is that with the soft tissue mass in the proximal thigh, the socket often is so high that it contacts the contralateral side perineum. If an articulation is going to be added to these longer limbs, many times, "outside hinges" are used in conjunction with an elastic extension assist for stability at initial contact and loading response. Single pivot, upper-extremity hinges are frequently used because of the size of the child at initial fitting. If there is room, a locking knee joint may be added initially to provide stability and can be unlocked during sitting. Frequently, polycentric knee joints are used for these children as they get older to address hip instability and control of the prosthesis, enable swing phase clearance because the linkages "shorten" the lower leg when the knee is flexed, and provide minimal femoral length discrepancy during sitting.

A myriad of prosthetic knees and feet can be used for these children, provided there is room for the components and that they are at an appropriate functional level to benefit from the components. Even with higher levels of involvement, children are frequently variable cadence ambulators and can take advantage of the high-technology components and components that can adapt to changes in speed and terrain (138,139). Figure 13.22 shows a foot with a shell that allows sandal wear.

When a knee disarticulation has been performed, as is typically the case with a complete absence of the tibia, prostheses similar to the "above-knee" prosthesis mentioned for the children with femoral absence exist. The knees and feet are used in a similar sequence and fashion. The main difference is the socket design. The child may have a relatively invasive socket at a young age to capture the limb and provide maximum stability. When the child matures, the socket will be trimmed much lower because the presence of all good hip musculature, including the hip adductors, will enable the child to control the prosthesis well and walk with only minor gait deviations.

Figure 13.22 College Park Industries TruPer Foot. Note that configuration of separated great toe on foot shell permits use of flip flops or sandals.

The child with a partial tibial deficiency will be fitted with a prosthesis that resembles a standard transtibial design. Once the tibiofibular synostosis has healed, the child can utilize most of the options of transtibial prostheses, including pin-locking liners and multiaxial dynamic response feet. The angular deformities that follow surgical reconstruction may prove challenging to the prosthetists in terms of the socket design. They may need to provide a means of donning the device that is atypical of transtibial designs and more like that of ankle disarticulations. A socket with a removable panel (door) may need to be created to permit the limb to successfully enter the socket. Closure is often provided for by straps or Velcro.

Frequently, congenital lower-extremity limb deficiency may present with odd combinations of absent portions of the extremity and deformities of the remaining segments. The deficiency may include proximal muscles, skin, nails, and parts of the joint. The child with bilateral PFFD, for instance, may also have upper-extremity limb deficiencies, which create challenges for donning/doffing clothes, prosthesis, and the use of prosthesis.

Treatments of varying levels of deficiency of acquired amputation are frequently individualized based on the level, number, and condition of the amputation(s).

Cases of traumatic limb loss will be treated in a fashion similar to adults, with the exception of potential growth and "overgrowth." Children with amputations secondary to sarcoma may be treated slightly different, as their limb volume will often fluctuate dramatically when they are undergoing chemotherapy and radiation therapy (55). Major concerns for fitting the child with a septicemic cause of amputation is the resulting condition of skin and bone (65). The child will most likely have experienced skin grafting procedures, and underlying bone will often progress at different rates than expected. These growth rates may be sporadic, delayed, or cause angulation deformities to occur (63).

Fitting Timetable

The lower limb–deficient child should be fit with a prosthesis when they are ready to pull up to a standing position (48). This usually occurs between 9 and 10 months of age. The goals in fitting a prosthesis at this early age are to allow for normal two-legged standing, provide a means for reciprocating gait development, and provide a normal appearance. The prosthesis should be simple in design, allow growth adjustment, suspend securely, and be lightweight. Historically, at an early age, the transfemoral prosthesis should not utilize a knee joint due to the complexity of operating a free knee, however, this philosophy is being reevaluated. Knee joints were usually added between 3 and 5 years of age, at times with a manual locking option (140). Knee units are can be added initially if an extension assist on the knee is utilized to help bring the knee into full extension prior to loading. Either an endoskeletal or an exoskeletal construction may be employed; each has advantages and disadvantages (138). Endoskeletal construction is good for growth consideration and is generally durable enough in most settings. The foam cover of the endoskeletal design requires more maintenance than an exoskeletal finish. The exoskeletal construction is robust and should be considered for those individuals who will test the limits of durability.

The child who acquires an amputation will be treated much the same as the congenital limb-deficient child, with a few exceptions. A child who undergoes an amputation will likely require a preparatory prosthesis while postoperative swelling subsides. The preparatory limb will probably be worn for approximately three months. In the case of the child who is undergoing chemotherapy treatment, it is useful to use a volume-adjustable socket.

Training

The preprosthetic period for the lower limb is mainly focused on addressing the information needs of the parents. In addition, an assessment of strength, coordination, joint range of motion, skin condition, and sensation should be performed.

Each child must be assessed as an individual, with consideration given to the child's age (both developmentally and chronologically), physical abilities, interests, and activities. The goal of physical therapy is to develop a normal pattern of gait, including stride length, step length, and velocity. The normal child does not establish heel-to-toe gait until about 2 years of age. At about 20 months, the normal child can stand on one foot with help; at 3 years, on one foot momentarily; at 4 years, for several seconds; and at 5 years, for longer periods. Toddlers tend to stand and ambulate with a wide-based gait, with their lower extremities externally rotated, abducted, and flexed. As their gait matures, these characteristics change to a more narrow-based, upright fashion (141). The prosthesis should incorporate these features in order to allow for normal gait development. Because the goal of physical therapy is symmetry of posture and movements during developmental activities, proper alignment, controlled weight shifting, and balance activities are emphasized for children with lower limb prostheses. Use of a polycentric knee unit (Fig. 13.23) allows a more normal cadence. Kinematic studies are demonstrating that co-contractions of the limb are reduced and may result in joint instability, so strengthening both agonists and antagonist muscles about the joint is important (142).

Functional goals for the child with bilateral lower-extremity amputations should be optimistic. Functional outcomes measured for the child with a lower-extremity prosthesis with the Pediatrics Outcomes Data Collection Instrument (PODCI) reflect excellent acceptance and use for both congenital and acquired amputations (143). As long as children have arms with which to balance, they should be expected to walk independently (144). Step-in-place training is appropriate pregait training for children (139). Weight control is a concern for the child with lower-extremity amputations. Dietary instruction should be emphasized early and often. Gait analysis has been performed on adults with amputations. Crutch walking, with or without a prosthesis, increases energy expenditure during gait. In groups of traumatic amputations, the oxygen cost progressively increases with each higher-level amputation. Amputees preserve their energy expenditure by decreasing their chosen

Figure 13.23 Polycentric knee units from Seattle Limb Systems and Hosmer-Fillauer.

walking speed. Children's effort levels have been reported for transtibial amputations between crutch walking, SACH foot, and the Flex-Foot. Chosen walking speed was higher for the children using the Flex-Foot, approaching normal. This study only involved five children, so statistical significance could not be determined. A slightly higher oxygen consumption occurred for children using SACH feet (115). The Carbon Copy II prosthetic foot and Seattle Foot are energy-saving designs that permit the athlete a more natural gait. The energy-storing feet are available for children (Fig.13.24).

Training Following Amputations

Following surgery, the remaining leg must assume the dominant role in all transfer and locomotor activities. Therefore, the sound leg should be evaluated for strength and, if necessary, an appropriate exercise program developed. It is difficult to instruct the young active child in specific exercises and positioning due to limited comprehension and attention span. If specific exercises are indicated, a therapist often needs to be creative with games and use of equipment to get the desired responses, such as using a prone scooter to maintain or work on hip extension (90). Edema control is accomplished using one of several options, which include Ace bandage wrapping, elastic shrinker socks, layers of elastic stockinettes, rigid dressing, and removable rigid dressing. It is important that the parent and child understand the proper technique for use of the edema control system. The edema control system should be worn 24 hours a day, only being removed for wound care and hygiene (145).

To avoid increasing the patient's anxiety level, the therapist should not dwell on phantom pain, but the patient should be made aware of the normal postoperative discomfort that is to be expected. The adaptation to prosthetic ambulation is dependent on the fit and comfort of the residual limb and socket/suspension. The therapist, working closely with prosthetist at this point, can identify the fixable and ensure continued use for the child to gain confidence and competency.

Figure 13.24 Ossur Modular Flex-Foot.

Play is the primary motivation for desired movements and activities. Parents should be instructed on how to care for the prosthesis and encouraged to maintain contact with the prosthetist for routine adjustments and follow-up. Often, the first sign that an adjustment is needed is noted when the child reduces wearing time or begins to limp.

Adolescents widen their sphere of mobility to include the community by using public transportation or by driving. The site of the amputation or limb loss will determine the degree of difficulty an amputee will have driving standard vehicles. In most cases, the person with a partial or full amputation of a limb will require adaptive driving equipment to compensate for the loss of ability to reach and operate driving controls. Most amputees are able to independently get into and out of a standard-size sedan. Current driving aids are available for the driver who has normal strength and mobility of upper extremities. Control systems used include push-pull control, push-right angle pull control, and push twist. Each has the acceleration and braking system connected to usable upper-extremity function. State licensure for driving and installation of equipment varies. Physicians should be aware of their responsibility in certifying the capabilities of a potential driver. The evaluation for driving potential as well as specific equipment modifications should be discussed and made available for the individuals with multilimb and complex limb deficiencies.

For children with amputations secondary to tumors, return to school may be difficult. In a study concerning the adjustment post-tumor amputation, 67% could not keep up in their classes (60,136). In addition to direct intervention for psychological support, many family support systems are available to the families and children with limb deficiency. Many clinics provide opportunity for the interaction and peer support of their population. Frequently, the parent-to-parent or child-to-child interactions surpass the effect of professional input for education, information, and resources (105,146). Resource guides are available and provide pragmatic information for the child and parents (106,107).

Children with complex limb deficiency, such as tetraphocomelia, benefit from the early introduction of power mobility. Movement provides a sense of independence and competence derived from exploring one's environment. When exploration is restricted, there is a diffuse and long-lasting impact. Motorized wheelchairs traditionally have been used when a child

is 5 to 6 years of age. Innovative seating systems have been developed for the 1- to 3-year-old child. Salient features include the following:

- A powered device
- Proportional control drive with an adjustable joystick used with the head, chin, or lower or upper-extremity buds
- Adjustable positioning seating in an upright frame into which inserts can be attached for growth
- Compactness, durability, portability, reliability, and safety
- Low profile with mounting potential for children to interact on a peer level

In addition to power mobility, other adapted mobility devices are available that are child- and environment-friendly.

Advancements in Lower-Extremity Prosthetics

The most dramatic changes in lower-extremity prosthetics is with regard to the components that are available for children. For years, there were few options to choose from for the child. SACH feet and friction knees were standard components used. Increased awareness on the part of the parents of children with limb deficiencies and amputations, along with pressure from the rehabilitation community, has influenced the manufacturers to recognize the need for improvements in this small, yet lucrative market. Children test the limits of many components by competing in recreational activities that range from neighborhood skateboarding to extremely competitive sports against their "able-bodied" peers. Children have benefited from the influx of smaller components that provide responsiveness and control to variable cadences while also providing compensations for variable terrain.

Professionals who are involved with the population of children with loss of limb quickly appreciate the possibilities that exist for an individual to compensate and to accomplish as much as anyone. Improved materials, technology, and greater availability of resources contribute to versatile prosthetic options. Involvement of a child and family with a comprehensive amputee clinic team provides therapeutic choices throughout the child's life. Close collaboration of physicians, family, and all professionals is essential for a cohesive and practical rehabilitation program. As in all pediatric conditions, the process of decision making, treatment options, and delivery of care is variable and should be discussed with the child and family. The moving target is always the growing, developing child, while our aim should be a healthy, happy, well-functioning child, adolescent, and ultimately adult.

PEARLS AND PERILS

Upper Extremities

1. The younger the child is at the time of amputation, the easier the transformation of hand dominance.
2. Children with high-level, bilateral upper-limb loss may benefit from a prosthesis for ADLs. Since limited body movements are available, the child may benefit from at least one hybrid or completely externally powered prosthesis. Prior to the consideration of prosthetic fitting, it is paramount that the child and family begin exploring the use of the child's lower limbs, as independence can also be achieved with feet.
3. At birth, the severely deformed upper extremity often detracts from the identification of more important systemic workup. Although there may not be any other underlying etiologies or comorbidities, it is essential for the clinic team to explore these possibilities.
4. It is important to teach parents about the loss of surface area corresponding to the absent limb. Active children with multiple limb loss have a reduced surface to radiate heat loss, so they may have an increase in sweating and flushing about the head and neck.

Lower Extremities

1. Children tend to do well with lower-limb prostheses, often requiring little or no formal gait training.
2. Limb-volume changes occur following amputation and can be controlled by rigid dressings in the postoperative period (145). Although the postoperative edema is not as great as that for the adult dysvascular patient, children will benefit from these rigid dressings for the control of edema as well as to initiate earlier ambulation and prosthetic fitting. A rigid removable dressing is illustrated in Figure 13.25. This is particularly important for children who have had a remnant or dysfunctional limb segment for which they have some psychological attachment. Early ambulation may serve as a distraction to the surgery and provide a new focus on skills of ambulation.
3. When fitting an ankle disarticulation prosthesis, the prosthetists should strive to create a prosthesis that permits near full weight bearing on the distal end of the child's residuum. This will ensure that the

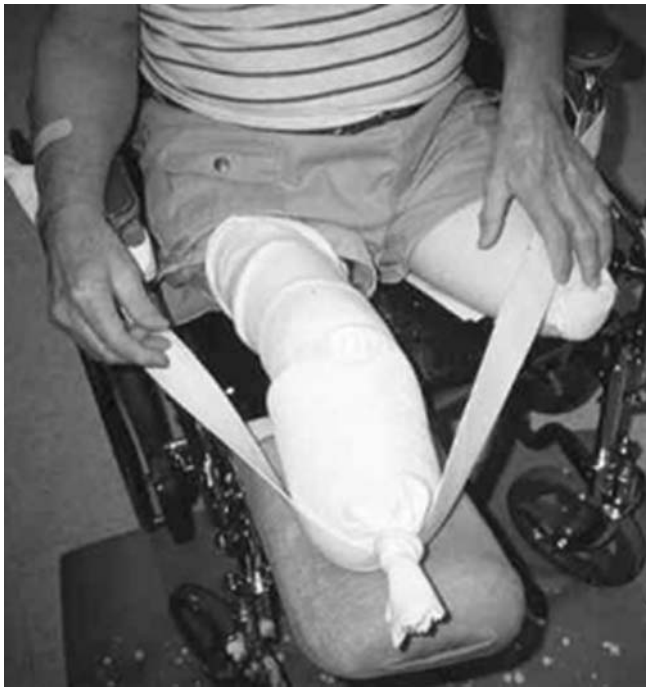


Figure 13.25 Individual with transtibial amputation and removable, rigid dressing. Note strap used to maintain compression on limb.

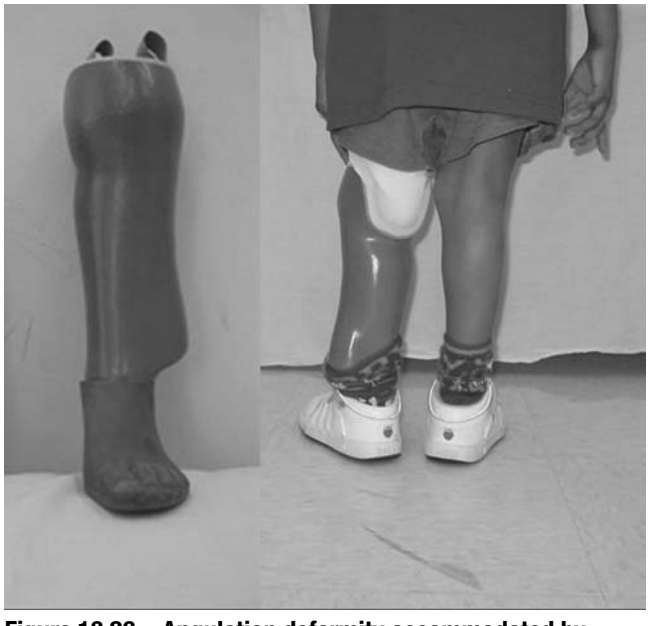


Figure 13.26 Angulation deformity accommodated by prothesis.

distal end of the residuum and the heel pad remain toughened. An end-bearing residuum of this length is beneficial for limited ambulation without the prosthesis (eg, using the bathroom in the middle of the night), as well as long-term fitting.

4. The surface area of the distal residuum will remain fairly consistent throughout the child's life; therefore, the child is encouraged to maintain a reasonable weight, so as not to lose the ability for distal end bearing.
5. Prosthetic fittings may be affected by angular deformities as the alignment of the device must be biomechanically appropriate and not necessarily the most cosmetic. Most of the angulation deformities can be accommodated in a prosthesis; however, it is not possible to provide a device that is advantageous to appropriate gait mechanics and satisfies the cosmetic expectations. The accommodation of angular deformity is illustrated in Figure 13.26.

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14

Orthopedics and Musculoskeletal Conditions

Kevin P. Murphy, Colleen A. Wunderlich, Elaine L. Pico, Sherilyn Whateley Driscoll, Elizabeth Moberg-Wolff, Melanie Rak, and Maureen R. Nelson

GROWTH AND DEVELOPMENT OF THE BONY SKELETON

The skeletal system develops from mesoderm and neural crest cells (1). Somites form from paraxial mesoderm and differentiate into sclerotomes, dermatomes, and myotomes. Sclerotome cells migrate from the somite and ultimately become chondrocytes. Remaining dermatome cells form the dermis. Myotome cells give rise to striated muscles of the backs of limbs (Fig. 14.1).

Limbs and respective girdles, the appendicular skeleton, are derived from cells of the lateral plate mesoderm. Limb buds appear in utero approximately day 26 for the upper extremities and day 28 for the lower extremities (2). The hand plate forms in the fifth week, with digitization of rays in the sixth week. Notches appear between the rays in the seventh week, failure of which results in syndactylism. During the seventh week, the limbs also rotate laterally in the uppers and medially in the lowers. This brings the thumb to the more lateral position in the upper extremity and the great toe to the more medial position in the lower extremity. Chondrification begins in the sixth week, followed by early ossification in the seventh week and subsequent joint cavity formation in the sixteenth week. By the eighth week, definite muscle formation is noted, as the embryo assumes a human appearance and basic organ systems are completed. The fetal period begins at nine weeks with rapid growth and changes in body proportion (3, 4).

Knowledge of the normal proportions and growth and development of the musculoskeletal system allows a firm foundation for the understanding of both congenital and acquired conditions requiring care in the developmental years.

Figure 14.2 displays the growth rates for boys and girls by age. About half of the individual's height is reached by age 2 and three-fourths by age 9. Prediction of adult height can be obtained by plotting bone age against current height to determine percentile value (Fig. 14.3). Following the percentile to skeletal maturation estimates final adult height. Paley height multipliers offer an even simpler way of estimating adult height at any child age (5). Predictions are less accurate for the younger child (Fig. 14.4). The reader is referred to more detailed references for tables displaying differences over time and growth and rate for standing, sitting and subischial lengths in boys and girls (6). The measurement of arm span provides an indirect control parameter for the measurement of standing height, particularly useful in those who are

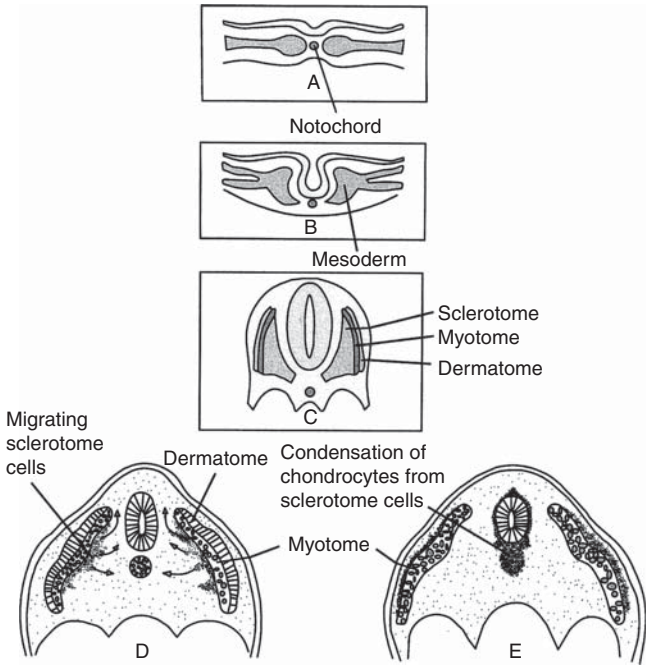


Figure 14.1 Trilaminar disc. Neural tube closure. Mesoderm differentiates into dermatome, myotome, and sclerotome. Migrating sclerotome cells become chondrocytes. Chondrocytes ultimately form vertebral bodies and arches.

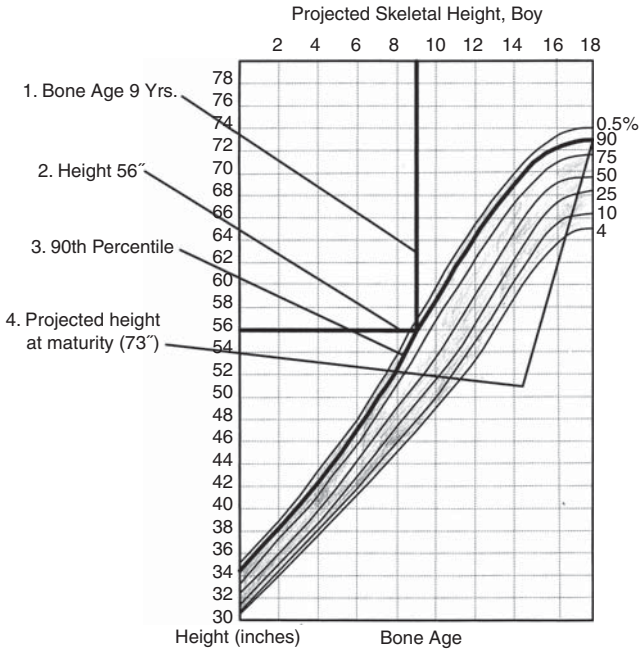


Figure 14.3 Prediction of adult height. Adult height predicted by plotting child's bone age versus current height to determine percentile value. Follow the percentile to skeletal maturation for estimate of final adult height.

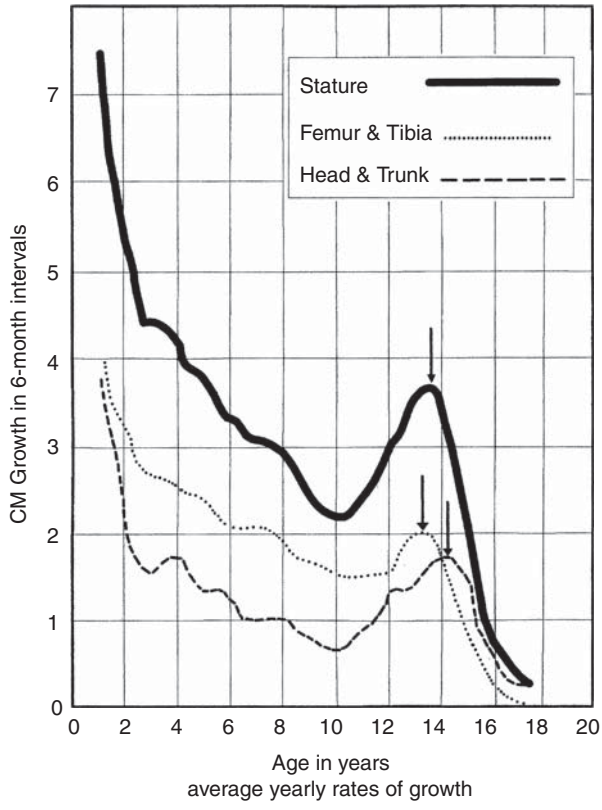


Figure 14.2 Greene and Anderson growth curve. (From Greene W, Anderson M: Skeletal age and the control of bone growth. *Instr Lect Am Acad Orthop Surg*. 1960;17: 199–217.)

nonambulatory. To measure arm span, the patient simply raises the arms to a horizontal position, and the distance between the tips of the middle fingers is measured with a tape measure (7,8). Standing height is about 97% of arm span. In children with spinal deformity, arm span is a good estimate of what standing height would be if there were no abnormal curvatures. It is well known that different proportions of the body grow and change at different percentages over the developmental years (Fig. 14.5).

CONGENITAL CONDITIONS

Minor limb deficiencies are relatively common in the upper and lower extremities. Syndactyly occurs in approximately 1 in 2,200 births, either as cutaneous with simple webbing of the fingers or osseous with fusion of the bones when the digital rays fail to separate between the fifth to eighth weeks of gestation (9). It is most frequent between the third and fourth fingers and between the second and third toes, and is inherited as a simple dominant or simple recessive trait. It can occur in isolation or as part of a syndromic condition. Surgical separation of the digits is more common with complete syndactyly for functional and cosmetic reasons. Polydactyly has an incidence of approximately 1 to 1.5 per 1,000 live births and is the most common congenital toe deformity (10,11). Eighty percent

Height Multiplier GIRLS Birth to 18 Years

Age (yr + mo)	*M*	*Age (yr + mo)*	*M*
Birth	3.290	8 + 6	1.254
0 + 3	2.759	9 + 0	1.229
0 + 6	2.505	9 + 6	1.207
0 + 9	2.341	10 + 0	1.183
1 + 0	2.216	10 + 6	1.160
1 + 3	2.120	11 + 0	1.135
1 + 6	2.038	11 + 6	1.108
1 + 9	1.965	12 + 0	1.082
2 + 0	1.917	12 + 6	1.059
2 + 6	1.815	13 + 0	1.040
3 + 0	1.735	13 + 6	1.027
3 + 6	1.677	14 + 0	1.019
4 + 0	1.622	14 + 6	1.013
4 + 6	1.570	15 + 0	1.008
5 + 0	1.514	15 + 6	1.009
5 + 6	1.467	16 + 0	1.004
6 + 0	1.421	16 + 6	1.004
6 + 6	1.381	17 + 0	1.002
7 + 0	1.341	17 + 6	–
7 + 6	1.309	18 + 0	–
8 + 0	1.279	Jonathan Paley et al., JPO 2004	

Height Multiplier BOYS Birth to 18 Years

Age (yr + mo)	*M*	*Age (yr + mo)*	*M*
Birth	3.535	8 + 6	1.351
0 + 3	2.908	9 + 0	1.322
0 + 6	2.639	9 + 6	1.298
0 + 9	2.462	10 + 0	1.278
1 + 0	2.337	10 + 6	1.260
1 + 3	2.239	11 + 0	1.235
1 + 6	2.160	11 + 6	1.210
1 + 9	2.088	12 + 0	1.186
2 + 0	2.045	12 + 6	1.161
2 + 6	1.942	13 + 0	1.135
3 + 0	1.859	13 + 6	1.106
3 + 6	1.783	14 + 0	1.081
4 + 0	1.731	14 + 6	1.056
4 + 6	1.675	15 + 0	1.044
5 + 0	1.627	15 + 6	1.030
5 + 6	1.579	16 + 0	1.021
6 + 0	1.535	16 + 6	1.014
6 + 6	1.492	17 + 0	1.010
7 + 0	1.455	17 + 6	1.006
7 + 6	1.416	18 + 0	1.005
8 + 0	1.383	Mature Height = Ht x M	

Figure 14.4 Paley height multipliers. Charts provide a simple method of predicting adult height for boys and girls.

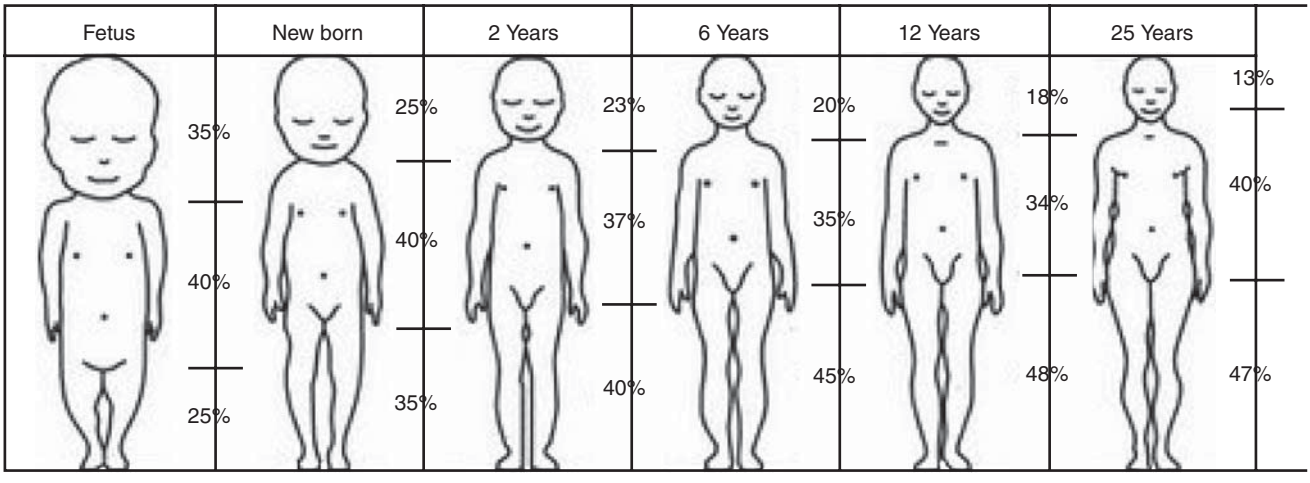


Figure 14.5 Proportions of the body as they change during growth. (Redrawn from Lowrey GH. *Growth and development of children*. 6th ed. Chicago, IL: MYB;1973.)

of polydactyly in the foot occurs with the fifth toe. Most often an isolated trait, an autosomal-dominant inheritance pattern has been identified with variable expressivity. Radiographic evaluation is necessary to define duplicated structures. Deferring radiography until after 6 months of age allows phalanges to ossify. Surgery around the age of 1 not only improves cosmesis, but also is helpful in facilitating shoe fitting.

Camptodactyly, translated from Greek, means "bent finger." The proximal interphalangeal (PIP) joint is flexed, most commonly digit 5. Incidence is felt to be less than 1% of the general population with equal gender distribution (12). Appearance in adolescence, often girls, is less common. Surgical reconstructions are for functional and cosmetic reasons.

Malformations of the radius are more common than those of the ulna and are associated with numerous syndromes (6,13). In children with limb anomalies, a multisystemic review is generally indicated because abnormalities in other systems are often present. Simple and multifactorial inheritance may all be causative in addition to teratogenic effects, such as maternal exposure to viral infections and chemical dependency such as alcohol (10). A failure of the scapular to descend from its cervical region overlying the first through fifth ribs results in Sprengel's deformity (14–17). Children often present with a shortened neckline. Lack of normal scapulothoracic motion and malpositioning of the glenoid causes limited forward flexion and abduction of the shoulder. An omovertebral bar is present in up to 50% of cases (25). The bar connects between the supermedial angle of the scapula and the cervical spine, and consists of fibrous cartilaginous tissue or bone. It is not uncommon to see other abnormal regional anatomy and syndromes that need to be screened for carefully, including scoliosis, spina bifida, rib anomalies, and Klippel-Feil syndrome (26). Renal and pulmonary disorders can also be present, and a renal ultrasound, if not already completed, is indicated. The condition can be bilateral in up to 30%.

Congenital radioulnar synostosis is a rare condition caused by failure of the radius and ulna to separate, usually proximal. The forearm is usually left in significant pronation with the condition bilateral 80% of the time (18). This condition is also associated with multiple other syndromes, which need to be carefully screened for (10). Children present for evaluation depending upon degree of functional deficit. Radiographs can be helpful when ossification is present. Magnetic resonance imaging (MRI) scans of the proximal radius and ulna can reveal more of a cartilaginous synostosis or a fibrous tether that has not ossified. Children with radioulnar synostosis without functional limitation should be observed. Surgical success to resect the synostosis is often limited with minimal functional gain (6). Rotational osteotomies for pronation deformities greater than 45 degrees can be helpful. Postoperative compartment syndrome of the forearm needs to be watched for (19).

Congenital dislocation of the radial head unaccompanied by other congenital abnormalities of the elbow or forearm is rare (6,20). Acquired dislocations account for six to eight percent of elbow injuries (21, 22). They are most frequent in children under the age of 10 (11). Typically, the injury involves the nondominant extremity with a fall onto the outstretched hand (23). Nursemaids elbow consists of a radial head subluxation from a sharp upward pull on the extended pronated arm in preschoolers. A generalized ligamentous laxity in children with large cartilaginous components of the distal humerus and proximal ulna, in addition to osseous instability with numerous secondary ossification centers and epiphyses, all contribute to the tendency for the pediatric elbow to dislocate. Posterior or posterolateral dislocations account for 80% to 90% of the injuries (11). Closed reduction with the patient under sedation is the treatment of choice. Longitudinal traction and flexion with supination will reduce the uncomplicated dislocation. Ulnar and median nerve entrapment needs to be ruled out clinically postreduction.

In about 5% of humans, there are minor variations in the number or proportions of vertebra (24). Osseous anomalies are felt to account for up to 6% of children who present with signs of torticollis. Individuals with cervical fusion are generally apparent on plain cervical radiographs, including flexion and extension views. The Klippel-Feil syndrome, sometimes called brevicollis, is characterized by short neck, low hairline, and restricted neck movement (25). Consisting of congenital fusions of the cervical vertebra, its incidence is approximately 0.7% (26). Failure of segmentation in the cervical spine most often characterizes the Klippel-Feil syndrome. Patients with Klippel-Feil syndrome or related conditions should have a renal ultrasound and cardiac evaluation (echocardiogram). Contact sports are contraindicated, as are similar, more aggressive activities.

Intraspinal anomalies need to be considered, especially in the presence of hairy patches, dimples, nevi tumors, or asymmetric or absent abdominal reflexes. In children with Down's syndrome, atlantoaxial instability may be identified in up to 13% (27,28), but only 1% to 2% has symptomatic instability that requires surgery. X-ray examination of the cervical spine in children with Down's syndrome should be obtained at about the age of 3 years and before such children enter competitive sports such as the Special Olympics. Repeat x-rays should be taken after the cervical spine has been completely formed, at around the age of 8 years and every decade thereafter across the lifespan,

as recommended by the American Association of Down Syndrome. The atlantodens interval (ADI) should be no greater than 4 mm in children 7 years of age and younger and no greater than 3 mm for children 8 years and older (11). ADI up to 5 mm has been accepted in the more traditional sense (29).

Clubfoot, talipes equinovarus, is a common term used to describe several kinds of ankle or foot deformities present at birth. The foot is generally in equinous, with forefoot and hindfoot varus and severe adduction (Fig. 14.6). As the most common birth defect, it carries an incidence ranging from 1:250 to 1:1,000 live births, depending on the population (11). The condition is one of the most treatable of birth defects, often leading to normal or near-normal athletic activities later in life (6). Multifactorial genetic inheritance, along with poorly understood environmental factors, may explain the bulk of etiology. Some clubfoot disorders are transient or apparent in nature and result simply from intrauterine crowding. Other conditions may occur in association with myelodysplasia, arthrogryposis, and particularly hip dislocation. Prenatal ultrasound can be effective in diagnosing intrauterine clubfoot, with no false-negative prediction and a true positive predictor rate of 83% (11). Recent treatment has focused primarily on the Ponseti technique (30,31,32). Range of motion should be maintained by passive exercise and therapeutic play, particularly into dorsiflexion and eversion. Persistent deformity into adulthood can result in unstable ankles, lateral sprains, and difficulty with weight bearing and other gross mobility tasks.

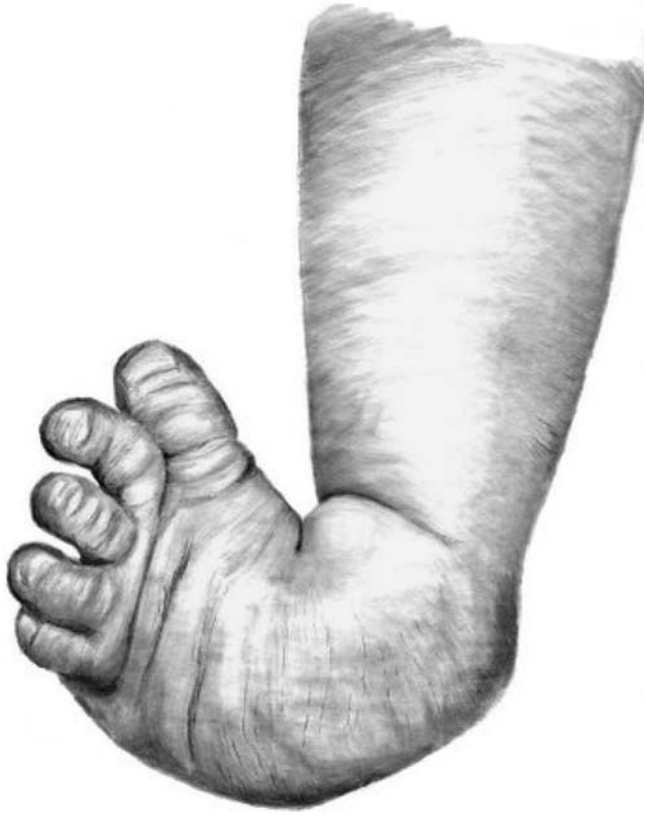


Figure 14.6 Club foot deformity. Associated forefoot supination, deep medial crease, and equinovarus of the hindfoot.

Metatarsus adductus can be seen in up to 12% of full-term births (11). Intrauterine crowding or positioning may be causative. Flexibility can be determined by fixing the hindfoot in a neutral position and gently manipulating the midfoot and forefoot to a more lateral position. Internal tibial torsion may be associated, making the thigh–foot angle worse. Serial casting may be helpful in children under 1 year of age. Careful attention should be given not to place the hindfoot in valgus or create a skew foot deformity. Surgery is rarely indicated, but can be done in the more rigid persistent deformities after the age of 5. Various forms of posterior medial release are available (6).

Flat feet or pes planus may be flexible or rigid (6). Flexible pes planus is usually asymptomatic, at least in the early years, and is the most common type found in children. Inexpensive scaphoid pads or medial inserts may help to create more plantigrade weight bearing in the child, but they do not correct the deformity. Extreme cases, such as in children with hypotonia, may require surgery after the age of 5 years in the form of a calcaneal lengthening once the bony cortices are more solid. Untreated progression may occur with compensatory hallux valgus, planovalgus, and secondary bunion and toe deformities. Pes planovalgus is associated with more active or shortened peroneal musculature, progressing over time, with the development of pain, particularly in later years. Rigid pes planus is a congenital deformity associated with other anomalies in 50% of cases (33). It is caused by failure of the tarsal bones to separate leaving a bony cartilaginous or fibrous bridge or coalition between two or more tarsal bones (34). Talocalcaneal coalitions tend to become symptomatic earlier, between 8 to 12 years, whereas calcanonavicular coalitions are more likely to be symptomatic between 12 and 16 years. Symptoms are insidious with occasional acute arch, ankle, and midfoot pain. The hindfoot often does not align in its normal varus position on tiptoe maneuvers (6). Patients are predisposed to ankle sprain secondary to the limited subtalar motion, and stress to the subtalar and transverse tarsal joints frequently causes pain. Computed tomography (CT) scans are diagnostic, and initial treatment is conservative with short-leg casting or molded orthosis and rest. If conservative care fails, surgical intervention is usually necessary. With all symptomatic pes planus, accessory navicular bones need to be considered (11). Rigid cavous feet may be associated with metatarsalgia, clawing, and intrinsic muscle atrophy. With a cavus foot, stresses are

increased across the joints, along with pressures on bony prominences and muscle strength being required to maintain posture. The result is pain, fatigue, and instability. The cavus foot may be caused by an underlying neurologic condition such as Charcot-Marie-Tooth disease, spinal dysraphism, Freidrich's ataxia, or spinal tumor. Custom molded inserts or orthosis may be helpful in providing arch support and decreased pain by relieving pressure off bony prominences and providing shock absorber effect. Cavous feet can often run in families, making family history critical. Clinical exam for flexibility with localization of the deformity to the forefoot or hindfoot should be completed. The Coleman block test for determination of hindfoot flexibility can be critical, particularly for any surgical repair in the more rigid and symptomatic deformity (35). Plantarfascia release is standard for all cavous foot procedures (36).

Congenital vertical talus is exceedingly rare (6). The navicular bone is dislocated dorsolaterally on the head of the talus. It is commonly associated with neuromuscular and genetic disorders, including trisomy 13, 14, 15, and 18 (37). Clinical features include a rigid convex plantar surface (rocker bottom) with hindfoot equinus and hypoplastic laterally deviated forefoot. Casting can initially have some benefit for contracted dorsolateral soft tissues, but only as a prelude to surgical intervention. A single-stage procedure is generally the consensus (6) and can involve talectomy, naviculectomy, subtalar arthrodesis, and triple arthrodesis.

Arthrogryposis multiplex congenita refers to a symptom complex characterized by multiple joint contractures that are present at birth. The clinical literature has delineated as many as 150 entities under this term (11,38). The incidence of arthrogryposis as a whole is approximately 1 per 3,000 live births. Amyeloplasia (which literally means no muscle growth) affecting all four limbs is less common, at approximately 1 in 10,000 live births (11). There are many different ways to divide up the arthrogrypotic conditions (6,39). A simple way is to divide the contracture syndromes into three different groups (6). Group number one involves arthrogryposis multiplex congenita, Larsen syndrome, and more or less total body involvement. Larson syndrome is a rare condition involving multiple congenital dislocations of large joints, a flat facies, and significant ligamentous laxity (40). Patients commonly have abnormal cervical spine segmentation with instability and can be associated with myelopathy. Group number two would include the distal arthrogryposis predominantly involving hands and feet. Distal arthrogryposis type II involves the presence of facial findings, whereas type I does not. Freeman-Sheldon syndrome is an example of a distal arthrogryposis type II, with a characteristic "whistling face" appearance (41). Group number three involves the pterygia syndromes. Pterygium comes from the Greek word meaning "little wing." Pterygiums can be isolated or multiple. Multiple pterygius syndrome is characterized by webbing across every flexion crease in the extremities, most prominently across the popliteal space, elbow, and axilla (42). Popliteal pterygium syndrome has features involving the face, genitals, and knees (43). A popliteal web is usually present bilaterally running from the ischium to calcaneus, resulting in severe knee flexion deformities. The diagnosis of arthrogryposis can be suspected with prenatal ultrasound. Absence of fetal movements of distal or proximal joints in combination with polyhydramnios is suggestive (44). The birthing process can be complicated by joint contractures, with neonatal fractures resulting. Perinatal fractures are common and believed to be secondary to hypotonia and rigid joints (45). Therapy should not be initiated in a newborn until such fractures are ruled out (46). Children who survive infantile arthrogryposis often have upper and lower extremity involvement in typical patterns. Common deformities of the upper extremities include adduction; internal rotation contractures of the shoulders; fixed flexion or extension contractures of the elbows, either wrist flexion and ulnar deviation or extension and radial deviation; and thumb-in-palm deformities. In the lower extremities, flexion, abduction, and external hip rotation contractures with unilateral or bilateral dislocations are noted. Bilateral dislocations of the hip are more often left alone, whereas unilateral dislocations, because of scoliosis risk, are more often surgically treated (6). Fixed extension or flexion contractures of the knees are also seen along with severe rigid bilateral clubfeet. In the most severe rigid clubfeet, not correctable with casting and conservative care, talectomy may necessary or talar enucleation in association with the posterior medial releases. Extension wedge osteotomies of the distal femur may be necessary to correct flexion contractures of the knee. There is always a well-recognized risk of neurovascular damage, with operative correction of knee flexion contractures needing careful consideration to avoid overstretching of the neurovascular bundle. Shortening osteotomies completed at the same time as the extension wedge osteotomy may minimize these risks. In the absence of degenerative neurologic conditions, individuals with arthrogryposis maintain their strength and range of motion over time (6). Surgical and rehabilitation goals are generally centered on self-help skills, such as feeding, toileting, and mobility skills such as standing, walking, and transfers using assistive devices as needed. Surgical procedures of the upper extremity are usually delayed until the child is old enough for a more definitive functional assessment to be completed. If both elbows are involved with extension, surgery to increase flexion may be best done on only one side.

Outcomes appear better if joint surgery is completed prior to the age of 6 to avoid adaptive intra-articular changes (11). Osteotomies for realignment are usually performed closer to skeletal maturity. Early mobility and avoidance of prolonged casting may result in improved range of motion and function postsurgery. Most individuals do not have intellectual impairment or sensory deficits. The children often have a keen natural ability to learn substitution techniques. A strong association between initial feeding difficulties and subsequent language development is known, which should not be misidentified as intellectual deficiency (47).

BRACHIAL PLEXUS PALSY

Birth brachial plexus injury occurs in between 1 and 2 per 1,000 live births in the United States. Babies with increased birth weights, multiparous mothers, and shoulder dystocia are at the highest risk for brachial plexus palsy (48,49). The most widely described mechanism of action for this is lateral stretch, which is logical secondary to the location of the brachial plexus, the high correlation with shoulder dystocia, and the positioning of the mother and infant (49). It has been described that between 50% and 95% of these infants will recover spontaneously. The goal of treatment of brachial plexus injuries is maximizing arm and hand function. Goals are normalization of limb function, with optimization of nerve regeneration and mechanical increase of elbow flexion and shoulder stabilization. This can be achieved through aggressive rehabilitation and microsurgical intervention (48).

For any nerve that is injured, classification makes evaluation and comparison clearer. The Seddon Classification of Nerve Injury is commonly used. Neurapraxia occurs with no lasting anatomical changes, with fibers preserved. This is exemplified by a football "stinger" injury. Complete resolution is expected. In axonotmesis, there is an interruption of neural continuity to some degree. There is an extremely variable level of deficit that is difficult to evaluate and predict the degree of recovery. Neurotmesis is the most severe injury, with total disruption of the elements of the nerve, and this will not recover. If it is preganglionic, or proximal to the dorsal root ganglion, it is called an avulsion. If it is postganglionic, or distal to the dorsal root ganglion, it is called a rupture (50). Both of these require surgical intervention for recovery.

There are also descriptors for the levels of brachial plexus palsy. Injury at C5–C6 is called Erb's palsy, sometimes called Erb-Duchenne palsy. This is the most common level of involvement, present in approximately three-fourths of those with birth brachial plexus palsy. Involvement of C8–T1 is Klumpke's palsy. It is debated whether Klumpke's can occur in a birth brachial plexus injury, though it definitely occurs in other types of brachial plexus injury. The reason for this question is whether it is anatomically possible to have a C8–T1 lesion alone without involvement of C5–C7. It appears that if there is an anatomical variation—for example, a rib, tendon, bony, or other anomaly that leads to C8–T1 compromise—this can occur in a birth brachial plexus injury. Otherwise, it appears that it cannot. Therefore, if a child presents with a C8–T1 birth brachial plexus injury, it may be from anatomical anomaly, but there are two other options to consider. Most likely, it was initially a complete brachial plexus involvement but there was quick recovery of C5–C7. This is likely, since the upper cervical root levels are relatively protected anatomically so C8 and T1 may end up with the most severe injury. It is also possible that a spinal cord injury has been mistaken for brachial plexus palsy. All of these are important to consider during evaluation. There also may be a complete brachial plexus palsy, including C5–T1, with total motor and sensory loss. There also can be a variety of levels involved between upper plexus and total plexus palsy.

Evaluation

Evaluation of patients with brachial plexus palsy includes clinical findings, electrodiagnosis, and MRI. There is debate about which of these is most effective. MRI is expensive and requires sedation to perform on infants. It has been found to correlate with surgical findings 70% of the time, electromyography 87% of the time, and clinical findings 60% of the time. The correlation was highest when all three of these were combined. MRI was effective only in those with C5, C6 root involvement (51).

Clinical exam consists of a history and physical examination. The history includes the birth number of the child, the birth weight, and presence of maternal diabetes during the pregnancy, along with the size of previous infants and the birth size of the parents. The motor and sensory findings at birth, along with any change up to the time of evaluation, are important. The use of vacuum or forceps may be indicative of any difficulty with delivery. The most common association is shoulder dystocia. Other useful information is whether there were signs of bruising or other injury, or whether there was involvement of the contralateral arm or the legs at delivery.

Physical examination begins with visualization of the arm to include the size and bulk. A cool temperature and blue color are sometimes noted. Sensory evaluation is critical to determine areas of involvement. Muscle stretch reflexes will be decreased or absent in the distribution of a brachial plexus injury.

The primitive reflexes are also important. Since the upper plexus has more frequent involvement, the Moro reflex, which shows shoulder abduction and elbow flexion, is valuable in assessing those active movements. Torticollis is frequently seen, and usually this is with the face turned away from the involved arm. Range of motion is an important part of the evaluation since contractures are commonly seen in shoulder adduction and internal rotation, wrist flexion, forearm pronation, and even at the elbow into flexion commonly in later months and years.

A key goal of the electrodiagnostic evaluation is to find subclinical nerve and muscle responses. The study must be individualized, with studies performed that are pertinent to each individual's examination. Sensory nerve conduction studies, motor nerve conduction studies, and electromyography are performed. Diagnostic evaluation should include nontraditional nerve conduction studies, and commonly not the classic median and ulnar nerves, due to frequent involvement of only the upper brachial plexus. Axillary, musculocutaneous, and radial nerves are among those useful for electrodiagnostic study. Sensory nerve action potentials (SNAPs) are important, as these are most sensitive to axonal loss (52). The presence of SNAP responses in an insensate area is indicative of a preganglionic lesion, due to the location of the sensory cell bodies in the dorsal root ganglion. Electromyography (EMG) may show activation of motor unit potentials in muscles with no clinical motor activity. Electromyographic evaluation is reported of being of some benefit, but underestimating the severity of lesions (53). It has been recommended to be performed early in the first few days, then with a repeat evaluation after several months to more accurately identify cases where there is reinervation occurring and therefore having earlier determination of the need for surgical intervention (54).

Plain x-rays may be useful. Other abnormalities may mimic a brachial plexus palsy, including a fracture of the clavicle or humerus. Osteomyelitis may also mimic this, and has actually been reported as inciting temporary brachial plexus palsy (55). Neurofibromatosis or other tumors may also cause it.

Treatment

Education is initiated when a family is first seen. Therapy should be started as soon as possible after diagnosis. Positioning instruction begins immediately, and range-of-motion exercises are generally initiated after two weeks. The wait is due to the fact that there is commonly noted to be pain with changing position of the shoulder for bathing or dressing in the first two weeks, so it appears that there is some tenderness after the initial brachial plexus injury, which is quickly resolved. It is also important to position the arm so that the baby will have maximal awareness of it. One way to accomplish this is with the use of a wrist rattle on the affected arm so that the baby's attention can be drawn to that arm by sound or vision, because the weakness of that arm usually limits it from being moved in front of the face spontaneously. It is also recommended to have the family replicate movements with the affected arm that the baby spontaneously does with the unaffected arm, such as bringing the hand to the mouth. It is important that the family realize that they need to perform the exercise program several times a day. It is also important not to have such aggressive range of motion in shoulder abduction or forearm supination that there is dislocation of the humeral head or radial head, respectively. Splinting is also commonly done by occupational therapy or physical therapy. Initially, there is frequently wrist drop, so splints may be made to provide optimal position of the wrist and fingers. Later on frequently there is an elbow contracture, so splinting is done to minimize that. Therapists also may do taping to help promote optimal positioning of the arm, particularly at the shoulder.

Electrical stimulation is sometimes done for brachial plexus palsy, though this is frequently not tolerated at a very young age. Over time it does become accepted by many young children. Most commonly, it is performed with surface electrodes to increase muscle bulk by use of sufficient stimulation to get a local muscle twitch for approximately 20 minutes twice daily. It has been shown that continuous electrical stimulation to denervated muscles with implantable electrodes will lead to improved muscle outcome after nerve regeneration (56). This has not been widely utilized and is not currently available on the U.S. market.

It has been proposed that the adverse affects of prolonged denervation leave intramuscular axons deteriorated to such low numbers such that even with successful nerve regeneration, it is impossible to reinnervate enough muscle fibers for sufficient force (57). There are also proposals that low doses of brain-derived neurotrophic factor (BDNF) may protect against this decrease in those who have late nerve grafts, though high doses are inhibitory (58).

Complications

It is important to monitor for secondary complications. These commonly include muscle atrophy and joint contractures. The affected arm frequently is shorter and has decreased circumference as well. Joints may become dislocated, and scapular winging is frequently seen. There may be torticollis, most commonly with the face turning away from the involved arm. General child development may be affected, including by lack of awareness of the arm. Similarly, body image may be affected. There can be ulcerations from trauma,

particularly in insensate areas. Pain is infrequent after birth brachial plexus palsy but not after later trauma.

Surgical Indications

Indications for timing of brachial plexus surgery for infants have been controversial. It has been shown that a longer time for recovery leads to a worse shoulder function and that those who regain elbow flexion after 6 months of age have worse function than those who regain it between 3 and 6 months (59). Those with recovery by 3 months have normal function. Those who had microsurgery at 6 months did better than those who spontaneously recovered elbow flexion at 5 months (60). Surgical intervention is commonly recommended for those having less-than-antigravity strength in elbow flexion at 6 months of age (61).

Later brachial plexus injuries are divided into supraclavicular and infraclavicular injuries, supraclavicular being 75% and infraclavicular 25%. Supraclavicular injuries are generally felt to be due to traction of the plexus (classically in a motorcycle crash), and these have a worse prognosis than infraclavicular injuries (62). There may be a fracture of the clavicle or cervical transverse process, and supraclavicular fossa swelling may be seen. Dorsal scapular nerve or long thoracic nerve injury may be present. Supraclavicular lesions may also be due to falls; large objects falling on a shoulder, such as a tree limb; skiing or climbing; or contact sports, including football (52). Other etiologies are backpacks that are too heavy, tumors and gunshot wounds, or lacerations or animal bites. Those who have ipsilateral Horner's syndrome and persistent pain have a worse prognosis (52).

Infraclavicular brachial plexus injuries are more commonly associated with fractures and dislocations about the shoulder or humerus, occurring more often in older adults. The posterior cord, axillary nerve, or musculocutaneous nerve are classically involved. Infraclavicular injuries are less severe and have better outcomes (63). Infraclavicular plexus injuries may also be due to falls, motor vehicle collision, or tumors (52). Gunshot wounds, stab wounds, and failed attempt at shoulder reductions may cause infraclavicular injuries as well (64). Brachial plexus palsy has been reported after axillary crutch use, anesthesia positioning (particularly with table tilt), and after bony fracture with malunion (65). For severe injuries later in life, recommendations are for surgical exploration and nerve grafting, most commonly at three to four months postinjury (64,66).

Surgery

Surgical interventions for brachial plexus palsy are varied. There may be electrical testing, including evoked potentials, and nerve conduction studies done to assess the nerves in the operating room to be as specific as possible with the procedures undertaken. Microsurgical repair yields results months later. Recovery is generally felt to proceed at the rate of approximately a millimeter a day or an inch a month. There is also felt to be more nerve growth factor available in younger beings so that both size and age have an impact in outcome. It is critical to have therapy postsurgery and to continue a faithful daily home program as well.

There are a variety of options for surgical procedures for brachial plexus injury. Neurosurgery may include neurolysis in which scar and fibrotic tissue are removed from nerve tissue. Direct nerve transfers have the advantage of quick recovery time due to short regeneration distance versus neurotization, which requires interposition of a nerve graft. The sural nerve and great auricular nerve are commonly used as donor nerve fibers for these grafts (67). More recently, end-to-side neurorraphy is performed for those who have some intact fibers for augmentation. The advantage of this is not requiring a sacrifice of any other nerves. Not uncommonly, synkinesis of newly innervated muscles with contraction of muscles innervated by the donor nerve may be seen, and is treated with therapy (68).

Some classic nerve procedures involve transfer from a functionally less important nerve to a distal denervated nerve. Common examples include taking intercostal nerves to the upper trunk or to the suprascapular nerve. Another classic surgery is the Oberlin procedure, which transfers one or several ulnar nerve fascicles to the musculotaneous nerve as it enters the biceps muscle (69). Transfer of the spinal accessory nerve to the suprascapular nerve is also commonly used for shoulder abduction. For approximately the last 10 years, contralateral C7 transfers have been performed both in adults and infants for those with multiple severe avulsions. This procedure has been shown to provide adequate elbow flexion as a result, and most patients have had only temporary sensory deficits on the ipsilateral C7 side (70). This procedure clearly illustrates the point that nerve grafts are not required to have their original source but can have function coming from a variety of intact neurological structures. This allows for greater flexibility and creativity in the surgeon performing the procedure, aiming for recovery of function.

Glenoid dysplasia with posterior shoulder subluxation is frequently a complication of children after birth brachial plexus palsy. It was commonly thought to be the result of a slowly progressive glenohumeral deformation due to muscle imbalance and possible physeal trauma, but it was found that posterior shoulder dislocation happened at a mean age of 6 months, with rapid loss of passive external rotation. There was no

correlation between the initial neurological deficit and the presence or absence of dislocation (71).

Many musculotendinous surgical procedures are performed for children with birth brachial plexus palsy. It has been shown that latissimus dorsi and teres major tendon transfer to the rotator cuff, along with musculotendinous lengthening, will provide improved shoulder function but no significant change in the bony position of the shoulder or humerus. This procedure does not decrease glenohumeral dysplasia (72).

With internal rotational contracture and glenohumeral joint deformity, along with significant abnormality of glenohumeral joint, a derotational osteotomy can result in improved shoulder function, along with improved internal rotation contracture (73).

Some children with birth brachial plexus palsy have been described to have arthroscopic release of shoulder deformity alone before 3 years, and for those over 3 years of age, arthroscopic release with latissimus dorsi transfer. They all show improved shoulder position, but they do have loss of internal rotation. Some of the children under 3 years do have a recurrence and require a second procedure with a latissimus dorsi transfer (74).

In adults, performing a glenohumeral arthrodesis, both in patients with upper plexus palsy with functional distal arm, as well as in those with total plexus palsy, has been shown to increase functional capabilities. The strength of the pectoralis major is a significant prognostic factor for outcome (75).

Performing wrist arthrodesis in adults with brachial plexus injury is done for improved function as well as pain relief. There will be limitations after having this procedure, and potential patients need to have full information in order to know what to expect prior to the procedure. There also remains some controversy of the ideal position to place the hand, which is generally placed in slight wrist extension and ulnar deviation in order to have the most powerful grip (65,76). A dramatic surgical procedure sometimes performed for children and adults with brachial plexus palsy is a free muscle transfer, most commonly performed with the gracilis muscle. The muscle is transferred with its vascular and nerve supply and attached to these in the arm. This procedure has been described as having reliable results for elbow flexion and wrist extension (65).

Pain

Pain has not been reported as a severe problem in birth brachial plexus injury, although with one study reporting biting of the limbs in less than 5% of the cases, it is possible that this is a manifestation of pain. Self=mutilation has been reported in youngsters after a birth brachial plexus injury. This study of 280 patients with a birth brachial plexus injury found that 11 of these children had self-mutilating behavior by biting or mouthing the affected arm. The age of onset was between 11 and 21 months, and the duration of the behavior was 4 to 7 months. This was more frequent in children who underwent surgery, with 6.8% of these children, and 1.4% of children who did not have surgery. It is unclear if this is due to surgery or the severity of the injury or a combination of these (77). It is also possible that this is a response to the unusual sensation of the recovering nerve, possibly a manifestation of what we see on examination as a Tinel's sign. It has been felt, however, that it is more likely biting with the resumption of nerve growth with sensation of tingling as there is recovery occurring, but this is not proven.

In those who have later traumatic or nontraumatic brachial plexus injuries, pain can be a significant problem. It has been described most commonly with avulsions as severe burning and crushing pain most commonly in the hand. This may develop days to months after the injury and almost always within three months. It is most commonly resolved within several years, but approximately 20% of those with pain have severe, long-lasting disruptive pain (78). This can be treated with transcutaneous nerve stimulation classically from C3–T2. Medications, including antidepressants and anticonvulsant agents, have been affective. Topical treatments, including topical lidocaine 5% pain patches, are sometimes useful. Nerve surgery is commonly effective in resolving pain (79,80). The author has seen children with traumatic brachial plexus injuries and severe pain complaints prior to their nerve procedure wake up postoperatively in the recovery room excited that the pain is gone. Amputation is not effective for resolving the pain (81).

REHABILITATION OF THE CHILD WITH RHEUMATIC DISEASE

Rehabilitation of the child with rheumatic disease requires an interdisciplinary approach that includes the child and family. Although most often the physiatrist is not the treating physician in rheumatological disease, they can play a key role in the comprehensive management of these conditions, along with other members of the rehabilitation team, to maintain or restore age-appropriate function and development, prevent deformity and contractures, and help manage pain.

Juvenile Idiopathic Arthritis

Juvenile idiopathic arthritis (JIA), formerly known as juvenile rheumatic arthritis (82), is the most common

rheumatic disease of childhood, affecting approximately 16–150 in 100,000 (83). In 1995, the International League Against Rheumatism (ILAR), together with the World Health Organization, reclassified chronic childhood arthritis (84); the second revision occurred in 2001 (85). Chronic childhood arthritis is now known as JIA and is divided into the following seven subtypes: systemic arthritis, oligoarthritis, rheumatic factor (RF)–negative polyarthritis, RF-positive arthritis, psoriatic arthritis, enthesitis-related arthritis, and undifferentiated arthritis. JIA occurs in children before the age of 16 years, persists at least six weeks, and has had other known conditions excluded; etiology is unknown, but seems to include genetic and environmental components (83,86).

Early arthritis may be manifested by swelling, warmth, and joint stiffness, typically worse at the beginning of the day then improving with activity. Symptoms usually fluctuate; uncontrolled inflammation leads to joint damage. Younger children rarely complain of joint pain, but may instead become irritable, stop walking or using an extremity, or regress in their behavior (87). Other symptoms include decreased appetite, malaise, inactivity, morning stiffness, nighttime joint pains, and failure to thrive (87). Enuresis may occur in a recently toilet-trained child (88). Later disease presents with reduced range of motion (ROM), contractures, overgrowth or undergrowth of affected limbs, and resultant disability.

A characteristic feature of chronic arthritis in children is the effect the disease has on bone and joint development (89,90). Local growth disturbances at inflammation sites can lead to overgrowth (secondary to possible inflammatory-mediated increased vascularization and growth factor release) or undergrowth (secondary to growth center damage or premature fusion of epiphyseal plates). Irregular traction on growing structures secondary to muscle spasms and periarticular fibrosis can also cause aberrant growth (89,90). Micrognathia, leg-length inequalities, and developmental hip anomalies are all possible results from these processes. Steroids can also contribute to severe growth effects, as well as osteoporosis (91).

The differential diagnosis of JIA is large (Table 14.1 provides a full differential diagnosis).

The assumption that JIA will universally resolve by adulthood is incorrect (92). Radiological joint damage occurs in children with systemic arthritis and polyarticular arthritis within two years, and in oligoarthritis within five years (93,94) Despite long-term persistence of disease activity in JIA, much improvement in functional outcomes has been made in the last decade (95,96). Indicators of poor outcome include greater severity or extension of arthritis at onset, symmetrical disease, early wrist or hip involvement, presence of RF, persistent active disease, and early radiographic changes (97).

Clinical Features of JIA Subtypes

Systemic JIA Systemic-onset JIA presents with many extra-articular features and represents 10% to 20% of all JIA (86). Diagnosis requires arthritis accompanied or preceded by quotidian fever (spikes >39 degrees Celsius once a day with return to normal between peaks) of at least two weeks' duration, plus one or more of the following: evanescent salmon-colored rash, generalized lymphadenopathy, hepatomegaly, splenomegaly, or serositis.

About 5% to 8% of children with systemic JIA develop a life-threatening complication known as macrophage activation syndrome (98) with persistent fever, lymphadenopathy, and splenomegaly, and there is profound depression in one or more of the blood cell lines (often initially platelets) with raised liver function enzymes and clotting abnormalities. Definitive bone marrow examination shows numerous well-differentiated macrophages actively phagocytizing hemopoetic elements (99).

In one-half of children with systemic JIA, the course follows a relapsing-remitting course, with arthritis accompanying febrile episodes, then remission once systemic features are controlled. Long-term outlook for these children is usually good. In the other half, the disease is unremitting, with resultant severe joint destruction, and is probably the most severe JIA subtype (83,100). Poor prognostic signs include the continued presence of systemic features and a platelet count exceeding 600,000/mm^3 six months after onset (87). At least one-third of children will develop severe arthritis (101).

Oligoarthritis. Oligoarthritis is classified into two subtypes: persistent (affecting not more than four joints throughout the disease course) and extended (affecting more than four joints after the first six months of disease). Characteristically, there is an early onset before 6 years of age of an asymmetric arthritis, usually in the lower limbs, and predominantly in females. Antinuclear antibodies (ANAs) are detected in substantial titres in about 70% to 80%, and they represent a risk factor for iridocyclitis. Children with the oligoarthritis subtype generally have the best outcome (83); however, sight-threatening, clinically silent uveitis develops in the first four years from diagnosis. Regular ophthalmology follow-up is essential (102).

Polyarthritis. Polyarthritis must affect five or more joints in the first six months of the disease. RF-positive polyarthritis mainly affects adolescent girls, with a symmetrical pattern, and is the same as adult RF-positive disease (89). By five years from onset, severe deforming arthritis is generally present (90). RF-negative polyarthritis is a more heterogenous group with more

14.1 Differential Diagnosis of Juvenile Idiopathic Arthritis

Pediatric Rheumatic Diseases
Systemic lupus erythematosus
Juvenile dermatomyositis
Scleroderma
- Localized (linear, morphea, etc.)
- Generalized (systemic sclerosis, CREST, etc.)

Mixed connective tissue cisease (overlap syndrome)
Juvenile ankylosing spondylitis
Acute rheumatic fever
Reactive or postinfectious arthritis
Vasculitis
- Kawasaki disease
- Henoch-Schoenlein purpura
- Behçets disease
- Wegener granulomatosis
- Polyarteritis nodosa

Autoinflammatory disorders
- Tumor necrosis factor receptor-alpha associated periodic syndromes
- Familial cold autoinflammatory syndrome
- Neonatal onset multisystem inflammatory disease
- Chronic infantile neurologic, cutaneous and articular syndrome
- Periodic fever, adenitis, pharyngitis and apthous ulcer syndrome

Fibromyalgia
Complex regional pain syndrome, type II

Infectious Diseases
Bacterial arthritis
Viral arthritis
Fungal arthritis
Osteomyelitis
Fasciitis/myositis

Neoplastic Diseases
Leukemia
Lymphoma
Neuroblastoma
Primary bone neoplasms

Hematologic Diseases
Hemophilia
Sickle cell disease

Noninflammatory Disorders
Trauma
Overuse syndromes
Osteonecrosis syndromes
Avascular necrosis syndromes
Slipped capital femoral epiphysis
Toxic synovitis of the hip
Patellofemoral dysfunction (chondromalacia patellae)
Diskitis

Miscellaneous Disorders
Inflammatory bowel disease
Sarcoidosis
Collagen disorders
Chronic recurrent multifocal osteomyelitis
Growing pains
Hypermobility syndromes
Foreign-body arthritis
Psychogenic arthralgias/arthritis (conversion reactions)

variable outcome. Approximately 20% to 40% of those affected are ANA-positive, and chronic uveitis is found in 5% to 20% (89); it is believed by some authors that this entity represents a later stage of early-onset oligoarthritis (103). Future versions of the ILAR classification of JIA may explore this more fully.

Psoriatic Arthritis. Psoriatic arthritis accounts for about 5% of JIA and requires the simultaneous presence of arthritis and the typical psoriatic rash, or if the rash is absent, arthritis plus two of the following: positive family history of psoriasis in a first-degree relative, dactylitis, and nail pitting. Psoriatic disease in children before the age of 5 years appears to be more difficult to control than in an older subset of children, with a median of 9.5 years (86).

Enthesitis-Related Arthritis. Enthesitis-related arthritis affects males after the age of 6 years (89, 90) and most children are HLA-B27–positive. The most common sites of enthesitis are the calcaneal insertion of the Achilles tendon, plantar fascia, and tarsal area. Arthritis commonly affects the joints of the lower extremities. Unlike other JIA subsets, hip involvement is common at disease presentation.

These children may progress to fulfill criteria for ankylosing spondylitis, reactive arthritis, or arthritis associated with inflammatory bowel disease. Uveitis is also a clinical problem in this subset, but it is usually sudden in onset, symptomatic, and more unilateral than in children with other JIA subsets (86).

Juvenile ankylosing spondylitis, not considered part of the JIA subclassification; mainly affects adolescent boys; is strongly associated with HLA-B7; and manifests as an asymmetric, often episodic, oligoarthritis in the lower limbs. Later on, bilateral sacroiliac joints become involved, and progression of the disease can lead to the characteristic "bamboo" spine on radiographic images secondary to ankylosis of spinal joints. In children, peripheral arthritis and

enthesitis present early in the disease, but sacroiliac and spine joints are not involved until many years later (104). Rehabilitation involves maintaining spinal ROM through extension exercises, strengthening hip extensors and quadriceps muscles, custom shoe inserts to relieve pain, and deep breathing exercises to maximize chest expansion. Because of the chronic course of the disease, the child and parents should not restrict age-appropriate social and recreational activities (104).

Inflammatory bowel-associated arthritis occurs in approximately 10% to 20% of children with ulcerative colitis and Crohn's disease. The arthritis usually affects a few joints and may be associated with spondylitis; erythema nodosum and growth failure may occur.

Undifferentiated Arthritis. This subset is not a separate entity, but is more of a catch-all category for those children who do not satisfy inclusion criteria for any category, or who meet criteria in more than one category.

Rehabilitation of the Child With JIA

Goals of treatment include controlling symptoms, preventing joint damage, achieving normal growth and development, and maintaining function and normal activity levels.

Treatment goals may vary during maintenance and acute flare-ups of the disease.

Resting a joint may be necessary during an acute flare-up to prevent aggravation of the disease process; activities that affect or excessively stress joints should be discouraged during acute flare-ups. Resting a joint may also be useful during the maintenance phase for joint protection. Rest periods may be necessary to reduce fatigue; resting in the prone position will help reduce hip and knee flexion contractures.

Splinting is used during a flare-up to provide alignment during a rest period. Functional splints may be used during flare-ups and maintenance phases if they provide joint relief and allow functional activities without stressing inflamed joints. Splinting can be used during the maintenance phase to promote local joint rest, support weakened structures, and assist function. To prevent flexion contractures, the upper extremity is splinted in a functional position as follows: wrist 15–20 degrees of extension, some finger flexion, 25 degrees at the metacarpophalangeal (MCP) joint, and 5–10 degrees at the PIP joint, with the thumb in opposition. Ring splints can be used for finger deformities. Knee immobilizers may be used to maintain knee extension at night; rotate on alternate legs for better compliance. Dynamic splints or serial casts can increase ROM. Foot orthoses can promote arch support and reduce pain in weight bearing.

Gentle ROM with passive extension greater than flexion two to three times a day is used to preserve joint ROM. Incorporating pain medication, progressive muscle relaxation, breathing exercises, biofeedback, massage, or doing the exercises in a nice, warm tub can greatly facilitate ROM exercises. Gentle ROM exercises should be done as tolerated during acute flare-ups to prevent flexion contractures.

Heat is an excellent modality in the maintenance phase to decrease stiffness, increase tissue elasticity, and decrease pain and muscle spasm. Hydrotherapy with temperatures 90–100 degrees Fahrenheit, fluidotherapy, paraffin, or moist heat can be used. Most children prefer heat to cold. Taking a hot bath or shower, sleeping in a sleeping bag, or using a hot pack (along with ROM exercises) may help relieve morning stiffness. Caution must be exercised in insensate areas to avoid burns. Ultrasound is contraindicated in children with open growth plates. Heat should not be used during an acute flare-up, as it increases the inflammatory response and causes further joint destruction.

Cold can be used during an acute flare-up for pain relief and to decrease swelling. It may also be beneficial during the maintenance phase for the same reasons. Cold should not be used over insensate areas or in those with Raynaud's phenomenon.

Adaptive strengthening exercises can be incorporated into play and recreational activities. Some examples include throwing a ball (strengthens elbow and shoulder), riding a bike (promotes knee and hip extension), and swimming (decreases weight bearing on painful joints). Incorporating general aerobic conditioning is also important and may include activities such as swimming, dancing, noncontact karate, and tai chi. Isometric strengthening exercises are fine during an acute flare-up, but vigorous exercise should be held until the acute process is over. Hydrotherapy can be combined with land-based physiotherapy in treating JIA (105).

Adaptive equipment can be used for joint protection, rest, and to minimize further joint destruction during both phases. Examples include adaptive utensils, adaptive pens and computer access, table and desk modifications (to prevent excessive trunk and neck flexion), zipper pulls, dressing sticks, long-handled brushes, elastic waistbands, Velcro closures, and larger buttons. Children should actively participate in functional activities of daily living (ADL) training in order to choose acceptable devices and improve their use.

Activity and ambulation should be encouraged as much as possible. A posterior walker for upright posture (with decreased flexion) and a standing program may be useful for functional mobility training if wheelchair use cannot be avoided. In children with JIA, custom-made semirigid foot orthotics with shock-absorbing posts have been found to significantly

improve pain, ambulation speed, self-rated activity, and functional ability levels compared to prefabricated off-the-shelf shoe inserts or supportive athletic shoes alone (106).

A presurgical joint rehabilitation program aims to strengthen the muscles needed for mobility in the postop period, train for future ambulation aids, and identify other joint involvement that may affect the rehabilitation process. Post-surgical rehabilitation fulfills those goals set in the pre-surgical rehabilitation program. Ambulation aids such as the platform walker may be used to better distribute weight bearing pressure on affected upper extremity joints after knee or hip surgery. In children status post-hip prosthesis, the acetabular component should be checked for loosening (as opposed to the femoral component in adults), especially if children are active.

Growth retardation can occur during periods of active disease; it may also be compounded by corticosteroid use. Maximize growth by promoting optimal nutrition. Children with JIA should eat a balanced diet with supplemental multivitamins, calcium, vitamin D, and sunshine secondary to the high risk of osteopenia. Plenty of (nonimpact) activity again should be encouraged.

Counseling for both the child with JIA and their family should be provided to maximize psychosocial and emotional well-being. Treatment goals also include addressing family, school, and vocation. Assisting in the preparation of a 504 plan for school accommodations enables a child with joint disease opportunity for more complete participation in his or her school life and academic career. Summer camps are a practical way of addressing peer support within adolescent rheumatology services; positive effects include increased control, self-esteem, physical fitness, independence from parents, self-management of health care, and an opportunity to meet others with a similar condition (107).

Specific Joints in JIA

Cervical Spine. Cervical spine involvement occurs more often in children with JIA than adults. Restriction of ROM, pain, and muscle spasms, which may present as torticollis, may be seen. A soft cervical collar to serve as a reminder for proper alignment and provide warmth may be helpful in acute pain with muscle spasm. Minimizing time in flexion is important. If the transverse ligament becomes weakened, atlantoaxial subluxation can occur. If subluxation occurs, a firm cervical collar should be worn during automotive transport.

Temporomandibular Joint (TMJ). This joint is affected in almost two-thirds of children with JIA (108) by causing pain in chewing and opening the mouth, stiffness, and micrognathia. Younger children will not complain of jaw pain, but will instead choose to modify their diet to avoid pain. Progressive jaw ROM exercises and modalites may help treat pain and stiffness. If the lower jaw does not develop properly, it may create an overbite, requiring orthodontist intervention and/or oral surgery. Mandibular and facial growth disturbances are more common in polyarticular types of JIA.

Upper Extremities. The shoulder is not commonly involved at the onset of disease. Approximately one-third of children with polyarticular or psoriatic disease may eventually develop shoulder involvement and loss of adduction and internal rotation affecting midline ADLs, such as grooming and toileting. The elbow requires at least 90 degrees of flexion range to perform ADLs such as eating, grooming, and reaching. Loss of more than 45 degrees of elbow extension limits the use of arms as levers to rise from a seated position and makes toileting and lower extremity dressing difficult. Wrist involvement is common in children; there is early loss of wrist extension with progressive flexion contracture. A nighttime resting wrist splint can maintain the wrist in 15 to 20 degrees of extension with the fingers in a few degrees of flexion; ulnar deviation can also be built in as necessary. Strengthening of wrist extensors and radial deviators is necessary to reduce wrist flexion and ulnar deviation contractures. Moist heat to reduce spasm and improve tissue elasticity followed by serial casting for 48–72 hours as tolerated may help reduce contractures by slowly increasing wrist extension as tolerated while controlling ulnar deviation and subluxation; commercially available dynamic splinting may also facilitate stretching. Should ankylosis be inevitable, the hand should be splinted in a neutral position for optimal function in self-cares.

Functional grasp may become limited as fingers lose both flexion and extension range. Flexion contractures of the metacarpal and proximal interphalangeal joints are often seen. The use of ring splints in metal or plastic can help control proximal interphalangeal flexion and extension seen in boutonniere and swan neck deformities, respectively. Fingers can be strengthened through play with clay and various adaptive putties.

Lower Extremities. In the lower extremities, flexion contractures occur at the knee and hip. Painful ambulation can lead to increased sitting, which in turn leads to increased flexion contracture, deconditioning, weakness, atrophy, and osteoporosis. Hip flexion contractures in children occur with internal rotation and adduction, compared with adults who tend to develop external rotation and abduction. Prone lying greater

than 20 minutes per day with the hips and knees extended and feet off the edge of the bed can help prevent these contractures. Other strategies include strengthening of the hip extensors, external rotators, abductors, and quadriceps, along with ROM exercises to stretch the hip flexors, internal rotators, adductors, and hamstrings. Hip extensors can be strengthened through swimming, aquatic therapy, and bicycling. Encouraging upright posture and ambulation, using a stander as necessary, is also helpful. Hip development may be assisted by the use of a stander; a prone stander can strengthen neck and hip extensors, while a supine stander maintains the knees in extension and allows upright weight bearing.

The knee is the most commonly affected joint in JIA; early involvement of the knee can cause quadriceps weakness that may not resolve. Knee contractures can lead to other joint contractures and further gait abnormalities. Bony overgrowth with resultant leg-length discrepancies are often seen. The knee can be maintained in extension using resting splints such as knee immobilizers and alternating legs nightly as needed to increase comfort and compliance. Dynamic splinting using an adjustable knee joint can be used to improve ROM and limit excessive flexion and valgus tendency. Because forced extension of the knee with a contracture can exacerbate posterior subluxation, caution must be exercised in using bracing and splinting. Active quadriceps strengthening should be done post-brace removal and also maintained with knee extension exercise or isometric exercises if too painful. Kicking, bicycling, and walking can also strengthen weak quadriceps muscles.

Multiple foot deformities can occur in JIA, including claw toe, valgus or varus hindfoot, and ankle plantarflexion contracture deformities. The midfoot is frequently affected, and can be quite painful and difficult to treat. Tenosynovitis that is difficult to discern from joint disease may occur. Molded foot orthoses can be used to reduce pain at the metatarsal heads and heels with weight bearing. A University of California at Berkeley orthosis can prevent or control varus and valgus deformities. A posterior leaf-spring ankle foot orthosis (AFO) or nighttime resting splint may be helpful to reduce loss of ankle dorsiflexion range and control varus and valgus. Ankle rotation exercises, balancing exercises, and raising the heel on a step can strengthen ankle muscles. Footwear should be comfortable and accommodate any foot deformities. High heels should generally be avoided, as they can help develop plantarflexion contractures and add to foot deformities. Flip-flops should also be avoided secondary to their lack of adequate support.

Inflammation causing bony overgrowth at the distal femur can cause a true leg-length discrepancy (LLD), leading to pelvic asymmetry and scoliosis. The increased blood flow from inflammation may alternatively cause early epiphyseal closure and overall limb shortening.

Medical and Surgical Treatments of JIA

Children with JIA are treated with more of an induction and maintenance approach, taking advantage of windows of opportunity to modify the disease course, usually under the guidance of a pediatric rheumatologist (109). Nonsteroidal anti-inflammatory drugs (NSAIDs) are used briefly in the initial phase (110).

Intra-articular steroid injections in affected joints using triamcinolone hexacetonide (preferred formulation in pediatric practice) (111) are frequently needed at disease onset or during the disease course. They may be substituted for NSAIDs in mono or oligoarthritis at times (83). Early use of intra-articular steroids in one or two affected joints may even have the potential to modify the course of JIA (87).

Methotrexate is used early on in the disease course as a second-line agent of choice for persistent, active arthritis (112), with improvement usually seen in 6 to 12 weeks. Parenteral methotrexate is superior to oral, especially at higher doses. Leflunamide may also be used if methotrexate is ineffective (113). Recommendations vary from 6 to 24 months of remission before tapering medications other than NSAIDs; the best method for tapering methotrexate is unknown (88). Approximately 70% to 75% of children with chronic arthritis achieve remissions with NSAIDs plus methotrexate (114).

The biologics (etanercept, infliximab, adalimumab, anakinra, abatacept, and rituximab) have all been demonstrated to be effective in treating inflammatory arthritis (88). Tumor necrosis factor (TNF) inhibitors (etanercept and adalimumab) are now approved for use in children (115) and are used after methotrexate. Infliximab has an efficacy similar to etanercept (116). Abatacept, a T-cell blocker, has been recently approved by the Food and Drug Administration (FDA) for use in children with JIA, and has promise for the TNF inhibitor nonresponders. Special risks in treating children with biologics include increased risk for infections (especially varicella), how and when to proceed with usual immunizations, long-term effects, and possibility of later malignancies or development of central nervous system demylinating disease (88). Early and aggressive treatment of JIA with newer agents holds unlimited promise for even better outcomes for children with JIA. Steroids are used as sparingly as possible to control inflammation in order to avoid long-term side effects such as weight gain, poor growth, and risk of infection. There is no systemic evidence that steroids are disease-modifying (92).

Children with JIA are at high risk of developing osteopenia secondary to the disease itself, to steroid treatment of the primary disease, lack of physical activity and weight bearing, limited sunshine exposure, and inadequate vitamin D and calcium. Calcium and vitamin D supplementation, sunshine, and encouragement of physical activity should be incorporated into the treatment plan.

Surgery is rarely used in the early course of the disease; however, surgery can be used later in the course to relieve pain, release joint contractures, and replace a damaged joint. Older children whose growth is complete or almost complete and whose joints are badly damaged by arthritis may need joint replacement surgery to reduce pain and improve function. Soft tissue releases may be needed to reposition malaligned joints or release contractures.

Infectious Disease With Arthritis

Infectious causes of arthritis include bacterial, viral or post-viral and fungal. Osteomyelitis and reactive arthritis can also be confused with JIA.

Septic Arthritis

Joint involvement in septic arthritis may be by hematogenous spread, direct extension from local tissues, or as a reactive arthritis.

Bacterial septic arthritis is usually monoarticular in children, but multiple joints can be involved. Children may present with fever, joint pain, and decreased joint mobility, especially in the knees, hips, ankles, and elbows. A child may not allow the affected joint to be touched and, sometimes, may not even allow the affected joint to be seen. An ambulatory child will refuse to bear weight on the affected extremity. Premature infants presenting with irritability, fever, and hips positioned in abduction, flexion, and external rotation should be checked for septic arthritis of the hip. Boys 3 to 10 years who present with hip or referred knee pain should be checked for transient synovitis. Ear infections are the most common source of bacteria leading to septic arthritis in children (117). Osteomyelitis or disciitis can develop in children with septic or reactive arthritis.

In all age groups, 80% of cases are caused by gram-positive aerobes (60% *S. aureus*; 15% beta-hemolytic streptococci; 5% *Streptococcus pneumoniae*), and approximately 20% of cases are caused by gram-negative anaerobes. In neonates and infants younger than 6 months, *S. aureus* and gram-negative anaerobes comprise the majority of infections.

Clinically affected joints require emergent aspiration and treatment. Aspiration of joint fluid is necessary for possibly identifying the agent and relieving pain. Joint fluid reveals increased white blood cells (WBCs), protein, and low-to-normal glucose. Radiographic findings progress from soft tissue swelling to juxta-articular osteoporosis, joint space narrowing, and erosion. Treatment consists of appropriate antibiotic therapy, joint aspiration to relieve pressure and pain, and physical therapy to maintain ROM.

Reactive Arthritis

Reactive arthritis is different from septic arthritis in that it is an autoimmune response triggered by antigen deposit in the joint spaces; synovial fluid cultures are negative. It is set off by a preceding infection, the most common of which would be a genital infection with *Chlamydia trachomatis* in the United States, usually in adult males (*118).* Reactive arthritis after *Yersinia* and *Campylobacter* can be associated with HLA-B27. *Yersinia enterocolitica* infection can show persistence of the organism in joint fluid, especially the knee. The main goal of treatment is to identify and eradicate the underlying infectious source with appropriate antibiotics, if still present. Analgesics, steroids, and immunosuppressants may be needed for patients with severe reactive symptoms that do not respond to any other treatment.

Lyme Disease

Lyme disease is caused by the spirochete, *Borrelia burgdorferi*, with transmission to humans via the deer tick, *Ixodes dammini*. Lyme disease is the most common tickborne disease in North America and Europe. The initial phase of Lyme disease (lasting about four weeks) consists of fever, fatigue, headache, athralgias, myalgias, stiff neck, and erythema migrans. Erythema migrans looks like a reverse target skin lesion, as it is a large, red lesion with a central clearing area; it occurs 1 to 30 days after the tick bite. The late phase, lasting months to years, is characterized by arthritis, cardiac disease, and neurological disease. Intermittent episodes of unilateral arthritis involve the knee most often; hip, shoulder, elbow, wrist, and ankle may also be involved. In 85% of children, the arthritis resolves before the end of the initial treatment; in 10%, a chronic inflammatory phase develops.

Other Rheumatic Diseases of Childhood

Systemic Lupus Erythrematosus

Systemic lupus erythrematosus (SLE) is a multisystem autoimmune disease with widespread immune complex deposition that results in episodic inflammation, vasculitis, and serositis. Children are more likely than adults to present with systemic disease; 20% of cases

begin in childhood. Females are affected 4.5 times more than males. One-third of children have the erythematous butterfly rash over the bridge of the nose and cheeks; this rash may occur after exposure to sunlight. Most children develop a transient, migratory arthritis of the extremities; radiographic evidence of joint deformity and erosion are not common. Pain may be out of proportion to joint findings on examination. Proximal muscle weakness may be a result of acute illness, myositis, or the result of steroid-induced myopathy. Long-term steroids also increase the risk of avascular necrosis of the femoral head.

Systemic features of SLE may include pericarditis or endocarditis; proliferative glomerulonephritis or other renal disease; seizures, psychosis, memory deficits, headaches, or behavior changes; pulmonary hypertension and/or hypertension. Nephritis occurs in ~75% of children with SLE and is the main factor for determining outcome. Hematuria, proteinurea, persistent hypertension, chronic active disease, and biopsy-proven diffuse proliferative glomerulonephritis are associated with a poor outcome. Ten-year survival is ~80%, although this number is lower in lower socioeconomic populations.

Management of SLE is symptomatic. Maintaining physical activity as much as possible, avoiding excess sunlight exposure, optimizing nutrition, and providing adequate social supports are key. For some children with open discoid lupus rash lesions, dressing changes and wound cares may be best facilitated with individualized whirlpool therapy, much like is used for burn wound cares.

NSAIDs are mainly used for arthritis and musculoskeletal conditions. Fever, dermatitis, arthritis, and serositis usually resolve quickly with low-dose steroids, whereas serologic findings may require weeks of steroid therapy. Hydroxychloroquine may be used for skin manifestations or in concert with steroids to lower the steroid dose. High-dose steroids, immunosuppressive agents, and biologic agents may be necessary for more severe disease manifestations.

Scleroderma

Systemic sclerosis is uncommon in children; linear and focal cutaneous involvement is most common in children. Girls between ages 8 and 10 years are more often affected; duration can last 7 to 9 years. Linear scleroderma presents with atrophic, erythematous skin areas, which later become fibrotic. This skin then binds to underlying subcutaneous tissues, and underlying muscle and bone also become involved. Children may have pain from these skin changes. Soft tissues can atrophy, leaving areas of asymmetry. Scleroderma en coup de sabre is a unilateral linear involvement of the face and scalp, often with loss of hair on the involved side, with loss of facial asymmetry. Systemic disease in children is uncommon. Physical therapy is necessary to prevent loss of ROM and contractures because of the cutaneous involvement. Soft tissue massage, moist heat, stretching, and ROM exercises help maximize joint mobility. Topical corticosteroids may be helpful in treating localized skin disease; systemic steroids, methotrexate, and physical therapy may alter the course of progressive disease.

Hematological Disorders

Hemophilia

Hemophilia is a bleeding disorder that affects about 18,000 Americans; each year, about 400 babies are born with the disease, and it occurs in 1 out of every 7,500 males. Of these, about 85% of cases are Factor VIII (hemophilia A) and 14% are Factor IX (hemophilia B).

In hemophilia, bleeding occurs without any recognizable trauma; spontaneous bleeding happens most often in the knees, ankles, elbows, and shoulders. Bleeding into the joints usually begins after a child begins to walk. As bleeding begins, the child may experience warmth or tingling in the joint. As bleeding progresses, there is usually a feeling of stiffness, fullness, and pain. The joint swells and may be warm and tender, causing synovial membrane thickening. Without treatment, hypertrophy of the synovium with its increased vascular supply, creates a cycle of more bleeding and destruction. Without intervention, fibrosis and arthritis sets in, making joint replacement at an early age the only option. Pain and swelling can also lead to decreased active joint ROM, further leading to contractures. Other complications include muscle atrophy, osteopenia, peripheral neuropathy, and compartment syndrome.

The main treatment for hemophilia is injections of cryoprecipitate. Acute hemarthrosis requires joint immobilization for 48 hours to prevent further bleeding. Once pain and swelling subsides, passive ROM should be started to prevent fibrosis and contracture development. Analgesics, anti-inflammatory medications, and aspiration of blood from the joint if overlying skin is tense are important in pain management. Joint function may be regained in 12–24 hours with early factor replacement, but may take up to two weeks for more blood reabsorption (119). ROM exercises can be done in the water to reduce stress on the joint while providing resistance; strengthening of specific muscle groups to maximize joint stability should be prescribed. Contact sports are generally contraindicated. Joint replacement is used in end-stage arthropathy; oftentimes, loosening occurs more often, especially in younger children.

Sickle Cell Disease

Joint involvement occurs in infancy in sickle cell disease. Bones and joints are often the site of vaso-occlusive

episodes, and chronic infarcts may result. One of the earliest manifestations of sickling in young children is dactylitis, or "hand–foot syndrome." An episode of painful swelling of the bones of the hand or foot may predict severe disease (120). Abnormalities of the vertebrae ("fish mouthing") are characteristic of sickle cell disease. Hyperplasia of the bone marrow may cause growth disturbances and osteopenia. Osteomyelitis is also more common and may be difficult to distinguish from infarction; radionucleotide imaging and bone aspiration are often necessary to diagnose bone infection. Multiple joints can be involved in septic arthritis caused by *S. aureus, E. coli, Enterobacter,* and *Salmonella.* More often, noninflammatory joint effusions of the knee, ankle, or elbow occur during crises. Chronic synovitis in wrists, metacarpal heads, and calcanei with resultant erosive joint destruction has been reported in children with sickle cell disease.

Avascular necrosis of the femur and, less often, the humeral head and temporomandibular joint can occur in sickle cell anemia (121,122). Avascular necrosis of the weight bearing joints (hip and shoulders) causes chronic pain and may require surgical intervention. Plain x-ray films may not detect early disease, and magnetic resonance imaging may be necessary. Early disease may improve with coring and osteotomy (123). Late disease requires joint replacement. Patients with sickle cell disease have an increased incidence of infection and failure of prosthesis.

Ischemic stroke is one of the most devastating problems in children. The optimal setting for the care of patients with sickle cell disease is a comprehensive center, with a multidisciplinary team to provide ongoing support.

Summary

The management of children and adolescents with chronic rheumatic disease is broad and multidisciplinary (110). Pediatric physiatrists can help provide supportive treatment to children with rheumatic disease by prescribing appropriate pain medications, exercise, bracing, and equipment to maintain or restore age-appropriate function and development. Such treatment can help prevent deformity and contractures; promote normal growth; and maximize physical, psychosocial, and cognitive development in children with rheumatic disease.

BURN INJURIES IN CHILDREN

Epidemiology

Burns can be a devastating cause of morbidity and mortality in children. The American Burn Association estimates that 500,000 burned adults and children require medical attention each year in the United States (124). Twenty percent to twenty-five percent of those hospitalized for burns are between 0 and 14 years of age (125). Males are more than twice as likely to become burned as females (124). Disabled children have a higher incidence of burns than their nondisabled counterparts (126). Almost half of all burns (124) and 80% of contact burns (127) occur in the home. Burns are the third leading cause of unintentional injury death in children 1 to 9 years of age (128). Morality is highest in those who are very young, have larger burn size, develop sepsis, and/or experience inhalational injury (129).

Burn Assessment

Burns can result from thermal, chemical, and electrical exposure. Most burns occur as a result of fire or flame (46%). Scald injury occurs in 32%, hot object contact in 8%, electrical in 4%, and chemical in 3% (124). However, predominant burn etiology varies with age. For example, scald injuries are most common in children under 4 years of age (129,130), and contact burns disproportionately affect children under 5 years of age (127,130). In older children, burns are more likely to occur as a result of playing with matches or other flammable material (130,131). Children, especially boys ages 10–14 years, have the highest injury rate related to fireworks (132).

A burn evaluation must include an assessment of the severity of the burn (Fig. 14–7). One may observe different burn severities within a single injury. Often, the center of the burn is more severely injured than the periphery.

Also critical in evaluating burn severity is an assessment of the amount of body area involved, or percentage of total body surface area (TBSA) burned. Increased mortality is associated with a larger TBSA burned, though survival rates have improved considerably over recent decades. In a study of 1,150 hospitalized children from 1991–1997, those with 0% to 59% TBSA burns had a mortality of 0%, and those with 60% to 100% TBSA burns had a mortality of 14% (133). Standard charts, such as the Lund and Browder charts, are available for estimating burned surface areas for children of various ages (Fig. 14.8). The standard adult "rule of 9s" (9% for each upper extremity and head, 18% for lower extremities and anterior and posterior trunk) applies to adolescents but does not apply to small children, who have relatively larger heads and smaller limbs. Another option is to estimate the palm size of the affected child as 1% of their TBSA although this approach may be less accurate (134). Of all individuals burned, the ABA estimates that over one-third have greater-than-10% TBSA burns and 10% have greater-than-30% TBSA burns (124).

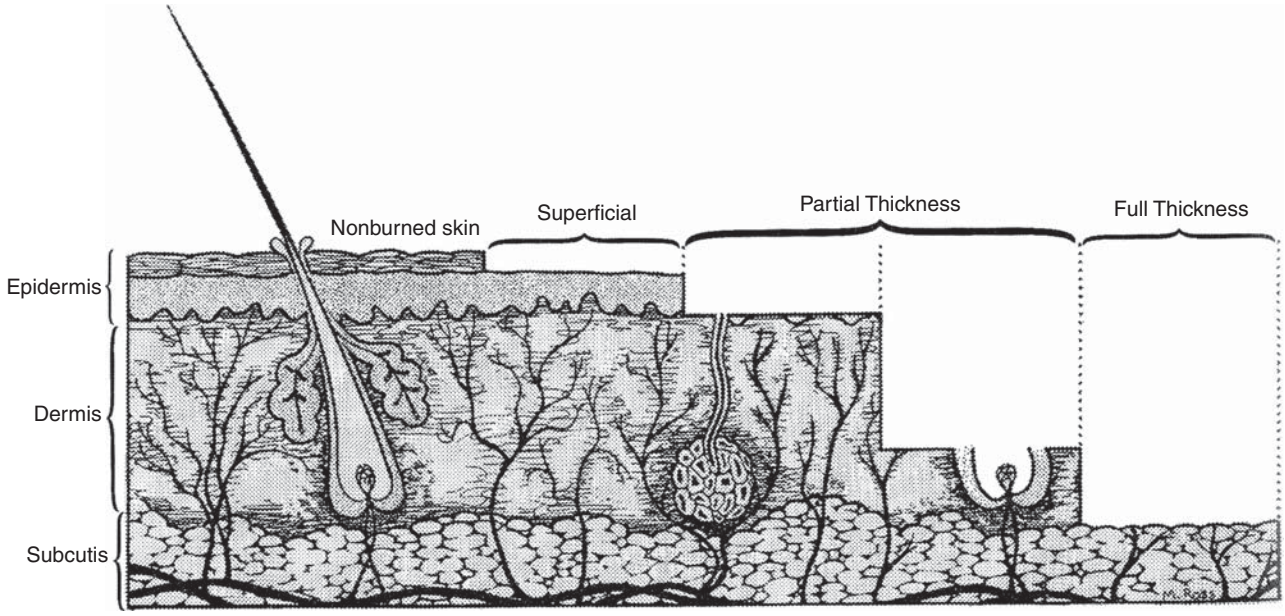


Figure 14.7 Classification of burns by severity.

Acute Burn Management

The decrease in burn morbidity and mortality seen in recent decades is believed to be related to improved acute care and management of individuals with burns. Pain control has been a major area of emphasis in the care of children with burns in the past decade (135). Immediate pain management includes using cool saline-soaked gauze or sheets over burned areas. Medications such as acetaminophen or ibuprofen may be used for smaller burns. Children with larger burns may require opioids such as morphine for pain control and benzodiazepines for sedation. With protracted, painful treatment in severe burns, tolerance to these medications develops, and very high doses may be required (136). More recently, protocols using ketamine or dexmedetomidine for sedation, amnesia, and analgesia have been described as safe and effective (137,138). Nonpharmacologic adjuvants for pain control include distraction, music and art therapy, relaxation, massage, hypnosis, and imagery (135).

Acute burns are cleaned with a mild soap and water. Ice or very hot or cold water should be avoided. Loose skin should be gently debrided. Controversy exists regarding whether blisters should be unroofed, though there is general agreement that needle aspiration should not be performed (134). Tetanus immunization is provided as needed. Intravenous antibiotics are reserved for those with wound infection and sepsis.

Current burn care practice utilizes topical creams, ointments, and/or semiocclusive dressings in order to promote moist healing and rapid epithelialization. Scar symptoms such as pain, itching, and tightening may be reduced with the use of these products, although the effect on ultimate scar appearance is unclear (139). Superficial burns require moisturizer only because intact dermis will protect against infection (134). There is no evidence to support the use of vitamin E cream or topical onion extract ointment in improving scar appearance (139,140).

Partial-thickness burns usually require once- or twice-daily topical antibiotic ointment such as bacitracin and polymyxin in addition to a nonadhesive dressing such as petroleum or bismuth-impregnated gauze. Full-thickness burns, in which infection is a more significant concern, are covered with an antimicrobial cream such as silver sulfadiazine 1% cream, silver nitrate 0.5% solution, or mafenide acetate 0.5% cream (134). Each has advantages and disadvantages. Larger, deeper wounds generally require dressing changes twice per day because of their increased risk of infection (134).

An alternative acute wound management strategy uses synthetic occlusive dressings, human allograft, or pigskin for smaller, partial-thickness burns. They adhere to the wound until epithelization occurs and are trimmed back daily. One author reported successful use of a single application of xenogenic (porcine) acellular dermal matrix for two weeks at a time after initial debridement (141). Other products, such as Acticoat, a silver ion–impregnated gauze in which ions are released over three to four days, allow for only twice-weekly dressing changes. Improved ability to manage pediatric burns on an outpatient basis using this product compared to silver sulfadiazine has been demonstrated (142).

Burns have increasingly been managed in outpatient settings with frequent wound checks or dressing

AGE vs. AREA

Initial Evaluation

UMC.519a, Rev 3.99

Area	Birth 1 yr	1–4 yr	5–9 yr	10–14 yr	15 yr	Adult	2"	3"	Total	Donor Area
Head	19	17	13	11	9	7				
Neck	2	2	2	2	2	2				
Ant. Trunk	13	13	13	13	13	13				
Post. Trunk	13	13	13	13	13	13				
R. Butlock	2½	2½	2½	2½	2½	2½				
L. Butlock	2½	2½	2½	2½	2½	2½				
Genitalia	1	1	1	1	1	1				
R. U. Arm	4	4	4	4	4	4				
L. U. Arm	4	4	4	4	4	4				
R. L. Arm	3	3	3	3	3	3				
L. L. Arm	3	3	3	3	3	3				
R. Hand	2½	2½	2½	2½	2½	2½				
L. Hand	2½	2½	2½	2½	2½	2½				
R. Thigh	5½	6½	8	8½	9	9½				
L. Thigh	5½	6½	8	8½	9	9½				
R. Leg	5	5	5½	6	6½	7				
L. Leg	5	5	5½	6	6½	7				
R. Foot	3½	3½	3½	3½	3½	3½				
L. Foot	3½	3½	3½	3½	3½	3½				
						TOTAL				

Cause of Burn _____

Date of Burn _____

Time of Burn _____

Age _____

Sex _____

Weight _____

Height _____

Date of Admission _____

Signature _____

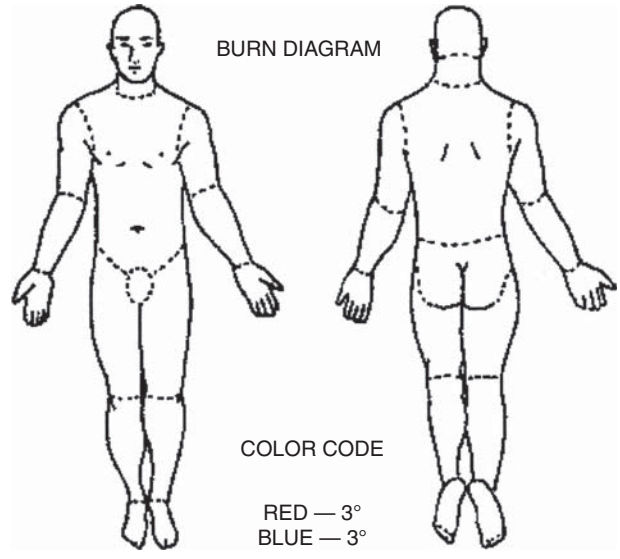


Figure 14.8 Lund and Browder burn chart.

changes (130,143). However, partial-thickness burns of greater than 10 % TBSA, full-thickness burns of greater than 2%, and any circumferential burns or burns of the hands, feet, face, or genitals require inpatient treatment (144). Transfer to a burn center is recommended for anyone with burns greater than 20% TBSA or in children under the age of 10 years with greater than 10% TBSA. In addition, individuals with burns complicated by other trauma, child abuse, or medical comorbidities, or burns caused by chemical or electrical exposure should also be transferred to a burn hospital (134,144). An estimated 125 hospitals have specialized burn centers in the United States (124).

About 10% of burn admissions in children are related to child abuse, and about 10% of all abuse cases include burn injuries (145). Features that should raise suspicion for child abuse include symmetric "dip" injuries of the limbs or buttocks, round cigarette or "dropped ash" burns, and prior history of repeated trauma, report of the child or sibling causing the burn, and accompaniment of the child by someone other than the parent (144). An investigator's checklist is available for use in suspected cases of deliberate burn injuries of children (145).

Chronic Burn Management

Whether undergoing ambulatory or inpatient management, rehabilitation is critical in achieving improved outcomes. Gait and mobility training with gait aids may be necessary. Other equipment and adaptive aids may help children and adolescents achieve increased independence in self-care skills. Range-of-motion and stretching exercises of areas affected by burns must begin in the acute care phase in order to help prevent contracture formation. Positioning to promote functional range of motion and prevent contractures is also important. Splints and other custom-molded orthotics are occasionally necessary to further advance this goal (146,147). Pillows, pads, and other bed-based apparatus may be helpful as well. Optimal positioning based on area burned is summarized (Table 14.2).

Children with severe burns may develop low bone density and an increased risk of long bone fracture due to prolonged immobilization, nutritional deficit,s and an alteration of the hormonal milieu (148). Intervention to improve bone density includes mobilization and improved nutritional intake of calcium and vitamin D (149). Treatment with growth hormone for the year following hospitalization in severely burned children has been observed to improve lean body mass, height, weight, strength, cardiac function, and bone mineral content. It is hypothesized that this may lead to a higher daily activity level and result in decreased contracture formation (150).

14.2 Positioning of the Pediatric Burn Patient

AREA INVOLVED	CONTRACTURE PREDISPOSITION	CONTRACTURE PREVENTING POSITION
Anterior neck	Flexion	Extension, no pillows
Anterior axilla	Shoulder adduction	90° abduction, neutral Rotation
Posterior axilla	Shoulder extension	Shoulder flexion
Elbow/ forearm	Flexion/pronation	Elbows extended, forearm supination
Wrists	Flexion	15–20° extension
Hands MCPs Ips Palmar burn	Hyperextension Flexion Finger flexion, thumb opposition	70–90° flexion Full extension All joints full extension, thumb radially abducted
Chest	Lateral/anterior Flexion	Straight, no lateral or anterior flexion
Hips	Flexion, adduction, external rotation	Extension, 10° abduction, neutral rotation
Knees	Flexion	Extension
Ankles	Plantarflexion	90° dorsiflexion

MCPs, metacarpophalangeals; IPs, interphalangeals.

Strength and endurance exercises are important and are facilitated by a physical or occupational therapist. Studies reveal that children with burns who participated in a resistance exercise program over 12 weeks had improved muscle strength, power, lean body mass (151), and pulmonary function (152) compared to those who participated in a standard rehabilitation program without exercise. There was also a decreased need for surgical release of burn contractures (153).

Mustoe and colleagues published clinical guidelines regarding scar management based on systematic literature review and expert consensus (154). The importance of prevention of hypertrophic scarring and keloids was emphasized. When a symptomatic scar has developed, the recommended treatment depends on scar classification (Table 14.3, Fig. 14.9).

Pressure garments have been the mainstay of scar treatment for decades. Proposed mechanisms of action include decreasing collagen synthesis by decreasing blood flow and realigning collagen bundles already present. A decrease in hypertrophic scar

14.3 Burn Scar Classification

Mature scar: A light-colored, flat scar

Immature scar: A red, sometimes itchy or painful, and slightly elevated scar in the process of remodeling. Many of these will mature normally over time and become flat, and assume a pigmentation that is similar to the surrounding skin, although they can be paler or slightly darker.

Linear hypertrophic (e.g., surgical/traumatic) scar: A red, raised, sometimes itchy scar confined to the border of the original surgical incision. This usually occurs within weeks after surgery. These scars may increase in size rapidly for 3–6 months and then, after a static phase, begin to regress. They generally mature to have an elevated, slightly rope-like appearance with increased width, which is variable. The full maturation process may take up to 2 years.

Widespread hypertrophic (e.g., burn) scar: A widespread red, raised, sometimes itchy scar that remains within the borders of the burn injury.

Minor keloid: A focally raised, itchy scar extending over normal tissue. This may Develop up to 1 year after injury and does not regress on its own. Simple Surgical excision is often followed by recurrence. There may be a genetic Abnormality involved in keloid scarring. Typical sites include earlobes.

Major keloid: A large, raised (>0.5 cm) scar, possibly painful or pruritic and extending over normal tissue. This often results from minor trauma and can continue to spread over years.

formation reduces the incidence of contractures as well as pain and itchiness (155). Pressure over scars may be achieved through traditional off-the-shelf or custom pressure garments. Care should be taken to avoid wound irritation from seams or zippers. Pressure is usually initiated after wound closure, and garments are worn 23 hours per day for 6–24 months (144), or until scar maturation. Garments should be replaced every 6–12 weeks in order to maintain compression. Over time, garment alterations may be necessary due to limb size fluctuation and growth of the child. The pressure required for effective treatment has not been scientifically established (155). Some have suggested 24–35 mmHg (154), while others have described improvement with pressures as low as 15 mmHg (156). Complications such as discomfort and skin breakdown occasionally result from the use of pressure dressings. High pressures may cause harm such as obstructive sleep apnea (157) or skeletal and dental deformity in children (158).

It is important to note that although pressure dressings are routinely used to prevent and treat burn scars, little scientific evidence exists to support their use (139,154,159). In fact, one prospective, randomized trial of the efficacy of pressure garment therapy showed no significant differences in burn outcome parameters using the Vancouver Burn Scar Assessment between one group that used pressure garments and another that did not (160).

There is some evidence to support the use of silicone dressings to prevent hypertrophic scars in those at risk as well as to improve scar elasticity in already existing symptomatic scars (139,161). Vigorous scar massage may help to keep scars supple. Massage techniques are performed by a skilled therapist and are taught to the patient or family. Exercise and scar modification techniques should continue over the 12–24 months necessary for scar maturation (144).

Additional management options for keloids and recalcitrant hypertrophic scars include the injection of triamcinolone (154). Topical steroids and vitamin E creams have not been successfully used for scar treatment (162). Radiotherapy, laser therapy, and cryotherapy have been used with mixed results (154).

In the acute period, a surgical referral should be made if the burn wound has not healed within a week. Skin grafting may be considered, with the goal of preventing later scar complications (144). Mature scars require surgical intervention when functional loss or cosmesis can be restored or improved. Later, when a hypertrophic scar, keloid, or contracture has developed, surgical interventions include scar excision, skin grafts, reorientation of scars using flaps, W- or Z-plasties, contracture release, and use of skin substitutes or tissue expansion. Surgical excision of hypertrophic scars and keloids without additional treatment results in a high rate of recurrence. Similarly, surgical correction of equinus contractures using the Ilizarov method resulted in recurrence rates of approximately 70% in younger children (163). However, surgical excision in combination with the use of adhesive microporous hypoallergenic paper tape, silicone gel sheeting, steroid injection, or even radiation therapy may decrease the likelihood of recurrence (154).

Outcome

The most common complication for burn survivors is abnormal or hypertrophic scarring, though the actual prevalence is unknown (164). Abnormal scarring may cause contractures and impaired function. Scar contraction may lead to growth restriction in a child, with resultant distortion of anatomical features and disfigurement. Based on a Medline review of 50 studies related to functional outcomes after burn injury, limited range of motion was reported in 0% to 5% of children with minor burns (mean TBSA 6%) and 47% with massive burns (>80% TBSA). One-third of the children with massive burns were dependent on others for assistance for activities of daily living years after

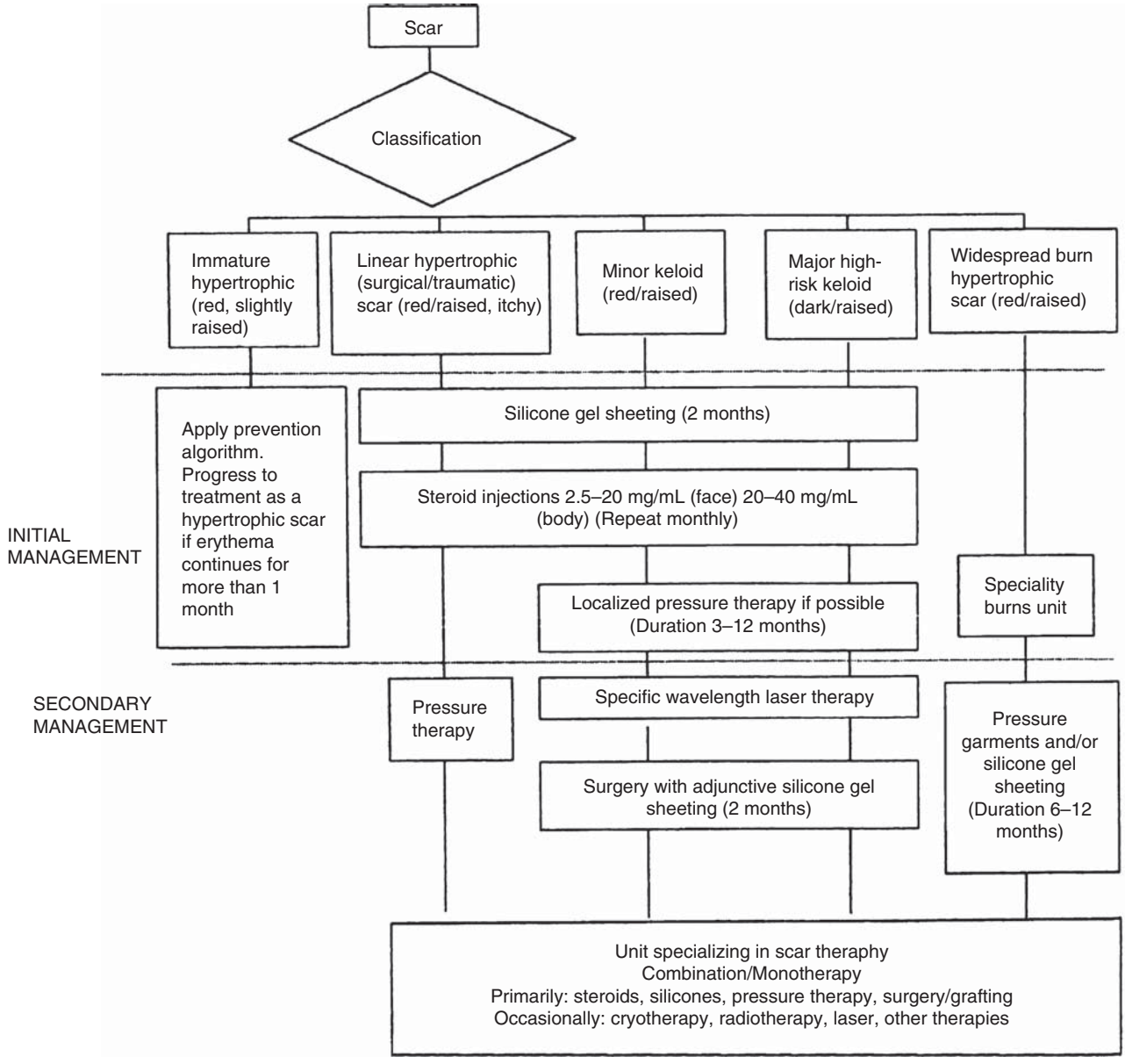


Figure 14.9 Complete burn management algorithm.

injury. It was felt that insufficient data exist to fully describe the burden of burn injuries (165). In a study of adult survivors of massive burns, quality of life was comparable to the general population. The strongest independent predictors of physical quality of life were size of full-thickness injury and hand function. Mental quality of life was best predicted by age at the time of injury (with younger age predicting a better quality of life) and perceived social support (166).

Following a burn injury, children have an increased incidence of psychological disturbance (167) and difficulty with behavior (165). Significantly higher levels of anxiety, phobias, and enuresis have been noted in this population, and 30% met criteria for post-traumatic stress disorder (PTSD) within six months of their burn (168). Pain, separation anxiety, and acute dissociation have been found to contribute to the development of PTSD (169). Feelings of depression and misery were reported by 79% of children with burns in another study (167). Self-esteem and confidence may decline. Fourteen percent to forty-three percent of individuals report dissatisfaction with appearance after a burn (165). Social reintegration may be difficult and social isolation prevalent, with a third having symptoms of antisocial disorder (167). One-fourth to one-third of children with burns experienced interference in playing with other children or seeing friends (165). Return to school and community activities may be eased by education provided to the child's peers and teachers prior to their return.

Prevention of Pediatric Burns

Public education campaigns have played a key role in reducing the incidence of childhood burns in recent years. An example of this is children's familiarity with the "Stop, drop, and roll" practice taught by firefighters around the country. Other tactics related to reducing burn incidence include reducing hot water heater temperatures to 120 degrees, turning pot handles to the back of the stove, using back burners on the stove, keeping irons off the floor, using smoke detectors and outlet covers, and proper storage of chemicals. Public educational materials related to fire safety are available through the Centers for Disease Control (CDC) and American Burn Association (124,170).

DEVELOPMENTAL CONDITIONS

Legg-Calvé-Perthes disease is osteonecrosis of the capital femoral epiphysis in children (11). The condition usually presents between the ages of 4 and 10 years, with a peak incidence of 5 to 7 years. Presentation has been seen as early as 2 years and as late as the late teens. There is a definite male preponderance, with a 4:1 ratio. The incidence of bilaterality has been reported as 10% to 12% (171,172), rarely simultaneous. There is no evidence that the condition is inherited. Limitations in internal rotation, extension, and abduction of the effected hip, with slight shortening of the leg, are common physical findings. Children presenting with knee pain always require a thorough examination of the hip, as this is a common referral pattern. Catterall classification (173) is graded over four degrees of involvement, depending upon the extent of necrosis across the capital femoral epiphysis. A Catterall classification I involves up to 25% of capital femoral epiphysis involvement; classification II, 25% to 50%; classification III, 51% to l<100%; and classification IV, 100% involvement. Bone scanning and MRI may provide the diagnosis before radiographs. Arthrography can be helpful in defining classification grades and extent of involvement of the capital femoral epiphysis (6). Laboratory tests include erythrocyte sedimentation rate, C-reactive protein, and white blood cell counts, often normal unless concurrent illness is present. Controversy exists about whether treatment of any type affects the natural history of the disorder, particularly when onset occurs under the age of 6. The short-term goal is reduction of pain and stiffness of the hip. The disease process is self-limited and may last for two to four years. NSAIDs are effective in reducing synovitis. Restriction of activity helps relieve pain, which at times may include non-weight bearing with crutches. Abduction orthosis and casting may be helpful at some point, with improved abduction and femoral head containment the goal. For abduction orthosis to function satisfactorily, the affected hip must be able to be abducted in extension to 40 or 45 degrees. After the orthosis is applied, an anterior-posterior (AP) radiograph of the pelvis is obtained to ensure that the affected femoral head is contained within the acetabulum. To be effective, the orthosis must be worn full-time. The use of the orthosis is continued until subchondral reossification is demonstrated on the AP radiograph (174,175). The important principal of treatment is based on femoral head containment within a spherical acetabulum so that at least theoretically, reossification is also spherical. Generally, the active phase of the disease that requires an orthosis is 9 to 18 months. Patients with Catterall classifications I or II can be treated conservatively, while levels III or IV often require more surgical intervention. Surgery, including proximal femoral varus osteotomy, may eliminate longer-term bracing and allow earlier resumption of activities in some children. The prognosis again is better with earlier detection under the age of 8 years and with less than 50% involvement of the femoral head. With increased involvement of the lateral femoral head, more extensive surgical options may be chosen. Femoral osteotomy can result in elevation of the greater trochanter accentuating the abductor dysfunction. Patients older than 9 years of age at onset with Catterall groups III and IV have unpredictable success rates, regardless of treatment methodology. Return to high-impact athletics is restricted until a pain-free status is found during clinical examination and radiographs show healing. Osteoarthritis later in life is often seen, with 50% of untreated patients showing severe changes by the age of 50 years (176). Other causes of avascular necrosis always need to be considered, including sickle cell anemia, femoral neck fracture, Gaucher's disease, slipped epiphysis, congenital hip dislocations, rheumatoid arthritis, and other collagen disorders, not to mention steroid therapy. Bilateral involvement may be confused with multiple epiphyseal dysplasia or spondyloepiphyseal dysplasia and can be differentiated by doing a skeletal survey. Acute transient synovitis of the hip (ATS) is the most common cause of hip pain in children and can present in a fashion similar to Legg-Calve'-Perthes disease (6). In fact, a transient ischemia may occur during acute synovitis of the hip with some rare reports of progression to Legg-Calve'-Perthes disease at a later date. The condition may develop at any time from toddler age onward, with a peak age between 3 and 6 years, and slightly more common in boys. At least half of the children with ATS have or recently have had an upper respiratory illness,

including pharyngitis or otis media. Trauma of a mild nature is frequently present. Annual hospital admissions for the diagnosis of ATS are reported between 0.4% to 0.9% (6,10,177). The actual incidence of ATS is likely higher, however, as many patients never seek medical attention, and a minority of patients are hospitalized once the diagnosis is made. A lower incidence in African Americans has been noted (178). Ninety five percent of the cases are unilateral. Annual risk of recurrence for a child with an affected hip is 4% (179). Viral etiologies are suspected. Common presentations include rapid onset of limping, unilateral pain involving the hip or groin with referral to the knee, and refusal to bear weight on the involved extremity in an otherwise healthy child. A low-grade fever may be present related somewhat to an associated upper respiratory infection. Septic arthritis needs to be excluded, as this is a much more serious joint- and limb-threatening condition. Radiographs are frequently reported as normal when compared to the opposite side, but may show some slight intracapsular effusion. Ultrasonography remains most helpful in detecting effusion (180) and may correlate with MR imaging and a positive radionuclear bone scan. ATS remains a diagnosis only after other conditions have been excluded. Laboratory evaluation may show normal to mild elevation of the white blood cell count and erythrocyte sedimentation rate along with the C-reactive protein. Other laboratory parameters are generally within normal limits. Upon hospital admission, aspiration of the hip joint should be performed if septic arthritis is suspected. Long-term follow-up of children with ATS has demonstrated some lifelong abnormalities, including increased coxa magna (defined as an enlargement of 2 mm or more of the proximal femoral epiphyses) in one-third of patients (181). A reactive increase in the blood supply to the femur with increased growth of the articular cartilage secondary to the transient inflammation may be associated with this finding (182). Long-term changes of degenerative arthritis in the hip have been reported in some individuals as well. The fundamental treatment consists of rest and age-appropriate NSAIDs. Partial weight bearing with crutches can begin with improvement in pain and full range of motion through the hip. Most patients will resolve of symptomatology within five to seven days, and recurrence is uncommon unless premature activity occurs. Persistent symptoms should be reevaluated, realizing that low-grade symptomatology can last, in some, up to several weeks. Full, unrestricted activity should be avoided until the hip is completely pain-free and there is no evidence of limping.

A slipped capital femoral epiphysis (SCFE) usually involves posterior inferior displacement of the epiphysis on the proximal femoral metaphysis. The term slipped capital femoral epiphysis is actually a misnomer. More accurately, the epiphysis remains in normal position within the acetabulum while the proximal femoral neck and shaft move anteriorly and rotate externally relative to the femoral head (183). The incidence of SCFE in the literature can vary between 1 and 61 per 100,000 persons (11). SCFE is approximately twice as common in boys than in girls and may be bilateral in up to 25% of cases, 5% of which occur simultaneously (6,184). More than 90% who develop late SCFE on the contralateral side are asymptomatic. Affected children are often large and overweight, and an association with endocrine factors such as hypothyroidism, hypopituitarism, hypogonadism, and excessive growth hormone has been reported. Body mass index (BMI) may be an accurate tool for assessment of risk for SCFE (185). Findings show that patients with SCFE had a statistically higher BMI during growth than normal developing children. It is more common in African American boys with accumulated risk that may be as high as 1 in 400 (11). It is also more common in northern environments, possibly related to less sun exposure and relative vitamin D deficiency. Laboratory studies have demonstrated that estrogen strengthens and testosterone weakens the physes (186,187). SCFE may be thought of as occurring because of physiologic loads across an abnormally weak physes (more common in peripubertal children) or abnormally high loads across a normal physes (more common in obese children). The increased prevalence of hypothyroidism in children with Down's syndrome is a likely explanation for the increased risk of this condition in these children (188,189). Mechanical factors appear important with an association of SCFE, with decreased femoral anteversion and femoral neck shaft angle (190). Age at presentation is typically between 12 to 16 years for boys and 10 to 14 years for girls. Presentations of SCFE outside of these age ranges should alert physicians to potential endocrinopathy or alternative conditions such as renal osteodystrophy. The two most common features of presentation are pain and altered gait. The pain may come on acutely (unstable SCFE), but more commonly builds over a number of weeks or months. As usual with hip pathology in children, pain occurs in the groin region and radiates to the knee and medial thigh. It is aggravated by walking and other high-impact activities. External rotation of the leg is common with some shortening and antalgic Trendelenburg's gait. Physical examination demonstrates a loss of internal rotation, diminished flexion, shortening of the leg, and atrophy of the thigh if the symptoms have been longstanding. Mild slips show displacement of the epiphysis up to one-third of width of the metaphysis, moderate slips up to two-thirds, and severe slips greater than two-thirds displacement. The displacement is best quantified on lateral radiographs,

which should not be lacking in the workup of a child with hip pain. Klein's line (or Trethowan's sign) is a line drawn along the superior surface of the femoral neck, which normally should pass through the lateral portion of the capital femoral epiphysis. If this line passes above the epiphysis, at least minimal slippage is present and further intervention required (191,192). Slip angle is another good way of measuring degree of severity on a true lateral radiography (193). A perpendicular line drawn from the base of the capital femoral epiphysis bisecting a line drawn through the midshaft of the proximal femur is measured. An angle less than 30 degrees is mild slippage, 30 to 60 degrees moderate, and 60 to 90 degrees severe. When SCFE is suspected, ambulation should not be allowed until an orthopedic surgeon sees the child. Other radiographic features include widening of the epiphyseal line (Salter I fracture-type appearance) with metaphysial changes including rarefaction and cysts. Bone scan and MRI can be helpful in determining the preslip stage as compared to the opposite uninvolved side (11). The current standard of treatment for SCFE is situ two cannulated screw fixation done on an urgent basis. The goal of treatment is to arrest further progression of the slip and to gain closure of the capital femoral epiphysis. Management of patients with unstable SCFE can involve minimal repositioning by an experienced orthopedist and two-screw fixation instead of one. Generally, the epiphysis is left in its displaced position because avascular necrosis is a 10% to 25% risk if manipulation is attempted. Spontaneous reduction of the slippage or controlled reduction by an experienced orthopedist under fluoroscopic guidance has not been associated with an increased rate of osteonecrosis in patients with unstable SCFE (11). Cortical bone grafts have also been used, crossing from the metaphysis to the epiphysis and resulting in epiphysiodesis. Spica casting is becoming a less common practice because of secondary complications in obese children and immobility for up to three months. After successful physeal closure, the proximal femur can remodel, particularly in children under the age of 10 years. Bony osteotomies can be indicated if further femoral head coverage is required despite more conservative care. Chondrolysis or acute cartilage necrosis may occur postoperatively in severe cases. If chondrolysis is present, most individuals go on to develop narrowing of the joint space with some degree of ankylosis, degenerative arthrosis, and pain. Total hip arthroplasty can be a consideration for older individuals. Weight bearing is generally avoided for at least six weeks after surgery followed by active assistive exercises and strengthening to restore lengthening, adduction, and internal rotation. Full identification of this condition while only minimal displacement is present and immediate surgery generally allow rapid mobilization and return to full activity with no sequelae. Prophylactic pinning of the contralateral hip is an area of ongoing discussion. In one recent study of 94 hips treated with prophylactic pinning, there were no significant complications (194). The risk of osteonecrosis and chondrolysis was felt to be virtually negligible when using insight two-screw fixation with improved imaging technology and radiolucent tables. Opponents of prophylactic pinning cite the complications and potential risks of pinning numerous hips that will never slip. They also point out that with appropriate patient counseling and close follow-up, most subsequent slips will be detected early while they are still mild and treatable. Currently, prophylactic contralateral hip fixation is recommended for patients with established metabolic or endocrine disorders, those with increased risk from radiotherapy or chemotherapy, and for children with SCFE who are younger than 10 years of age. Once the triradiate cartilage is closed (around the age of 14 to 16 years) and when Risser lines appear, the risk of contralateral slip is felt to be negligible (11). SCFE differs from other pediatric disorders of the hip such as Legg-Calvé-Perthes disease and developmental dysplasia of the hip, in that SCFE occurs at an age when the majority of the acetabulum has been developed and thus less acetabular adaptation to deformity of the femoral head can occur. All of this speaks to early detection and early treatment, particularly in those children of elevated risk.

Developmental dysplasia of the hip (DDH) is the most common disorder of the hip in children and the musculoskeletal condition, causing the highest level of concern for the pediatric practitioner (11). Dysplasia of the hip (mostly involving the acetabulum) occurs in approximately 1 in 100 births, with frank dislocation in approximately 1.5 births per 1,000. DDH is not always detectable at birth, and thus the term "developmental" rather than "congenital" has been chosen by the Pediatric Orthopedic Society of North America. The term DDH is felt to more accurately reflect the variable presentation of this complex disorder. Dysplasia refers to an underdeveloped acetabulum, subluxation to hip still in partial contact, with the acetabulum and dislocation to femoral head not contained in the acetabulum. The dislocated hip should be detectable clinically in the newborn period by four to six weeks. Teratologic hip dislocations (atypical) occur in utero and are not reducible on neonatal examination. Atypical dislocations are present about 10% of the time and are more commonly associated with other chromosomal or neuromuscular conditions, such as myelomeningocele, arthrogryposis, or Ehlers-Danlos syndrome. Typical DDH occurs in an otherwise normal infant and may take place in utero, perinatally, or postnatally (Table 14.4). Risk factors associated with DDH are listed in Table 14.5. DDH predominates

14.4 Classification of Developmental Dysplasia of the Hip

CLASSIFICATION	DESCRIPTION
Atypical (10%) or teratological	Primarily malformed acetabulum or femoral head in utero associated with myelomeningoceole, arthrogryposis, Ehler-Danlos or other syndromes
Typical (90%)	Otherwise normal infant but varying degrees of hip morphology and placement
Subluxed	The femoral head and the acetabular cartilage are in contact, but not correctly centered
Dislocatable	The femoral head can be dislocated with maneuvering
Dislocated	The femoral head is completely out of the acetabulum

14.5 Risk Factors Associated With Developmental Dysplasia of the Hip

Caucasian
Hip swaddling in extension (Native American, Lapland)
Female: Male ratio 6:1
Breech birth
Positive Family History
Primaparity
Ligamentous laxity
High birth weight (>4000gm)
Congenital muscular torticollis
Metatarsus adductus
Oligohydramnios
Hip asymmetry (limited abduction of one or both hips)
Congenital knee dislocation/recurvatum.

in the left hip (60%), but often bilateral involvement can be discovered. Bilaterality can be most difficult to diagnose with the absence of asymmetry as a helper. Beware of bilateral DDH when thigh skin folds extend past the anus and decreased absolute abduction is present on both sides (6). In the older child, bilateral involvement may be detected only by hyperlordosis and a waddling gait. First-born females presenting with breech have the highest risk for DDH at 8% (11). Risks for DDH in subsequent pregnancies is 6% when neither parent has a positive history and 12% when one parent is with positive history. The presence of idiopathic clubfeet do not obligate special screening (195), but this may be helpful in a small percentage. Ultrasound screening of newborn hips continues to be with some controversy (196). The technique is sensitive for detecting abnormalities of the newborn hip, but has poor specificity in detecting patients with DDH who require treatment. Cost-effectiveness has yet to be shown. Neonatal hips with immaturity or mild dysplasia that have no instability do not benefit from early treatment, as more than 95% of such hips spontaneously normalize (197,198). Certainly infants with risk factors (see Table 14.5) need to be screened in ultrasound followed by careful clinical examinations until the child reaches walking age. The alpha angle is measured from the vertical reference through the iliac bone and tangential to the osseous roof of the acetabulum. This angle represents the hard bony roof and reflects acetabular depth (11). The beta angle is created by the vertical reference through the iliac bone, intersecting with a line drawn through the labrum representing the cartilaginous roof of the acetabulum. The beta angle indirectly reflects the lateral position of the femoral head. A normal alpha angle is greater than or equal to 60 degrees and a beta angle less than 55 degrees (199). DDH classification is based on the Graf method (200), with varying severity having type I, a normal hip and type IV, a severely affected dislocated hip. After 4 to 5 months of age, when the ossific nucleus of the femoral head has generally appeared, radiographic screening replaces ultrasound in evaluation of infants with DDH. Parameters for monitoring hip dysplasia in this age group are represented in Figure 14.10. Measurement of center-edge angle becomes useful in the patient who is more than 5 years of age and particularly useful in the adult patient (201). Center-edge angles are less than 20 degrees (angle between a vertical line drawn through the center of the femoral head intersecting with the tangential line drawn through the lateral acetabular edge) are concerning for unwanted lateralization of the femoral head. Clinical examination with repetitive follow-up continues to be the mainstay of diagnosis for DDH (6). The infant should be quiet and comfortable so the muscles about the hip are relaxed and supple to exam. In early infancy, instability is the most reliable sign (193). Instability declines rapidly with age, over 50% within the first week. Stiffness, shortening, and limited abduction become much more prominent by 2 to 3 months of age. Initial instability may be the result of maternal or fetal hormonal laxity, genetics, and intra- or extrauterine malpositioning. The longer the femoral head remains in a subluxated or dislocated position, the more likely progressive change in acetabular anatomy will occur. A hip that is reduced at rest but subluxated or dislocated by adduction, flexion, and posterior pressure has a positive Barlow's maneuver. Concurrent acetabular dysplasia may or may

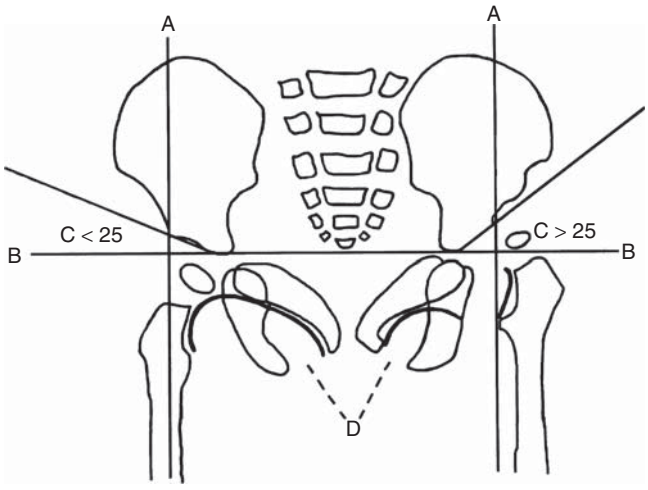


Figure 14.10 Radiographic evaluation in developmental hip dislocation. (A) Perkin's vertical line: perpendicular dropped from the lateral acetabular margin. (B) Hilgenreiner's line, through the Y cartilages. The femoral head should lie in the lower medial quadrant formed by the intersection of the two lines. (C) Acetabular index: the angle formed by a line through the acetabular roof and Hilgenreiner's line; normal below 25 degrees. (D) Shenton's line: the arc appears broken in the presence of dislocation. The abnormal hip appears on the right.

not be present (197,198). Barlow tests often become negative by 2 to 3 weeks of age as maternal or fetal hormonal influences diminish. Hips that are dislocated can be reduced back into the acetabulum by abduction and forward lifting of the thigh producing a palpable "clunk." A hip that is reduced this way has a positive Ortolani's sign and is often accompanied by acetabular maldevelopemnt. Hip "clicks" are short-duration, high-pitched sounds that are common, benign, and need to be distinguished clearly from "clunks." "Clicks" and asymmetrical thigh folds are common in normal infants and generally benign (193). A positive Galeazzi's sign may be seen in infants with DDH, noting a decrease in height of the involved knee with the hips flexed supine to 90 degrees. In infants older than 3 months, limitation of motion and apparent limb shortening predominate. The dislocated or subluxed hip develops tightness in the adductor muscle groups with limited asymmetric abduction. Again, this is much easier to detect when unilateral than bilateral. Parental or family reports of an infant with unusual positioning of legs or crawling warrants investigation. In older ambulatory patients, Trendelenburg's limp, waddling gait, and hyperlordotic posture require evaluation. Fatigue, pain, and instability can still occur in adolescence.

For the infant with a positive Barlow's sign and normal ultrasound at 4 to 6 weeks (no evidence of instability on stress maneuvers) with clinical stability returned, no treatment or radiograph follow-up is recommended (11). Serial clinical examinations of the hip should continue by the primary care physician until the child reaches walking age.

For children with dysplasia and an abnormal ultrasound or persistent subluxation, treatment is with the Pavlik harness. Follow-up clinical examinations should be completed at least every two weeks, with serial ultrasound studies at least monthly. The Pavlik harness needs to be adjusted at least every two weeks for the rapid growth evident in this young infantile population. Failure to adjust the Pavlik harness can cause additional acetabular pathology, including a now-dislocated hip that was previously reduced. The anterior adjustable straps for the Pavlik harness are set to keep the hips flexed at approximately 100 degrees. Excessive flexion and tightening needs to be avoided, as additional problems can be caused such as femoral nerve palsy. The posterior straps are meant to encourage gentle abduction of approximately 45 degrees. They should be loose enough to allow two to three fingerbreadths between the knees when the knees are held flexed and adducted. Forced abduction should be avoided to minimize any complication of osteonecrosis. The child can be weaned from the Pavlik harness over a three- to four-week period once ultrasound parameters become normal along with stability on clinical examination. Treatment with the Pavlik harness for neonatal acetabular dysplasia is more than 90% successful. Follow-up is still required with AP radiographs through the growing years, with a 10% risk of deformity necessitating clinical follow-up into adolescence. Fixed hip abduction orthosis replaces the Pavlik harness in children over 6 months of age, generally because of strength and size (202). Early screening and repetitive clinical examinations have been shown to significantly reduce surgical procedures and hospitalizations, including late presentation of DDH in this population (203,204,205).

A hip with reducible dislocation (Ortolani's sign positive) may still be initially treated with the Pavlik harness under 2 months of age. Ultrasound imaging is required to document hip centering over the acetabulum once in the harness (206). Clinical and ultrasound follow-up is weekly, with a careful clinical examination at three weeks, prior to further decision-making. If the hip is not reduced, continuation of the harness until normal exam, ultrasound, or radiographs have been obtained can occur (206). If instability is present on exam but reducible, a fixed abduction brace or spica casting should be considered (6). If the hip is not reduced, traction, adductor tenotomy, or closed or open reduction including arthrogram and spica casting need to be considered (201). Treatment with the Pavlik harness is effective in achieving reduction of a reducible hip in 85% of patients with a low incidence

of osteonecrosis (less than 5%) with early treatment (11). Persisting with the Pavlik harness when reduction is not achieved by three to four weeks may cause additional femoral head deformity or posterior fixation, and make closed reduction difficult or impossible (193). The Pavlik harness is not appropriate for teratologic dislocation.

The preferred method of treatment in children 18 months of age or younger is closed reduction, provided it can be achieved without undue force (11). The preliminary use of traction for three to four weeks before attempting closed reduction is becoming less common (207,208). The quality of reduction is confirmed by arthrography and objectively defined by the width of the contrast column remaining between the femoral head and the acetabulum, including the status of the limbus. Stability of the reduction needs to be assessed in all planes by determining the stable zone and safe zone. The stable zone is defined as a difference between the maximum abduction of the hip and the minimal amount of abduction before hip dislocation (6). The safe zone is the range of motion where the hip is safe from excessive abduction that can cause osteonecrosis and adduction that may facilitate dislocation. The safe zone is generally 15 degrees less on the limits of motion defined in the stable zone. With the stable and safe zone in mind, reduction is maintained in a bilateral hip spica cast (202). The hips are generally maintained in about 0- to 100 degrees of flexion, with abduction less than 60 degrees. Reduction of the hip or hips in the spica cast must be confirmed, usually by CT scan (6). Immobilization in the spica is continued for approximately three to four months with cast changes about every six weeks. With clinical stability achieved and visualized on radiographs, abduction bracing can be used subsequently until a normal acetabulum is achieved.

When treatment with the Pavlik harness and/or closed reduction fails, surgical reduction is required, more commonly after the age of 18 months (206). Often, the decision to perform open reduction is made in the operating room following arthrography and failed closed reduction. The purpose of open reduction is to remove obstacles to reduction, achieving increased stability and clinical outcome. Intraoperative arthrography can be helpful in defining specific anatomic blocks to reduction and choosing the best surgical approach. Obstacles to reduction include the iliopsoas tendon, which is interposed between the femoral head and acetabulum and often must be released. Capsular constriction needs to be released in addition to the transverse acetabulum ligament when blocking a deep concentric reduction (193). Pulvinar fatty fibrous tissue filling the depth of the acetabulum is removed with a rongeur. The ligamentum teres, when hypertrophied, is usually removed, the vascular contribution of which is felt to be minimal. The limbus is generally left alone, as it will remodel and form the labrum postoperatively, an important structure for hip stability and longevity (6). Numerous pelvic and femoral osteotomies are available, with choice based on the pathology and the experience in surgical preference (206). Femoral shortening can relieve pressure on the femoral head and acetabulum, reducing cartilaginous pressure and the risk of osteonecrosis (209,210). Derotational femoral osteotomy can be helpful if excessive anteversion is present requiring extreme internal rotation to maintain reduction. Secondary producers, including redirectional femoral and pelvic osteotomies, are more common after the age of 2 in an effort to maintain concentric reduction and minimize the risk of osteonecrosis. Remodeling of the hip and acetabulum is most predictable under the age of 4, less predictable between the ages of 4 and 8, and unpredictable after the age of 8. Secondary procedures should be performed if at all possible prior to the age of 8 for best outcome (206). Failure to achieve reduction in the older child results in a permanently subluxed hip with marked gait deviation and susceptibility to osteoarthritis and pain syndromes. Long-term outcomes can include joint arthrodesis and the need for total hip arthroplasty in the younger adult (6). Spica casts applied after surgery need appropriate infantile care for hygiene, toileting, and positioning and mobility devices such as scooters, carts, and accessible toys. The importance of early diagnosis and treatment of developmental hip dysplasia cannot be overemphasized: The results are generally good with appropriate intervention and disastrous if neglected.

Traumatic hip dislocations in children are relatively rare, and when they occur, they are usually posterior (11,211,212). The mechanism is usually traumatic, with a direct blow to the knee with hip and knee flexed, as occurs with a fall during ground impact or dashboard contact injury in a car accident. Some dislocations have occurred during mini rugby, in which players kneeling on the ground have had someone fall on top of them. Avascular necrosis may occur in up to 10% of cases. Sciatic nerve palsy is rare, but needs to be ruled out. Only 5% of all traumatic hip dislocations occur in patients younger than 14 years. Males account for approximately two-thirds of these dislocations, with more than 99% being unilateral. Posterior hip dislocation is an emergency that requires immediate referral to an orthopedic specialist (6).

Overuse syndromes are generally conditions caused by unresolved submaximal stress in previously normal tissues. They involve microtrauma resulting from chronic repetitive insults to the musculoskeletal system. With focus on single sports early in life, these injuries have become more prevalent in the pediatric athlete (6,193). Growth cartilage seems to

be more susceptible to stress and overuse than adult cartilage. Growth cartilage is present at three different sites: the physes, the joint surface, and the major muscle–tendon insertions or apophyses. Little League elbow comprises a group of pathologic entities in and about the elbow joint in young developing pitchers. The injury may include medial epicondular fragmentation and evulsion, osteochondritis of the capitulum or radial head, and delayed closure of the growth plates around the elbow (213). The mechanism of injury appears to be repetitive valgus strain applied to the elbow by throwing (214). Guidelines for young pitchers include participation in only three to four innings per game, fewer than 90 pitches per outing, fewer than 200 pitches per week and mandatory rest periods between appearances (11). Stress injuries to the distal, radial, and ulnar physes are commonly found in gymnasts (6,214). X-rays demonstrate widened epiphyses, cystic changes, and beaking of the distal metaphysis (215, 216). Some risk of distal, radial, and ulnar growth arrest exist. Overuse injuries around the pelvis and hips are common and may be seen along the iliac crest; ischial tuberocities; and anterior, superior or inferior iliac spine. Sometimes, late diagnosis of the avulsion of the ischial tuberocity is mistaken for an osteosarcoma. An avulsion may occur with a hamstring tear in a child sprinting during sporting activities or other recreational pursuits. Bones grow faster than muscles in children, and with associated growth spurts and limited stretching and warm-up activities, epiphyseal avulsions are more common (214). Treatment of overuse syndromes generally involves conservative modalities and rest, followed by strengthening and stretching of muscle imbalances and gradual return to activity as tolerated. The snapping hip syndrome in children is an entity most commonly associated with iliotibial band irritation of the greater trochanteric bursa on hip flexion, extension, and internal rotation (6). It can also occur with the iliopsoas snapping over the lesser trochanteric process (193). Osteoitis pubis is more common in adults, but may be occasionally seen in older teenagers with high-mileage running (6,217).

The most frequently injured area in childhood and adolescent athletics is the knee (6,11,193). The collateral ligaments of the knee, especially the medial collateral ligament, are frequently injured in sports. An isolated injury to the medial collateral ligament usually may be treated successfully without surgery in the immature athlete. Anterior cruciate ligament (ACL) injury in the immature athlete has always been considered a relatively rare occurrence (6). Increased participation in organized sports, along with improved imaging techniques such as MRI, appears responsible for the reported increased incidence of this injury (218,219). Girls are two to nine times more likely to disrupt their ACL than boys (220), with a soccer injury a common scenario. Often, these injuries are associated with avulsion of the anterior tibial spine. Anterior cruciate ligament reconstructions in children, when performed, need to consider early closure of the distal, femoral, or proximal tibial physes or other growth disturbances with grafts that might cross the growth plate (221). Concerning growth disturbances include limb length discrepancy and angular deformities. Autogenous patellar tendon grafting appears to be the surgical choice, not to exclude other surgical considerations of autologous iliotibial graft or hamstring autograft or allograft. Over-the-top femoral graft placements (graft passed through the interchondular notch of the femur) have been reported by some authors with good success and efforts to spare excessive physeal penetration (222,223,224). Additional physeal-sparing reconstructions with minimal risk of growth arrest have been reported, with good success in the younger child (under 12) and adolescence (225). Conservative care, including rest, elevation, ice and derotational bracing, are recommended initially under a rehabilitation program that can take two to three months. Surgical repair is considered thereafter for ongoing instability and to minimize additional cartilaginous and meniscial injuries. An isolated meniscial tear in a child under the age of 10 is unusual. Surgery is used only if conservative measures fail. The choice is often repair of the meniscus rather than surgical resection because of the increased potential in children for cartilaginous healing.

The elbow continues to be the most commonly injured joint in children (6). Acquired dislocations account for about 8% of elbow injuries and are most frequent in children under the age of 10 years (11,226,227). Typically, the injury involves the nondominant extremity with a fall onto the outstretched hand (228). Nursemaid's elbow consists of radial head subluxation from a sharp upward pull on the extended pronated arm in preschoolers. A generalized ligament dyslaxia of children with large cartilaginous components of the distal humerus and proximal ulna, in addition to osseous instability, with numerous secondary ossification centers and epiphysis all contribute to the tendency for the pediatric elbow to dislocate. Posterior or posteriolateral dislocations account for up to 90% of the injury and can be reduced through numerous conservative techniques (6,228,229). With nursemaid's elbow, typically the child will not move the arm and holds it in a slightly flexed and pronated position. Radiographs are usually not indicated, as the injury is more subluxation of the annular ligament rather than true joint subluxation. Longitudinal traction and additional pronation followed by flexing the elbow above 90 degrees and then fully and firmly supinating the form produces reduction in most cases. A click or snap is often felt as the annular ligament repositions (6).

Shoulder injuries remain relatively uncommon in the overall picture of injuries to the pediatric musculoskeletal system (11). When they occur, they include separation of the acromioclavicular joint from direct trauma, osteolysis of the distal clavicle (mostly in weightlifters), and cervical clavicular injuries in the young thrower (6,230). Rotator cuff injuries remain less common in the younger athlete. Conservative treatment for musculoskeletal injury in children includes rest, ice, compression, and elevation (RICE) in addition to NSAIDs such as Telectin, naproxen (Naprosyn), and ibuprofen (Children's Motrin, Children's Advil). Appropriate equipment, coaching, recreation environments, and training often prevent sports injury, with safety remaining the primary consideration.

Osteochondritis dissecans is a condition resulting in partial or complete separation of a segment of normal highland cartilage from its supporting bone. Depending on the separation, cartilaginous or osteochondral intra-articular fragments may form (193). Mechanical symptoms may arise within the joint such as catching or locking. Although it has been more than 100 years since Konig (230) coined the term osteochondritis dissecans, the cause remains unclear. Five theories commonly suggested are ischemia, genetic predisposition, abnormal ossification, trauma, and cyclical strain (6). The condition most commonly affects the knee (lateral aspect of the medial femoral condyle in 70% of patients, lateral femoral condyle in 20%, and the patella in 10%) or can be seen in the elbow (11). Treatment of osteochondritis dissecans remains controversial. Intact lesions can often be treated symptomatically, with or without activity modification or immobilization (6). Free fragments often require surgical removal. Drilling techniques are commonly utilized and can help stimulate new bone formation healing and return of mobile bodies to their original donor sites (11). Long-term sequelae can be garnered in up to 25% with atypical lesions, older age, effusion, and larger lesions.

Chondromalacia of the knee needs to be distinguished from the more serious osteochondritis dissecans. Chondromalacia is a term used to describe anterior knee pain of undetermined cause in the younger athlete associated with softening of the articular cartilage beneath the patellar surface. The pain is frequently worse with squatting and climbing stairs, and is associated with a high-riding patella or malalignment. Patellar dislocations can occur in association and are usually lateral and associated with genu valgum, external tibial torsion, and general ligament dyslaxity. The subluxation of the patella is usually reducible, but can be painful. Exercises to strengthen the quadriceps, particularly the vastus medialis and the use of patella tracking braces, may be helpful. Surgical stabilization of the medial patellar tissues and lateral retinacular release can be helpful in more difficult cases.

Osteochondrosis is characterized by a disturbance in endochondral ossification, including both chondrogenesis and osteogenesis, in a previously normal endochondral growth region (6). The term osteochondrosis is preferred, as not all conditions are inflammatory, making the term osteochondritis inappropriate (193). Osteochondrosis is idiopathic and has been reported in nearly every growth center of the body, including apophyses, epiphyses, and physes. Their eponyms are generally named according to the region of the body and growth center involved (193). Most osteochondroses have well-defined natural histories and generally predictable outcomes (231). Freiberg's disease involves collapse of the articular surface in subchondral bone, usually of the second metatarsal (232,233). Kohler's disease involves irregular ossification of the tarsal navicular joint with localized pain and increased density. Freiberg's disease is more common in girls between the ages of 12 and 15 years, whereas Kohler's disease occurs in younger individuals age 2 to 9 years and is frequently reversible with conservative care including orthoses and casting. Apophysitis is relatively common at the knee, foot, and ankle, all secondary to traction overuse and microtrauma. Apophysistis at the inferior pole of the patella is called Sinding-Larsen-Johansson syndrome. Osgood-Schlatter disease involves apophysitis at the tibial tuberocity, and Sever's disease involves apophysitis at the posterior calcaneus. These conditions generally occur around the age of 10 to 15 years of age, a few years earlier in girls, and are generally treated conservatively with the RICE protocol. Care should be taken not to overgeneralize treatment, however, as each condition can be different and require special attention. LaNec disease or ischial pubic synchondrosis, for instance, can be confused with a bone tumor if not careful and subsequently overtreated (193). Heel cups may be helpful with Sever's disease in addition to short periods of casting and/or splinting. Stretching of the quadriceps and hamstrings can be helpful with Osgood-Schlatter disease in addition to knee sleeves or knee straps. NSAIDs are often prescribed as well. Pain-free strengthening of weight bearing soft tissues using more closed kinetic chain techniques may be best. Chondromalacia needs to be differentiated also from the osteochondroses in the young person with anterior knee pain. Chondromalacia is associated with softening of the cartilage beneath the patellar surface and often a high-riding patella or malalignment. The pain frequently gets worse with squatting and climbing stairs, and benefits with conservative treatment under the RICE protocol. Osteochondrosis of the vertebral end plate is known as Scheuermann's disease. The incidence of Scheuermann's deformities in the general population ranges between 0.5% and 8%, with an increased prevelance in males (6,234).

It is distinguished from postural roundback by its more rigid structural characteristics. Symptoms are common during the early teenage years and in most instances decrease in late adolescence (11). When three or more consecutive vertebrae are wedged more than 5 degrees, radiographic criteria for Scheuermann's disease are met (235). The radiographic picture includes irregular vertebral endplates, protrusion of disc material into the spongiosum of the vertebral body, Schmorl nodes, narrowed disc spaces, and anterior wedging of the vertebral bodies. The cause of Scheuermann's disease again is unknown, but thought by some to fall within the spectrum of repetitive microtrauma and fatigue failure of the immature thoracic vertebral bodies. An increase in the incidence of disabling back pain in adults has been reported and may lead to surgery in this older age group (11,236). More severe pain is reported in patients with kyphotic deformities greater than 75 degrees. Cardiorespiratory conditions may occur in patients with severe deformities (kyphosis greater than 100 degrees). Atypical Scheuermann's disease (237) or thoracolumbar apophysitis is named because it does not meet the usual radiographic criteria for Scheuermann's disease established by Sorenson (238). This phenomenon is usually seen at the thoracolumbar junction and may be the pediatric equivalent of an adult compression fracture. There is a 2:1 male-to-female predominance, with a peak age of incidence between 15 and 17 years. When Scheuermann's disease is associated with pain in the presence of one or more irregular vertebral bodies, physical exercise is prohibited. A thoracic lumbosacral orthosis (TLSO) or Milwaukee brace is used for more severe involvement. Sometimes bracing is required for three months to achieve pain control. Conservative care, including traditional RICE protocol, gentle flexibility routines, and NSAIDs, can be helpful. For correction of spinal deformity with bracing, a mobile kyphotic deformity is required in addition to at least a year of growth remaining in the spine (11). In most cases, brace treatment must be continued for a minimum of 18 months to have an effect on vertebral wedging. Severe involvement progressing to more rigid kyphosis, greater than 75 degrees, may require spinal fusion, both posterior and anterior (11).

Intervertebral disc injuries in children and the young athlete are uncommon (11). In contrast to the selective motor and sensory deficits often observed in adults with disc herniation, athletes under 20 years of age have pain and tenderness localized generally to the midline and, to a lesser extent, over the course of the sciatic nerve (239). Of surgically treated disc herniations, only 1% to 2% percent occurs in the pediatric population. Many of these children have underlying congenital anomalies, including transitional vertebrae, spondylolisthesis, and congenital spinal stenosis. Spondylolysis has never been found in the newborn. Its occurrence increases between the ages of 5.5 and 6.5 years to a rate of 5%, close to the frequency of 5.8% in the Caucasian population (240). The condition involves a fracture to the paras interarticularis and is more common in athletes involved with repetitive flexion-extension and hyperextension activities of the lumbar spine. Oblique radiographs of the lumbar spine show the classic "scotty dog" sign (241). A positive stork leg test with careful hyperextension of the lumbar spine is often present. Common sports associated with this condition are collisional in nature: gymnastics, weight lifting, and figure skating. Involvement is generally at the L5–S1 level, but can occur at other levels. In the absence of disc herniation or spondylolisthesis, radicular symptoms are uncommon. Treatment often consists of activity limits, stretching of tight hamstrings, and lumbar corsets or bracing in carefully selected individuals. Nuclear medicine bone scans can be particularly helpful in identifying these lesions and eventual healing, which can take up to nine months (242). The incidence of back pain in backpack users of school age has been noted in up to 74% of individuals (243). Heavy backpack use, female gender, and larger body mass index were all associated with back pain. Back pain from backpacks needs to be considered readily in all individuals, particularly those with spondylitic conditions and regular daily use (244). Spondylolisthesis, or slipping forward of the vertebral body, may occur during childhood, with a prepubital peak incidence and promoted by hyperlordosis. Grading of spondylolisthesis is according to the classification developed by Meyerding (245). The superior border of the inferior vertebrae is divided into four equal quadrants, with slips in each quadrant accounting for one grade. Surgical treatment is necessary in the presence of neurologic signs or forward slipping of the vertebral body beyond 50% of its width. Other apophyseal injuries in the spine include slipped vertebral apophysis or endplate fracture (246). This condition may mimic a herinated lumbar disc and is often associated with heavy lifting. Commonly, the inferior apophysis of L4 is displaced into the vertebral canal along with some attached disc material (11). Radiographs reveal a small bony fragment pulled off the inferior edge of the vertebral endplate. A CT scan or MR imaging reveals an extradural mass. Surgical excision can provide excellent relief of symptoms in those in whom conservative care has failed. Epidural steroids may be used for individuals in whom initial nonsurgical treatment is unsuccessful. Strains of the lower back are less common in children in view of the open iliac apophysis. Children with iliac apophysitis usually have a beltlike pain along the muscular attachments of the superior iliac crest (247). Lumbar interspinous process bursitis, or "kissing spines," also needs consideration in the

young patient, especially those participating in gymnastics or other activities involving hyperextension of the thoracolumbar spine.

Discitis is a rare condition (occurring in less than 1%) that also causes back pain in children (206). It can be divided into septic and aseptic types. Between the vertebrae, the notochord expands to form a gelatinous center of the intervertebral disc called the nucleus pulposis. This nucleus is later surrounded by circularly arranged fibers from sclerotome-derived mesodermal cells called the annulus fibrosis. The nucleus pulposis and the annulus fibrosis together constitute the intervertebral disc. The intervertebral disc is vascular in children up to 7 years. Around the age of 7, the disc begins to develop some of the end arteries common to the adolescent and adult. From the age of approximately 13 years, all end arteries are thought to be formed and thus, the disc becomes avascular. It may well be that the more vascular nature of the disc is a major reason why discitis occurs almost solely in children (248,249,250,251). Positive cultures are generally more common in younger children, with *Staphylococcus aureus* by far the most common finding (206). A slower, indulent form of discitis may develop in a child from brucellosis or tuberculosis. A skin test for tuberculosis may be helpful. Trauma might cause release from the disc tissue enzymes such as phospholipase A2, known to be a potent inflammatory simulator, which could, in theory, cause inflammation. Viral causes are also thought to be present and likely make up a substantial component of the aseptic variety. High fever, toxemia, elevated white blood cell counts, positive blood cultures, and bone scans in a child under the age of 3 who refuses to sit or stand is a common history. The diagnosis must be considered in a child with just mild illness who has abdominal pain or refuses to walk for unclear reasons. Pain frequently occurs at night, and children are usually not systemically ill (252). An MR imaging scan shows involvement of the disc space and vertebral bodies one level above and below. The two most serious diseases in the differential diagnosis include vertebral osteomyelitis, rare in children, and spinal tumors. Biopsy of the disc space may be necessary, particularly in an adolescent suspected of abusing drugs. Vancomycin may be the treatment of choice or other staphylococcal antimicrobials. When there is no response to early antibiotic therapy, aspiration or biopsy should be performed, followed by culture-specific antibiotic treatment (11). Immobilization of the child may or may not be helpful. Hematogenous spread is the most common cause of vertebral osteomyelitis, with *Staphylococcus aureus* the most common organism. Vertebral osteomyelitis generally involves the more anterior aspects of the spine and may be associated with paravertebral collections. Tuberculous spondylitis or Pott's disease remains common worldwide and is still seen in some neglected areas of the United States (253).

Gait abnormalities, although frequently benign, can be a great source of parental concern. The child's whole posture needs to be looked at carefully, particularly from the waist downward, because malalignment of any lower extremity joint may stem from another. Figure 14.11 shows anteversion of the femoral head and neck on the femoral diaphysis in addition to coxa valga and coxa vara. The normal angle of the femoral neck and shaft at birth is approximately 160 degrees and decreases to approximately 140 degrees at 5 years and 120 degrees at adulthood. At birth, the normal anterior femoral neck angle relative to the transcondylar line of the distal femur is approximately 40 degrees. This angle decreases to approximately 25 degrees by age 5 and 15 degrees in adulthood (254,255). An increase in the anteversion angle is frequently associated with in-toeing and increased internal rotation best assessed with the child lying prone. Figures 14.12 and 14.13 show normal degrees of internal and external rotation throughout the lifetime within two standard deviations. The degree of femoral neck anteversion is generally thought to be

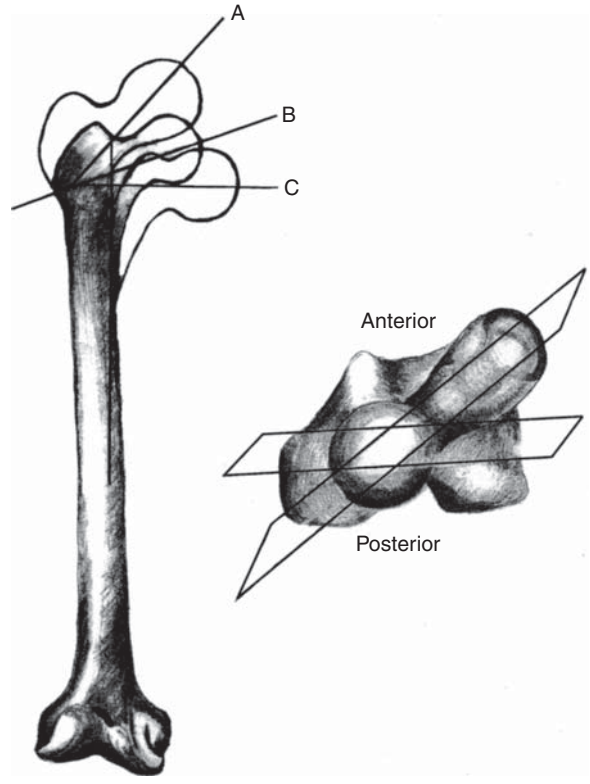


Figure 14.11 Angle of neck shaft and anteversion of the femur: (A) increased, coxa-valga, (B) mormal, (C) decreased coxa vara. The smaller diagram shows a top view relating a plane from left to right through the greater trochanter and femoral head referenced to the transcondylar femoral axis distally.

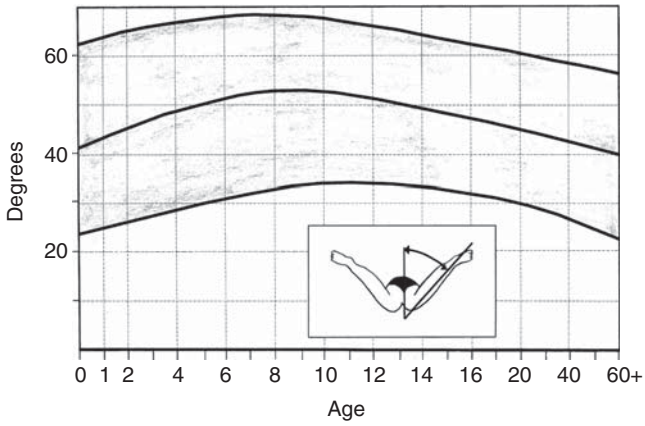


Figure 14.12 Hip internal rotation assessed with the child prone. Normal ranges are shaded. (Adapted from Ref. 256 with permission.)

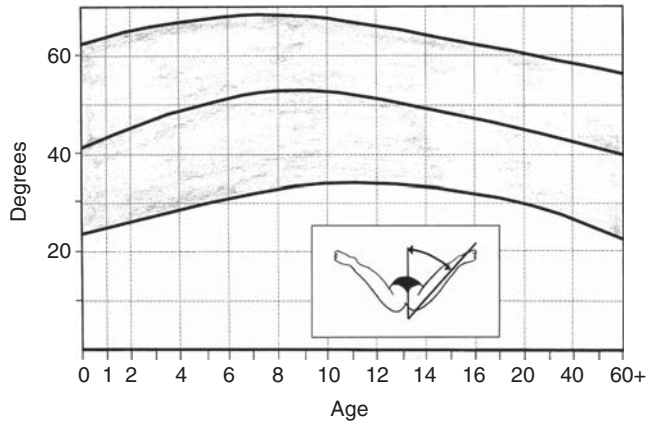


Figure 14.13 Hip external rotation assessed with the child prone. Normal ranges are shaded. (Adapted from Ref. 256 with permission.)

about 20 degrees less than full internal rotation of the hip (6). An estimate of anteversion can be measured by trochanteric palpation with the child prone on the examination table. The degree of internal rotation measured at the point where the greater trochanter is most prominent on the lateral surface of the hip is the estimate of anteversion. In-toeing may persist into adulthood, but often improves with time in the physically normal child by the age of 8. Exercises to strengthen the external rotators of the hip and physical and verbal cues to out-toe and compensate may, at times, offer benefit. This benefit is achieved through facilitating motor memory and improved compensatory strategy to increase out-toeing and not the result of any change in the bony anteversion. Severe in-toeing not correcting over time, associated with falls and significantly limited external rotation, can be corrected surgically. Surgery is deferred at least beyond the age of 6 years and frequently after 10 years, when there is less chance of postoperative derotation of the surgically corrected torsion. Surgery should not be taken lightly, and good indication should be present along with well-educated parents and child to justify the risk.

Excessive hip external rotation with minimal internal rotation, often tested with the child lying prone with hip extended (see Fig. 14.13), is associated with femoral retroversion (opposite of anteversion). This condition can be seen more common in children with low tone and increased joint laxity, such as those with Down's syndrome and Ehlers-Danlos syndrome. Gait is with excessive out-toeing, and familial traits may be present. Most rotational variations in children resolve spontaneously with time and minimal intervention (257,258). Careful examination is required to exclude more serious disorders.

Tibial torsion, both internal and external, may occur as compensation for the femoral version or by

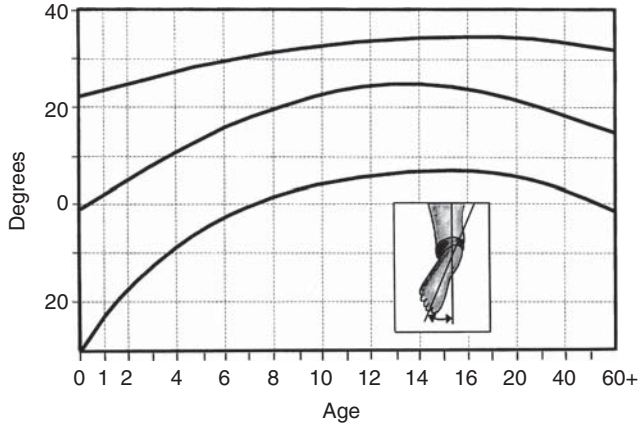


Figure 14.14 Rotational status of the tibia assessed by evaluating the child in the prone position. Foot placed in plantigrade neutral position. (Adapted from Ref. 256 with permission.)

themselves, causing in-toeing and out-toeing. The transmalleolar axis may be palpated in prone and knee-flexed positions. The lateral malleolus is approximately 5 to 10 degrees posterior to the medial malleolus in the toddler and increases to approximately 15 degrees by adolescence (259). Figure 14.14 (256) shows the normal degree of thigh-foot angle over the lifetime within two standard deviations. Dennis-Brown bars have been found to have essentially no effect in altering tibial torsion and have generally fallen into disuse for this condition (6). In measurement of the thigh-foot angle, the foot is placed into the plantigrade and hindfoot neutral position with palpated talonavicular alignment. This helps eliminate other, more intrinsic foot conditions, such as metatarsus varus and adductus, that can otherwise confound the thigh-foot angle measurement. Figure 14.15 shows normal foot progression angle over the lifetime (256). All rotational abnormalities of the lower extremities have influence on the

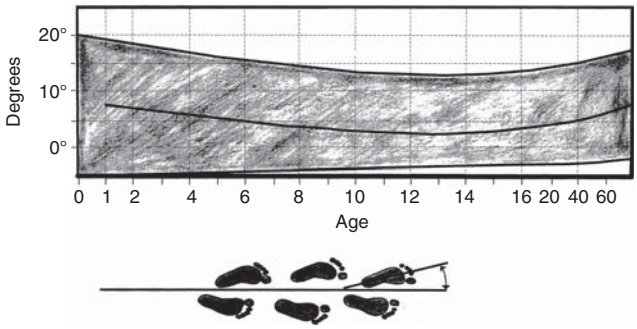


Figure 14.15 Foot progression angle. Normal ranges shaded. (Adapted from Ref. 256 with permission.)

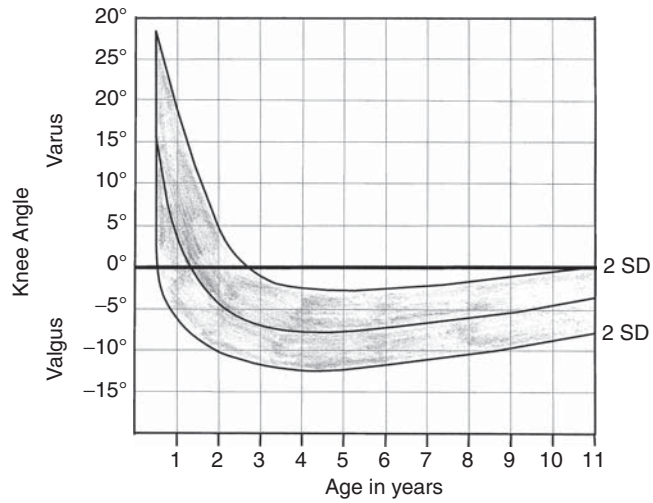


Figure 14.16 Normal values for knee angle measured in valgus and varus. (Adapted from Ref. 256 with permission.)

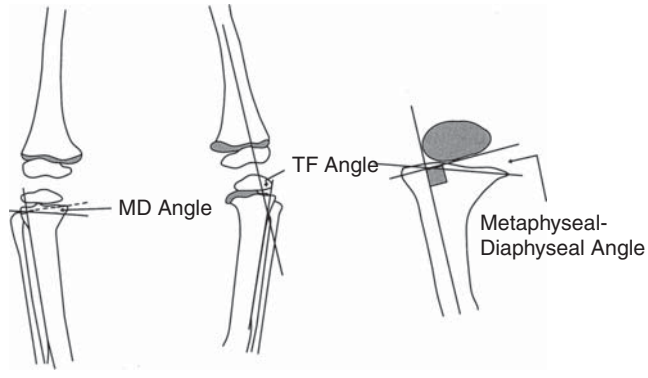


Figure 14.17 Measurement of metaphyseal-diaphyseal angle and tibiofemoral angle.

foot progression angle. Flat feet, or pes planus, is no exception. Flexible pes planus is usually asymptomatic, at least in earlier years, and more common than its rigid counterpart in children. Inexpensive scaphoid pads or medial inserts may help to create more plantigrade weight bearing and improve foot progression angle, but they do not correct the deformity. Extreme cases such as in children with hypotonia may require surgery after the age of 5 years in the form of calcaneal lengthening once bony cortices are more solid. Untreated progression with increased foot progression angle may occur along with compensatory hallux valgus, planovalgus, and secondary bunion and toe deformities. Pes planovalgus is associated with more active or shortened peroneal musculature, progressing over time, with the development of pain particularly in later years. Rigid pes planus is a congenital deformity associated with other anomalies in 50% of cases, as discussed earlier in this chapter.

Angular deformities of the femoral-tibial alignment are also a source of frequent concern for parents and families. At birth, the infant has a bowlegged posture with a genu varum of 10 to 15 degrees (260,261). The bowing gradually straightens so that the femoral-tibial alignment is neutral or 0% by 12 to 18 months of age (6). Continued growth results in a peak valgus angulation of 12 to 15 degrees by the age of 3 to 4 years (11). Subsequent growth reduces the genu valgum to normal adult values of approximately 5 to 7 degrees by the age of 12 years. At any age there is a fairly wide standard deviation of normal (256). Figure 14.16 shows the normal variation of valgus and varus at the knee up through adulthood values (256). Measurements between the medial and femoral condyles or intermalleolar distance help to quantitate the deformity (256). The most common cause of genu varum in children is physiologic bowlegs. Children with this condition have genu varum that persists after the age of 18 months, usually resolving before the age of 3 years. X-rays show symmetric growth plate anatomy and medial bowing that involves the proximal tibia as well as the distal femur. Measurement of the metaphyseal-diaphyseal angle in addition to the tibiofemoral angles is helpful in the differential diagnosis (Fig. 14.17) (262,263,264). The differential diagnosis includes infantile tibia vara or Blount's disease, hypophosphatemic rickets, metaphyseal chondrodysplasia, focal fibrocartilagenous dysplasia, and trauma to the epiphysis. Blount's disease occurs in children with no apparent abnormality at birth, having a typical history of genu varum worsening with gait before the age of 2 years. The less frequent juvenile onset may occur between 4 and 10 years and the adolescent form over 11 years. The condition is more frequent in African Americans and girls, and is seen with obesity and in children walking at an early age (265). The condition is also more common in certain geographical locations such as the southeastern part of the United States (193). Classic radiographic changes associated with Blount's disease and tibial varum are seen in the Langenskiold classification (266). Blount's disease is believed to result from abnormal compression of the medial aspect of the proximal tibial physes, causing retardation of growth

in that area or increased growth laterally of the proximal tibia or fibula (6,267). In juvenile Blount's disease, the etiology is less clear and may relate more to malalignment, leading to the characteristic changes visible on radiographs. Patients with metaphysial-diaphysial angles greater than 16 degrees have been reported to experience progression of the angular deformity (268). Early and continuous bracing in Langenskiold stage I and stage II disease (266) can achieve good results (11). Bracing should not be initiated after 3 years of age, nor should brace treatment be continued if Langenskiold stage III changes develop (269,270). The authors' preference is a medial upright knee ankle foot orthosis (KAFO) with valgus promotion padding through the center of the knee axis and free-swinging knee and ankle. Modified KAFOs preventing knee flexion have also been promoted (6). Proximal valgus osteotomies may be required for severe persistent angular deformity after the age of 3 years, along with consideration of Ilizarov techniques (11). Stapling of the lateral physis (often both tibia and femur) are also considerations, particularly in the adolescent prior to cessation of growth. Increased fragmentation, declination, and beaking of the medial–proximal epiphysis generally indicate the need for surgery. Surgical complications can include compartment syndrome with persistent neurovascular compromise. Careful postsurgical follow up of the child is required to prevent unnecessary over- or undercorrection. Graphs for timing of hemiepiphysiodesis are available and can be helpful in experienced hands (271).

Genu valgum, or "knock-knees," is a concern in children who are developing peak valgus alignment around the ages of 3 to 4 years. Almost 99% of the time, this valgus is benign in nature, correcting toward adult values by early adolescence. X-rays show symmetric growth plates with no particular abnormalities. Observation is the treatment of choice in these individuals. Children who have genu valgum with a femoral–tibial angle greater than 20 degrees require follow-up, but generally the problem resolves spontaneously. If abnormal genu valgum persists into the teens, correction by hemiepiphysiodesis or stapling of the medial physis may be effective (11,272). Staples that are placed extraperiosteal for varus or valgus deformity allow for growth to resume once removed. Rebound phenomena can be anticipated, undoing some of the corrected valgus or varus. Overcorrection slightly in anticipation of this problem, especially in children under the age of 12, needs to be considered (193).

Idiopathic toe walking is a common condition in children under 3 years of age. By 3 years of age, children should walk with a heelstrike (273,274,275). Persistent toe walking beyond this age is abnormal (6). Little is known about the natural history of idiopathic toe walking, with most individuals improving or showing resolution prior to the age of 6. Persistent toe walking in the older child and young adult can result in leg pains, more activity-related, and frequently in the anterior tibial or knee regions. Toe walking can diminish or cease with time, as body mass becomes too large to be supported by the triceps surae or as a result of secondary development of external tibial torsion (276). Toe walking developing sometime after birth can be associated more with problematic conditions, such as muscular dystrophy, dystonia, tethered cord syndrome, central nervous system neoplastic processes, or autism (277). A family history is often positive along with that of prematurity and a slight male predominance (277).

Leg pains in children are generally benign, but need to be followed carefully for signs of progression or persistence despite conservative care. Conservative care, generally involving the RICE protocol, NSAIDs, and warm baths and massage, often relieves most of the discomforts. A pattern of increased pain with activity or recreational pursuits is common. If improvements are not noted within a few weeks of conservative care, additional workup is required to rule out other, more concerning etiologic entities. Workup should include radiographs, hematology and metabolic parameters, erythrocyte sedimentation rate (ESR), possible nuclear medicine scan, and Lyme's disease titre along with other rheumatologic markers.

In children who toe walk, walking is generally not delayed as a developmental milestone, and when this occurs, conditions like spastic diplegia should be considered. A few beats of clonus at the ankle can be helpful in differentiating associated mild diplegia from idiopathic toe walking. Nonoperative treatment, including heel cord-stretching routines with the calcaneus midline or inverted, can be helpful when performed on a regular basis along with dorsiflexion-strengthening exercises. Stretching a tight heel cord with the hindfoot in valgus can contribute to midfoot breakage while being ineffective in lengthening plantarflexion soft tissues. Articulating AFOs with plantarflexion blocks or posterior leaf-spring types can be helpful in maintaining position both day and night. Serial casting can be an option for resistant equinous deformity not felt to be surgical at the time. Casting should occur with maximal dorsiflexion as tolerated, again with the heel in a neutral or slightly inverted position. Two or three sets of short-leg casts of the walking nature, lengthening the heel cord, can result in greater passive dorsiflexion. Short-term weakness of the anterior tibialis and dorsiflexors can be anticipated postcasting requiring additional strengthening intervention. *Clostridium botulinum* toxin A injections can be helpful also in weakening partially the plantarflexors, facilitating improved stretch into dorsiflexion along with relative strengthening of active

dorsiflexion. Orthotics can be weaned over three to six months once toe walking has resolved and improvements obvious. Nighttime splinting can be discontinued in the absence of recurrent toe walking. Surgical intervention, including heel cord lengthening and/or gastrocsoleus recession, is reserved for those who have failed conservative trial. Toe walking after the age of 6 years often does not improve, and heel cord contractures can worsen (6). External tibial torsion can progress further developing as compensation for lack of foot-flat contact. The torsion may be severe enough with excessive external foot progression angle to warrant corrective osteotomy. Surgical lengthening is performed sufficient to obtain 10 degrees of dorsiflexion with the knee extended (6,11). Overlengthening of the heel cord can be disastrous, resulting in persistent crouched gait and associated pain syndromes and limitations. In more severe and chronic equinous deformities, posterior ankle capsular release may be required. Short-leg casting postoperatively is common up to six weeks followed by custom-molded AFOs for up to two months thereafter. Home exercise, along with physical therapy for gentle heel cord stretching and strengthening ankle dorsiflexion, is mandatory or recurrent equinous deformity can be anticipated. Long-term outcomes of surgical lengthening in skilled hands are generally positive when recommendations are followed with satisfactory heel-toe walking over the lifetime (278,279).

SCOLIOSIS

Overview

Scoliosis is a frontal plane deformity of the spine of >10 degrees, with frequent coexistence of rotational deformity. It is the most common pediatric spine deformity, and is classified into congenital, idiopathic (subtypes: infantile, juvenile, adolescent), neuromuscular and functional types (Table 14.6). While the etiology, onset, prognosis, and treatments vary between classifications, the possible outcomes of severe untreated scoliosis are the same: respiratory compromise, seating compromise, pain, gait impairment, difficulty with activities of daily living, and psychological distress (280,281). Understanding the natural history and available interventions is important in helping patients achieve long-term comfort and functionality.

Embryology, Growth, and Maturation

Spinal development is a complex process, which begins in the first month of gestation when mesoderm cells

14.6 Type of Scoliosis With Categorical Description

TYPE OF SCOLIOSIS	CAUSE	SEX/AGE OF ONSET (YR)	COMMON ASSOCIATED CURVES	COMMON CHARACTERISTICS
Functional	Nonstructural, secondary to leg length discrepancy, herniated disk, trauma, arthritis	Any	None	Resolves with correction of underlying cause
Congenital	Failure of somite formation or segmentation 60% have other anomolies	Birth, but delayed diagnosis possible	None	Progressive tendency, surgery more likely
Idiopathic				
Infantile (<1%)	Positional contributions	Male, < 3 yr	Left thoracic	May resolve spontaneously
Juvenile (19%)	Etiology unknown	Male=Female, 3–10 yr	Any	Aggressive traits, surgery more likely
Adolescent (80%)	Multifactoral, polygenetic	Male>Female,females progress more often, >10 yr	Right thoracic	10% require treatment (bracing > surgery)
Neuromuscular	Upper or lower motor lesions, myopathic processes	Any age	Long sweeping typical	Aggressive, less responsive to bracing, progress after maturity

surrounding the notocord begin to differentiate into sclerotomes. These will ultimately form vertebral bodies and arches. Injury in early gestation often affects other nearby organs, primarily the cardiac, renal, and gastrointestinal systems. Approximately 60% of those with spinal anomalies have other congenital malformations, so abnormalities in these areas are essential to screen for (282).

Unlike limb growth, vertebral growth is nonlinear. Two major growth spurts typically occur: the first before the age of 3, and the second during puberty. The relationship of scoliosis to growth has been well established, and screening programs and surgical interventions best planned with these in mind. The Tanner stage and/or the Risser's sign classifications are helpful in predicting growth spurts, the progression of scoliosis, and the cessation of growth (193).

While race, heredity, physical activity, physical disability, and nutrition may affect growth, growth typically accelerates girls at Tanner stage 2 and in boys at Tanner stage 3 (283).

The use of Risser lines, seen by posterior anterior (PA) radiographs of the iliac crest, assist in staging skeletal maturity and predicting future growth (Fig. 14.18). The Risser system is based on ossification of the iliac crest proceeding from the lateral to medial, and extends from grade 0 (no ossification) to grade 5 (complete fusion to iliac apophysis) (284,285). Risser 1 represents the period of most rapid skeletal growth, and correlation of the Risser's sign with the degree of a scoliotic curve can be predictive of curve progression (see Fig. 14.18).

Curve Classification and Naming

Scoliotic curves are named by their direction, location, and magnitude (Fig. 14.19). The curve's *convex apex* (most laterally deviated vertebrae from the sacral line) indicates its named direction and location, and measurement by the Cobb angle provides its most reliable magnitude (Fig. 14.20) (286). If more than one curve exists, the largest-degree curve is designated as major and the others minor. Curves over 60 degrees are associated with restrictive lung disease.

Rotation of the spine, commonly present with scoliotic curves, is measured using a scoliometer when the child is bending forward, or radiographically by the Nash-Moe method or by CT scan (287,288). Rotational deformities may complicate surgical correction.

History, Physical Exam, and Treatment Overview

The scoliosis exam will vary, depending on the patient's age and associated diagnosis, but important general information must be collected from all patients. A positive family history is particularly pertinent in congenital and idiopathic scoliosis, and the presence of back pain may indicate a serious discitis or tumor. Rapid curve progression, bowel and bladder changes, recent trauma, associated weight loss, muscle weakness, or joint pain can point to other serious primary processes such as spinal cord syrinx or tethered cord, spinal fracture, rheumatologic disease, osteoblastoma, or hip deformity.

Reflexes, strength, range of motion, general posture, and gait must be examined. Seating systems and assistive devices should be assessed, as improper walker or crutch height and truncal weakness with poor seating support can affect spinal position in children with disabilities. Examination of the skin for café au lait spots, webbed neck or low hairline, and

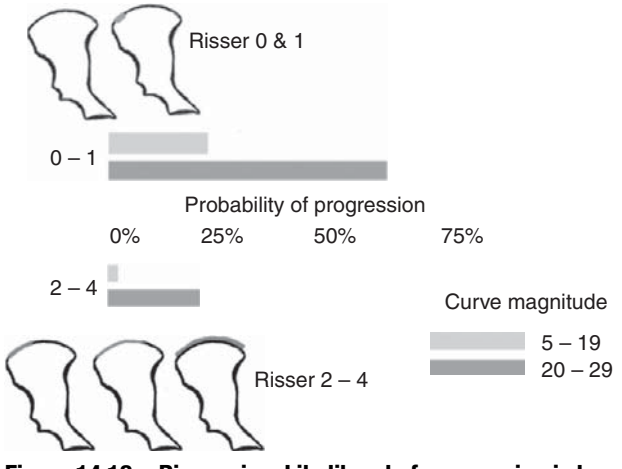


Figure 14.18 Risser sign. Likelihood of progression is based on the Risser sign and curve magnitude. (From Ref. 375)

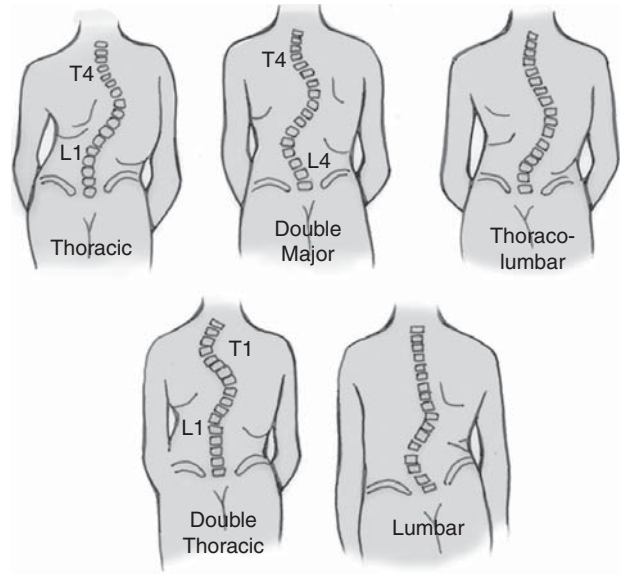


Figure 14.19 Classification of scoliosis. Scoliosis is classified into general categories by level. (Adapted from Ref. 284).

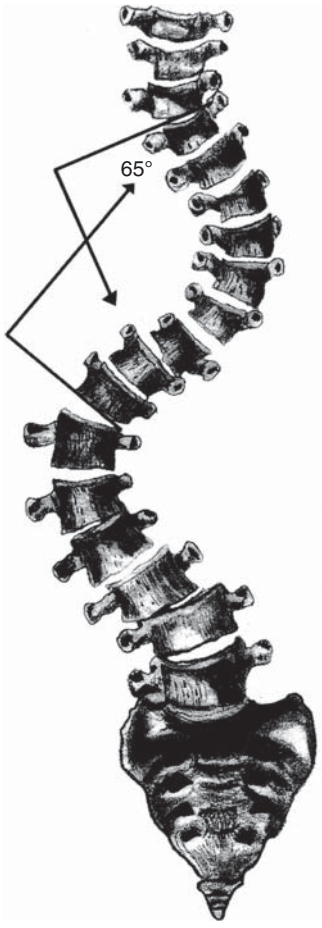


Figure 14.20 The Cobb method of measuring curvature in scoliosis. The angle measured is formed by perpendicular lines drawn through the superior border of the upper vertebra and the inferior border of the lowest vertebra of a given curve.

hairy patches or skin dimples may lead to recognition of disorders such as Klippel-Feil, spina bifida occulta, or neurofibromatosis. Excessive height, arm span, or joint hyperextensibility may signal a connective tissue disorder of which scoliosis is only a presenting symptom. Leg length, straight-leg raise, and range of motion should be checked, as length discrepancies, asymmetry, contracture, or pain may point to hip dysplasia or an underlying neurologic disorder such as hemiplegia or herniated disk.

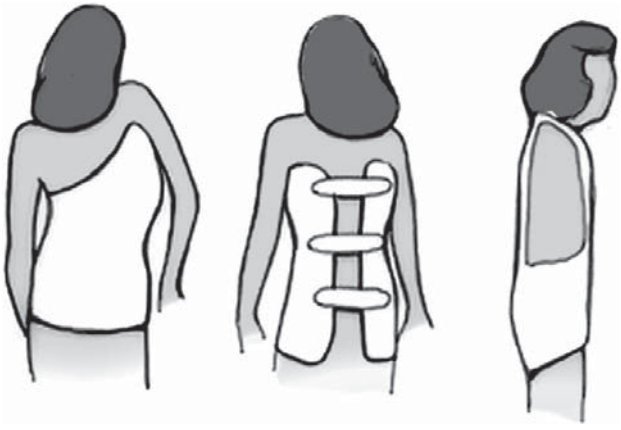
Examine the back for pelvic obliquity, elevation of either iliac crest, or asymmetry of the scapula or shoulder girdle. Forward bending (Adam's test) with the feet and palms together may show asymmetrical prominence of the rib cage (vertebra rotate into the convexity of the curve), which can be measured with a scoliometer. A rotation of over 7 degrees warrants further investigation. Side bending may help assess the flexibility or rigidity of a curve, which is important when considering treatment options.

Curve Documentation. A full spine PA radiograph is usually appropriate for screening purposes, although certain curves (ie, congenital, infantile) may require CT or MRI evaluation. Curves with significant rotational components or kyphosis may require lateral views. Radiographs should be taken standing, if possible and, if wearing an orthosis, both in and out of the brace. Cobb angles, which have proven reliable in tracking curves, should be followed every three months to a year, depending on the rate of progression (286). Serial evaluations should continue until growth is complete, although neuromuscular curves often progress after maturity, so continued screening is warranted (284).

General Treatment Options. Orthotic management is not appropriate, effective, or recommended for all forms of scoliosis. Long-term bracing, while reducing curve progression and maintaining flexibility, needs to be considered carefully with respect to function, social development, and self-esteem (285).

Curves less than 40 degrees are typically compatible with bracing, but their location affects brace choice. Curves with apex at or below T7 are typically managed with a soft or rigid TLSO, which allows more functional activity than the Milwaukee brace used for curves above T7 (Fig. 14.21). This brace often incorporates a chin and head pad, is more restrictive, and is less well tolerated. It is recommended that both braces be worn 16–24 hours a day to be effective (282). Removing the brace for an hour or two per day to accommodate athletic or recreational pursuits is not uncommon. A Charleston- or Providence-style brace, which bends the body away from the curve and is worn at nighttime, may be more tolerable, but is likely to be less effective unless it is used for a thoracolumbar curve of less than 35 degrees (see Fig. 14.21) (282).

Surgical Interventions. Achieving a balanced spine (head and shoulders over sacrum), a solid arthrodesis, and a reduction in the deformity are the primary goals of surgical intervention. Surgical techniques vary by type of scoliosis. Titanium instrumentation is recommended when it is clear that MRI imaging will be needed in the future, given the frequency of coexisting organ and developmental abnormalities seen in some children. Continuous intraoperative spinal monitoring (somatosensory-evoked potentials, motor-evoked potentials) is important for preventing neurologic injury during surgery (288). Complications of surgery vary with patient diagnosis, curve size, and a multitude of other factors, but infection, pseudarthrosis, anemia, hypotension, and hardware failure are the most common.



Charieston	TLSO	Milwaukee
Least obtusive	Moderate	Most difficult to accept
Nightime only Most curves	16–22/24 hours Most curves	16–22/24 hours High curves

Figure 14.21 Types of braces. These are common braces and generalizations about their use. (Adapted from Ref. 284.)

Types of Scoliosis

Congenital Scoliosis

Congenital scoliosis accounts for approximately 20% of all scoliosis and is due to prenatal disruption of vertebral formation (hemivertebra, wedge vertebra) or veterbral segmentation (block vertebra, unilateral bar). A single hemivertebra is the most common anomaly. A positive family history may be present, with 5% to 10% of siblings having a similar disorder (285).

While congenital scoliosis may not be clinically evident until later in life, problems related to defective organogenesis may lead to its early detection. Abnormalities of the trachea, esophagus, renal tract, gastrointestinal tract, lungs, heart, radius, ear, lip, and palate often accompany congenital scoliosis. Up to 25% of children may have renal disorders, 10% may have cardiac problems, and 30% may have spinal dysraphism *(*282,289*).* Scoliosis is a primary symptom in VATER syndrome (vertebral defects, anal atresia, tracheoesophageal fistula, radial and renal dysplasia) and thoracic insufficiency syndrome. Immediate surgical referral is required if a congenital spine abnormality is identified.

Unilateral, unsegmented bars that restrict growth on one side of the spine while the other grows normally, especially in the thoracic area, produce curves that are the most likely to progress. If in the cervical area, torticollis may be a presenting symptom (288). Unsegmented block vertebra, as seen in Klippel-Feil syndrome, generally do not produce a progressive scoliosis, but restrict range of motion (284). Avoiding activities that place these patients at risk (diving, contact sports) is important.

Imaging MRI of the brainstem and entire spine provide the best evaluative tool to visualize not only bony abnormalities, but spinal cord dysraphism (diastematomyelia, lipoma, hydromyelia) that may coexist, yet not be evident on routine radiographs (289). Myelography is rarely used. Close monitoring every three to six months until age 4 and again in the adolescent years is essential (282).

Treatment Typically, orthoses are ineffective, except perhaps in small-degree, long, flexible curves. If an orthosis is used, the family, physiatrist, and therapist must work to encourage the child's acquisition of developmental skills through adaptive activities that accommodate their reduced spinal range. Maintaining cardiovascular health and endurance is especially important prior to surgery.

Approximately 50% of children with congenital scoliosis require surgical intervention at an early age, before spinal rigidity or secondary pulmonary deficiencies occur (284). Surgical options are aimed at prevention of deformity. They include hemivertebra excision, convex growth arrest (hemifusion), fusion with instrumentation and allograft, or instrumentation without fusion (sparing growth). Due to concerns over the loss of spinal height, and the impact that fusion of the thoracic spine may have on long-term pulmonary function, numerous nonfusion technologies have been developed.

Congenital Kyphosis Congenital kyphosis is most common at T10–L1 and due to a failure of vertebral segmentation and/or formation. It may accompany myelomeningoceole or spinal dysraphism, and progressive deformity may lead to paralysis. If the curve is less than 50 degrees, it is most often treated surgically by posterior fusion, but anterior-posterior fusion or kyphectomy may be necessary if the curve is larger (282).

Idiopathic Scoliosis

More than 80 percent of scoliosis cases belong in the idiopathic category, which is subgrouped into three types defined by age of onset (Table 14.6). All differ significantly in demographic distribution, progression, and treatment type.

Infantile

Infantile scoliosis is rare and is not related to a vertebral defect, but has an unknown etiology. It occurs within the first three years of life and often spontaneously resolves.

Left thoracic curves are common in infantile scoliosis, and boys are predominantly affected.

Plagiocephaly, developmental dysplasia of the hip, and congenital muscular torticollis are often associated, so radiographs of the spine and hips and MRI of the brainstem and spinal cord should be obtained (288). Neuromuscular disease, congenital scoliosis, and intraspinal pathology (Chiari malformation, tumor) must be ruled out.

Large curves in infants over a year of age may progress and require bracing or body casting. Curves over 40 degrees require surgery to avoid cardiopulmonary compromise. VEPTR or "growing rod" technology may be used in curve management. If fusion is needed, trunk height will be lost and anterior-posterior procedures will be needed to prevent the risk of crankshaft deformity.

Juvenile

Juvenile scoliosis appears equally in males and females between the ages of 3 and 10 years, and is unfortunately aggressive, with about 70 % of children requiring treatment—either bracing (50%) or surgical intervention (50%) (282). Tumors or spinal abnormalities may be causative, and an MRI of the spine and brainstem, along with radiographs, are necessary for a thorough evaluation. Progression is related to age of onset (Fig. 14.22).

Bracing may be effective, but the social impact of long-term bracing in this age group may be significant, and surgical intervention with "growing rod" systems is recommended once the curve reaches over 40–50 degrees. In a child older than 8 years and categorized as Risser 0, if a fusion is done, an anterior-posterior fusion will typically be performed to avoid crankshaft problems.

Adolescent

Idiopathic scoliosis developing after age 10 is the most common form of scoliosis, and occurs in about 25/1,000 adolescents. The incidence is greater in females, as is the tendency to progress with a right thoracic curve.

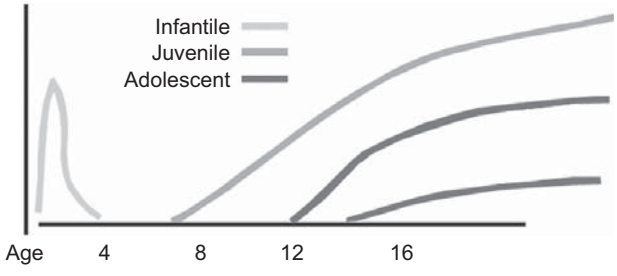


Figure 14.22 Natural history of idiopathic scoliosis. Progression is related to the age of onset of the scoliosis. (Adapted from Ref. 284.)

A left thoracic curve in an adolescent male is suspect, and causality should be further investigated. The exact etiology of idiopathic scoliosis is unknown, but genetics play a role, as about 30% of patients have a positive family history, and there is 50% concordance among twins (282,285). A multifactorial etiology related to growth hormone and melatonin production, connective tissue and muscle structure, and platelet function have all been postulated, but research is inconclusive (290). Fortunately, most curves are fairly benign and only about 10 % require treatment other than observation (288). More than 90% can be controlled effectively with bracing for curves between 20 and 40 degrees. Effective control means no progression of curve beyond 5 to 7 degrees after onset of bracing (285). Progression also relates to age of onset (see Fig. 14.22).

Curve Progression. Curve progression depends on several factors, including age of onset, curve size, and level of skeletal maturity. Young (<12 years), premenarchal, skeletally immature (Risser <2) females with large curves (>20 degrees) are most at risk. In the past, spinal and abdominal exercises were recommended to help reduce curve progression, but while these help maintain range of motion and fitness, there is no evidence that they prevent scoliosis (291).

Curves less than 25–30 degrees that are not progressing can be observed serially, especially if the child is approaching skeletal maturity. Brace wear, while difficult in adolescence, is still a standard of care for reducing curve progression, although there is no level 1 evidence in favor of bracing. A randomized, controlled, multicenter, National Institutes of Health (NIH) study is currently underway. Bracing is recommended for curves over 20 degrees in a skeletally immature patient in whom only 5–10 degrees of progression has been noted over a six-month period. Wearing the orthosis 16–24 hours a day is suggested until skeletal maturity is reached, at either Risser stage 4 in girls or stage 5 in boys, or two years postmenarche (292). Brace wear is discontinued in lieu of surgical intervention generally if the curve exceeds 40 degrees or more rapid progression is noted.

Treatment in adolescence is important because psychological distress and social limitations have been noted in adult females who have disfiguring curves greater than 40 degrees (281). Curves less than 40 degrees at skeletal maturity do not tend to progress in adulthood, but curves over 50 degrees, especially in the thoracic area, can. Progression of just 1 degree per year in adulthood can lead to degenerative changes that may become painful, and rigid curves in osteopenic adults are difficult to treat surgically.

Surgical Intervention. Operative interventions vary with the number, location, and size of curves and the child's

skeletal maturity. For the typical right thoracic curve of 40–50 degrees in a skeletally immature female, a posterior spine fusion often is recommended. For thoracolumbar or lumbar curves, anterior fusion may offer an advantage of reducing the number of levels fused.

Anterior-posterior fusions are often needed for severe curves over 60–70 degrees, for rigid curves (do not improve to less than 50 degrees in bending), and for skeletally immature children who are at risk for crankshaft deformity (282). However, the use of new and stronger pedicle screws may allow the anterior portion of the fusion to be deferred. In symptomatic adults with untreated idiopathic scoliosis, anterior-posterior fusions can be required for correction, with possible spinal cord decompression taking place as well.

Neuromuscular Scoliosis

Curve Types. Scoliosis in neuromuscular disease is common. These curves occur with quadraplegic cerebral palsy (up to 70%), muscular dystrophy, or quadraplegic spinal cord injury (up to 90%); are long and sweeping; begin early; and progress quickly, affecting pelvic symmetry. They are fairly unresponsive to bracing, and may require extensive fusions to slow progression (282,293). Young, nonambulatory patients with thoracolumbar curves are at greatest risk for curve progression. A comparison of the surgical hospitalizations of children with neuromuscular scoliosis to those with idiopathic curves shows their stays to be longer, more complicated, and more costly (294). These children often have neurological, pulmonary, cardiac, or gastrointestinal comorbidities that affect their ability to wear spinal orthosis or undergo surgery, so careful surveillance of curve progression is important.

In children with myelomeingocoele, rapid progression of scoliosis may be indicative of a tethered cord, worsening hydrocephalus, or hydromyelia. In children with neurofibromatosis, intraspinal tumors may have developed. In both instance, MRI versus radiographs should be obtained.

Nonsurgical Management. Orthosis are often utilized in idiopathic curves of less than 40 degrees to delay progression. However, in neuromuscular scoliosis, while orthoses may improve trunk control and sitting posture, they less often slow curve progression and do not prevent the need for surgical intervention (293). Medical conditions such as rib cage deformity, pulmonary disease, gastroesophageal reflux, or insensate skin may make brace wear difficult, and the presence of coexisting feeding tubes, intrathecal baclofen pumps, or vagal nerve stimulators may complicate fit even further.

Orthotic wear is often abandoned unless it is useful for sitting support, for improving head position, or for stabilizing a flexible thoracic kyphosis. Often, a soft foam orthosis rather than one of rigid orthoplast, will be more tolerable to the patient, have less adverse impact on pulmonary function, and yet still provide adequate positioning support (295,296). The benefits versus disadvantages need to be fully explained to families before these expensive custom braces are fabricated.

Spasticity is a common issue in many children with cerebral palsy and neurologic impairment, and the use of peripheral botulinum toxin injections and intrathecal baclofen are fairly commonplace for its treatment. For scoliosis, botulinum toxin injections in the concave-side paraspinals for counterparalysis in progressing curves have been reported (297). It may provide some short-term benefit in patients who are progressing rapidly and in whom surgery must be delayed for medical reasons, especially if bracing is done concurrently. Intrathecal baclofen therapy has not been noted to have a significant impact, either positively or negatively, on curve progression (298). Due to the entrance of the intrathecal catheter at the thoracolumbar junction, care needs to be taken when fusing the spine post-pump placement, or when inserting the intrathecal catheter after a fusion, to avoid complications such as disruption of the catheter, infection, or a dural leak (299).

Surgical Intervention. Surgical interventions for neuromuscular curves differ from those of idiopathic curves due to their continued progression after maturity, the likelihood of concurrent pelvic obliquity, the osteopenic bone that must support instrumentation, and the length of the curves that are often present. Sublaminar wires, pedicle screws, and hooks are often used to provide segmentally stability, although Luque-Galvaston rods may be used for large curves or when pelvic obliquity is present (288,300). Lengthier fusions that extend from T2 to the pelvis are common in nonambulatory patients, although pelvic stabilization is avoided if possible in ambulators to reduce problems related to limiting lordosis. Posterior fusions are preferred, as this bone is more stable and there is more difficulty accessing the anterior spine because of the diaphragm. Anterior fusions also produce sympathectomies and are associated with superior mesenteric artery syndrome (285). Anterior-posterior fusions are often done for severe curves (>60 degrees), although this may involve a two-stage procedure and may not improve correction that significantly. While safe, effective, and at times necessary due to a patient's medical stability or surgeon's skill, staging can increase cost and length of hospital stay (301).

As neuromuscular curves over 50 degrees may continue to progress at a rate of 1.5 degrees per year even after maturity, the long-term advantages of early

surgical intervention need to be discussed so that valid anticipatory guidance can be given (282). Advancing age, reduced bone quality, more rigid curve, limited respiratory reserve, and impaired skin integrity can adversely affect outcome. Functional goals of maintaining sitting tolerance, cosmesis, transfer capabilities, pulmonary and gastrointestinal function, and skin integrity are typical long-term concerns that should be considered.

In children with spina bifida, partial or complete vertebral body resections or fusions may be necessary to achieve stability (282). Children with Marfan's syndrome and Freidrich's ataxia, often have curves that are shorter and can be treated as though they were idiopathic curves (282). Achondroplasia may result in thoracolumbar kyphosis, lumbar stenosis, and lordosis. When these children experience pain or neurologic deficit, decompression and fusion is often necessary (282).

In children with Duchenne's muscular dystrophy, scoliosis is often relentless and progresses at up to 8 degrees per year. The use of oral steroids such as deflazacort to slow the decline in muscle strength and delay nonambulatory status can significantly attenuate the development of scoliosis and need for spinal surgery (300). Surgical intervention needs to be timed to maximize pulmonary status (FVC >35%) and curves of 20–30 degrees are often corrected in order to improve seating and respiratory function early on (282,302). The increased risk of anesthesia-induced malignant hyperthermia needs to be recognized in this population.

Preoperative nutritional and health optimization and perioperative infection and pain control are important to successful spinal surgery. Postoperative nutritional supplementation, pressure sore vigilance, pulmonary toilet, gastrointestinal motility, aspiration prevention, and rapid upright sitting posture and mobility to prevent deep venous thrombosis are needed. These issues as well as adequate discharge planning need to be addressed proactively by both the physiatrist and the surgeon in order to achieve best outcomes.

Despite the challenges of surgical correction in children with neuromuscular scoliosis, studies show that curve degree, lung function, seating position, and activities of daily living may all improve postoperatively, potentially improving quality of life and caregiving abilities (280).

Functional Scoliosis

Functional or "secondary" scoliosis is a flexible, nonbony curve secondary to leg length discrepancy, herniated disk, spondylolisthesis, discitis, muscle spasm, trauma, arthritis, or hip disease. Treatment of the underlying problem typically resolves the curve.

Leg Length Inequality

Leg length inequality is common, with estimates of up to one-third having a 2-cm or less discrepancy measured between the length of their legs (6,303). There are two basic types of leg length discrepancies: true and apparent. True leg length discrepancy is present when bilateral leg length measurements between the greater trochanter and the medial malleolus demonstrate shortening on one side. Apparent leg length discrepancies are present when bony lengths are the same but joint alignment or pelvic femoral asymmetry is present (eg, adductor spasticity, pelvic obliquity). Apparent discrepancies can best be measured using a tape measure from the umbilicus to the medial malleolus of either side.

Radiographic measurement is the most reliable. The scanogram technique avoids magnification by taking separate exposures of the hip, knee, and ankle so that the central x-ray beam passes through the joints, giving true readings from scale (Fig. 14.23) (304,305). CT scanogram is still the standard, reducing errors from angular deformity (306). If the examination is done specifically for this purpose, economic cost can be competitive (multiple sections unnecessary) and radiation exposure less with microdose technique (307,308). Causes of true leg length discrepancy are many and can be classified by growth retardation versus growth stimulation (6,20). Growth retardation has included conditions such as congenital hemiatrophy, developmental hip dysplasia, Legg-Calve'-Perthes disease, slipped femoral capital epiphyses, polio, achondroplasia and dyschondroplasia, and severe burns. Causes by growth stimulation include congenital giantism, Wilm's tumor vascular abnormalities such as Klippel-Trenaunay-Weber, thrombosis or femoral or iliac veins,

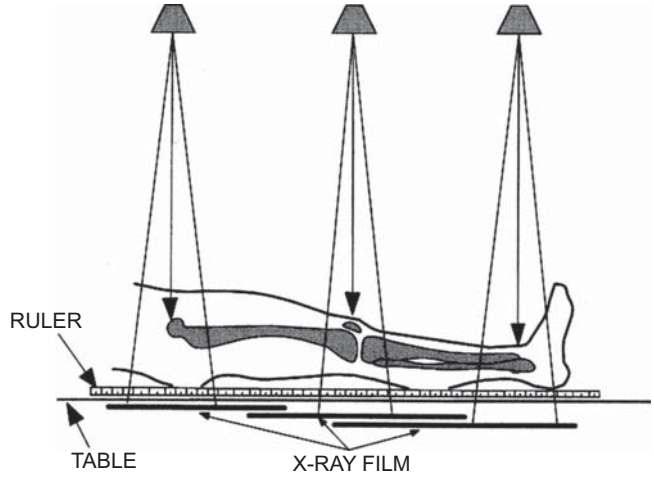


Figure 14.23 The scanogram technique avoids errors of magnification and is preferred for children who can remain still for three exposures.

and traumatic arterial venous aneurysms. Tumors such as giant cell, neurofibromatosis, and bony fractures can cause other growth retardation or growth stimulation. The child with true hemihypertrophy needs to be screened every four months for the possibility of Wilm's tumor (6,309) up through the age of 8 and every 6 to 12 months through the age of 10. Eighty percent of Wilm's tumors present prior to the age of 8, with an average age at presentation of 3 years. The tumor may be associated also with aniridia (lack of an iris in the eye) and secondary metastases to the skeleton. A firm, nontender abdominal mass may be palpated. Damage to the growth plate with trauma and epiphysiodesis, including fractures with marked-over riding of fragments, tend to cause more growth retardation.

Treatment objectives include obtaining leg length equality, producing a level pelvis, and improving function. Leg length discrepancy of less than 1.5 cm is usually just observed. Shoe lifts can be utilized for differences up to 3 cm. Horizontal alignment of the iliac crest or sacral base in the standing position should also be witnessed with appropriate shoe lifts in place. Early attention should be given by the age of 7 or 8 to observe and record the pattern of growth and appropriately time the growth plate arrest.

The Greulich-Pyle norms for skeletal maturation of the hand (310) and the charts of Green-Anderson (311) are used for prediction of future growth and the timing of surgery when stapling epiphysiodesis of the longer side is considered for true discrepancies between 3 and 6 cm. Stapling techniques across the physis produce a tethering effect and can be removed later once equalization has been achieved (312,313). Surgical epiphysiodesis is an all-or-nothing procedure that completely and permanently arrests physeal growth. The principle is to produce a symmetrical bony bridge that tethers the physes and prevents future growth (314). Epiphysiodesis is most commonly performed two to three years prior to maturity (girls age 11 or 12 years; boys age 12 or 13 years).

Shortening procedures can also include removal of a section of bone for limb equalization performed in adults or adolescents who are no longer growing (11). Charts of Green and Anderson are displayed in Figures 14.24 and 14.25, respectively. Total leg length versus skeletalage for boys and girls are shown respectively. Plotting of leg length versus skeletal age is critical in the timing of any surgical procedure projecting limb length equalization into the future (315,316). The Green and Anderson studies provide good documentation for the general population studied, but no guarantees for children of other races or genetic stock. Additional and more specific determination of leg length discrepancy can be obtained through three additional methodologies (6,11). These include the arithmetic method, the growth remaining method, and the straight line graph method—not described further, as such is beyond the scope of this text. Growth discrepancies beyond 6 cm are best treated by limb lengthening through such methods as Wagner or Ilizarov (6,11). Unlike epiphysiodesis, leg-lengthening procedures can be performed at almost any skeletal or chronologic age. Discrepancy greater than 15 to 20 cm should consider combined shortening and lengthening procedures in addition to amputation. Codivilla first reported mechanical bone lengthening in 1905 (317). Subsequent advancement in limb lengthening has been by the method of Ilizarov (318), whose biologic principle of distraction osteogenesis has revolutionized the surgery. Ilizarov's circular external fixation system is complex, but provides for multilevel correction, including angular deformities and lengthening simultaneously (Fig. 14.26). Corticotomy technique is utilized with care so as not to disturb the medullary cavity contents so that they may make their greatest contribution to osteogenesis during lengthening (6,11).

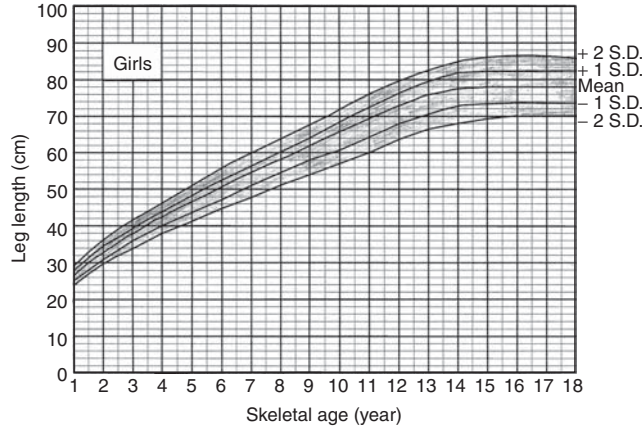


Figure 14.24 Grafts showing total leg length vs. skeletal age for girls. It provides useful analysis of leg length data, allowing a projection into the future on the basis of present status.

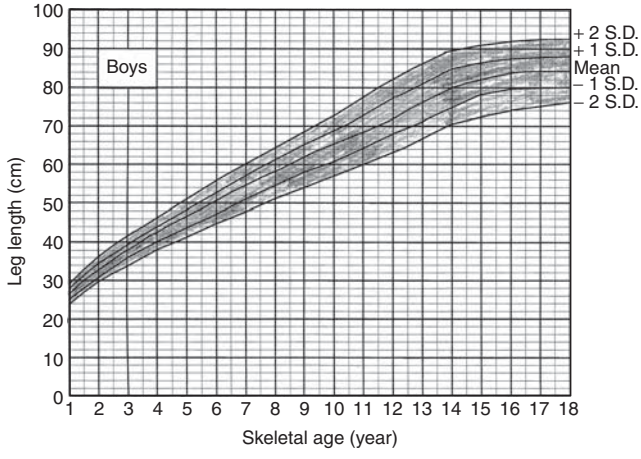


Figure 14.25 Grafts showing total leg length vs. skeletal age for boys. It provides useful analysis of leg length data, allowing a projection into the future on the basis of present status.

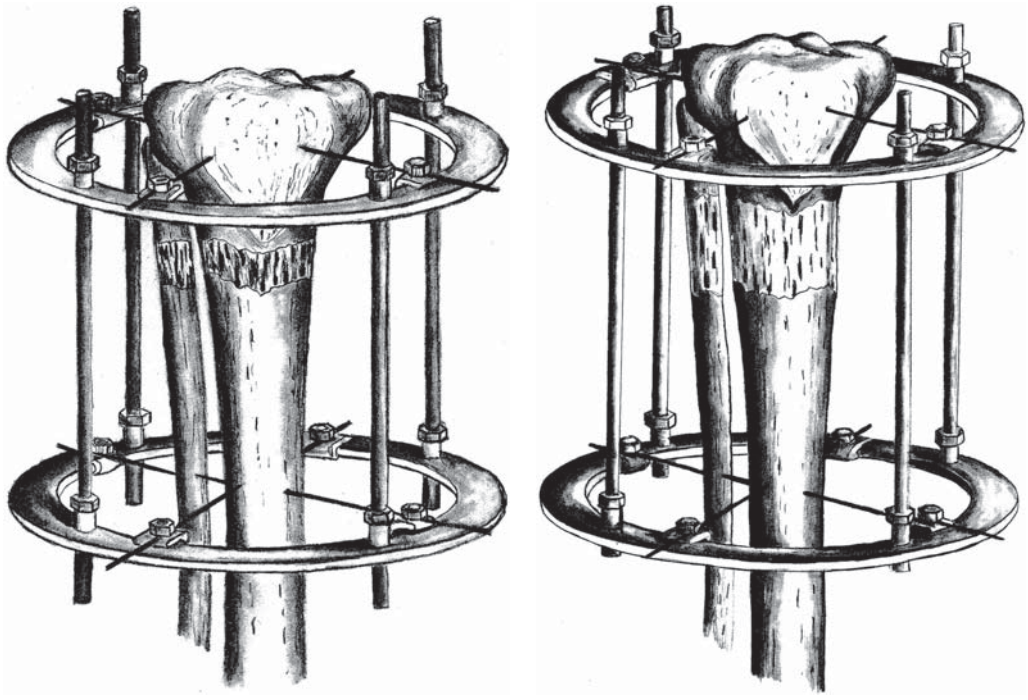


Figure 14.26
(A) Sequential metaphyseal lengthening. (B) Elongation through the active metaphysis promotes osteogenesis and strength by the large cross-sectional area across the lengthening gap.

Elongation through the metaphyses promotes osteogenesis because metaphyseal bone is so active and promotes strength by the large cross-sectional area. The lengthening process begins approximately 5 to 10 days after surgery. Lengthening of 1 mm per day or approximately 1 inch per month is recommended (6,11). External fixators are worn until the bone is strong enough to support the patient safely. This usually takes about three months for each inch. A normal lifestyle during treatment is encouraged. Some children even go swimming with the external fixator in place. Complications include pin tract infections (most common), fracture, axis deviation, delayed union, and soft tissue contractures. A child whose family is not capable of sustained follow-up may be a poor candidate for limb lengthening. Significant patient and family education needs to occur, including preoperative and postoperative phases, preparing the child and family both physically and emotionally for the long treatment. Counseling services may prove helpful. Rehabilitation services are most helpful, including frequent physical therapy visits for any successful long-term outcome.

CONSTITUTIONAL OR INTRINSIC BONE CONDITIONS

Constitutional conditions of bone may be divided into five categories:

- Defects of tubular bone or spinal growth
- Disorganized cartilage and fibrous components
- Abnormal bony density or structure
- Metabolic conditions usually affecting calcium or phosphorus metabolism
- Extraskeletal disorders

Defects of Tubular Bone or Spinal Growth Present at Birth

Achondroplasia

More than 350 conditions (319) may be defined in these mostly inheritable groups of skeletal dysplasias of which achondroplasia is the most common. Achondroplasia is an autosomal-dominant disorder; with approximately 85 % new mutations, it is the most common of the skeletal dysplasias (320).

Clinical Features. The diagnosis of achondroplasia is made clinically with characteristic features on radiograph. These conditions are often associated with shortened trunk, narrow thorax, and variant body proportions, including enlarged head size with frontal bossing, hypoplasia of the midface, short limbs and fingers, lordotic lumbar spine, and bowed legs. Although typically normal in intelligence, secondary to their size, these individuals are often looked upon and treated as younger than their stated age. Secondary to transitory muscular hypotonia, early motor milestones are frequently delayed in infancy. Visual spatial learning issues or deficits similar to other children with compensated hydrocephalus may

be observed. Physical therapy that includes exercises with these babies prone are also important in order to minimize thoracolumbar gibbus (320).

Prevention: Obesity. There are specific weight charts for those with achondroplasia (321). Obesity is also correlated with the increase in cardiovascular-related deaths (322). Lumbar-region symptomatic spinal stenosis may be seen in achondroplasia; it is aggravated by obesity. Signs of this include lower back and leg pain, and may be observed in 50% of those with this condition. There may be weakness, altered deep tendon reflexes, paresthesia, and later, claudications. Early treatment includes anti-inflammatory medication and corticosteroid injections to treat lumbar radiculopathy, with one-third eventually needing lumbar laminectomy (323). Kyphoscoliosis is common.

Prevention: Ear, Nose and Throat. Tonsillectomy and ventilation ear tubes may help prevent conductive hearing loss. Otitis media may be recurrent secondary to shortened eustachian tubes secondary to midface hypoplasia. This is a significant problem in approximately 40% of those with achondroplasia. Often, there are too many teeth than can be accommodated and teeth need to be pulled or the jaw needs to be expanded (323). This is necessary for dental alignment.

Precautions, Monitoring, and Surgical Intervention. This group requires precautions with regard to atlantoaxial instability. The instability may be from maldevelopment of the odontoid, transverse ligament laxity, or longitudinal ligament abnormalities. MRI of the brain, including the cervical junction and the spinal cord, is recommended between the ages of 6–12 months. Signs of cervical cord compression (myelopathy) are increased reflexes of the lower extremities, clonus, severe hypotonia, central sleep apnea, and sudden death. Polysomnography is used to demonstrate the central sleep apnea (323). Referral to the appropriate specialist is necessary for evaluation and treatment.

Hydrocephalus, if present in achondroplasia, must be carefully evaluated and may need surgical intervention (324). Head circumference must be monitored every six months while growing (especially in the first two years of life), and symptoms of increased cranial pressure must be evaluated (320,323). MRI of the cervicomedullary junction, as well as CSF flow studies, may be normal with a neutral neck position. With flexion and extension of the cervical spine, complete blockage of CSF flow in the former and posterior cervicomedullary compression in the latter may be demonstrated (324). Flexion and extension imaging is warranted if there are mild to severe symptoms and signs present, as surgical options such as ventriculoperitoneal (VP) shunt or decompressive surgery can be corrective (324).

Treatment. This may include human growth hormone therapy. The long-term sequelae of this are unknown. Parathyroid hormone has been shown to improve bone growth and mitigate the effects of FGFR3 mutations found in achondroplasia (325). Limb lengthening is a possibility, but has many risks involved, including infections as well as soft tissue, nerve, and joint damage. This remains controversial (323). There is an increased number, compared to the general population, of sudden deaths thought to be caused by foramen magnum stenosis in children under 5 years of age, and an increase from cardiac disease and neurological diseases, including drug overdose and suicide in older patients (322). Reports of depression, low self-esteem, poor body image, and chronic pain need to be addressed. The key to successfully treating this condition is a multidisciplinary team that includes the rehab specialist, occupation and physical therapists, social worker and/or psychologist, neurosurgeon, cardiologist, and orthopedist.

Disorganized Cartilage and Fibrous Components

Fibrodysplasia

Fibrous dysplasia is a condition characterized by the presence of expanding fibro-osseous tissue in the interior of affected bones. It is characterized by cancellous bone being replaced by fibrous tissue. Primarily this is a lesion of the growing skeleton.

Clinical Characteristics. Fibrous dysplasia may cause pain or limping gait, extremity length discrepancy, bowing, or fractures. This may be associated with endocrine abnormalities such as Albright's syndrome, which consists of the triad of multifocal bone involvement, precocious puberty, and cutaneous pigmentation.

Diagnostics. Radiographic lesions typically are sharply marginated with sclerotic bone and appear as ground-glass or lytic expansile lesions of the diaphysis or the metaphysis.

Treatment. Treatment typically includes observation. Surgery may be necessary for those lesions causing progressive deformity, pain, fracture, or impending fracture.

Abnormal Bony Density or Structure

Osteogenesis imperfecta

Osteogenesis imperfecta (OI) is a heritable bone disorder with abnormal bone quality or quantity (326).

Characteristics. Fractures are the hallmark of OI. The number of fractures in a lifetime vary from a few to several hundred. There are numerous associated clinical findings, but phenotype can vary greatly even within families with the same genotype (327). Short stature is common, as are a relative macrocephaly and triangular facies (328). Cognition is normal. People with OI tend to have a high-pitched voice. In addition to fractures, musculoskeletal findings can include scoliosis, muscle spasms, and hypermobility. Multiple bone microfractures can lead to bowing and increased fracture risk (329). Respiratory failure is the leading cause of death in OI (330). Basilar impression, an abnormality of the skull base, can cause compression and neurologic compromise (331). Skin tends to be fragile, leading to increased bruising (328). With fractures and bruising, some children with mild OI may be difficult to distinguish from those sustaining nonaccidental trauma (326,328). Hypercalcuria and renal calculi can occur, as can aortic dissection and mitral valve prolapse (328). Hearing loss may require amplification or surgery. Dentinogenesis imperfecta can be present.

Sillence described four types of OI (326,327,332, 333,334,335), as outlined in Table 14.7. There is overlap in the clinical presentation, particularly Sillence types III and IV. In recent years, new types of OI have been described based on unique structure found on bone biopsies as well as clinically distinguishing features (326). Previously, these patients had been described as having type IV OI, but were found to have normal type I collagen.

Rehabilitation. Infants can be positioned to encourage active movement while decreasing fracture risk (336,337,338). Improvement of head control can be encouraged by prone lying or lying on a reclining parent's chest or shoulder. Towel rolls can be used to avoid excessive hip abduction while supine or support the infant's back in side-lying. Diaper changes should be done by rolling the infant, not by lifting the legs (339). Lifting the infant should be done with a wide base (eg, hands spread apart), not under the arms.

Range-of-motion exercises should be active (337). Aquatic therapy has been recommended to increase strength and mobility (337,340). Weight bearing can improve bone strength. Clamshell bracing may be used to provide support for weight bearing (336,338,341). Long leg bracing may be shortened later (336). There has been a trend for less bracing recently, as infants have been starting early intervention programs and sit in their first year (336).

Sports and recreation activities may be added in the school-age or teen years (342). However, high-impact activities such as gymnastics, aerobics, martial arts, hiking, and contact sports are not recommended (343). The Osteogenesis Imperfecta Foundation published a book that provides detailed therapy recommendations and rates the relative risks and benefits of various sports (344). They also provide excellent resources for patients, families, physicians, nurses, and therapists (345).

Children should stand or walk daily (343). They may benefit from playing a wind instrument or singing to improve pulmonary health (343). Independence with activities of daily living can be gradually increased. Children should avoid staying in their wheelchairs for the entire day. Armrests can be removed from manual wheelchairs to decrease forearm bowing (338). Adolescents can learn to manage their medical care, drive, and transition to college or work (346).

Medical Interventions. Multiple drugs have been tried in OI without success until the bisphosphonates were shown to increase bone density, decrease risk of fractures, increase mobility in some patients, and decrease pain (347,348). Side effects such as transient fevers and discomfort were relatively mild for most children. Markers of bone turnover decreased. Some studies have shown benefit for infants and toddlers (349,350). There are concerns that prolonged use could cause decreased bone healing (348). Long-term risks are unknown. Some studies (351,352), but not all (353), have shown improved function with bisphosphonates treatment. The optimal drug and dosing has not been determined (354).

Surgical Interventions. The risk of fracture has been found to increase significantly when long bone angulation was 40 degrees (341,355). Intramedullary rods can improve fracture risk but can migrate into joints (356). As a child grows, the bone "unprotected" by the now too-short rod can break. Telescoping rods have been used, but still have risks. Some surgeons have found fewer surgical complications in children treated with bisphosphonates (357).

Outcomes. Despite the fractures, surgeries, and mobility impairments common in OI, people with OI rate their quality of life well (358,359). A recent study showed that children with OI rated higher than the reference norms on the psychosocial summary of the Child Health Questionnaire (359). Adults with OI often attend college and have employment similar to the general population without disabilities (358).

Metabolic Conditions Usually Affecting Calcium or Phosphorus Metabolism

Rickets

Rickets is caused by vitamin D deficiency that results in osteomalacia, the delayed or inadequate mineralization of osteoid in mature cortical and spongy bone (5). Rickets

14.7 Types of Osteogenesis Imperfecta

TYPES OF OSTEOGENESIS IMPERFECTA	SCLERA	GENETIC ANOMALY	SEVERITY	DISTINGUISHING CHARACTERISTICS
I	Blue	Reduced Amount of type I collagen	Usually mild Few fractures	Most common form, usually without bony deformities; hearing loss may be main disability
II	Blue	Abnormal structure of type I collagen	Most severe, often fatal in perinatal period	Severe respiratory compromise often
III	Blue or gray, may fade	Abnormal structure of type I collagen	Severe bone fragility	Progressive long bone deformities; very short stature; may have respiratory insufficiency
IV	Blue or gray, may fade	Abnormal structure of type I collagen	Variable, often moderate	Bowing of long bones is usually less severe than in type III
V	White	Unknown	Moderate to Severe	Hypertrophic callus; fusion of intraosseous membranes; "meshlike" bone biopsy
VI	White	Unknown	Moderate	Extremely rare (8); characteristic bones biopsy with mineralization defect (1)
VII	White	Mutation of Cartilage-associated protein (CRTAP) gene	Moderate to lethal	Rhizomelia coxa vara is common; may resemble types II or VI recessive
VIII	White	Mutation of LEPRE 1 gene leads to abnormal propyl 3-hydroxylase activity	Severe or fatal (8)	May resemble type II or III; bone is under mineralized; recessive

is a rare condition in the United States. However, it may be found in higher numbers in dark-skinned breastfed babies who are unsupplemented and all breastfed babies who themselves and/or their mothers have little to no exposure to the sun on a daily basis. Typically, this becomes problematic after 6 months of age.

Clinical Characteristics. The clinical features of nutritional rickets include early-onset craniotabes, rachitic rosary (costochondral junction enlargement), and thickening of the wrists and ankles. As rickets continues, clinical findings include progressive bowing of the legs; poor linear growth; and abnormal serum calcium (ionized calcium is the most accurate test), phosphate, and alkaline phosphatase levels. In severe cases, the baby may have seizures.

Diagnostics. The diagnosis of rickets is made with radiographic demonstration of metaphyseal flaring, cupping, and decreased mineralization of the distal metaphysic, as well as laboratory evidence of elevated alkaline phosphatase.

Treatment. The treatment includes supplementation of vitamin D and/or formula. If left untreated, permanent deformities may ensue.

Mucopolysaccharidoses

Mucopolysaccharidoses are hereditary progressive conditions secondary to the accumulation of the mucopolysaccharides (MPS). The underlying problem is a defect in the degradation of MPS leading to

accumulation in lysosomes (vacuoles found in almost all cells) (360). There is marked heterogeneity within each of the groups, and life expectancy for some can reach into the fifth decade (361). In general, the later the clinical onset, the slower the clinical picture (360,362).

Clinical Characteristics. Typically, the facial features are coarse. Dwarfism is present to some degree in all these entities. Odontoid hypoplasia can be serious and lead to tetraplegia. Atlantoaxial instability frequently requires fusion, as it is a major complication of this condition, causing spinal cord compression with resulting tetraplegia. Fingers are short and stubby, and hands are wide. Carpal tunnel syndrome (CTS) is common. Typically, the presenting complaint for CTS is difficulty with fine motor tasks, not pain (6). Progressive spasticity and mental deterioration eventually occurs in most types. Kyphosis can appear early and is usually marked. Blindness may result from optic atrophy. Corneal clouding is a common finding in MPS (362). Deafness may occur as well.

Specific Types. Mucopolysaccharidoses are usually divided into six groups. In MPS I, the most severe form is Hurler's syndrome and the mildest form is Scheie's syndrome (MPS IS). The mode of inheritance is autosomal recessive for all groups except MPS II Hunter's disease, which is X-linked recessive. Type III is Sanfilippo A, B, C, D. This is the most common MPS (362).

Type IV Morquio's disease (mucopolysaccharidosis type IV) is characterized by normal intelligence and gross motor milestones early in life. Over time, gait may progressively worsen with severe genu valgum, ligamentous laxity, severe pes planus, and increased sternal protrusion. The chest deformity can be restrictive and cause cardiorespiratory symptoms (362).

Type VI Maroteaux-Lamy has the facial features typical of MPS; intellect generally remains normal; and obstructive sleep apnea, corneal clouding, and deafness are common. VII Sly type can present as hydrops fetalis, or life expectancy can be into the second decade. There can be a wide variability of cognitive ability.

Rehabilitation. A multidisciplinary approach is essential for management of these highly variable MPS. Specific rehabilitation issues may include hand and wrist bracing in neutral to help avoid carpal tunnel syndrome, leg braces for the lower extremities to help avoid contractures and deformities, and TLSO for the back to help avoid scoliosis. Aids for functional independence are essential.

Medical and Surgical Interventions. Bone marrow transplant (BMT) can alter the severe nature of MPS (360). Enzyme replacement therapy (ERT) is available for MPS I, II, and VI (359) and may be beneficial for type VII as well (362). The ERT does not cross the blood–brain barrier or enter the joint space/cartilage or cornea. Earlier BMT in MPS is thought to have the best possibility of good results. Consent for this treatment is often difficult when the child is doing well. VP shunt can help manage hydrocephalus in type I to help preserve intellectual function (362). Similarly, in type III, VP shunting can help with behavioral changes in some patients. Genetic consultation is important secondary to prolonged life expectancy.

Extraskeletal Disorders

Sickle Cell Anemia

Sickle cell anemia has been discussed earlier in this chapter. The reader is referred to outside references for additional discussion as needed.

Chronic Kidney Disease

Children with chronic kidney disease (CKD) are at great risk for short stature. Adequate nutrition may be problematic. With failing kidneys, erythropoietin production is inadequate and anemia may result. These children may be resistant to their own elevated growth hormone (GH) and may require recombinant human GH subcutaneously (363). Renal osteodystrophy is a term that describes the bone disorder spectrum in CKD. It is most commonly associated with a high turnover bone disease secondary to hyperparathyroidism (363). Osteitis fibrosa cystica is the pathologic skeletal finding in this condition. The excessive parathyroid hormone is a response to correct the hypocalcemia by increasing the bone resorption (363). Clinically, these patients have muscle weakness, bone pain, and fractures from minor trauma. Rachitic changes as well as varus and valgus deformities of the long bones and slipped capital femoral epiphyses may be seen in growing children. The x-rays demonstrate subperiosteal resorption and widening of the metaphyses in the hands, wrists, and knees (363). Medical management for this condition is by a nephrologist. Diets include low phosphorus, phosphate binders, vitamin D, and non-calcium–based diets for those who are prone to hypercalcemia. Recombinant human erythropoietin subcutaneously and iron orally or intravenously are important treatments for anemia (363).

Summary

As mentioned at the onset of this constitutional bone condition section, typically, these individuals have normal intelligence. They may, however, be perceived

differently, especially if they are smaller than their chronological age. Age-appropriate interventions are key in this group. It is important to know the key features in these groups as well as serious complications. As these conditions generally have increased risk of atlantoaxial instability, these individuals should be restricted from contact sports and other high-risk activities (364).

MUSCULOSKELETAL PAIN AND CHILD ABUSE

Musculoskeletal pain in children is variable. Depending on age and verbal and cognitive abilities, assessing pain in the pediatric patient may present additional challenges. Children under 5 years may have difficulty describing pain. A scale with faces illustrating different emotions may help children describe how they feel (365). At the age of 6 years, children can usually score their pain on a level between 0–10 by increasing severity (364).

Complex Regional Pain Syndrome

Complex regional pain syndrome, also known as reflex sympathetic dystrophy, is a condition that usually involves one limb and more commonly the lower extremity in children.

Clinical Characteristics. Complex regional pain syndrome is characterized by pain, hyperesthesia, edema, cold or warm extremity, cyanosis, mottling of skin, limited range of motion, and patchy bone demineralization.

Diagnostics. Unlike adults, who usually have an inciting event such as a fracture, surgery, prolonged immobilization, or vascular insult, children usually do not have a clear event that precipitates the condition (366). There appears to be a sympathetic nervous system reflex arc mechanism of action. The majority of children with this condition are teenage girls around 12–13 years of age. Radiographs are useful to rule out a fracture or osteomyelitis (367). Regional nerve blocks may be both diagnostic and therapeutic. The diagnosis should be considered with trauma and pain that is out of proportion to the stimulus and worsened with use.

Treatment. A multidisciplinary approach is useful. The earlier the recognition and treatment, the more rapidly recovery is possible. Once contractures and atrophy set in, this is a much more difficult entity to treat. Some advocate medications such as calcium channel blockers, beta blockers (propranolol), and tricyclic antidepressants such as amitriptyline.

Fibromyalgia

The etiology of fibromyalgia in children and adolescents is unclear.

Clinical Characteristics. Diffuse musculoskeletal pain involving the neck, back, and upper and lower extremities is common in fibromyalgia in children and adolescents. Sleep disturbance, headaches, fatigue, and problems with peer relationships (368) are common among those diagnosed.

Diagnostics. Polysomnography is frequently positive while other tests are negative (369). There is an increase in children whose mother has the condition; this may be cultural rather than genetic. Females are more affected, and the onset ranges from around 11.5 to 15 years. Children with fibromyalgia can have fewer trigger points than adults, although the exact number is uncertain.

Treatment. Education and psychological interventions are the first line of treatment.

Back Pain

Back pain in children is relatively uncommon. Usually, the child with back pain presents with a muscular strain-type pattern related to poor posture, activities at school or home, or other recreational or sporting pursuits (6). When carrying backpacks of greater than 10% to 20% the body weight of the child or adolescent, musculoskeletal strain is common. Children and adolescents generally do well with strategies such as decreasing backpack weight, making sure the backpack is level to their shoulders, carrying the backpack on both shoulders, and using proper body mechanics when picking up items from the ground. With prompt adherence to these guidelines, only a small percentage of children and adolescents go on to have chronic symptomatology. Conservative intervention with physical therapy, correction of biomechanics, postures, equipment, and sporting environment are often all that is required for resolution of symptoms. NSAIDs along with the RICE protocol are utilized as well. Back pain that is not improving within two to four weeks of conservative care needs to investigated in a much more serious manner. Unlike the adult, chronic back pain in children can be met with serious pathologic entities, including neoplasm, infection, and noninfectious inflammatory disease (11). A full discussion of back pain in children is beyond the scope of this text, and the reader is referred to other sources (6,11,206). As mentioned previously in the text, backpacks are being utilized more often in children and adolescents, particularly to and from school and other recreational environments.

Referred Back Pain

Many conditions can produce referred back pain. These include pyelonephritis, pneumonia, endocarditis, cholecystitis, pancreatitis, osteomyelitis, pelvic inflammatory disease, and other more general conditions affecting the muscles, as well as conditions such as inflammatory arthritis. Sickle cell pain crisis can cause back pain. Conditions that usually have the presenting complaint of nighttime pain are osteoid osteoma, a benign bone tumor that is the most common neoplasm, and ankylosing spondylitis. For malignant neoplasms of the spine, 90% are secondary sites and not primary tumors. Functional pain issues also present the clinical challenge of ruling out underlying, more serious disease. A good history and physical exam often points out inconsistencies.

Child Abuse

There are approximately 1,200 deaths from child abuse or nonaccidental trauma annually. Approximately half of these deaths happen in the first year of life. About half of the children who died were known to their local child protective service agencies. The most common injuries were soft tissue followed by fractures. It is estimated that one out of four fractures in children under 1 year of age are from abuse. The most common fracture for children with just one fracture is the femur, followed by the humerus, followed by the skull. Posterior rib fractures are found in up to 30% of abused children, with the majority found in those under the age of 2 years.

Initial action depends on whether the suspicion is great enough to warrant making a report to Child Protective Services (CPS) (370). It is essential to obtain a detailed history, including the mechanism of injury, and to look for inconsistencies. Knowledge of child development is essential. Suspicion is increased if the injuries are inconsistent with the child's developmental level or mechanism of injury, blamed on the victim's siblings, or not witnessed. Children, for example, generally cannot roll over until the age of 4 months. Most children that fall off a piece of furniture have a fracture risk of less than 2 percent. Therefore, a history of a 3-month-old rolling off a piece of furniture and sustaining a severe injury should raise suspicion of child abuse. Multiple injuries in various stages of healing should increase suspicion.

With suspected child abuse, physical exam includes an ophthalmological examination for retinal hemorrhages as well as a head-to-toe examination that also looks for skin bruising, swelling or deformity of extremities, malnourishment, and poor hygiene. Photos are useful for clinical documentation of any abnormalities and frequently document skin marks, bruises, welts, or burns. An AP and lateral films are necessary for any extremity that is tender, has swelling, or has limited range. A radionucleotide study can be an added help to the skeletal survey. Remember that there are no pathognomonic fracture patterns, but high suspicion fractures include posterior rib fractures; metaphyseal corner fractures; sternum, scapula, or spinous process fractures; bilateral acute long-bone fractures; complex skull fractures; fingers in nonambulatory children; and multiple fractures in various stages of healing. The most common type of fractures involved with child abuse are transverse, followed by spiral fractures, followed by avulsion fractures, followed by oblique fractures. Those fractures with low specificity include clavicle fractures, simple skull fractures, and isolated long-bone fractures (371). When child abuse is suspected, the physician is legally obligated to file a report with the appropriate child protection agency. Adequate supportive measures and counseling should be in place before returning any abused child to the home. When in doubt, temporary foster placement should be seriously considered.

Tumors of the Bone

The prevalence of bone tumors in the United States is approximately 7 children per million under the age of 15 years. Although rare, with approximately 400 cases diagnosed per year, osteosarcoma is the most common primary malignancy of bone during the adolescent growth spurt (rapid bone growth) (309). There is a slight preference for boys. It is followed by Ewing's sarcoma, with approximately 200 cases diagnosed per year. Ewing's sarcoma is more common in those younger than 10 years of age (372). However, both tumors present more commonly in the second decade of life. Osteosarcoma may develop from irradiation treatment of Ewing's or other malignancies. Tumors may mimic various pain syndromes throughout the body. Primary bone tumors common to the upper extremities include Ewing's sarcoma of the scapula, osteogenic sarcoma of the proximal humerus, and osteoblastomas and chondroblastomas common in the diaphyses and epiphyses of long bones (373). The most common presenting manifestations of osteosarcoma are pain, limp, and swelling. Similar presentation may be found in Ewing's, as well as weight loss and fever.

Diagnostics. The timing of the presentation complicates the differential diagnosis. The symptoms may be attributed to a growth spurt, sprain, or sports injury. Those presenting with osteosarcoma are usually taller than their peers. A complaint of pain that awakens a child or adolescent from sleep is suggestive of malignancy. The most common location of the osteosarcoma is the distal femur, followed by the proximal tibia

and proximal humerus. Symptoms not responding to conservative treatment require further investigation, specifically with a radiograph. A sunburst pattern or Codman's triangle (lifting of the cortex by new bone formation) are classic radiographic findings found in two-thirds of those presenting with osteosarcoma. With Ewing's, a permeative "moth-eaten appearance" is demonstrated on x-ray. If suspicion of tumor is high, and radiograph is negative, seen with medullary osteogenic sarcoma, MRI should be obtained of the entire long bone, as no pattern on x-ray is pathognomonic (374). Laboratory tests, including a complete blood count (CBC), will usually be normal. Elevated sed rate, alkaline phosphatase, or lactic dehydrogenase levels may be found. Early diagnosis is key, as the prognosis is better if there is less spread of the disease. Metastasis to the lungs remains the most likely cause of death. Additional primary bone tumors to the lower extremities include those of the long bones. These include histiocytosis X in the diaphysis and esosinophilic granuloma in the epiphysis. Tumors more common in the area of the pelvis include osteoblastoma, aneurysmal bone cyst, and fibrous dysplasia. Additional metastatic tumors to the lower extremities include neuroblastoma and lymphomas of various types.

Treatment. This requires wide resections of the long bone and adjuvant chemotherapy. Once diagnosed, further workup and treatment is necessary at a center with expertise in managing these tumors.

Rehabilitation. Physical activity and contracture management are important rehabilitation issues during acute treatment. Chronically, residual limb skin integrity, prosthesis management, and contracture management are important issues when managing this patient population. Team management is critical, led by the pediatric rehabilitation medicine specialist in the comprehensive care of patient, family, and loved ones.

PEARLS AND RESOURCES

Growth and Development/Congenital Conditions

Pearls

- An embryologic alteration of the musculoskeletal system often is a superficial marker for embryologic alterations in other organ systems.
- Deferring radiography until a minimum of 6 months in age, allowing bones to ossify, is generally a good idea .
- Malformations of the radius are more common and associated with more syndromes than malformations of the ulna.
- To be a clubfoot, there must be hindfoot varus and adduction.
- Cavus feet always need an explanation and can be a superficial sign of an underlying neurologic diagnosis.

Resources

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Brachial Plexus Palsy

Pearls

- The most common risk factors for a baby having a birth brachial plexus palsy (BBPP) are shoulder dystocia, large birth weights, and multiparous mothers.
- It is critical for the caregivers to have the baby/toddler see/use the arm with BBPP as much as possible to minimize learned disuse.
- A key difference between acquired BPP and BBPP is pain in the former and no pain in the latter (at least after the first two weeks).
- Neuropraxia has no permanent anatomical changes and will resolve; axonotmesis (partial) and neurotmesis (complete) anatomical severance each has long-lasting sequelae.
- Sensory nerve conduction studies in someone from an insensate area with intact SNAP indicate a preganglionic lesion.

Resources

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Children With Rheumatic Disease

Pearls

- Juvenile idiopathic arthritis (JIA) occurs in children before the age of 16 years, persists at least six weeks, and has had other known conditions excluded.
- Treatment of children with JIA is a team approach that focuses on eliminating inflammation, promoting developmentally appropriate function and activity, and minimizing complications.
- JIA or its treatment can cause local or systemic abnormalities, including micrognathia, leg-length inequalities, muscle atrophy, short stature, osteoporosis, and increased risk of infection.
- Enthesitis, uveitis, rash, fever, or lymphadenopathy may be the earliest signs/symptoms in children with JIA and/or juvenile ankylosing spondylitis.
- Pediatric physiatrists can play a key role in the comprehensive management of the child with rheumatic conditions to maximize age-appropriate function, prevent deformity, and manage pain.

Resources

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Burn Injuries

Pearls

- Children under 5 are more likely to sustain scald or contact burns, while older children and adolescents are more likely to sustain burns from flames.
  - □ Refer to a burn center when a child has chemical, electrical, or third-degree burns; partial-thickness burns to 10% total BSA; burns to sensitive body parts; inhalation injury; or complicating medical factors.
- About 10% of burn admissions in children are related to child abuse, and about 10% of all child abuse cases include burn injuries.
- Silicone dressings may help prevent hypertrophic scars in those at risk as well as to improve scar elasticity in already existing symptomatic scars.
- The most common complication of burns is abnormal or hypertrophic scarring that may cause contractures and impaired function.

Resources

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Developmental Conditions

Pearls

- Observation is the rule in Legg-Calvé-Perthes disease, with minimal pain, good ROM, and a strong lateral bony column.
- Overweight, adolescent, altered gait, and hip pain—think slipped capital femoral epiphysis (SCFE).
- Typical DDH, if not detected and aggressively treated prior to 18 months, portrays a much higher risk of lifelong disability and degenerative arthritis.
- If idiopathic toe walking doesn't improve within a few months of aggressive conservative care, a lifelong presentation is likely present.
- In-toeing in the otherwise able-bodied child generally gets better over time, with or without correction of the bony torsional abnormalities.

Resources

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Scoliosis

Pearls

- If pain is associated with scoliosis, check for other etiology, including neoplasm and infection. Scoliosis in children is typically not painful.
- Steroids reduce the incidence of scoliosis in Duchenne's muscular dystrophy, as well as delay loss of muscle strength and ambulation.
- Surgical fusion into the pelvis is generally performed in the nonambulatory child and those with significant pelvic obliquity.
- Atypical left thoracic curves in individuals with idiopathic scoliosis require investigation for spinal pathology, including MRI imaging.
- Scoliotic curvatures 50 degrees or greater have a tendency to progress, averaging about a degree per year over a lifetime.

Resources

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Leg Length Inequality

Pearls

- Leg length discrepancy of 2 cm or less is common, treated with a shoe lift or observed, depending upon functional preference.

Resources

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Constitutional or Intrinsic Bone Conditions

Pearls

- Multiple epiphyseal dysplasia and Legg-Calvé-Perthes disease both involve the capital femoral epiphysis, but Legg-Calvé-Perthes disease is never symmetrical and rarely bilateral.
- Involvement of the atlantoaxial and atlantooccipital articulations always need to be considered in conditions of constitutional bone.
- Little people of adult age almost always have normal intellect and psychosocial being requiring treatment as such, despite a physical stature that might suggest otherwise.
- Macrocephaly in individuals with achondroplasia needs to be followed carefully, including serial head circumferences and surgical referral for symptomatic hydrocephalus.
- Despite atlantoaxial instability, progressive spasticity, and mental deterioration all common to individuals with a mucopolysaccharidoses syndrome, life expectancy can reach into the fifth decade.

Resources

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Musculoskeletal Pain and Child Abuse

Pearls

- Back pain in children, although relatively uncommon, if not improving within a couple weeks of

conservative care needs aggressive investigation for potential life-threatening etiologies.

- Multiple injuries in various stages of healing or those inconsistent with developmental level, blamed on siblings or unwitnessed, should increase suspicion for child abuse.

Resources

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15

Aging With Pediatric Onset Disability and Diseases

Margaret A. Turk, Lynne Romeiser Logan, and David Kanter

Aging is a fact of life, and although many may not be well prepared for typical aging changes, current marketing suggests this is not an unexpected event. However, aging with a disability can be an overwhelming and alarming situation, especially for those experiencing changes in function or health at an earlier-than-expected time. These changes can also mean the difference between living alone with minimal to no support and requiring a more restrictive living environment, including a move to an institutional setting, at a young age.

For years, children with disabilities and their families have been told that health and functional status, mobility, and musculoskeletal problems essentially stabilize by early adulthood. However, as more people with lifelong mobility and other impairments live through their adult years, it is apparent that mobility, functional status, and musculoskeletal changes commonly continue into adulthood. In fact, questions and concerns about mobility, function change, and pain are common among the majority of adults with mobility impairments caused by any etiology (1).

Despite the personal accounts and experiences of those with disabilities, their families, and many clinicians, there are no longitudinal studies on disabilities and few surveys or statistics that can document these aging changes and risk factors for them. Present statistics estimate that the number of Americans of all ages with disabilities (broadly defined by impairment, functional limitation, or participation restriction) exceeds 40 million, and may be closer to 50 million (2). However, these are estimates using multiple national surveys, cross-analyzed in an attempt to cover all ages and living situations. Many of these surveys exclude those living in institutions or assisted living programs (where a number of adults with congenital or childhood-onset disabilities may live), and many exclude young children or adults younger than retirement age. There are no national surveillance programs that monitor the trajectory of aging with a disability by specific disability condition, by severity, or by age of onset. Data do identify that more infants, children, and young adults are surviving with conditions that were at one time fatal, and children and young adults are completing and surviving long-term risk treatments (eg, chemotherapy, radiation, surgery). Approximately 500,000 children and youth with special health care needs turn 18 years annually (3). Thus, there is an increasing population of adults with disabilities, with accompanying risks for long-term complications and disabilities. As well, there have been declines in a few health conditions in childhood that contribute to adults with disabilities statistics. The incidence of spina bifida dropped from 24.9 to 18.9 per 1,000 live

births with the use of folic acid supplements in women of childbearing age (4). Lead exposure, a risk factor for neurodevelopmental problems, has dropped significantly, with reported lead levels now below 2% (5). These and other advances in medical care and public health practices will change the face of the type of disabilities seen in adults with early-onset disabilities in the future.

Table 15.1 identifies the leading chronic health conditions as causes of activity limitations, reported through the National Health Interview Survey 2002–2003. As is noted, listed chronic conditions do not list disability types typically identified in medical rehabilitation systems as identified by diagnosis, ICD-9 codes, or diagnostic-related groups, but rather by more generalized conditions. As noted, we have little information that is disability-specific or that can offer details about a specific disability over a life course. In comparing the listed function and medical conditions for those younger than 17 years, there is no commonality of conditions at age 18 years and older other than mental illness or emotional problems. The pediatric chronic conditions are largely cognitive and mental health–based and, for adults, are related to typical health conditions such as cardiovascular or pulmonary conditions. The only estimate of adults with early-onset disabilities is by Verbrugge and Yang (6) using data from the 1994 National Health Interview Survey Disability Supplement, Phase 1, suggesting that 7% to 9% of adults reporting a disability had onset before the age of 20 years. The surveillance data available imply that the prevalence of disability diagnoses typical of rehabilitation program settings is in a minority, and usually not the primary focus of public health, surveillance, and policy programs.

All health and function information regarding aging in congenital and childhood-onset disabilities that is known is derived from existing databases developed for service or financial reasons, case studies and series, limited survey information, cross-sectional studies, opinion pieces, and the like. Much of the conventional wisdom in this area has been communicated through the network of persons with disabilities and, more recently, through books and texts. There is minimal information regarding the impact of commonly practiced interventions over a lifetime, including environmental approaches to barriers. Health care providers receive minimal education regarding disability and/or aging with a disability during undergraduate and graduate education. Therefore, health care providers and consumers have limited knowledge from which to base decisions regarding adult health issues and anticipated changes in function.

This chapter will provide a conceptual framework regarding aging as it relates to congenital and childhood-onset disabilities, review general issues of health

15.1 Leading Chronic Health Conditions Reported Through the National Health Information Survey as Causes for Activity Limitation (2002–2003)

CHRONIC CONDITION	NUMBER OF PEOPLE WITH ACTIVITY LIMITATIONS		
	Under 5 years	**5–11 years**	**12–17 years**
Speech problem	10.7	18.5	4.6
Asthma or breathing problem	8.2	8.4	8.3
Mental retardation or other developmental problem	7.0	10.2	9.6
Other mental, emotional, or behavioral problem	2.7	12.0	14.2
Attention deficit or hyperactivity disorder	2.1	17.6	21.8
Learning disability	2.9	23.3	33.9
	18–44 years	**45–54 years**	**55–64 years**
Mental illness	12.9	23.1	24.1
Fractures or joint injury	7.0	15.5	20.6
Lung	5.0	12.6	25.6
Diabetes	2.5	13.4	33.4
Heart or other circulatory	5.9	28.4	74.3
Arthritis or other musculoskeletal	22.2	61.9	100.7

Adapted from Ref. 10.

and function across early-onset disabilities, discuss lifelong functional status and health issues of adults with specific early-onset disabilities, and consider the issues surrounding health care access and transitioning from pediatric to adult care services.

LIFESPAN PERSPECTIVE

Improved medical care, increased life expectancy, and better services for lifelong care and support in society have provoked an interest and need for long-term future planning. This includes transitions in care from typical nurturing pediatric care systems to more traditional adult self-directed services. Retrospective reviews and anecdotal experiences also question some long-held beliefs of "use it or lose it" to one of "conserve it to preserve it (7)." Choice of health care providers for adults with early-onset disabilities and special health care needs is often limited by insurance and expertise.

Clinicians with an understanding of the natural history of disabling conditions can be helpful in monitoring and keeping vigilance for and prevention of some general health conditions and aging or secondary conditions seen in disability. This public health model of prevention also includes tertiary prevention with the use of environmental modifications and technologies and removal of barriers to participation. There are general aging, associated conditions, secondary conditions, and health concepts that are helpful in understanding a lifespan perspective.

Aging is a developmental process. It begins at birth and continues to death. Typically, however, children and adolescents are said to develop, whereas adults, especially adults over 50 or 60, are said to age. During the early stages of aging (infancy, childhood, adolescence), attainment of skills and capabilities is on the rise; in the middle stages (adulthood), maintaining and retaining function is the focus. Over a normal lifespan, natural physiological declines are not truly preventable, although they may be accelerated or slowed by a variety of individual genetic factors, personal behaviors (eg, diet and exercise), health care practices, and environmental conditions. Aging changes in motor performance seem to be accelerated in some adults with early-onset disabilities, with earlier-than-typical manifestation of slowed or decreased motor performance and pain complaints. Persons with disabilities follow a course of aging, although likely with a slower and lower attainment of skills and a smaller capacity to adjust to acute or intercurrent health or medical and surgical intercedents (Fig. 15.1). So the emphasis here is on *aging with a disability*, not aging into disability.

There is also a need to appreciate the different time dimensions at play, as noted by Campbell (8) and

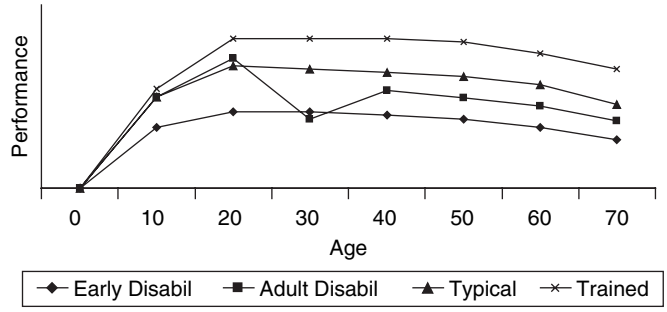

Figure 15.1 Conceptual model of aging and performance. Performance is a conceptual quotient of multiple skills. The trained person will achieve a higher level of performance than typical and, assuming ongoing exercise, will have a slower decline with age. With the onset of disability in adult years, there is loss of skill then improvement, but often not achieving the previous typical level. Those with early-onset disabilities do not achieve full "performance" and are slower to achieve the maximum level.

Krause and Adkins (7). These include the typical aging process, as noted previously, age of onset of disability in relation to developmental maturity (congenital onset versus adolescent onset), the number of years spent with a disability (hemiparesis onset at age 5 years versus age 17 years), cumulative effects of medications or treatments (long-term steroid use), and era of disability onset (cerebral palsy onset in 1950s versus 1990s, including different treatments, opportunities, and attitudes). Anticipated aging changes and treatment strategies will be modified by these temporal concepts.

Secondary conditions are defined as "any additional physical or mental health condition that occurs *as a result* of having a primary disabling condition (2,9)." The initial concept (10) and intended use (11) distinguishes secondary health conditions from the social and economic consequences that may follow a primary disabling condition (societal limitations and barriers—for example, poverty with disability, social isolation, limited transportation). There are key common features of secondary conditions (9):

- Causal relationship to the primary disability—the primary disability is a risk factor for the secondary condition
- Preventable or modifiable conditions
- Variability in expression and timing of manifestation
- Capability to increase the severity of the primary condition
- Potential to become the primary health concern

Many secondary conditions are linked across several primary disabling conditions through common physiologic processes or functional characteristics. As an example, disabilities with sensation changes and

immobility are risk factors for pressure ulcers, such as spinal cord injury, spina bifida, multiple sclerosis, and severe brain injury. Three common secondary conditions noted through cross-disability studies are fatigue, chronic pain, and depression (1,12–14).

Secondary conditions are distinct from associated conditions or residual deficits and comorbidities. Associated conditions describe elements that result from the defect, injury, disease, or pathology, although the expression may be variable. These conditions are the residual from the original pathology, and are often present at the time of diagnosis of the primary disability, although by development or evolution may not be expressed or expressed fully at initial diagnosis. For cerebral palsy or other brain injuries, the list of associated conditions includes seizures, spasticity, learning disabilities, intellectual disability, sensory problems, and oral motor and communication problems. These conditions may not be present for all people with the specific disability, are fairly well known to require monitoring by clinicians, and their presence is confirmed through typical timely evaluation. Persons with a primary disabling condition may have any combination of associated conditions, all of which will affect their ultimate functional capabilities. Comorbidities are other medical conditions unrelated to the primary disabling condition, and not a feature of the primary disability. As an example, persons with cerebral palsy may also develop diabetes mellitus or colon cancer should they have the risk factors or genetic predisposition for these conditions. As research continues, especially through longitudinal studies, links may be identified between primary disabilities and specific health conditions.

Health is a concept only recently considered to be an important goal for people with disabilities, and is the absence of disease or illness, beyond the disabling condition. Health perception is an individual determination, and is affected by personal expectations, experiences, sense of vulnerability, support, and locale. How people with disabilities self-rate their health has been in question (15). This self-concept may also direct consideration of engagement in typical health and wellness activities. Often, the health of persons with disabilities is perceived as poor by clinicians and providers when individuals report a positive perception of their own health. This health provider concept may limit the offer of screening or health promotion opportunities. Perception of health in adults with disabilities may be related to time of onset, with report that adults with early-onset disabilities may identify better health than those with adult-onset disabilities (16,17). Research further suggests that adults with disabilities likely have a different construct of and self-rating process for health (18). In general, persons with nonprogressive disabilities should be considered healthy, with a shift of the health care model from an illness and disability paradigm to one of wellness and prevention or early identification of secondary conditions, aging issues, and/or comorbidities.

GENERAL KNOWLEDGE REGARDING HEALTH AND PERFORMANCE

A body of literature has accumulated regarding health, aging, and secondary conditions for adults with disabilities and for some specific disabilities of early onset. Most research has focused on disabilities and impairments that have higher prevalence rates (eg, cerebral palsy); are easy to associate with a disability group (eg, spina bifida, Down syndrome); benefit from organized, dedicated service programs (eg, muscle diseases); and therefore have attracted research funding to generate a significant body of knowledge about the condition. The literature includes a combination of scientifically observed and anecdotal information as the database, often involving a "convenience" sample and a cross-sectional approach, with conclusions drawn from patient reports, clinical observations, and ICD-9 codes; none of these are standardized measures of individual characteristics or outcomes. Most studies identify health issues or concerns, with few challenging prevention or intervention strategies. Each factor in the interaction of disability and aging or secondary conditions has the capability to become a "negative feedback loop" (19) that may lead to further disability or a new health condition. There are studies using cross-disability groups that may have a higher representation of certain disability groups or may be small sample sizes, and consequently generalization to other disability groups should be considered with caution. In like manner, prevalence rates for some aging, secondary, and health conditions in disability-specific studies cannot be applied to all disability groups.

Pain is a common health condition for adults with disabilities, as noted earlier, and may be seen earlier in early-onset disability groups, especially those with mobility impairments. Pain is also a common complaint in adults without disabilities, and there is an expected response from health care providers, including evaluation and treatment. This should also be the expectation for those with disabilities, especially at younger ages. Any significant decrease or loss of motor skill, change in continence, change in typical activities, direct pain complaint, or "sluggishness" (20) requires further evaluation. Common musculoskeletal etiologies include poor ergonomics and biomechanics in tasks (secondary to deformity or limited motor control), underlying weakness and therefore overuse, hypertonia depending on the primary disability, and degenerative joint disease. Neurologic etiologies may

also need to be considered, including general neuropathies, focal neuropathies (eg, carpal tunnel syndrome, ulnar entrapments), radiculopathies, and myelopathy or stenosis. Appropriate evaluation should be completed to determine the treatment strategy. Typical treatment strategies should be implemented and modified as needed, given the disability and improvement noted. Management may include traditional noninvasive interventions (eg, analgesics, nonsteroidal anti-inflammatory drugs [NSAIDs], therapy modalities), more aggressive pain management strategies (eg, manual medicine, trigger point injection, massage, spinal injections), and reevaluation of functional activities or positioning that may predispose to the pain complaints. For spasticity-related problems, use of tone management techniques can be helpful, including oral antispasticity medications, use of botulinum toxin injections for focal problems, or intrathecal baclofen. Surgical interventions should also be considered, and will require preplanning for rehabilitation, living arrangements, and supports postprocedure.

There are anticipated health and performance changes with aging. The risk for additional health problems should be monitored and addressed as with the general population. However, people with disabilities are often not afforded typical screening as in the general population. Iezzoni et al reported those with mobility impairments did receive pneumonia and flu vaccines, but were less likely to receive other preventive services. Women with severe mobility impairments in particular were less likely to receive Pap smear and mammography screening (21). Women with disabilities had less knowledge about cardiovascular risks and no screening for risk factors, despite their higher risk with low activity levels (22). However, Cooper described a minor modification in office-screening techniques for adults with intellectual disability that improved identification of risk factors and health needs, with improved health determinants (23). Additional health risks and conditions can affect general performance.

Performance changes with aging include decrease in strength, balance, flexibility, coordination, and cardiopulmonary function, to name a few. The impact of these known aging changes on a person with mobility impairment is not well understood. Use of equipment, modifications to environment or activities, and joint protection all contribute to maintaining function over time. It is, however, known that persons with mobility impairments use more energy to perform mobility activities than their nondisabled peers. Therefore, exercise and activity to improve performance and maintain those improvements would seem intuitively obvious. In fact, there is scientific evidence that exercise and activities are effective for people with mobility impairments and that these activities can be managed through home programs and health clubs, not just traditional physical therapy programs (24,25). Simply continuing typical activities, even though considered "strenuous," will not increase strength, conditioning, or performance. Exercise and activities should be a part of a health maintenance program for adults with mobility impairments.

DISABILITY-SPECIFIC HEALTH

There is increasing information about specific early-onset disability conditions and adults' health and expectations for functional performance with aging. This chapter will highlight those conditions commonly managed by pediatric physiatrists, or those for which we have useful information. There is actually considerable information available for clinicians; however, as has been noted, the quality of the study or report is often at the case series level, usually involving a convenience cohort with single-point assessments or follow-up interview contact. Table 15.2 identifies the more common health conditions and management strategies for the disabilities described in this chapter. Nonetheless, it does begin to provide a picture of the health of adults with childhood-onset conditions and the need for modifications to our health monitoring and interventions for some conditions.

Cerebral Palsy

Cerebral palsy (CP) is the most common condition that pediatric physiatrists will manage, although it is not the most common reason for childhood disability, as noted earlier. There are estimates of about 500,000 people in the United States with CP. Over the past 10 years, there has been increasing information available about the life course in CP, and adult issues and health are better defined.

The health of adults with CP is generally good. Although cerebral palsy may affect multiple organ systems, in general, the long-term health problems are related to pain, fatigue, and the musculoskeletal system (see Table 15.2).

Mortality

Mortality for people with CP appears to be related to severity of impairments. This is very clear in the pediatric population, but less so for adults who have survived into their late twenties and thirties. There is also an obvious cohort bias when comparing mortality data from those born prior to the 1980s to mortality data of a younger adult population, and it is not clear that the information about the adults of today may be used specifically for predicting life expectancies.

DISABILITY	COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Cerebral Palsy	Pain	Routine exercise	Exercise prescription
	Fatigue	Monitor and query routinely Work simplification Ergonomic evaluations Energy conservation	Query/evaluate sleep; manage as needed Evaluate for pain etiology and treat Modify equipment or workplace Evaluate mental health and manage Progress to pain management program
	Musculoskeletal Contractures Hip pathology Knee pathology Foot or ankle pain Back pain	Monitor and query routinely Joint protection strategies Routine exercise Biomechanic and ergonomic assessments	Focal musculoskeletal evaluation Tone management Modify equipment. workplace, biomechanics of function Physical therapy prescription Adjust orthoses
	Osteoporosis/fractures	Routine exercise Calcium/vitamin D supplement Fracture and fall prevention; education	DEXA evaluation Consider treatment when multiple fractures Exercise when appropriate
	Neurologic Spasticity Seizures Spinal stenosis Nerve entrapments	Routine monitoring Adjust medications with reported change Query for changes; high index of suspicion for pathology	Tone management; medications, BTX injections, ITB Seizure management Radiologic evaluation Electrodiagnosis Surgical referral when appropriate
	Urinary conditions Incontinence UTIs	Monitor and query routinely	Urodynamic evaluation Scans/radiographs Medications and CIC when needed Urology referral as appropriate
	Respiratory conditions Infection Sleep apnea	Routine monitoring Immunization Query sleep hygiene	Scoliosis evaluation Sleep study and management Specialty referral as needed
	Gastrointestinal Constipation GERD Obstruction	Monitor and query routinely; recognition of severity Nutritional management	Adjustment to bowel program regimen Specialty referral when appropriate
	Deconditioning Falls	Routine exercise Education and prevention	Therapy prescription; focus on strength and aerobics Reconsideration of equipment

	Mental health	Routine monitoring Query of support, living arrangements	Specialty referral as appropriate Referral for psychological and social support Use of community resources
	Sexual functioning	Provide with education; appropriate modality for level of function Assist with environmental modification for routine assessments as able Assure pregnancy high risk needs are met	Following pregnancy, support may be needed in the home
	Health maintenance	Monitoring (see Table15. 4)	
Spina Bifida	Urologic/renal disease	Routine monitoring; UTI frequency, renal scans, urodynamics	Appropriate management, consideration of alternatives for treatment
	UTIs Incontinence Vesico-ureteral reflux End-stage renal disease Bladder cancer	Maintain routine urology appointments	With change consider neurologic evaluation as cause
	Musculoskeletal	Routine exercise, especially strengthening posterior shoulder	Focal musculoskeletal evaluation
	Shoulder pain/overuse Scoliosis	Monitor and query routinely Joint protection strategies	Evaluate for neurologic change with new symptoms Modify equipment (possible power wheelchair), workplace, biomechanics of function
	Joint pain Osteoporosis/Fracture	Routine exercise Biomechanic and ergonomic assessments Calcium/vitamin D supplement Education and fall prevention	Therapy prescription
	Neurologic Hydrocephalus Chiari malformation	Routine monitoring Query for changes Maintain neurosurgery appointments	Neurosurgical evaluation Post-surgery, may require rehabilitation admission Cognitive and functional assessments post-intercurrent events to assure safe community living
	Tethered cord Epilepsy	Routine neurology appointments with active epilepsy	
	Obesity	Monitor weight	Nutrition referral
		Routine exercise	Exercise prescription
		Nutrition management	
	Pressure ulcers	Frequent position change Monitor skin, nutrition, equipment, change in function	Modify positioning or pressure relief equipment Assure good nutrition Appropriate care for ulcer staging May need change to tone management Surgical referral

Continued

15.2 Aging Health and Performance Changes (Continued)

DISABILITY	COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
	Pulmonary restriction	Monitor for infection	Evaluate for neurologic change with new symptoms
	Pulmonary infection	Immunizations	Consider sleep study or O_2 supplement
	Bowel incontinence	Monitor and adjust program for change	Evaluate for neurologic change with new symptoms
	Lymphedema	Monitor, use of compression and elevation at first sign	Referral for lymphedema program and prescribed compression garments
	Latex allergy	Limit exposure to latex	Modify equipment if needed Acute event treatment Appropriate recognition in medical record, personal acknowledgement
	Mental health	Monitor routinely	Specialty referral as appropriate Referral for psychological and social support Use of community resources
	Sexual functioning	Provide with education; appropriate modality for level of function Assist with environmental modification for routine assessments as able Urology referral for fertility/performance Assure pregnancy high risk needs are met	Following pregnancy, support may be needed in the home
	Health Maintenance	Monitoring (see Table 15.4)	
Spinal cord injury	Urologic/renal disease	Routine monitoring; UTI frequency, renal scans, urodynamics	Appropriate management, consideration of alternatives for treatment
	UTIs Renal calculi Incontinence Reflux	Maintain routine urology appointments	With change consider neurologic evaluation as cause
	Musculoskeletal	Routine exercise, especially strengthening posterior shoulder	Focal musculoskeletal evaluation
	Shoulder pain/overuse	Monitor and query routinely	Evaluate for neurologic change with new symptoms
	Scoliosis	Joint protection strategies	Modify equipment (possible power wheelchair), workplace, biomechanics of function
	Other pain complaints	Routine exercise	Therapy prescription
	Osteoporosis/fracture	Biomechanic and ergonomic assessments Calcium/vitamin D supplement Education and fall prevention	Adjust orthoses, footwear

	Neurologic	Routine monitoring	
	Spasticity	Query for changes	Tone management; progress to more aggressive strategies
	Autonomic dysreflexia	Adjust medications with reported change	Evaluate for neurologic change, painful symptoms, bowel/bladder etiologies, fractures, pressure ulcers with more frequent AD symptoms
	Pulmonary conditions	Monitor for infection	Evaluate for neurologic change with new symptoms
	Ventilator dependency	Immunizations Consider diaphragm or phrenic nerve pacing	Consider sleep study or O2 supplement
	Pressure ulcers	Frequent position change Monitor skin, equipment, change in function	Modify positioning or pressure relief equipment Appropriate care for ulcer staging May need change to tone management Assure good nutrition Surgical referral as appropriate
	Bowel incontinence	Monitor and adjust program for change	Evaluate for neurologic change with new symptoms
	Latex allergy	Limit exposure to latex	Acute event treatment Appropriate recognition in medical record, personal acknowledgement
	Mental health	Monitor routinely	Specialty referral as appropriate Referral for psychological and social support Use of community resources
	Sexual functioning	Provide with education; appropriate modality for level of function Assist with environmental modification for routine assessments as able Urology referral for fertility/performance Assure pregnancy high risk needs are met	Following pregnancy, support may be needed in the home
	Health Maintenance	Monitoring (see Table 15.4)	
Limb deficiency	Overweight or obesity	Monitor weight and nutrition Routine exercise	Exercise or therapy prescription Referral to nutritionist if indicated Modify prosthesis as needed
	Pain	Routine exercise Monitor and query routinely Work simplification Ergonomic evaluations Energy conservation	Focal examination and evaluate/treat Exercise prescription Modify equipment or workplace Progress to pain management program Adjust prosthesis as needed
	Deconditioning	Education and falls prevention	Therapy prescription; focus on strength and aerobics
	Falls	Routine exercise	Adjust prosthesis as needed Consider other equipment

Continued

15.2 Aging Health and Performance Changes (Continued)

DISABILITY	COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
	CVD/PVD	Reduce risks Routine exercise Monitor as indicated	Referral and management as indicated
	Health Maintenance	Monitoring (see Table 15.4)	
Intellectual disability	CVD	Routine monitoring	Exercise prescription
	Obesity	Routine exercise Nutrition management	Referral for nutritional consultation
	Respiratory disorders	Routine monitoring Immunizations	Consideration of sleep apnea, need for O_2 supplement
	Epilepsy	Routine Neurology appointments Query and monitor	Assist with change in community living arrangement as needed
	Osteoporosis/fractures	Routine exercise Calcium/vitamin D supplement Fracture and fall prevention; education	DEXA evaluation Consider treatment when multiple fractures Exercise when appropriate
	Poor oral health	Monitoring Assist with environmental accessibility if able	
	Mental health	Routine monitoring Reduce life events Query of support, living arrangements	Specialty referral as appropriate Referral for needed supports Use of community resources
	Sexual functioning	Provide with education; appropriate modality for level of function Assist with environmental modification for routine assessments as able Assure pregnancy high risk needs are met	Following pregnancy, support may be needed in the home
	Health maintenance	Monitoring (see Table 15.4)	

Down syndrome	Mental health	Monitor	Behavior management
	Alzheimer dementia	Reduce life events such as moves	Medications as needed
	Depression		Specialty referral as appropriate
			Referral for needed supports
			Use of community resources
	Endocrine	Routine monitoring	Medication management
	Hypo- or hyperthyroid	Annual TSH monitoring	Diet management
	Diabetes		
	CVD	Reduce vascular risk	Evaluation, treatment per study
	Mitral valve prolapse	Routine exercise	
		Monitor	
	Celiac disease	Monitor	Management per gastroenterologist
	Hearing loss	Monitor	Consider amplification if appropriate
	Sleep apnea	Query and monitor	Sleep study and management
	Musculoskeletal	Calcium and vitamin D	Evaluate pain complaints appropriately
	Arthritis	Routine exercise	Medications as appropriate
	Osteoporosis	Monitor for urologic change, dysphagia, spasticity, weakness, bowel changes, pain	Therapy prescription
	Atlantoaxial instability		Consider treatment if multiple fractures
			Full evaluation of performance changes; radiographs and referral as appropriate
			Referral to neurosurgery with acute loss
	Obesity	Routine exercise	Referral for exercise program
		Nutrition management	Referral for diet management
	Respiratory infections	Monitor	Medications, possible O_2 supplement
		Immunizations	
	Sexual functioning	Provide with education; appropriate modality for level of function	
		Assist with environmental modification for routine assessments as able	
	Health maintenance	Monitoring (see Table 15.4)	

DEXA, dual energy X-ray absorptiometry; BTX, botulinum toxin; ITB, intrathecal baclofen; UTI, urinary tract infection; CIC, clean intermittent catheterization; GERD, gastroesophageal reflux disease; AD, autonomic dysreflexia; CVD/AVD, cardiovascular disease/atherosclerotic vascular disease; TSH, thyroid-stimulating hormone.

Through a large database in California defined by financial and service support needed and especially representative of the more severely impaired individuals with CP, survival of higher-functioning adults was close to that of the general population (26). Strauss et al also reported with this same database that older subjects who had lost the ability to walk by age 60 years had poorer survival and that those who had the most severe disabilities rarely survived to age 60 years (27). A later report by Strauss et al noted improved survival for adults with gastrostomy tubes in particular over a 20-year period (28), indicating improvements in treatment and care of the most fragile individuals with high levels of impairment. Additional information from this database, weighted towards a more severely impaired cohort, reports standardized mortality ratios, noting a higher mortality in general at 8.4, and as high as 13.8 in the most severe group (29). There was a decrease in this discrepancy with age, which may indicate a healthy survivor effect and increasing mortality in the general population. Respiratory etiologies as cause-of-death standard mortality ratio was 15, which is lower than is generally thought, and the highest overall for all ages was intestinal obstruction.

Reports from abroad also identify life expectancies for adults with CP to be close to the general population for those with mild to moderate impairments. The Western Australia Cerebral Palsy Registry noted the strongest single predictor of mortality was intellectual disability, with survival exceeding 92% for IQ/DQ scores >34 (30). This study noted motor impairment severity increased the risk of early mortality, with mortality declining after age 5 to 15 years, and remaining steady at 0.35% for the next 20 years. Providing insights on era of disability onset, Hemming et al reported on adults with CP in the 1940–1950 birth cohort in the UK. Assuming survival to age 20 years, almost 85% survived to age 50 years compared to 96% of the general population (31). Again comparing to the general population, many of the deaths noted in ages 20s–30s were respiratory, and deaths in ages 40s–50s were circulatory conditions and neoplasms. Few deaths in adulthood were attributed to CP, although the nervous system was implicated more than in the general population. The notion of increased neoplasm as cause for death rates is echoed by the large California database noting a three-times-higher rate for breast cancer in CP than in the general population, and this may be related to severity as well as poor screening (29). Survival rates for children of today may not necessarily be extrapolated from any of these studies.

Health and Functional Status

The general health of adults with CP is self-reported as good or satisfactory to excellent (32,33), and this can be comparable to that of the community at large (16). In a population-based study of adults with cerebral palsy in a mid-sized metropolitan area, persons with cerebral palsy were generally healthy (based on clinical information and self-report), but noted worries and concerns about their health status and futures (34). Self-perceived health ratings and life satisfaction may be related to the presence of pain or functional changes over time, but not to the severity of impairment (35–37). Despite reports of good health, a Canadian publication notes adults with CP attended outpatient physician visits 1.9 times higher than age-matched peers (38).

The functional status of adults with CP is not static over time, and with aging there can be modest decreasing function, as there is for the general population. A number of studies, both in the United States and abroad, with small to large convenient samples, have noted that about a third of subjects report modest to significant decreases in walking or self-care tasks (16,27,39–41). Changes in dressing and walking with relative sparing of other self-care or social activities were reported in two of these studies (16,27). Day et al used the large California database to determine the probabilities of loss or gain of walking skills into adulthood for those with CP (42). They noted that by age 25 years, there would unlikely be any improvement in walking skill and most would not change over the next 15 years, although there could be some decline. Therefore, the reason for even modest decreasing skill is not clear and may be related to progressive neurologic problems (eg, cervical spine stenosis, radiculopathy), lack of environmental modifications, pain, no access to or participation in exercise or activity programs, aging, or other medical conditions.

Decreased independence (increased need for assistance) in mobility and self-care is a common complaint of adults with mobility impairments. The reasons for change are varied, and may include those related to age changes (eg, decreased endurance, flexibility, strength, or balance), progressive pathology or secondary conditions (eg, pain, contractures, spasticity, osteoporosis and fractures, stenosis), or personal choices (eg, use of powered mobility to conserve energy). The change in mobility is often a response to a secondary condition or age-related change. Falls may also be such a response. Significant change in mobility or falls should not automatically be accepted as a part of a congenital or childhood-onset disabling condition in adult years; treatable etiologies should be sought.

It has been suggested through cross-sectional and convenience samples that adults with congenital or childhood-onset disabilities may show musculoskeletal or performance changes typical of advanced aging earlier than their nondisabled peers (32,26,43). These observations require confirmation through

longitudinal controlled studies. While risk factors may predispose a person to these changes, they are, as yet, unproven. If these earlier-than-expected aging changes are confirmed, they should be considered secondary conditions.

Pain and Fatigue

Pain is the most consistent health condition reported by adults with cerebral palsy (17,32,44,45). It has been reported in a number of samples of adults with CP at a variety of ages to be 30% to 80%, with activity limitation from this at >50%. For this reason, it will be covered as a separate topic. Pain may be present for a variety of reasons; it may be acute, recurrent, or chronic. Increased spasticity, weakness, falls, or progression of contractures or deformities can result from pain, particularly when pain is not reported because of communication difficulties or severe intellectual disability. Because of the high prevalence, the health care provider should try to elicit complaints or indications of pain, and evaluation, diagnosis, and intervention should ensue. Pain is often the reason for a change in function, living arrangement, or social interaction.

Pain is usually identified by proximity to a joint, and less often a limb. Most people report "arthritis" as the etiology of these pain complaints; however, these pains may originate from either joints or muscles. A good history and clinical exam will help sort out the issues and direct appropriate treatment. Back, leg, and hip pain complaints are common in persons with cerebral palsy (46,47). There are usually more pain complaints in those with spasticity (46). It has been reported that fatigue often incites pain, and exercise most commonly relieves pain (46,48).

Fatigue is a common complaint of adults with CP, and is associated with pain (49). It is also associated with deterioration of skills and low life satisfaction, with no association with any specific type or severity of CP. As noted, it may incite pain. The fatigue may also be associated with the reported coping strategies sometimes used for chronic pain by adults with CP (50). Sleep disruption should also be questioned since it is commonly seen with pain and fatigue. Anecdotally, the pain/fatigue complex appears to respond positively to directed pain management, good sleep hygiene, medications, and exercise.

Appropriate management includes early identification of the problem and its source. Common musculoskeletal etiologies include poor ergonomics and biomechanics in tasks (secondary to deformity or limited motor control (41)), underlying weakness and therefore overuse (51), hypertonia (52), and degenerative joint disease (53). Typical management strategies should be offered, and referral for additional interventional, orthopedic, or neurosurgical consultation should be considered. However, adults with CP tend to self-manage their pain complaints (54), and for those who seek medical care, report is minimal improvement and few options offered (55).

Musculoskeletal and Neurologic Conditions

Contractures. Contractures are a common secondary condition, and reported in multiple case series. Their impact on functional status or general health care needs is variable. Increasing contractures, particularly when associated with pain or increased spasticity, may be an indication of progressing pathology. Aging changes include decreased flexibility, and the clinician must distinguish pathological causes of increasing contracture through appropriate diagnosis.

Osteoarthritis. Because of the significant pain complaints that adults with CP offer, it is often stated that there is an early onset of osteoarthritis. Conceptually, this has been explained by unusual and possibly increased forces on joints that may have malalignment and/or deformity, and associated with underlying weakness and poor motor control (32). In fact, health care providers often will make a presumed diagnosis of "arthritis" for pain complaints in adults with disabilities. Clinically, it is not surprising to find significant arthritic changes with radiographs of painful joints, and sometimes at young adult ages. However, the presence of early-onset arthritic changes has been documented by case reports, and studies that report arthritis among subjects base this information on self-report of arthritis or presence of pain. Often, the pain complaint is not evaluated fully, and may have an etiology in soft tissue injuries or problems and not degenerative changes within the joint. There may, in fact, be premature osteoarthritis, but it has not been documented definitively. Of importance is the recognition of pain, appropriate evaluation, and treatment.

Hip Pathology. Degenerative changes have been noted radiographically in dislocated and subluxed hips, not always related to weight bearing activities, in persons with cerebral palsy (44,56). Use of tone reduction strategies may be helpful. Femoral head resection as a treatment strategy for control of pain in hip disease for persons with cerebral palsy has been suggested; however, pain often persists or recurs postoperatively (57–60). Total hip and knee replacements as a treatment option for pain from severe arthritis in adults with cerebral palsy are becoming more common; however, as their lifelong efficacy remains unknown (61–64), revision may be anticipated with placement at younger ages.

Knee Pathology. Knee contractures are common in those who do not walk and in those who walk with obvious

knee flexion and crouch. Not all knee contractures are painful. Tone management may improve range, function, and pain. Patella alta may develop over time, and pain or chondromalacia may result. Joint laxity may also be present. Modalities, exercise, kinesiotaping, and other interventions may be helpful. There are advocates for patellar tendon advancement surgeries, with or without distal femoral extension osteotomies, in adolescents and young adults to improve pain and restore knee function in gait, confirmed on gait analysis (65).

Foot or Ankle Pain. Again from biomechanical factors, contractures and pain may develop. Typical interventions may assist including orthoses, but not all bracing or shoe inserts are helpful, and biomechanics must be taken into account. Plantar fasciitis with appropriate treatment should be considered.

Spine Pathology. In people with cerebral palsy, severe motor impairment is associated with scoliosis and other deformities (66). Scoliosis may progress during adulthood, and those at 50 degrees or greater at skeletal maturity may deteriorate more rapidly (67). Scoliosis can cause seating and pressure problems, impaired respiratory function, and pain (52,67,68), and may be associated with windswept hips and pressure sores (52). It has been reported that spinal fusion improves the quality of life for those with CP (69).

Spinal stenosis must be ruled out whenever significant functional change is noted, particularly for change in or loss of walking skills, increased leg spasticity, change in bladder habits, neck pain, vague sensory changes, and (late) change in arm and hand function (70–72). A tethering effect on the spinal cord also may occur, resulting in cranial nerve changes. Some early reports noted a higher risk in those with an athetoid or dyskinetic component (73,74); however, more recent reports show these problems are present in spastic forms of cerebral palsy as well. While it is generally held that stenosis is due to early spondylosis and compression, there may also be a predisposition to it in those with a congenitally narrow canal, especially at C4–C5 (70,73). Diagnosis is made through imaging studies, while comparative evoked potentials may also be helpful in determining neurologic function. Surgical decompression may prevent further, often catastrophic, loss of function, but does not assure return of lost function, particularly in cases of longstanding compression with spinal cord atrophy. Recurrence at levels above or below surgical correction may be noted (75,76). Postoperative management planning should accommodate changes in functional capabilities and care needs. The presence of an athetoid movement component will affect postoperative spine stabilization and possibly head positioning and neck mobility. When no surgical intervention is undertaken, a frank discussion of possible respiratory compromise and the future need for ventilator assistance should be provided.

Peripheral Neurologic Compression. Radiculopathies may be a cause for painful complaints, and appropriate evaluation and treatment should ensue. It is most important that treatment strategies are based on the person's history of function, that there is effective input from that person or their care provider, and that practical outcome goals are identified. Although not as common as a musculoskeletal etiology, nerve entrapment is also a cause of pain. The most common nerves and areas of entrapment as reported by adults with CP are the same as those susceptible to compression in the nondisabled population: the median nerve at the carpal tunnel and the ulnar nerve in the hand distally and at the elbow. Compression points are often related to use of crutches, transfer techniques, propelling wheelchairs, or existing deformity. Work-related or positional activities may also cause entrapments, just as in the nondisabled population. There is no reported increased incidence in CP. All hand pain or sensation change does not represent nerve entrapment. Often, these complaints are actually problems of repetitive motion or are position-related. While they may be ascribed to carpal tunnel syndrome, they often respond poorly to surgery (77). Appropriate testing (including electrodiagnostic testing) is necessary to determine their etiology. Where treatment options are similar for disabled and nondisabled adults, some modification of management will be required if functional independence is changed by or during treatment.

Osteoporosis. Osteoporosis has been documented in at least 50% of children and adults with cerebral palsy (78,79). The aging process may exacerbate this issue, as does anticonvulsant use and mobility impairment. Pathologic fractures occur typically in the long bones, but frequency data vary and no large studies of people with cerebral palsy have been reported. Low serum 25-OH vitamin D concentrations are not identified as a cause in most cases described in the literature (79). Typical screening devices, such as the Simple Calculated Osteoporosis Risk Estimation (SCORE), do not accurately identify osteoporosis risk in women with disabilities (80); therefore, bone mineral density testing and counseling on fall risk is important for both women and men with disabilities. Dual energy x-ray absorption (DEXA) scans must be read with caution, since contractures often skew results. Recommendation is to use the scan results of the distal femur, as is used in children with CP and contractures (81). Use of bisphosphonates is described, but the functional improvement derived from these drugs over the long term is unknown.

Additional Health Conditions

There are no comorbidities known to be associated with CP. As noted, general health is good. A recent study of adults living in group homes from upstate New York notes increasing health conditions with age for adults with CP as would be expected: cardiovascular, respiratory, and hearing/vision (82); this has been replicated in Taiwan and Israel (83,84). Of interest is that in comparison to U.S. national norms, there are fewer cardiovascular risk factors than seen in the general population; either this is a healthier population or there has not been effective screening and monitoring. In looking more critically at this population, the severity of the CP was related to increasing health problems with aging more than the diagnosis of CP alone (85). Vision and hearing problems may have been present early, and as anticipated, there is an increase in vision and hearing problems with age (82).

Dental issues are reported for adults with CP (46). Medications, nutrition problems, poor dental hygiene, and difficulty with access to dental care all contribute to the ongoing problems into adulthood.

Previously known associated conditions will persist into adulthood. Dysphagia will continue, and monitoring is required. Constipation also persists, and adjustments to bowel programs may be needed. Gastroesophageal reflux is often reported, but has not been present at increased rates. Intestinal obstruction is reportedly common in CP, and in an upstate New York cohort living in group homes, adults with CP had an increased rate compared to other adults with developmental disabilities (85).

Urinary incontinence may also continue, and assurance must be made that there is no dyssynergia or overflow with retention. Rosasco et al reported adults with CP had a higher incidence of urinary tract infections (UTIs) that was related more to severity than the presence of CP (85), compared to other adults with developmental disabilities living in group homes in upstate New York. Neurogenic bladders in adults with cerebral palsy are only infrequently associated with upper tract pathology (86). Some women report that incontinence consistently occurs at a particular point of their menstrual cycle, often associated with increased spasticity (87). Urinary incontinence can be effectively addressed through well-established diagnostic and intervention approaches. There are no available data that assess the adverse impact of urinary incontinence on social integration in cerebral palsy, but anecdotal support for this association is abundant. In both men and women, urinary incontinence should be identified and addressed, regardless of age or other conditions.

Respiratory problems have been implicated as cause of death early in life and in early adulthood, as was noted earlier. Use of vaccinations may be helpful, along with vigilance and monitoring. Respiratory problems may increase with progressive scoliosis, and aspiration from gastroesophageal reflux disease (GERD) or dysphagia must be recognized. Sleep disorders related to pulmonary problems should be considered with progressive scoliosis, especially with complaints of poor sleep, morning headache, or daytime sleepiness.

There has been suggestion that obesity is a problem in CP, and yet there are no studies to support this. In fact, a small study of adults with CP identified mean body fat percentages and body mass indexes were within normal range, although 40% had heights below the fifth percentile for age and gender. Fifty-five percent reported dysphagia (88).

Sexual Functioning

Women's sexual health and functioning is better described than men's. Women with CP typically have limited participation in health maintenance activities such as routine pelvic examinations, Pap smears, and breast examinations (33,89). Office visit planning is required for those with significant motor impairments to assure a complete examination. Attitudinal barriers of health care providers often limit services and education. However, women with CP are typically able to conceive and carry pregnancies to term without the expectation of major complications related to their CP. Use of contraceptives has not been well studied, and consideration of thrombotic effects must be considered in choice of options. A commonly offered contraception is nonestrogenic formulations such as Depo-Provera, although long-term effects are not well defined (87,90). Women with CP report fewer sexual encounters as compared to other women with disabilities (17,91). Women with early-onset disabilities also experience high levels of sexual desire compared to other women with disabilities, postulated as being related to reduced social opportunities, frustrated satisfaction of sexual urges, discouragement of childhood sexual expression, or perceived social stereotypes (91).

Men with CP also should receive information on sexual functioning, including information on contraception and protection. There have been no reported problems with sexual functioning or fertility.

Spinal Cord Dysfunction

Spina bifida (SB) and spinal cord injuries (SCIs) are the most common etiologies for spinal cord dysfunction (SCD) in childhood, although infectious, rheumatologic, demyelinating, and tumor etiologies are also seen. The incidence and prevalence of SCD in general is low in a pediatric population. Earlier chapters have identified the decreasing incidence of both SCI and SB.

The prevalence for both, and for SCD in general, are only estimates, and are well below estimates for intellectual disabilities (ID) and CP. It is also estimated that life expectancy is increasing, and therefore, it is important to understand the lifelong health and functional issues of adults with childhood-onset SCD. SCD usually involves multiple organ systems at a high level; these medical conditions are fairly well described; and consequently, there may be more medical monitoring than in other conditions. There is significant overlap in the long-term management of those with SCI and SB, although there are disability-specific health issues and risks. This section will highlight what is known about the health of adults with childhood-onset SCI and SB independently. For both subsets, adults are presenting with health challenges, such as renal dysfunction, musculoskeletal problems, neurologic complications, pulmonary conditions, pressure ulcers, and sexuality and reproduction issues (see Table 15.2).

Spina Bifida

Mortality

As noted, in general, both early and late survival has improved over the past 20 years. There are few databases that maintain statistics for specific disability diagnosis groups, but there are databases that involve specific sites of care for programs serving people with SCD. Today, children born with an open SB have at least a 75% chance of living into early adulthood. There is a high correlation of childhood death with hindbrain dysfunction and posterior cervical decompression, requiring tracheostomies and gastrostomies (92). Common causes of death in adulthood are renal failure and causes related to the central nervous system (CNS), with continued hindbrain dysfunction and unrecognized shunt malfunction (92,93).

Functional Status and Mobility

There are no large studies to identify change in function over time. Most studies identify mobility based on defect level without regard for American Spinal Injury Association (ASIA) levels or declaration of complete or incomplete function. Lower lesions are associated with higher walking abilities, with or without aids (92,94). A small cohort of adults with sacral-level myelomeningocele was noted to have maintained their walking abilities for low-sacral lesions, and almost 90% maintained walking in the high-sacral group (95). Complications reported included scoliosis, osteomyelitis, amputations, and spinal tethering. A single small study in the Midwest identified that across the spectrum of SB, mobility decreased from early childhood to early teen years (92). At least three-fourths who walked during their teen years continued walking as adults.

The associated cognitive effects of SB influence the level of functional independence in adults. A small cohort report showed that most young adults with hydrocephalus and lesions at L2 or above were dependent for sphincter control, locomotion, and self-care, with an additional number requiring assist with transfers and social interaction and communication (96). Those without hydrocephalus or with hydrocephalus and lesions below L2 required assist with sphincter control only. An additional small study reported more difficulties in independence and quality of life, with increasing numbers of shunt revisions (97).

Perceived health for a group of young adults in the Netherlands was related to physical functioning, as would be expected using a tool standardized for the general population, not for disability (98). Of interest was that the domains associated with emotional health did not differ from the population group. Using the Life Satisfaction Questionnaire, again in the Netherlands, highest proportion of dissatisfaction was with financial situation, partnership relations, and sex life, and those with hydrocephalus were less satisfied with self-care ability and partnership relationships than those without hydrocephalus (99). Overall, the presence of SB does not appear to be an important determinant of life satisfaction.

Urology/Nephrology

Urinary and renal issues are common health problems for those with SCD. Renal damage and renal failure are among the most severe complications in SB (100), and contributes to early and late mortality.

In general, typical strategies for management of neurogenic bladders are used with goals of preventing UTIs, preventing renal calculi, managing detrusor pressures to prevent upper tract problems, monitoring renal function to prevent renal failure, and assuring continence. Clean intermittent catheterization (CIC) is an effective long-term management strategy for properly selected persons with neurogenic bladders from SB (100,101), usually concomitant with medications. However, there is no consensus for the evaluation, follow-along studies, and general management or management of bacteriuria among SB programs responding to a national U.S. survey (102), and there is no data about long-term outcomes. Renal function, as measured by creatinine (Cr) clearance, intravenous pyelogram (IVP), ultrasound, or scan, has been found to be normal in 47.7% of patients with SB and abnormal in 46.1% (94). In patients with lumbar-level SB who undergo CIC and are dry between catheterizations, only 38% have normal renal ultrasound and Cr clearance greater than 1.5mg/dL (103). This correlates

well with the fact that renal failure is the leading cause of death among patients with SB despite proper management and follow-up (93).

For adults with SB, almost 60% of hospital admissions are for urologic reasons, with neurologic problems accounting for almost 21%, and dermatologic problems almost 20%. Of the urologic admissions, almost half of these were for conditions such as UTI and renal calculi (104). In one study, urinary tract stones were responsible for about 30% of all renal complications (93). Repeated UTI, along with pyelonephritis and an already compromised kidney, can lead to acute renal failure with loss of nephrons. Unfortunately, by the time the serum creatinine begins to rise, the patient will have already lost up to two-thirds of their nephrons (105). It is important to note serum Cr is dependent on muscle mass, so in adults with SCD who often have low muscle mass, the serum Cr may not be indicative of the true renal function (106).

A study comparing long-term urologic outcomes among children and adults with neural tube defects noted the type of neural tube defect influenced the urologic outcome (107). Neurogenic bladder was seen in practically all those with myelomeningocele (MMC), with caudal regression syndrome (CRS) at >50% and spinal lipoma (SL) at <50%. Vesicoureteral reflux was most common in MMC, with CRS surprisingly close behind. The incidence of renal agenesis was highest in CRS. Subjects with SL were best controlled with CIC and medications.

There have been a variety of surgical procedures to assist with acute and long-term management of neurogenic bladders developed and/or promoted over the past 20 years in those with SCD. However, aside from a few retrospective cohort studies regarding specific interventions (108–110), there are no large or randomized controlled studies to identify best treatment strategies or factors that may indicate the procedure of choice (111,112). Furthermore, there are no reports of the effectiveness of surgical interventions over a lifetime. A small study from a center in British Columbia noted no significant increase in health-related quality of life in SB patients who underwent reconstruction for incontinence, compared to those who did not (113).

Moderate hypertension and proteinuria can also increase the risk of progression of renal dysfunction, eventually leading to chronic renal failure in patients with SB. For this reason, as well as the cardiovascular protective effects, even moderately elevated blood pressure should be treated. An angiotensin-converting enzyme (ACE) inhibitor should be considered, except in cases of advanced renal disease, due to the risk of hyperkalemia and further advancement of the renal disease (114). There are increasing numbers of children with SB who have undergone renal transplantation, with or without lower urinary tract reconstruction (115), and mortality and morbidities into adulthood are not well characterized.

Bladder cancer has been reported in adults with long-term SCI, and it has also been reported in adults with spina bifida (116). The characteristics appear to differ from adult-onset SCI patients with younger age onset, variable tumor histology and advanced stage, and poor survival. A case report cautions about recognizing pseudotumors of the bladder in SB and SCI (117).

Urinary incontinence in adults with childhood-onset SCD can persist into adulthood, and can be a socially limiting condition. Up to 80% of adults with SB can achieve social urinary continence (44), although a survey of persons identified in a state registry as having spina bifida reported only a slight majority of the adults had achieved independence in urinary management (118). For adults with SB, incontinence has been shown to be associated with partial employment or unemployment (119). There has been an attempt to begin to explore the issues of incontinence and quality of life. For adults with SB, urinary or fecal incontinence does not appear to play a major determinant role in health-related quality of life (120).

Many patients with SCD receive regular urologic follow-up as children, but not necessarily as adults. Adults with SCD who do not have urinary calculi or urinary incontinence are often assumed to be urologically stable. However, many adults with SB (and likely SCI) have been found to have urologic abnormalities, such as abnormal renal ultrasound or elevated serum Cr, that put them at increased risk of further renal problems, especially in the upper urinary tract (103). Many of these patients were also found to have increased pressure (>40 cm H_2O) in their bladder with the storage of urine at normal volumes.

Musculoskeletal

Level of motor function and musculoskeletal abnormalities are typically the areas of concern during growth and development, and often changes are not anticipated during adult years. Pain is a common complaint, and may be related to musculoskeletal issues, although in SB, tethered cord must be considered.

Overuse syndromes are common for wheelchair users, and have been identified in adults with SCI at shoulders, wrists, and hands. In a comparison with adult wheelchair users, those with childhood-onset disabilities had fewer shoulder pain complaints than those with adult-onset disabilities, even though lifestyles were no different (121). Shoulder pain in adults and adolescents with SB is not as common as in adult-onset SCI wheelchair users, although older SB subjects had more pain than younger ones (122). It is important to identify the risk for shoulder pain, recognize the onset, evaluate, and treat appropriately.

Scoliosis is common in SB, and is a common contracture noted in adults. It rarely progresses in adulthood. Spinal fusion has usually been performed prior to adulthood, but does not appear to improve the quality of life for those with SB (69). A combined anterior and posterior approach is reported to be more effective in older adolescents and adults with pelvic obliquity (123). Seating difficulties, back pain, and pressure ulcers arise from the scoliosis and pelvic deformities. Adults with SB report back pain less frequently than those with SCI, although, in general, pain complaints increased with age. In adults with SB, back pain may presage tethered cord.

Hip dislocation is related to thoracic or high lumbar neurologic-level abnormalities, and hip contractures notable in high neurologic level, but also in thoracic and high lumbar levels (124). There are no published reports of hip or knee pain in adults with SB, although this should not be unexpected in those who walk, given muscle imbalances and poor skeletal alignment. Charcot joints can be seen given the lack of sensation and muscle imbalances, especially in adults with SB, especially with lower-level defects (95).

Osteoporosis with associated fractures has become an area of interest and evaluation. For adults with SB, the high incidence of renal dysfunction is an added component for osteoporosis, given that renal dysfunction can lead to impaired bone mineralization (105). Renal dysfunction can also lead to metabolic acidosis as well as hyperparathyroidism; there may also be hyperphosphatemia, which can enhance the secondary hyperparathyroidism. This often necessitates the requirement for a low-phosphate diet and may also include taking phosphate-binding agents. For those with SB, bone mineral density is one to two standard deviations below the normal population, without a difference between ambulatory and nonambulatory patients (125). Treatment is not definitive. There are proponents for managing with calcium and vitamin D and/or using bisphosphonates, although no long-term information is available. Continued walking with muscle activity and weight bearing has a positive effect on bone mineral density in those with SB (125). Again, long-term follow-up is not available to identify dosing to achieve and maintain improvements.

Fractures may be more concerning than the risk factor of osteoporosis. Few studies detail incidence and prevalence. In an SB program cohort in upstate New York, where the vast majority is adults and late adolescents, the overall fracture prevalence was 200/1,000, most common during adolescence and least likely during adulthood. In comparing adult and childhood fractures, there was no significance to sex, body mass index (BMI), defect level, functional independence, shunted hydrocephalus, epilepsy, or other congenital anomalies (126). It has also been noted that patients with a higher level of defect have more of a risk for fractures (125,126). Most of the fractures reported in SB involved the tibia or femur, with 75% occurring in children after casting for an orthopedic procedure (127). Postorthopedic procedure and fracture management must be tailored to the situation. Environmental modifications to prevent fractures may be more effective than pharmacologic interventions (126).

Neurologic

Adults with SB are at an increased risk for neurologic complications because of the pathophysiology of their disability. Among the neurologic abnormalities seen in SB, the most common for which to monitor are hydrocephalus, Arnold-Chiari malformation/hydrosyringomelia complex, and tethered spinal cord. The vast majority with hydrocephalus have some form of shunting, possibly contributing to the increased survival rates seen today. However, shunt malfunctions are not uncommon, are often unsuspected, and can lead to significant morbidity and mortality (128). Recommendation is that adults with SB have routine neurologic evaluations and periodic computed tomography (CT) scans to monitor the shunt (44), with report that only 40% of adults with SB with a shunt have regular follow-up (129). Symptoms often seen with a shunt malfunction include headache, vomiting, lethargy, or change in mental status, with other neurologic sequelae also possible (44). Chronic headaches may be seen in adults with SB, and recurrent hydrocephalus or shunt malfunction must be excluded through intracranial pressure (ICP) monitoring if necessary (130). In the absence of increased pressure, further treatment options should be considered for pain management. Presence of hydrocephalus is associated with more dependence for self-care (including bladder and bowel care), for mobility, and for communication and cognitive assist into adulthood (96).

Adults with SB can have worsening neurologic symptoms from progression of an Arnold-Chiari malformation, with or without the hydrosyringomyelia complex. Presenting symptoms in adults with SB may not be those seen typically with brainstem compression, but may include upper limb weakness, sensory symptoms or reflex changes, ataxia, and lower cranial nerve palsies (94). Outcomes postsurgical intervention vary, including some level of recovery, stabilization of symptoms, further deterioration, and even death.

Tethering of the spinal cord can be seen at any age for those with SB who report changes in bladder or bowel habits, increase in leg weakness, change in sensory level, onset or increase of spasticity, report of pain (usually backache), or progression of deformities. In adults, an antecedent event such as trauma to the back or buttocks often initiates symptoms. Prominent

changes for adults are diffuse leg pain with referral to the anorectal area, and changes in bladder or bowel habits, often difficult to detect given reconstructive surgeries; progressive deformity usually is not noted, as is reported in children (131). Studies report that tethering, cord thinning, lipomas, cavities within the cord, and diastematomyelia are common in this population, with or without symptoms, so identification on scan may not be definitive (132). Treatment consists of conservative management of symptoms with monitoring or neurosurgical intervention. Neurosurgical intervention is usually associated with improvement in pain, urinary symptoms, and weakness, and poorer outcomes are associated with repeat procedures (133); however, not all outcomes are good.

Epilepsy may remain an active problem in adulthood for those with SB. Seizures are associated with shunts. Most series identify program cohorts with <15% requiring active seizure management with anticonvulsants (92,94).

Additional Medical Conditions

Pulmonary conditions may be seen in adults with childhood-onset SCD, although is not typically reported in SB cohorts. Restrictive lung disease occurs as a consequence of scoliosis, and decreasing pulmonary function with age in the general population is well documented. For adults with SB, changing pulmonary function may indicate further neurologic progression of a Chiari malformation.

Obesity is a reported medical condition in motor disabilities in general. It is commonly seen clinically in an SB population, although it is not mentioned in reported series of adults with SB. There are higher levels of body fat in adults with SB who do not walk (134), and there is an association of increased body fat with previous hydrocephalus (135). Obesity can often be an associated factor with onset and management of pressure ulcers. Appropriate nutrition and adequate exercise and activity should be a lifelong goal in persons with disabilities.

Pressure ulcers are a commonly occurring secondary condition in adults with SCD related to their impaired protective sensation. For adults with SB, there is an association with higher level and may be an association with hydrocephalus (136,137). Osteomyelitis is a complication of recurrent or chronic pressure sores, and may ultimately require amputation for management (118).

Gastrointestinal conditions can be seen in adults with childhood-onset SCD. Usually, they are chronic rather than new or late-onset problems, unless related to progressive neurologic conditions. Adults with SB also report problems with fecal incontinence in about 50% of reported cohorts (92,137). Constipation and diarrhea may continue through adulthood; megacolon can develop if management is inadequate. It has been noted that assistance is commonly required for bowel management in adulthood (96,118,138). Bowel continence is often difficult to achieve, and lack of continence can influence ability to participate in community activities. Of concern is assuring appropriate evaluation and management of acute abdominal symptoms; a case series of children and young adults notes etiologies included underlying neurogenic bladder or bowel, shunt, and complications from previous surgeries and a substantial mortality rate (139).

Latex sensitization/allergy is an important issue for adults with SB, and the rate may be higher for adults than children (140). The risk of sensitization increases with more surgical procedures being performed (141); the percentage of patients sensitized to latex ranges from 2.97% to 64.5%. Radioallergosorbent testing has been found to be more sensitive with a higher negative predictive value and more accurate than skin prick testing. The prevalence of latex allergy in the SB population is almost 19%, while the prevalence of latex sensitization is 32.4% (141); therefore, every effort should be made to limit exposure to latex.

Lymphedema is not reported in any large cohorts; however, it is clinically present and often associated with pressure ulcers. Simple over-the-counter compression garments are not useful, and most adults with SB are unable to apply daily Ace wraps. The edema is often responsive to lymphedema wrapping followed by tailored compression garments. There are two reports of severe and unresponsive lymphedema in adult women with SB—one responsive to suction-assisted lipectomy (142), and the other progressed to a diagnosis of lipedema, which has no successful treatment regimen (143).

Sexual Functioning

The number of adults with SB and childhood-onset SCI are increasing; therefore, the health care community can no longer ignore dealing with the medical and social issues of sexuality (144). A recent report noted that sexual education was received at school, and far less at home or by health professionals (145). Urinary incontinence may limit sexual participation (145), although this is not a consistent report (146). Higher neurologic level and presence of hydrocephalus was associated with less participation for both genders, but more problems with sexual functioning for men (137,145). There is no published data regarding contraception, but for women, contraception or suppression can be offered considering risks (eg, thrombotic risk, lack of sensation for intrauterine devices [IUDs]), side effects, and need for follow-up (147). Sexual education should be offered, with consideration for cognitive impairments when appropriate.

Many men with SB are able to achieve erections, but only about 53% are able to ejaculate (144). As anticipated, a lower defect gives men a greater chance of being able to sustain an erection, and there are normal testosterone levels. Erectile dysfunction is treatable with medications, although men with SB in a study did have some adverse effects after taking sildenafil, including dyspepsia, nausea, headache, flushing and nasal congestion, hematologic changes, and UTI. The dyspepsia was treated with antacids, and the UTI was treated with antibiotics. The remainder of the adverse events did not require treatment (148).

Women with SB had fewer problems with sexual functioning and were able to maintain pregnancies. Arata reported that there was no increase in back pain, no changes in neurologic or motor function, and no changes in bowel or bladder function during or following pregnancy (149). There were two commonly seen secondary conditions during pregnancy: UTI—but only in women who did not have normal voiding patterns—and pressure ulcers sometimes requiring hospitalization. Women with SB also had more emergent and elective C-sections than in the normal population. Women with SB were also found to have more antenatal admissions than women without SB, and it was noted that women with SB using wheelchairs exclusively had an average of 2.8 admissions antenatally per pregnancy, with an average stay of 25.8 days, while women with SB who walked had an average of 1.9 admissions antenatally per pregnancy, with an average stay of 17.3 days. More women with SB are admitted with preeclampsia than in the normal population, but given the incidence of renal dysfunction in this population, the prevalence is not overly high (149). Further study is needed to fully address the possible complications of pregnancy and childbirth in patients with SB. Pregnant women with SB may be evaluated through a high-risk pregnancy service.

There is no information specifically regarding typical gynecologic screening and prevention practices for women with childhood-onset SCD; however, national data regarding women with mobility impairments, especially those requiring use of a wheelchair, clearly demonstrate minimal participation, likely due to environmental and attitudinal barriers. Pregnant women with childhood-onset SCD should be at least evaluated through a high-risk pregnancy service.

Childhood-Onset Spinal Cord Injury

Mortality

Using data from the National Spinal Cord Injury Statistical Center over a 30-year period, it has been determined that life expectancy for adults injured as children appears to be slightly lower than that of those with comparable functional levels incurred through SCI as adults (150). More specifically, for those injured at a young age with incomplete injuries and minimal deficits, there is about an 83% chance of normal life expectancy, and for those with high cervical injuries without ventilator dependence, the estimate is about 50% of normal.

Life Satisfaction

Adults with childhood-onset SCI show relatively high satisfaction with life and relate this to independent living, education, income, satisfaction with employment, and social/recreation opportunities (151,152). Medical complications adversely affect satisfaction, especially presence of pressure ulcers, severe UTIs, and spasticity (152,153). Those with paraplegia are more satisfied than those with tetraplegia, and there appears to be no gender difference (151). Depression symptoms have been reported in adults with childhood-onset SCI, and are associated with medical complications, social participation, and incomplete injury (154). Life satisfaction is not associated with level of injury, age at injury, or years with disability (152).

Of interest is that adults with childhood-onset SCI self-perceptions are not reported to be as significantly altered as clinicians anticipate (155,156) and, therefore, are enriched by services and providers that emphasize education, employment, and long-term health management (152).

Urology/Nephrology

The most common reported health complication for adults with childhood-onset SCI was UTI (157). Typical strategies for management of neurogenic bladders are used, as previously noted, and CIC continues to be the typical management. Adults with childhood-onset SCI also frequently receive reconstructive lower tract surgeries; however, the decision factors determining best treatment options have not been determined. There are studies reviewing specific interventions (108), but there is no information regarding long-term effectiveness of surgical options.

Adults with childhood-onset SCI have some association of urologic complications that relate to age or years with disability, and consequently, regular urologic follow-up is recommended. In a large study of adults followed at Shriners Hospital for Children in Chicago, Vogel reports older age at interview and longer years with disability were associated with orchitis or epididymitis (157). Also, greater impairment was related to UTI, severe UTI, and renal stones. Severe UTIs were also related to poor life satisfaction (153). Although not reported in this cohort, bladder cancer and pseudotumors of the bladder may also be present.

Musculoskeletal

For adults with childhood-onset SCI, pain at any site was the most common complaint, and shoulder pain was noted in almost half of the respondents in interviews, as reported by Vogel at al (158). As was noted earlier, overuse syndromes must be considered, especially at the shoulder. In general, for adults with childhood-onset SCI, longer years with disability and increasing age are associated with shoulder pain (158). Etiology must be identified, and evaluation and treatment are essential. An outpatient physical therapy program or a home exercise program for shoulder pain, with or without impingement, in SCI (159) have been shown to be effective in pain management.

For adults with childhood-onset SCI, younger age at injury and longer years with disability has a correlation with scoliosis (158,160). More severe and frequent scoliosis has been reported in paraplegia and complete lesions, and lordosis has been noted to be greater in paraplegia and incomplete lesions (160). There is no evidence that bony injury at the time of childhood-onset SCI influences the development of scoliosis or lordosis (161).

For adults with childhood-onset SCI, younger age at injury and longer years with disability were associated with hip subluxation, and older age at injury was associated with elbow and ankle pain (158). Back pain may be seen in about 20% of patients unrelated to scoliosis, and ankle pain and elbow contractures are associated with tetraplegia, and hip contractures with paraplegia (158). For those who walk, presence of hip or knee pain should be questioned, and for any pain complaint, appropriate workup and management should ensue.

There are no reports detailing osteoporosis in adults with childhood-onset SCI; however, there is scientific research that identifies osteoporosis as a common secondary condition in SCI. As noted, the most effective treatment has not been established, and dosing parameters for medications or other strategies are unknown. Case series have advocated for the use of cycling with functional electrical stimulation (FES) to improve bone mineral density (162).

Adults with childhood-onset SCI report fractures associated with increasing age and longer years with disability (158). Those with lower cervical injuries tend to have more pathological fractures than the other groups.

Neurologic

Neurologic sequelae for adults with childhood-onset SCI appear to be limited by report in the literature. The presence of autonomic dysreflexia (AD) is not related to increasing age, age at injury, or years with a disability. AD is associated with greater neurologic impairment and is a common health condition for adults with childhood-onset SCI (157). Spasticity is seen in >50%, older age at injury is associated with spasticity, and longer years postinjury notes spasticity or neurologic changes (158). Monitoring for changes in function and adjustment to spasticity or other management must be part of routine medical care, with consideration for all possible options, including injections, pain management, medications, and surgical considerations.

Additional Medical Conditions

Pulmonary conditions may be seen in adults with childhood-onset SCI. Restrictive lung disease occurs as a consequence of scoliosis, and the addition of weakness or paralysis of secondary respiratory muscles may further increase risk for recurrent respiratory infections (150). Survival for childhood-onset SCI requiring ventilator support has improved in recent years, with reported survival up to 23 years (163). Deaths in this cohort were related to respiratory complications, followed by unknown and suicide. There have been rare unscheduled hospitalizations, and life satisfaction is associated with better mental health.

Obesity is a reported medical condition in motor disabilities in general, but it is not mentioned in several series of adults with childhood-onset SCI. Appropriate nutrition and adequate exercise and activity should be a lifelong goal in persons with disabilities.

Pressure ulcers were reported in just less than 50% of adults with childhood-onset SCI, were more common in men, and more common in greater neurologic impairment (157).

Gastrointestinal conditions are not common, other than neurogenic bowel–related issues. Bowel incontinence is reported in >50% of adults with childhood-onset SCI, and is seen with older age and greater impairment, although not with increasing years with disability.

Latex sensitization/allergy is seen in SCI, but seemingly not as frequently as SB. It is unclear what the incidence of latex allergy is in the childhood-onset SCI population, although it is known that women more commonly report a latex allergy (157).

Osteoporosis. There is no published data about osteoporosis in adults with childhood-onset SCI differing from adults with SCI. Treatments studied have included bisphosphonates and functional electrical stimulation (FES) exercise, although there is no definitive treatment suggested by the research findings. Fractures are the complication, and are reported with increasing age (158). Another bony deformity, heterotopic ossification (HO), is not reported as significant in this population, and decreased with age in a study of adults with SCI (164).

Sexual Functioning

There are less data about men and women with childhood-onset SCI. Although the general information available about adults with SCI can be helpful, it is not clear if it can be generalized. It is known that semen quality decreases at about two weeks postinjury, which could imply decreased fertility for adult men with childhood-onset SCI (165). Fertility is also affected by bladder care (166).

There are no menstrual cycle difficulties known for women with childhood-onset SCI (167). A multicenter study of women's self-reported reproductive health after SCI, likely adult-onset injuries, reported complications from pregnancy, labor, and delivery to be more frequent than what was noted preinjury, and delivered babies of low birth weight (168). Women reported increased bladder spasms, muscle spasms, and autonomic symptoms at some time during their menstrual cycle. Experience of orgasms and methods of contraception varied. The effects of menopause are unknown.

There is no specific information about typical gynecologic screening and prevention practices for women with childhood-onset SCI; however, national data concerning women with mobility impairments, especially those requiring use of a wheelchair, clearly demonstrate minimal participation likely due to environmental and attitudinal barriers. Risks for use of contraception options are not known; however, combined hormone oral therapy carries a risk for thrombophlebitis; progestin-only medications have early irregular bleeding and long-term suppression effects; and intrauterine devices with lack of sensation require vigilance for correct placement and risk of rare complications such as perforation, infection, or ectopic pregnancy (147). Given the information self-reported by women with SCI, pregnant women should be at least evaluated through a high-risk pregnancy service.

Limb Deficiency

Pediatric-onset limb deficiency is not uncommon, with 4/10,000 in upper extremity congenital limb deficiency alone. In addition, lower extremity hemimelia, traumatic amputations, and childhood cancers are associated with pediatric limb deficiency. Very little is known about aging with this disability. However, certain comorbidities and secondary conditions are typical for this group (see Table 15.2). Weight control is important to prevent osteoarthritis (169,170). One author describes increased velocity and lower effort in elderly amputees if a locked knee is used (171). Changes in gait or use of upper limb prostheses with aging in this population may be due to a variety of typical disorders of aging, including arthritis, sensory deficits, muscle weakness, or heart disease. Typical surveillance for these disorders is important to maintain ambulation status.

Intellectual Disabilities

Intellectual disability is a common reason for disability in childhood, although less prominent in adult surveillance. People with intellectual disabilities experience age-related health impairments at a higher rate and earlier age than people without disability (172). Depending on the etiology of their disability, they may be at much higher risk for both secondary conditions and comorbidities. These conditions can be life-threatening or life-altering. Some may be prevented or treated if identified early. Down syndrome (DS) will be discussed as a separate entity, as more is known about aging with this condition. Strategies for minimizing functional limitations will be highlighted. Rehabilitation surveillance and treatments will be discussed (see Table 15.2).

Intellectual Disability

Individuals with intellectual disability (ID) are living longer and experiencing most of the same illnesses as the general population (173). Their life expectancy remains somewhat less than the general population, but has steadily increased with the move away from institutionalized care (174). Community-based health care for people with ID is not well organized, and people with ID experience poorer health than the general population (175).

Cardiovascular

Janicki and colleagues noted that cardiovascular disease (CVD) and respiratory diseases were more common causes of death in the elderly with ID than in the general population, with cancers in a less prominent role (173). Although there have been discussions of significant rates of chronic health conditions and general poor health for adults with developmental disabilities, more recent studies of adults receiving state or national support in New York state, Taiwan, and Israel (82,83,176) note gradual increases in health conditions, but not with higher incidence than in the general population, and in some cases lower.

Obesity

In a cross-disability study of a South Carolina primary care practice that included almost 50% adults with developmental disabilities (DD), there was a lower odds ratio for coronary artery disease, cancer, and obesity for adults with DD in comparison to those without

disabilities and compared to other disability groups (177). Although obesity was reported as low in the South Carolina study, other studies report obesity as being more common in adults with developmental disabilities. Obesity in people with ID is higher, compared to those age-matched without ID (35.4% vs 20.6% in one survey) (178). Other researchers have found twice as many people with ID to be obese as those without ID within the same community (179,180). Those with mild ID have more obesity than those with severe ID, and there can be a move out of the obesity state (181). The combination of increased obesity and mortality due to CVD lead to a recommendation of increased surveillance and prevention strategies for obesity-related disease.

Respiratory

Several authors describe respiratory ailments as important factors in morbidity and mortality of aging adults with ID (172,173,182). Janicki and colleagues identified pneumonia as the most prevalent cause of death due to respiratory illness and second only to CVD (173). Sleep apnea due to obesity is mentioned as a comorbidity and may require separate screening or sleep studies.

Health Maintenance

People with ID require the same screening for cancers, diabetes, hyperlipidemia, hypertension, bone density, and ophthalmologic and hearing disorders as the general population. Communication about the results of these screenings and plans for treatment of any abnormalities may need to be through a proxy. Prevention strategies for diseases related to obesity may need to start earlier than in the general population. Preexisting conditions of epilepsy and poor oral health should be monitored closely (183). GERD and *Helicobacter pylori* infection is increased in prevalence and undertreated in people with ID (184,185). Symptoms of GERD should be queried in people with ID and treatment undertaken, as with the general population. Osteoporosis also is more prevalent in people with ID, with precipitating factors of small size, hypogonadism, and anticonvulsant therapy (186–188). Fractures are associated with frequency of falling. Screening for osteoporosis and falling should commence during early adulthood, with follow-up depending on the results.

Mental Health

Mental health impairments are prevalent in elderly people with intellectual disability. Estimates vary from 20% to 70%, depending on which assessments were used and the exact population studied (189–192). Dementia, depression and general psychiatric symptoms are all more prevalent in the elderly population with ID. Each of these groups also had high numbers of health comorbidities, such as CVD, sensory impairment, and mobility problems. Researchers note that life events, such as relocation, were more frequent in adults with ID than in comparison groups (191). Medication review is a priority for clinicians treating people with ID. Polypharmacy is a significant problem for people who may not have adequate understanding of the need to report side effects or efficacy of medications. Medications should not be prescribed unless a system is in place to ensure compliance, safety, and monitoring of efficacy (183). Surveillance for mental health problems in aging people with ID should be a priority, along with treatment of physical comorbidities, which may contribute to or appear as mental health concerns.

Sexual Functioning

People with ID are often not afforded typical education, contraception options, or sexual health screening. They face a high risk of sexual abuse, are unaware of protection from sexually transmitted diseases, and are generally unsupported in attaining healthy sexual relationships (147,193,194). Women are often prescribed suppression therapy (194,195). Sterilization for women with ID is more common abroad, and related to severity and living arrangement (196). Women and men with ID can be provided with education and support for sexual functioning, and regular health screenings can be accomplished with modifications and support (147).

Down Syndrome

More than half of people with Down syndrome (DS) will survive to age 50, and half of those will be alive at age 65 (197). Most people with DS are living in the community with family or in supported living. They require increased health care surveillance as they age due to higher prevalence of numerous clinical conditions. Access to appropriate health care may prove difficult for people with DS, as they may have difficulty with communication or behavior and typical primary care practices may not meet their needs. Specific health screening programs have shown a dramatic increase in recognition of unmet health care needs (23). Rehabilitation clinicians can assist families and primary care physicians to provide optimal maintenance of function throughout life.

Mental Health

Mental health problems in people with DS have been well described in the literature. An elderly (>65) group was well described by Cooper and colleagues

as having increased dementia, anxiety, and depression when compared to a younger group (190,198). Symptoms of Alzheimer's may be seen as early as age 35 and will be noted in 75% of people with DS by age 60 (191). A variety of causes have been postulated for the high incidence of Alzheimer's/dementia in people with DS, including antioxidant stress (199) lower bioavailable estradiol in women (200), and decreased alpha and beta secretase activity (201).

Treatable comorbidities, which may look like Alzheimer's, must be ruled out. These include hypothyroidism, visual and hearing impairments, depression, and epilepsy, all of which are significantly more common in DS than in other populations with Alzheimer's (202). Likewise, systemic illness, infection, drug effects, and alcoholism must also be eliminated as possible treatable causes of Alzheimer's symptoms (203).

Depression may cause decreased function in people with Down syndrome (177,198,203,204). Experiences of loss may trigger depression, as may changes in work or living situations. Depression may be treated with counseling; however, training or experience with this population will be needed for counseling to be effective. Treatment may also include medications. The use of selective serotonin reuptake inhibitors (SSRIs) in DS has been anecdotally described, but no randomized controlled trials have been reported to date (203,205–208).

Endocrine System

Thyroid disease is well described as a comorbidity of DS (23,172,173,189–191,197,202–204,209–212). Hypothyroidism is found in 15% to 50% of adults with DS (197,202,203,213–215). Thyroid-stimulating hormone levels should be assessed annually in patients with DS (203).

Diabetes mellitus may have a higher prevalence in adults with DS, but is rarely discussed in the literature (202,203,216,217). McDermott and colleagues found fewer developmentally disabled adults with diabetes than control adults in a large primary care practice (177). Typical yearly testing and treatment as needed should suffice for surveillance.

Otolaryngology

Hearing loss is extremely common in people with Down's syndrome and may not develop until adulthoo(189,197,203,218,219). Poor hearing may exacerbate preexisting communication difficulties and present as behavior problems. Auditory testing is recommended at least every two years in adults with DS.

Sleep apnea is also a common problem for adults with Down syndrome (220–224). The cause is likely multifactorial, with obesity (225), central (brainstem respiratory control) mechanisms (222), and obstructive (221,223,226) sources all implicated. Sleep apnea is associated with worsened cognitive skills (227), and may be successfully treated in a variety of ways (226,228,229). A sleep study is indicated to identify the cause and therefore predict the successful treatment for sleep apnea.

Musculoskeletal

Premature arthritis has been reported in adolescents and adults with Down syndrome and may be associated with joint subluxations and dislocations (197,230). Hip instability may occur or worsen in adults with DS and is associated with decreased ambulation status (231). Foot pain and arthritis may be associated with severe pronation and atypical gait; however, very little research has been done in this area (232). X-rays are indicated if ambulation status deteriorates. Treatment may begin with NSAIDs, but further evaluation and possible referral is indicated if typical arthritis pain relief strategies are not sufficient to maintain function.

Osteoporosis is also more common in adults with DS and is found in both men and women at a significantly younger age than in the general population (233). Long bone and vertebral compression fractures are common (234). Decreased physical activity, short stature, early menopause, low muscle tone, and increased incidence of thyroid disease may all be factors in osteoporosis in DS (200).

Atlantoaxial Instability

One to two percent of individuals with DS will have cervical subluxation or symptomatic atlantoaxial instability (AI) (235). Routine monitoring via x-ray is no longer recommended, but vigilance for progression is recommended. Concerning symptoms include new torticollis, weakness, neck pain, change in gait, change in bowel or bladder function, increased reflexes, or other symptoms of spinal cord compression (236). Presentation of these symptoms requires immediate stabilization and referral for surgery consideration (197,235,236). Outcomes from surgery are not always acceptable (237,238).

Cardiac

Nearly half of infants born with Down syndrome will have a structural heart anomaly. Most of the typical congenital heart abnormalities will have been corrected in infancy. Increased incidence of mitral valve prolapse in adults with DS has been reported (197,203,209). Careful auscultation should reveal any

change in heart murmurs, and electrocardiogram and chest x-ray can follow.

Cardiovascular disease (CVD) is not well studied in people with Down syndrome. As people with DS live longer, become more obese, and less active, it is reasonable to expect to see increasing rates of CVD (173,225,239). Several authors have noted decreased cardiovascular capacity in people with DS (240–244). A 2005 Cochrane review of exercise training programs for people with DS revealed only two small trials of good quality. Of these, only maximal treadmill grade was improved after the training program. Other studies have investigated components of fitness such as leg strength and capacity, as noted previously. Small uncontrolled trials not included in the Cochrane review have shown only limited aerobic improvement with exercise training programs (243,245).

Obesity

Obesity is a lifelong issue for many people with Down syndrome. As many as 70% of adults with DS are reported to be obese (225,246,247). Health promotion and group exercise classes have been successful at significantly reducing body fat percentages in short-term programs (240,241,243,245,248–250).

Sexual Functioning

There is little published information regarding sexual functioning in adults with DS. It has long been held that males are infertile and females are fertile or subfertile based on histology of gonads and serum levels (251,252). There are case reports of men and reports of small series of women who have been fertile (253,254). The male offspring are reported to have no abnormalities, congenital or genetic. In contrast, the female offspring are reported to have DS, be chromosomally normal, or have other congenital defects or ID. The need for education and counseling, monitoring for sexual abuse, and social support is obvious.

Health Maintenance

People with Down syndrome require the usual screenings for testicular and cervical or breast cancer and hypertension. Celiac disease is now recognized as a common condition associated with DS, and monitoring should be a part of health maintenance (255,256). Dental health is important, as gingivitis and periodontal disease are more common in people with DS (257–260). Cataracts and keratoconus both occur with increased frequency in people with DS. Regular ophthalmologic examinations are indicated to evaluate for these conditions. Health care screening and promotion programs have demonstrated improved detection of symptoms and compliance with health recommendations (172,203,225,248,249,261). Sexual health should not be ignored, and often contraception or suppression is prescribed for women for hygiene problems with menstrual cycles (147,195).

Williams' Syndrome

Williams' syndrome (WS) is caused by a gene deletion on chromosome 7. It is rare, occurring in 1 of 20,000 live births (262). Devenny and colleagues have been following a group of 15 adults with WS, some of whom have participated in a 15-year longitudinal study on aging in adults with ID. The participants with WS demonstrated early and rapid decline in long-term episodic memory not found in other adults with ID. Verbal short-term memory was better than their peers with ID and did not decline with age (262,263). No association was found with physical or mental comorbidities. Because Williams syndrome has only been clearly described within the current generation of adults, few people have been extensively studied, and we do not yet know the causes of the apparent precocious aging noted in this population.

TRANSITIONS AND ACCESS TO HEALTH CARE

Transition of Care to Adult Services

Improved medical care and increasing numbers of adults with childhood-onset disabilities has lead to much interest and concern about the transitioning of care of young adults from a family-centered pediatric approach to a self-directed adult care model (2). Pediatricians often will maintain care for their patients well into adulthood, especially for those with complex medical conditions (264). A consensus policy statement, adopted by the American Academy of Pediatrics, American Academy of Family Physicians, and the American College of Physicians—American Society of Internal Medicine, states that the transition of care should "maximize lifelong functioning and potential through the provision of high-quality, developmentally appropriate health care services that continues uninterrupted as the individual moves from adolescence to adulthood." (265) Barriers to transitions cited include lack of adult provider training, poor communication between pediatric and adult providers, and need for self-direction navigating the adult system (266). There have also been suggestions for specific elements to support a transition, such as preparation, flexible timing, care coordination, transition clinic visits, and interested adult care providers; however, this remains theoretic (267). At present, the

science is at an early stage of development (266,268). Adolescents with early-onset and chronic health care needs have received an organized level of care, and maintaining coordination of often complex care is an important part of quality health care over a lifetime.

There have been reports of successful transition of service models. Successes related to planned and evaluated transitions (269), personal health records management (269,270), and provision of education on health and needs (271). There remain questions regarding shared responsibilities for the transition (272–274), need for protocols (275), and timing for planning and implementation. Pediatric physiatrists can often provide the stability for this transition. Table 15.3 identifies challenges for transitioning health care from pediatric to adult systems of care (276).

Access to Health Care

Access to health care for young adults has been problematic for funding reasons as well as transition-of-care difficulties. Lack of insurance has been highlighted, and is as common among young adults without disabilities as those with disabilities, as noted through the National Health Information Survey (277). However, adults with disabilities had eight times greater odds of reporting unmet health care needs and six times greater odds of having no usual source of care, compared to those without disabilities. The majority of young adults with disabilities reported a gap in their insurance coverage, and many were uninsured over a three-year period (278).

Access also involves environment, attitudes, and systems. Architectural barriers have been addressed through the Americans with Disabilities Act, although accessible health care providers' offices and accessible examination and procedure tables continue to be available on only a limited basis. Attitudinal barriers are more difficult to remedy, and involve both consumers and providers. Rehabilitation clinicians may need to ask more direct questions of their patients regarding secondary conditions and additional health concerns to better identify conditions and begin management. Physiatrists can act as a resource for primary care providers, who likely have limited knowledge regarding persons with lifelong disabilities. Consumers with communication or cognitive impairments (eg, hearing impairment, speech production impairment, brain injury, ID) may need more time to communicate, require an interpreter, or require personal preparation time for the appointment in order to have their needs conveyed; modification of appointment times, with preplanning and written lists of concerns, can often be helpful. Consumers may seek help only late in the course of an acute medical condition or change because of previous difficulties managing the system. Specifically, consumers report that their routine health care providers know little about their disability and its impact on health and function (43).

15.3 Characteristics That Affect Successful Transition of Care*

Simple transition	Complex transition
Single condition	Multiple conditions
Few medications	Multiple medications or allergies
No cognitive impairments	Profound intellectual disability
No physical impairments	Physical impairments
No behavior concerns	Serious behavioral issues
Mentally healthy	Mentally ill
Effective family support	Family ineffective
Few physician consultants required	Multiple subspecialties involved
No nursing care needs	In-home skilled nursing and special equipment and supplies

Adapted from Ref 282.

Health and Wellness Agenda

As a result of the steady improvement in medical care and social support systems during the last 50 years, persons with disabilities are healthy, conducting active and productive lives, and generally living longer. The medical paradigm must now shift from that of illness and disease to one of health and wellness. The health care delivery system must view persons with disabilities through a typical health maintenance and preventive medicine approach. This requires a change in attitudes and care models. Both prevention and promotion strategies should be employed: prevention of activities that lead to illness and disease (eg, smoking cessation, dietary discretion, routine laboratory and examinations, protected sexual activity) and promotion of activities that improve general well-being (eg, stress management, exercise) adapted to meet individual requirements and performance (Table 15.4) (279,280). However, positive health behaviors require social, health, and community resources. The more resources a person has, the more likely that individual will engage in health promotion and protective behaviors (281). Again, access is an important issue. Availability of information in appropriate modalities and the education of consumers are important. To participate in positive health behaviors, one must be interested, be ready to make changes, have the needed resources, and have a supportive environment. Early involvement of adolescents with mobility impairments in health promotion activities may pave the way for maintaining these behaviors into adulthood.

Since musculoskeletal conditions are the most common age-related changes and secondary conditions that

15.4 Health Preventive Screening Services*

HEALTH CONDITION	RECOMMENDATION FOR GENERAL POPULATION	MODIFICATION NEEDED
Hypertension	>18 yrs and annually	None
Immunizations	Follow schedule	None
Cardiac, vascular diseases	Men: >35 yrs; possibly 20 yrs with CAD risks	None
Lipid	Women: >45 yrs with CAD risks; possibly 20 yrs with risks	
Abdominal aortic aneurysm	Men: age 65–75 yrs if ever smoked	Accessible procedure environment
Cancer		
Colorectal	Men and women, screening >50 yrs	Accessible procedure environment
Women's health		May need 1:1 assist
Breast	Annual mammogram >40 yrs	Accessible office exam table and procedure environment
	Clinical exam, every 3 yrs 20s–30s, annual >40 yrs	May need 1:1 assist
	Self-exam option >20 yrs	
	MRI only with high risk, annually	
Cervical	Screening begins 3 yrs postintercourse, not later 21 yrs	Accessible procedure environment
	Age 30 yrs, with 3 normal Pap tests, screen 2–3 yrs	May need 1:1 assist
	>70 yrs, 3 normal Pap tests and no abnormals or risks may discontinue	
	D/C after total hysterectomy and no risks	
Prostate	Offer PSA and digital exams >50 yrs, not required	Office exam table accessibility
	High risk, test 40 yrs; if normal, begin routine 45 yrs	
	>75 yrs not required	
Metabolic		
Obesity	Screening for all, with counseling and behavior interventions offered	Requires accessible scale
Diabetes mellitus	Screening for asymptomatic sustained blood pressure >135/80 mm Hg	None
Mental health		
Depression	Screening if able to diagnose, treat, follow-up	May require modification to queries; requires support to diagnose and treat
Dementia	Insufficient data to recommend in general population	Important to question in DS
Violence	Not recommended for general population	High incidence of violence and abuse in disability; offer opportunity to discuss
Tobacco use	Recommend regular screening and offer cessation interventions	None
Exercise	Unclear that screening is effective in the general population	Exercise is an important activity for those with motor impairments; has been shown to be effective for improved performance, pain control, weight management
Aging		
Vision	Presbyopia, cataract, macular degeneration, and glaucoma increases with increasing age—unclear screening is effective	Accessible examination
Hearing	>50 yrs, hearing decreases; unclear if screening is effective	Accessible examination

CAD, coronary artery disease; MRI, magnetic resonance imaging; D/C, discontinue; PSA, prostate-specific antigen; DS, Down syndrome.

Adapted from Ref 286.

affect performance, it would seem most reasonable to view typical physiatric strategies and interventions as preventive management techniques. Use of adaptive equipment, energy-conservation techniques, joint protection, and ergonomic positioning may enhance function, decrease musculoskeletal complaints, and possibly prevent or delay some functional changes. Personal attitudes (of the person with a mobility impairment or their personal support system) may have to change before a person with impaired mobility will consider such assistance or be supported in considering the value of employing supportive (less independent) techniques.

Exercise is a well-known health-promoting behavior, and its effects are positively demonstrated in persons with disabilities (24,282–286). Benefits of a regular exercise program include improved fitness, weight reduction, improved mood, and improved sleep. It is also known that persons must be judicious in participating in exercise programs, given the issues of fatigue and pain. Of course, care must be taken in prescribing exercise for persons with impaired mobility; they should participate in an appropriate program of exercise or activity, especially keeping in mind their risk factors for musculoskeletal injury. Jogging or running started by young adults without disabilities more often resulted in discontinuation of exercise because of joint pain than for persons who started a similar exercise program in their middle years, leading one to believe that long-term, high-impact exercise may result in pain. Aquatics programs can eliminate the wear and tear to joints. Adults with cerebral palsy tend to report perceived changes in balance and then fear of falling, which usually improves with a general fitness program. Exercises, including strengthening exercises, are not contraindicated for persons with spasticity. Generally, adults and young adults with developmental disabilities do not participate in routine fitness or exercise programs. This may be as much from limited knowledge in this area as from attitudes of care providers and persons with disabilities relative to exercise as a self-directed, nonmedical, or leisure activity. Consideration of exercise programs at home, in a health club, or as part of an individual recreation program (with or without modifications) must be initiated earlier than adulthood to achieve long-term participation. And, just as in the nondisabled population, priorities for persons with mobility impairment should include exercise and fitness.

SUMMARY

Adults with early-onset disabilities are generally healthy. Not all adults have serious health problems, and many now recognize the aging process as a natural course of events. The most common age-related changes and secondary conditions involve physical performance and the musculoskeletal system. Prevention strategies require knowledge of expected changes, recognition of changes that alter function and require intervention, and an understanding of interventions that positively impact on function. This requires that a person with a disability have access to knowledgeable health care providers. Physiatrists may offer that knowledge through direct clinical service or indirectly functioning as a resource in the community. Environmental, communication, attitudinal, and systems barriers must be overcome in order for health care providers and people with disabilities to work together for the best possible outcomes.

It is time to reconsider the model of illness and disease for persons with lifelong disabilities. Particularly in the realm of mobility, a health and wellness model should be developed. Use of prevention strategies must be considered in childhood and adolescence to address the more frequent secondary conditions. Programs of fitness and exercise have been proven beneficial in nondisabled groups and disability groups alike. Health promotion strategies should be employed for persons with congenital and childhood-onset mobility impairments.

PEARLS

- Most adults with early-onset disabilities are healthy with aging. Significant or acute loss of function should not be expected, and evaluation must ensue.
- Adults with early-onset disabilities view themselves as healthy, although this is dependent on the number of health conditions. Life satisfaction is usually not associated with disability. This is within the context of measurement instruments that have not been standardized for those with disabilities.
- Urinary/renal issues for adults with childhood-onset SCD are of primary concern. However, management of pressure ulcers and lymphedema can be most problematic.
- Consider newer tone management options to manage pain or improve function, with concomitant therapy. With decreased tone, additional focused therapy can improve function.
- Pain is common in adults with childhood-onset disabilities. All pain is not arthritis, and there can be many etiologies. Never miss the opportunity to question, evaluate, diagnose, and treat. Although most pain is musculoskeletal in origin, if there is no improvement, consider neurologically based etiologies, such as stenosis, tethering, or entrapments.
- Exercise can improve performance, and any person with a disability can participate, with modifications. Do not just consider therapy—home- and community-based programs can be effective.

Best Resources

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16

The Assessment of Human Gait, Motion, and Motor Function

James J. Carollo and Dennis J. Matthews

Instrumented gait analysis has evolved into a recognized objective evaluation that is important in surgical and rehabilitation therapy planning for the child with an abnormal walking pattern. The technology related to gait and motion analysis has improved significantly in recent years, enabling the collection and analysis of large amounts of data obtained simultaneously from a variety of specialized measurement instruments. The resulting quantitative description provides a comprehensive snapshot of the subject's movement pattern at a particular point in their development or at discrete intervals in their treatment. The clinician can use this information to describe the complex physiological interactions that lead to abnormal movement and motor control, and better understand their impact on gait, movement, and other functional activities.

A clear understanding of instrumented gait analysis data and the ability to perform a meaningful interpretation that is clinically relevant remains a challenge for many physicians. This may be attributable to the specialized nature of the gait analysis report or the false perception that an extensive biomechanics background is required to integrate movement data into the clinical decision-making process (1). More frequently, however, the underutilization of modern gait analysis techniques in pediatric rehabilitation is related to the difficulty associating gait measurement deviations seen in the report with specific functional deficits during the walking cycle. Fundamental to making this connection is a clear understanding of the functional demands of normal gait. Recognizing the essential features of normal, efficient locomotion provides the basis for identifying the *absence* of these features in the child with gait dysfunction and, when applied systematically, can provide a strategy for clinical gait analysis (2).

Therefore, the goal of this chapter is to familiarize the clinician with basic gait analysis principles by focusing on the inherent functional requirements of normal locomotion. This provides a framework for using specific gait measurements to pinpoint the joint or muscle system responsible for a particular functional deficit, which can then be the target of appropriate clinical interventions.

NORMAL GAIT IS CYCLICAL AND SYMMETRIC

The principal goal of locomotion is to propel the body forward as efficiently as possible. The most natural way to accomplish this task is to employ a bipedal gait pattern, where the base of support alternates from one leg to the other. Inman has described the cyclical alteration of each leg's support function and the existence of a transfer period when both feet are on the ground

as *essential features* of normal locomotion (3). Since normal gait assumes no biomechanical advantage provided by either limb, a natural consequence of these essential features is the existence of a repeatable pattern that is both cyclical and symmetric. Figure 16.1 illustrates one complete gait cycle, or *stride*, and includes the *time periods* and *temporal events* associated with foot/floor contact that necessarily arise from changing the support limb. Temporal events are specific moments in time that divide the gait cycle into discrete time periods of specific duration, and are identified by the stick figures along the top of Figure 16.1. Typically, a cycle begins when one foot makes contact with the walking surface (initial contact) and ends when that same foot strikes again. This is the functional definition of a stride. Using such a convention allows a stride to be *time-normalized*, where a specific stride location is expressed as a percentage of the total cycle time or *stride period*. Time normalizing the gait cycle facilitates comparing subjects with different stride lengths, stride periods, and walking speeds on the same scale. Figure 16.1 illustrates the time periods and temporal events relative to the shaded ipsilateral side. If a subject's gait pattern is normal, the stride would be cyclical and symmetric inherently, and so be equally ascribed to either side. The temporal event of foot off (sometimes referred to as *terminal contact*) separates the gait cycle into stance and swing periods. Typically, stance period accounts for 60% to 62% of the total gait cycle and swing period takes the remaining 40% to 38%. We have intentionally refrained from using the terms "stance phase" and "swing phase" here to avoid confusing these intervals with the *phases* of gait to be introduced in a later section, although in common practice, the terms can be used interchangeably.

Stance period includes two intervals of double limb support at the stance/swing transitions, each lasting approximately 10% to 12% of the gait cycle at typical walking speeds. These are generally described as initial and final double support, but can also be identified in the context of the leading limb as right or left double limb stance period. The duration of the double limb support periods decrease with increasing walking speed, reaching zero at the moment running begins. The time interval between the initial and double support periods is defined as the single support period, and is the same duration as the swing period of the opposite limb. Assuming normal symmetry, any reduction in double limb support time is absorbed by a proportional increase in single limb support time, but since single limb support always corresponds to the contralateral swing period, the overall stance period decreases, reaching 50% at the initiation of running when double limb support reaches zero. When both limbs' primary temporal events of foot-strike (initial contact) and foot-off (terminal contact) are represented on the same time scale, the duration of each time period is easily illustrated. These general terms for temporal events are applicable, regardless of gait pathology. Other terms are routinely used to identify temporal events marking the transition from swing period to stance period (heel strike, forefoot initial contact, foot flat) and stance to swing (toe-off, push-off), but should only be used when they clearly describe the observed foot/floor contact pattern.

While period durations relative to a single side are easily described when the gait cycle is represented on a linear scale, left/right symmetry may be more easily conceptualized when the gait cycle is wrapped around a unit circle (2,4,5), as shown in Figure 16.2. For typically developing children and adults, ipsilateral and contralateral initial contact and foot-off will occur directly opposite each other around the circle, or 180 degrees out of phase. This graphically illustrates

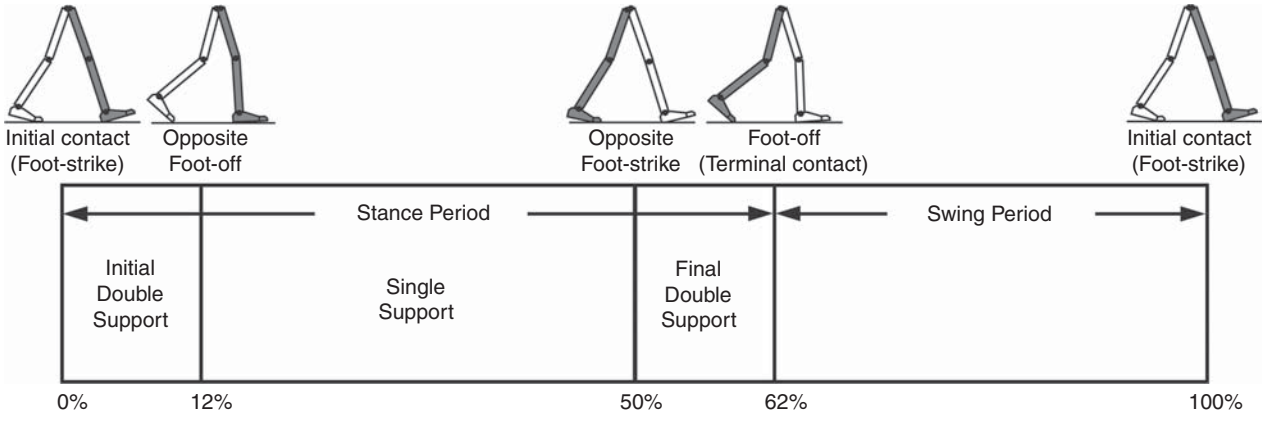

Figure 16.1 A typical gait cycle normalized in time, and represented on a linear scale from 0–100% of the total stride. This repeating cycle begins with initial contact and ends with the next initial contact of the same foot. The stick figures shown on top represent temporal events associated with foot-to-floor contact. They divide the cycle into swing and stance periods, one period of single support and two equal periods of double support.

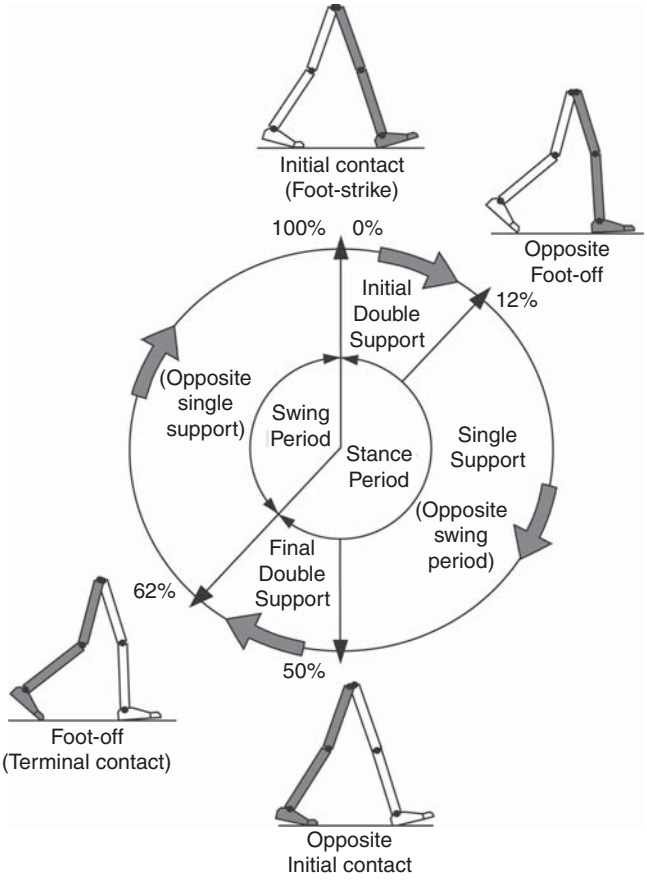

Figure 16.2 A typical gait cycle normalized in time, but wrapped around a continuous unit circle to illustrate symmetric phase relationships of temporal events and time periods. The beginning and end of the cycle occur at the 12 o'clock position. The temporal events of initial contact and foot-off for each leg are typically opposite each other on the unit circle, and the single support period of one limb is equal to the swing period of the opposite limb.

that the resulting time periods must be of equal duration for left and right single support, initial and final double support, and left and right swing periods. Any disruption in the natural sequence of temporal events anywhere along the cycle as a result of physical impairment, weakness, or spasticity will result in incorrect timing for the events that follow. This necessarily leads to a loss of symmetry that can be quantified by comparing the timing of temporal events between sides. Changes in symmetry reflected in the gait period durations is an index of gait pathology, and measuring this simple quantity can be quite useful for evaluating treatment performance over time.

Since the duration of the swing period and leg length determine the distance covered by the swinging limb, deviation from normal symmetry and timing will give rise to differences in step length on each side, and subsequently total distance traveled per gait cycle. By definition, step length and stride length are not synonymous. Step length is the distance (in the direction of progression) from a point of ground contact of the trailing foot to the next occurrence of the same point of ground contact with the leading foot. It is measured during initial double support and named for the leading limb. In contrast, stride length is the distance from initial contact of one foot to the next initial contact of the same foot, corresponds directly to the stride period, and is equivalent to the sum of successive left and right step lengths. Recognizing that speed is defined as the ratio of distance per unit time, step length, stride length, cadence (steps per minute), and walking speed are mathematically related by simple formulae:

$$\text{walking speed (m/s)} = (\text{cadence} \times \text{stride length})/120$$
$$\text{or}$$
$$\text{step length (m)} = (\text{walking speed} \times 60)/\text{cadence}$$

These basic outcome measures of overall gait performance, including the timing measures previously described and other quantities such a stance/swing ratio, are collectively known as *temporal-distance* or *temporal-spatial* parameters. They can provide considerable insight into the overall effect of subtle gait abnormalities on walking performance. For example, children with cerebral palsy may experience foot clearance problems during limb advancement due to excessive ankle plantar flexion or decreased knee flexion during swing period. Evidence of this could be found in prolonged single support times on the more normal or less involved side, and a reduced stance period, step length, and stance/swing ratio on the more involved side (6). If the source of the limb advancement problem can be attributed solely to the excess plantar flexion, the simplest intervention would be to prescribe a solid or leaf-spring ankle foot orthotic (AFO) with a rigid plantar flexion stop to restrict excess plantar flexion during swing. Evidence that this intervention improved gait performance could be found in more symmetric single limb support times and step lengths, a more normal stance/swing ratio, and a higher walking speed.

While clinical motion laboratories routinely compare a patient's temporal-spatial measures to age-matched normative values, caution should be used when interpreting these results. Temporal-spatial parameters of cadence and stride length are directly related to walking speed (7), and since humans routinely walk at a variety of speeds, simple deviations from reference values alone may *not* be indicative of gait pathology. Rather, reduced values for these measures may simply reflect the need to adopt a speed appropriate to the terrain, the required task, or the size of the room (8). A person's natural gait is also dependent on the environment, with studies showing

that subjects walk faster on a long walkway compared to a short one, and typically walk faster in outdoor studies compared to indoor studies (9). This lack of consensus regarding normal values supports the convention adopted by most clinical laboratories to compare patient results to their own laboratory-collected references, where these environmental factors can be consistent for all subjects. Nevertheless, while it is "normal" to walk at a variety of speeds, it clearly is abnormal to walk asymmetrically, so side-to-side differences in temporal/spatial measures within a particular patient should always be investigated.

When comparing temporal-spatial parameters in children, even greater care must be exercised, since several age-related differences arise from the close relationship of these measures to leg length and gait maturity (10). Sutherland has shown that in typically developing children, heel-first initial contact, sagittal plane knee flexion wave, reciprocal arm swing, and an adult joint angle pattern are acquired prior to the development of mature temporal-spatial parameters (11). All of these adult gait characteristics arise before the age of 3 years in most children (6). Because of this, Sutherland believes that gait maturity is best judged by the following five features, which he calls "determinants of mature gait (11)." These are: duration of single support, walking speed, cadence, step length, and ratio of pelvic span to ankle spread (P/A ratio). Notice that in addition to the first four measures that are fundamental temporal-spatial parameters, an anthropometric measure (P/A ratio) has been added, mainly to address the increased hip adduction common in the immature child's gait. In general, walking speed, step length, single support, and P/A ratio increase linearly with advancing age, with the greatest changes occurring during the first four years of life (6). Cadence decreases significantly between the ages of 1 and 2 years, after which it gradually continues to decrease (10). By age 4, the interrelationship between temporal/distance measures is fixed, although stride length and walking speed continue to increase with increasing leg length. Muscle phasic alterations in the early walkers are generally characterized by prolonged activation periods and subsequent longer periods of agonist/antagonist cocontraction around the joints of the lower extremities (12), most likely caused by neurologic immaturity associated with incomplete myeliniation (6). Despite all these age-related differences, the fundamental elements of a repetitive gait cycle are in place at a very early age. For this reason, asymmetric temporal/spatial measures can be used as indicators of gait pathology in both children and adults.

Because normal gait should be cyclical and symmetric, the existence of even small amounts of step-to-step variability may be an indication of gait pathology. Gait is most variable in the toddler, but gradually stabilizes as the child reaches adolescence (9). Hausdorff and colleagues have shown that the coefficient of variation for stride time in typically developing 3–4-year-olds is approximately 6%, but decreases to 2% in 11–14-year-olds (13). In the elderly, increased variability is associated with increased risk of falling, with speed variability the single best predictor of falls (9). These examples provide further evidence of the importance of a cyclical and symmetric gait pattern and how variations in symmetry and cycle times reflected in the temporal-spatial parameters of gait may be associated with gait pathology.

TYPICAL COMPONENTS OF AN INSTRUMENTED GAIT ANALYSIS

The phrase instrumented gait analysis (IGA) is often used to describe the application of computerized measurement technology to clinical gait analysis for the purpose of enhancing the interpretive power of the analysis beyond what can be discerned using observational and physical examination methods alone. The specialized nature of the systems used to perform an IGA typically requires a dedicated motion laboratory with specialists from clinical and technical disciplines to guide the patient through the testing procedures, make the required physical and anthropometric measurements, and record and process all data (Fig. 16.3). Analyses typically require 2 hours of patient contact time and between 8 and 12 hours of processing and analysis time, depending on the complexity of the patient referral and the number of measurements required to answer the clinical question. It is not within the scope of this discussion to comprehensively describe the full set of measurement tools available for clinical gait analysis

Figure 16.3 A motion laboratory clinical specialist works to place reflective markers on a subject while the technical staff prepares to record data for processing.

in children. For this, the reader is referred to several excellent descriptions that are widely available (5,14,15,16,17,18,19,20). However, since it is important for the discussions that follow, we will briefly introduce the primary measures used, some tips for their practical application, and give examples of typical recordings as a reference.

In addition to the temporal-spatial parameters described in the last section, the primary measurements comprising IGA are gait kinematics, kinetics, and dynamic electromyography (16). While there are certainly additional areas of measurement and many useful instruments that can be included in a comprehensive IGA, these three measurement categories are commonly accepted as the minimum necessary for clinical evaluation of the patient with gait dysfunction, and have been identified by the Commission for Motion Laboratory Accreditation (CMLA) as required for laboratory accreditation (21).

Gait kinematics is a general term that refers to measurement of the linear and angular displacements, velocities, and accelerations of body segments throughout the gait cycle. Generally expressed in terms of the joint angles between each limb segment, these quantities are most often described three-dimensionally using anatomical planes relative to the more proximal segment, but also includes the global position of the pelvis (pelvic tilt, obliquity, and rotation) and foot (foot progression angle) relative to a fixed laboratory coordinate system located in the middle of the walkway. Modern kinematic analysis systems use an assortment of markers or targets that are attached to the subject at strategic locations and can be tracked by specialized cameras or electromagnetic detectors (Fig. 16.4). The kinematic measurement system identifies the position of the targets from multiple perspectives in three-dimensional space using a high sampling rate (≥100 Hz) as the subject walks through a calibrated measurement volume. This determines a unique trajectory for each target, which can then be reconstructed by the computer utilizing a kinematic link-segment model to produce a three-dimensional animation of the walking subject within the virtual environment of the computer display (Fig. 16.5). From this mathematical representation of the subject, kinematic graphs and interactive reports can be produced to facilitate the clinical analysis of the child's gait pattern.

Kinematic measurement systems rely heavily on motion-capture technology and specialized software that fortunately have found a major market in the video game and motion picture industry. This has had the positive effect of substantially lowering the startup cost of these systems in recent years, making the technology more available to the clinical community and improving the accuracy, precision, camera resolution, and processing speed. These advances have also increased the complexity of the kinematic

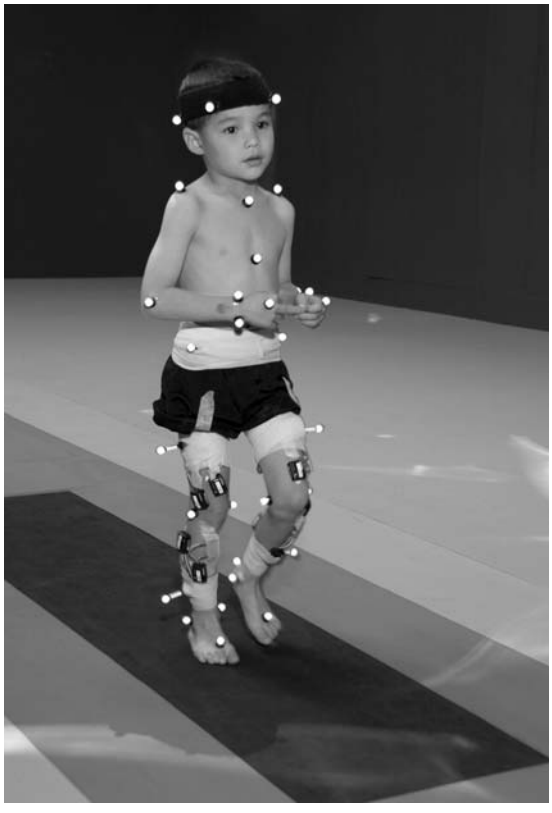

Figure 16.4 Subject with reflective markers or targets placed at strategic anatomic locations walks through a modern motion analysis laboratory. The location of the targets depends on the mathematical requirements of the limb-segment model used to calculate the kinematic values needed for analysis. This subject is using a full body model based on the modified Helen Hayes marker set.

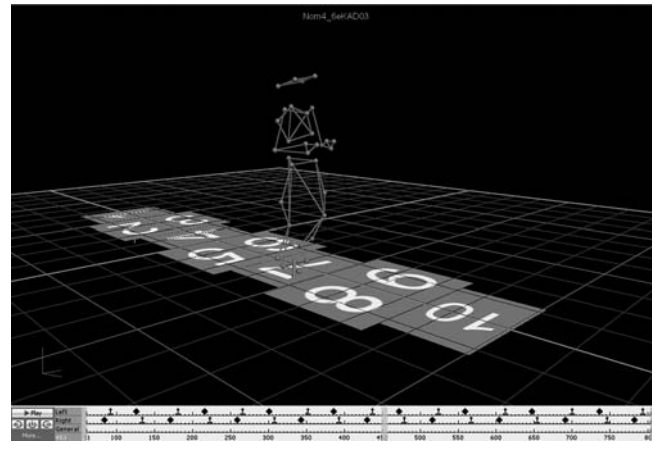

Figure 16.5 Three-dimensional animation of the walking subject within the virtual environment of a computer display.

models that can be implemented, which offers the promise of more comprehensive and anatomically correct descriptions of motion. However, it may also introduce new challenges since increased model complexity necessitates greater software complexity. Furthermore, the requirement for model validation with each new software release necessitates regular laboratory procedural changes, and may introduce data discrepancies when patient results are compared over time using different models. Recognizing these potential technical concerns, gait kinematics represent an integral component of clinical movement analysis and are essential for analyzing the child with gait dysfunction. Figure 16.6 shows a set of three-dimensional kinematic graphs associated with a sample of typically developing 12–13-year-old subjects used as a normal reference in our laboratory. We will discuss these kinematic graphs in more detail when discussing critical events in a later section.

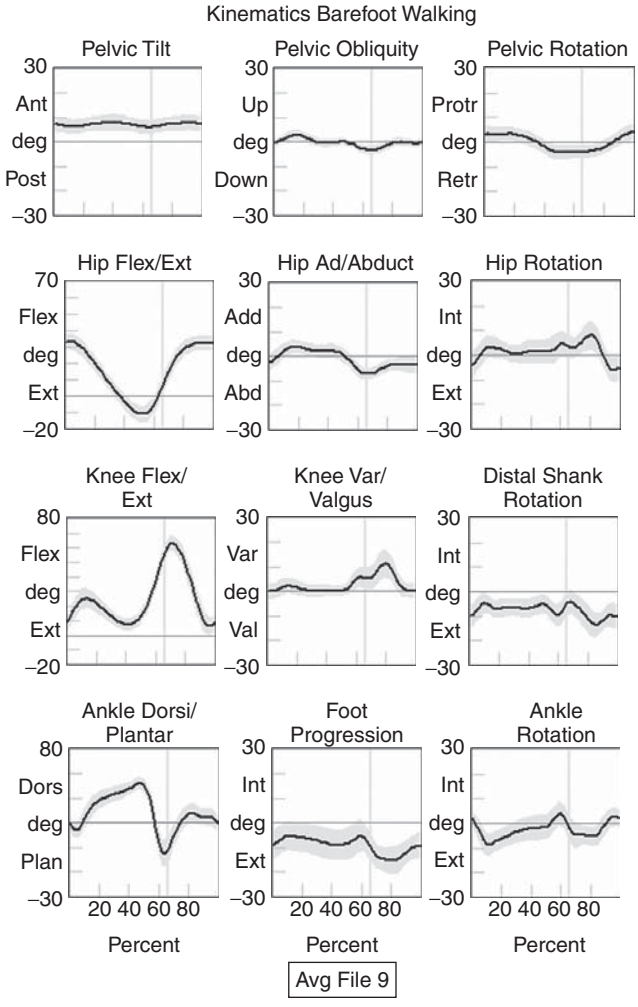

Figure 16.6 Normal three-dimensional kinematic graphs constructed using a sample of typically developing 12- to 13-year-old subjects. These data are used as a reference for comparing kinematic data from clinical subjects. The dark line is the average of all subjects and the gray band represents +/–1 standard deviation.

While measurements of gait kinematics provide a quantitative description of body segment and joint movement during walking, gait kinetics focus on describing the forces that cause these movements and the calculated quantities that arise when forces and three-dimensional kinematics are combined into a mathematical model of the body. Since joint and muscle forces cannot be measured directly from the walking subject, the forces due to foot/floor contact are measured using a specialized instrument known as a *force platform* embedded in the walkway. The force platform measures the vertical, fore-aft shear, and medial-lateral shear components of the *ground reaction force* (GRF), which is the force vector acting at the supporting surface that is equal and opposite to the sum of all muscular, gravitational, and inertial forces generated by the body in motion. Since a force platform measures the magnitude and direction of the GRF as a single resultant vector quantity, only one foot can be in contact with the platform at a time for a valid measurement. In order to measure multiple foot strikes from both feet, the subject either needs to walk multiple times across a single platform or the laboratory needs to include a *force platform array* with multiple platforms in different orientations so several clean foot strikes from both sides can be recorded in as few a number of passes as possible. A larger force platform array reduces alterations of gait characteristics in children with neuromuscular diseases in several ways. Installing multiple force platforms into the walkway reduces the number of trials required and thus minimizes the risk of fatigue. Furthermore, having multiple force platforms lessens the possibility of "targeting," which will alter the subject's characteristic gait pattern. Figure 16.7 shows the large 10-platform array of 60 cm × 40 cm force platforms currently used in our laboratory, and illustrates how rotating the long axis of each platform sequentially 90 degrees can accommodate a wide variety of stride lengths and step patterns for children and adults.

The direct measurement of the individual force components and the vector sum of the GRFs has historically been used to evaluate gait kinetics and facilitate a more qualitative pre-/postsurgical comparison. The most useful clinical application of gait kinetics, however, is when it is combined with GRF measurement and a kinetic model of the lower extremities to calculate joint kinetics, specifically joint moments and powers (22). The most common way to accomplish this is to apply an "inverse dynamics" model of the lower extremity using the anthropometric dimensions of each segment (typically seven segments, including the pelvis and both thighs, shanks, and feet) and estimates of each segment's center of mass and inertial

Figure 16.7 Large 10-platform array of 60 cm by 40 cm force platforms used in The Center for Gait and Movement Analysis at The Children's Hospital in Aurora, Colorado. The "hopscotch" pattern of the platform array permits the recording of several individual foot-strikes from both feet in a single walking pass. For illustration purposes each platform is shown without its protective floor covering, which caused the platforms to blend into the surrounding walkway when applied.

quantities. The forces at each joint can then be solved sequentially, starting from the GRF at the floor and working proximally, using the linear and angular forms of Newton's 2nd Law:

Linear: force = mass × acceleration ($F = ma$)
Angular: joint moment = moment of inertia × angular acceleration ($M = I\alpha$)

By convention, joint moments can be considered either *external* or *internal*. External moments reflect the forces acting on the body through the skeleton that arise from the GRF, and since they reflect an external biomechanical load, are sometimes called *demand* moments. Internal moments describe the force generated by the muscles and ligaments acting on the skeleton to balance the external moments, and because they are counteracting an external load, are sometimes called *response* moments. Aside from their different functional descriptions, external and internal moments for the same joint are of equal magnitude and differ only in their mathematical sign. The joint moments described in a typical IGA report are internal moments, but this should always be confirmed since the sign and direction of the curves will be reversed if they actually describe external moments. Joint moments are vector quantities that describe the net torque around each joint but do not provide the individual force contribution from each agonist/antagonist pair or from individual muscles. Nevertheless, the net moment around the joint is quite useful because the magnitude and sign of the curve at any instance in the cycle can illustrate if one half of the agonist or antagonist pair is dominating at a specific point in the gait cycle. Net moment values can aid in clinical interpretation of gait by comparing them to reference values for typically developing children and by observing changes in the values before and after treatment. In addition, net moment values are helpful in understanding how a child may be compensating at a given joint for weakness or limited range of motion at an adjacent joint. Figure 16.8 shows the sagittal plane kinematics, sagittal plane joint moments, and total joint power for the hip, knee, and ankle from a sample of typically developing 12–13-year-old subjects that we use as a normal reference.

Once the three-dimensional moments at each joint have been calculated, joint power at any time in the gait cycle is the product of the joint moment and the corresponding angular velocity (instantaneous slope of the joint angular displacement curve from kinematics) at each percent interval of the gait cycle:

joint power = joint moment × joint angular velocity
$(P(t) = M(t) \bullet \omega(t))$

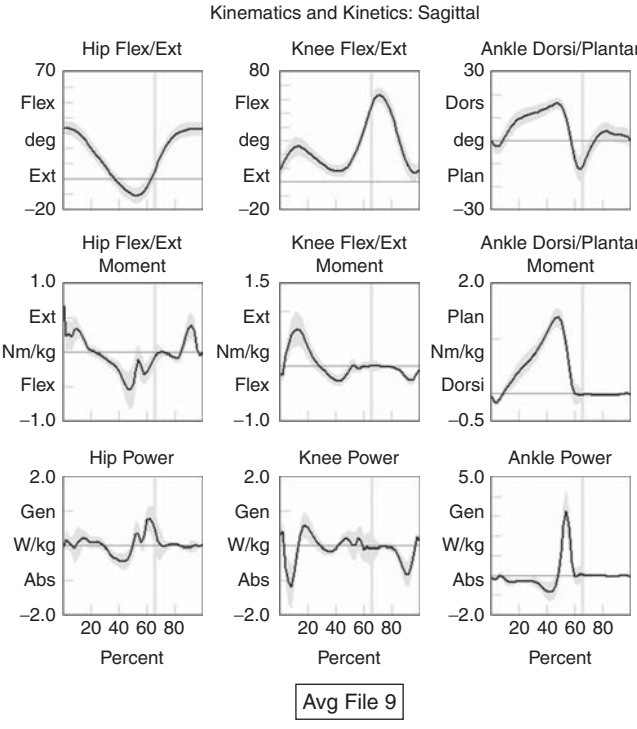

Figure 16.8 Graphs of sagittal plane kinematics, sagittal plane joint moments, and total joint power for the hip, knee and ankle constructed using a sample of typically developing 12- to 13-year-old subjects. These data are used as a reference for comparing kinetic data from clinical subjects. The dark line is the average of all subjects and the gray band represents ± standard deviation.

Just as with joint moments, joint power reflects the net power at a joint and not the individual power generated by a particular muscle or agonist/antagonist pair. However, unlike kinematics and joint moments that simply quantify the motion at a particular instant (kinematics) or calculate an estimate of the force dominating the joint related to muscle function (joint moments), joint power provides insight into the biomechanical mechanisms responsible for specific movements and, in a sense, quantifies the actual "motors" driving a particular gait pattern. In this way, joint power curves are extremely useful to identify when a particular joint is generating power (positive indicates concentric contraction) or absorbing power (negative indicates eccentric contraction) to analyze the transfer of power or energy from one joint to another and for understanding how one joint can compensate for disability at an adjacent joint. It should be pointed out here that although joint power is perhaps the single most informative biomechanical variable that can be obtained from an IGA, it does have limitations (23). For one thing, power is technically a single *scalar* quantity describing all planes of a joint combined, unlike displacement, velocity, and joint moment, which are directional vector quantities with individual component values for each anatomical plane. While in most cases the greatest contribution can be assumed to arise from the sagittal plane, the lack of a true directional component (especially at the hip) may lead to incomplete clinical interpretations. Another issue is that since extensive use of mathematical modeling is required to arrive at the joint power values, there are numerous assumptions made in the process and great opportunity for errors or artifacts to influence the final curves. These issues should be considered when utilizing any kinetic variable for clinical decision making. However, they should not hinder the use of this information since these estimates cannot be obtained in vivo by any other means and still provide considerable insight into the functional causes of gait abnormalities.

Electromyography is an important tool for evaluation of muscle and neurologic function and is well understood by the pediatric physiatrist. When used in the context of IGA, the purpose is slightly different from the conventional application. The primary objective of EMG in clinical gait analysis is to identify periods of muscle activation during walking so that decisions can be made regarding the appropriateness of muscle timing for agonists pairs as they selectively activate and deactivate during the gait cycle. This is the reason that we refer to this as *dynamic electromyography* or d-EMG, since the focus is on the phasic response of muscle during walking or some other functional activity. Since the subject won't be in a stationary position for the test, the instruments and technical procedures are also different from conventional diagnostic EMG. Dynamic electromyography requires a bipolar arrangement of electrodes and miniature differential amplifiers with high common mode rejection ratio (CMRR) placed close to the site of the recording to ensure the EMG signal isn't overwhelmed by motion artifact while the subject moves (Fig. 16.9). Differential amplifiers with high CMRR (typically greater than 100) amplify voltage differences between the inputs and reject common voltages that may arise from movement of the electrodes or the soft tissue vibration that occurs with foot contact. Surface electrodes are the most commonly used electrode type for recording d-EMG from the pediatric patient to avoid the emotional trauma and change in gait pattern that indwelling electrodes often cause. Typically, the active portion of each electrode in the bipolar pair should be small and the pair should be placed as close together as possible along the long axis of the muscle ($\leq$1 cm diameter, $\leq$2 cm separation) to minimize the effect of crosstalk from surrounding muscles. Unfortunately, surface electrodes are only suitable for recording muscle groups that are directly subcutaneous; if there is a need to evaluate deeper muscles individually, fine-wire electrodes made of a bipolar pair of 50-micron platinum wire must be inserted directly into the muscle of interest using a 25–28-gauge needle. When required, this is the most invasive aspect of an IGA, and should be used only when necessary in the pediatric patient and after all other data have been collected, since the level of patient cooperation and the likelihood of a typical gait pattern decrease considerably after a needle stick. In practice, most of the muscles of interest to the pediatric physiatrist can be successfully

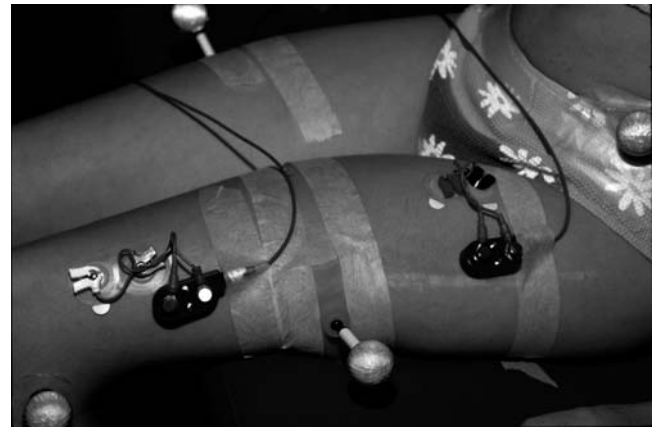

Figure 16.9 Patient with bipolar surface electrodes and small instrumentation amplifiers for recording dynamic EMG while the subject walks. This illustrates the electrode placement for the left vastus lateralis (distal location) and left rectus femoris (proximal location). Each electrode is connected to an instrumented backpack and then hardwired to the recording instruments. A wireless EMG recording system using similar electrodes but with individual transmitters for each muscle is shown in Figure 16.4.

recorded using the surface electrode approach if proper procedures to minimize crosstalk and reduce motion artifact are followed.

Before the EMG recording can be used for clinical interpretation, the raw data must be filtered, processed, and time normalized so periods of muscle activation during the gait cycle can be identified. A good reference for processing guidelines is available from the International Society of Electrophysiology and Kinesiology, where they state that surface electrode recordings should be bandpass-filtered between 10 Hz–350 Hz and fine-wire recordings filtered between 10 Hz–450 Hz. This maximizes the signal, minimizes the noise, and reduces motion artifact. In modern systems, the filtered EMG data are sampled by analog-to-digital converters, and further processing is performed by computer using specialized software or in concert with the motion-capture system. Data can be presented as a continuous recording of "raw" EMG, an ensemble average of several cycles of EMG normalized to the gait cycle, or as linear envelopes reflecting the EMG magnitude throughout the gait cycle after rectification and integration of the raw EMG signal. In our laboratory, we also have developed a system to superimpose the EMG signal over the observational video recording of the walking subject to screen for faulty EMG recording during the analysis and to better understand the interaction between observed movement and muscle activation (see Fig. 16.10, right side). Regardless of how these data are presented, the goal is to use the EMG recording to identify periods of abnormal muscle activity and determine if this activity is responsible for abnormal movement patterns presented by the patient. Typically, the patient's activity is compared to a normal EMG reference, and deviations from normal are scrutinized for their contribution to the overall movement pattern. Figure 16.11 shows filtered and time-normalized EMG for 12 muscles of the lower extremity from a 15-year-old typically developing subject used as a laboratory reference, along with published normal EMG activations represented as solid black bars at the bottom of each graph. The high-magnitude sections of the EMG recording for each muscle correspond to the published normal values, confirming that this typically developing subject has a normal adult activation pattern.

When EMG recordings are combined with the kinematics, kinetics, temporal-spatial parameters, radiographs, and the physical examination results, a comprehensive snapshot of the subject's walking pattern is revealed, providing an empirical basis for identifying the functional cause of a gait abnormality. To use these data successfully, however, we must return to the normal gait cycle to understand the functional requirements for walking, since these requirements are a natural consequence of subdividing the cycle on the basis of function.

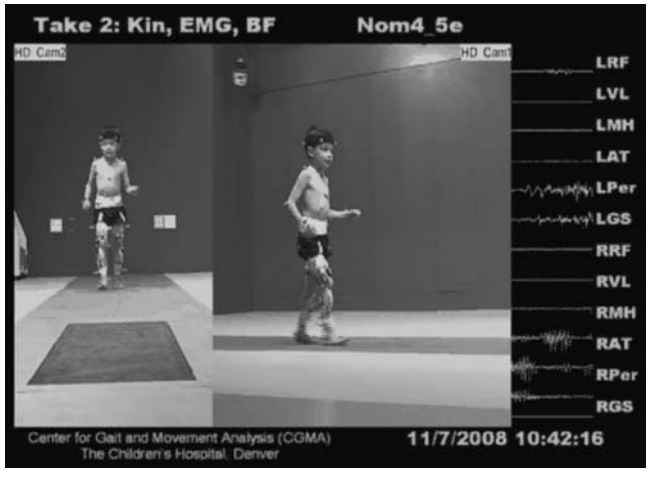

Figure 16.10 Biplane high-definition video recording with superimposed real-time EMG traces of six muscles bilaterally from a typically developing subject used as a reference at CGMA. The raw, unfiltered EMG recording provides immediate feedback on the quality of the EMG signal and the synchronization of muscle activity with observed movement.

IMPAIRMENT IDENTIFICATION IS FACILITATED BY SUBDIVIDING THE GAIT CYCLE

While a repetitive gait cycle arises from the alternating base of support found in all bipeds, the existence of this cycle provides great opportunity for clinical and biomechanical analysis of a child with gait dysfunction. In particular, a repetitive cycle lends itself to natural subdivision, which in turn, leads to a sequence of events that must be performed in order and with the correct timing for efficient walking to occur. The earlier section titled "Normal Gait Is Cyclical and Symmetric" discussed *temporal* subdivisions of the gait cycle delineated by foot/floor contact and their use in comparing limb symmetry, measuring outcomes, and the general characterization of overall gait performance. The focus of the current section is to describe another type of gait cycle deconstruction, one based on *functional* subdivisions. For this approach, the functional prerequisites of walking are identified, and this provides a framework for subdividing the gait cycle into functional divisions (24). It is then possible to use the measurements available from IGA to identify quantitative differences at each joint and the specific functional abnormalities that occur at critical moments in the gait cycle (25,26).

Functional Prerequisites for Walking

In their landmark paper published in 1953, Saunders, Inman, and Eberhart (27) described six gait subdivisions that they referred to as the "determinants" of normal

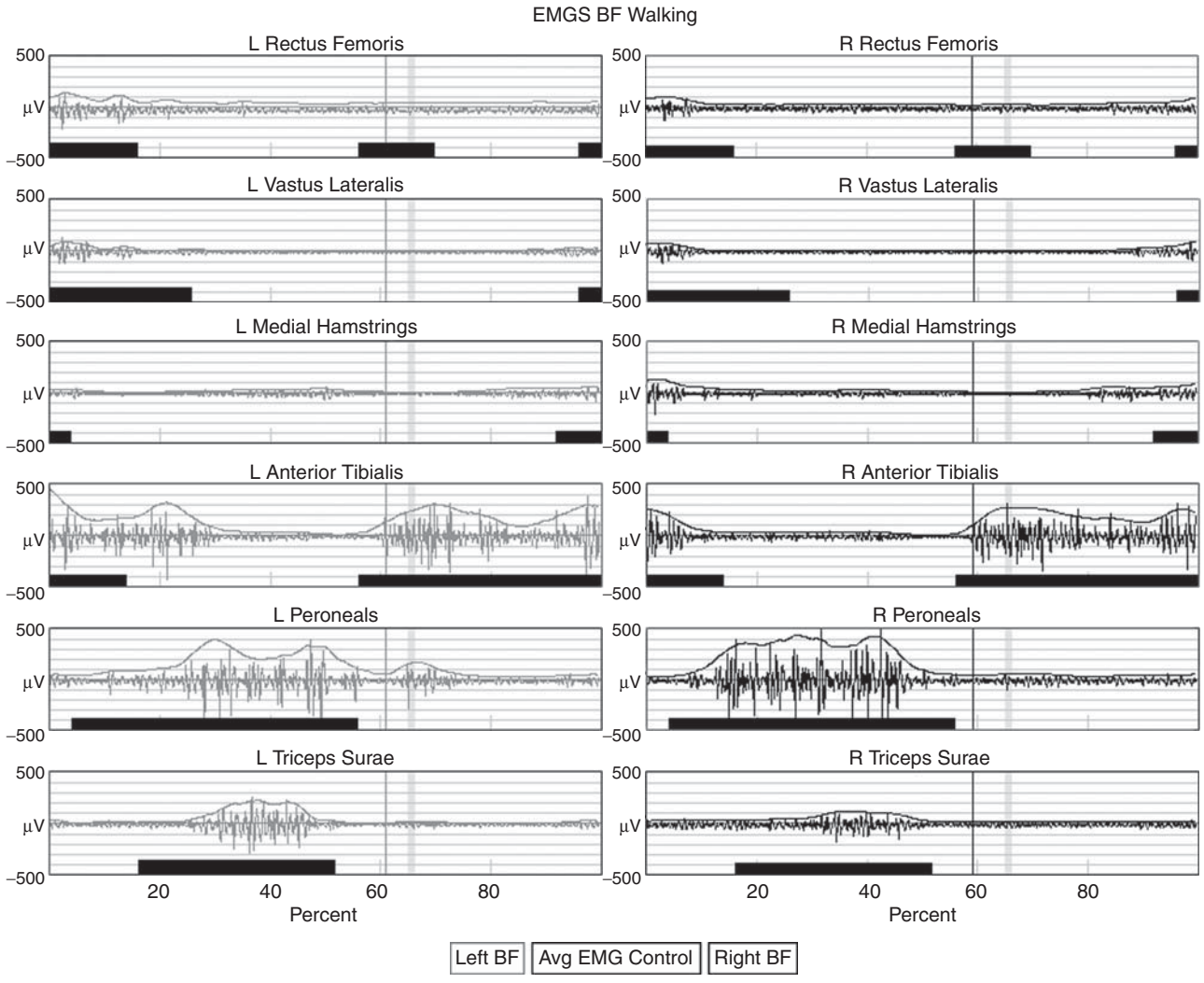

Figure 16.11 Filtered and time normalized EMG for 12 muscles of the lower extremity from a typically developing 15-year-old subject. The black bars at the bottom of each graph are constructed from published normal EMG activations and are used as reference values. The smooth curve above the EMG activation is a processed EMG signal obtained by rectifying and integrating the raw EMG. Notice that each EMG activation pattern above a baseline level is contained within the reference bar indicating a normal EMG pattern. In this typically developing subject, there is very little EMG activity from the rectus femoris during initial swing.

gait. This treatise was significant in that it was perhaps the first formalized delineation of the gait cycle that explained how coordinated movements of the hip, knee, and ankle at specific points in the cycle led to efficient forward progression. Each determinant's influence on the three-dimensional path of the whole body center of mass (COM) was described using simple theoretical models, and the cumulative effect of all six determinants led to a smooth, low-amplitude trajectory that was assumed to be consistent with optimal, efficient locomotion. Specifically, Inman and colleagues believed that minimizing vertical and horizontal motion of the COM would maximize walking efficiency, since unnecessarily raising and lowering the COM would be wasteful from a potential energy perspective. By changing functional limb length with the addition of joints to an initially jointless model of the lower extremities and pelvis, each determinant served to smooth different portions of the COM trajectory, effectively raising the COM during double support and lowering it during single support. While it is true that unnecessarily large and abrupt movements of the COM reduce gait efficiency, some of the specific determinants identified by Inman and colleagues have now been discredited (28,29,30). The improved accuracy and temporal resolution of kinematic measurement instruments over the last 50 years have uncovered problems with the timing of some of the theoretical mechanisms described in the original paper, and in the case of longer step lengths, larger COM displacements are not necessarily associated with decreased gait efficiency.

While the relevance of specific determinants may now be in dispute, the real impact of this work is that it inspired generations of investigators to consider

biomechanical explanations for gait dysfunction and led a few students of Dr. Inman's to develop clinically applicable gait cycle decompositions derived from the functional requirements of walking. In a later monograph, Inman described two basic functional requisites of walking that he deemed necessary for any form of bipedal gait, no matter how distorted by physical disability or assisted by prosthetic or orthotic devices (3): continuing ground reaction forces that support the body and periodic forward movement of each foot from one position of support to the next.

These essential features of walking give rise to a periodic gait cycle that must always be present for continued locomotion. An orthopedic resident of Dr. Inman's, Jacquelin Perry, recognized that the physical demands of supporting the body against gravity varied, depending on whether the stance limb was accepting the initial impact or continuing to carry the weight of the body during single support. To address this, she developed the notion of three functional gait *tasks* (31): weight acceptance, single limb support, and swing limb advancement. Dr. Perry considers weight acceptance the most demanding of the three functional gait tasks since it requires the stance limb's musculature and bony and ligamentous structure to provide shock absorption, initial limb stability (stiffness), and maintenance of forward progression. Preparation for the demands of weight acceptance begin late in swing period, when prepositioning of the leading limb occurs to correctly align the foot to accept weight at initial contact. The physical demands are lower for the task of single limb support, despite the fact that one leg alone has the complete responsibility for supporting body weight, maintaining whole-body stability (balance), and restraining forward momentum. This reduced physical demand during the task of single limb support is due to the inherent passive stability provided by the knee ligamentous structure and the force balance at the hip as body weight moves forward (31). An essential functional requirement for this task is strong eccentric contraction of the calf musculature to control the tibia (and subsequently the rest of the stance limb) as it rotates over the fixed base of support provided by the foot. When the task of single limb support ends and swing limb advancement begins, the physical demands increase once again, since the three goals of weight transfer, limb advancement, and foot clearance must all be accomplished. Similar to the weight acceptance task, important preparatory actions must begin before the swinging limb is lifted from the supporting surface at the end of stance period to meet the functional demands of swing limb advancement.

Findings from other investigators support the existence of these three fundamental gait tasks, although each investigator has used a somewhat different terminology when describing them (Table 16.1). Winter has characterized walking as an extremely complex motor control task that requires three elements: support control to prevent collapse against gravity (32); balance control of the head, arms, and trunk (HAT) acting as an inverted pendulum (33); and safe and coordinated lower limb movement during swing for minimum foot clearance and gentle heel contact (19). Dr. Winter and colleagues have also stated that the goals of these tasks

16.1 Functional Subdivisions of the Gait Cycle Attributed to Different Investigators

Investigator	Inman	Perry	Winter	Gage
Subdivision nomenclature	Requisites	Tasks	Motor Control Tasks	Prerequisites
Functional subdivisions	Continuing ground reaction forces that support the body	Weight acceptance	Support control to prevent collapse against gravity	Stability of the weight bearing foot throughout stance
		Single limb support	Balance control of the HAT	
	Periodic forward movement of each foot from one position of support to the next	Swing limb advancement	Safe and coordinated limb movement during swing to achieve ■ Minimum foot clearance ■ Gentle heel contact	Clearance of the non–weight-bearing foot during swing Appropriate prepositioning of the swinging foot in preparation for initial contact Adequate step length Energy conservation

HAT, head, arms, and trunk.
Source: Refs. 3, 19, 31–33, 35.

can still be accomplished after disease, injury, or loss of function because of the inherent redundancy of lower extremity musculature and rapid adaptability of the central nervous system (34). It is interesting that Perry and Winter have identified essentially the same three gait tasks, despite approaching the study of gait from two different perspectives: clinical analysis of pathologic gait and biomechanics of human movement, respectively. This lends support to the existence of these three elements and warrants using them to understand functional deficits in subjects with gait pathology.

Gage has expanded on this description by identifying five elements essential to walking that he has referred to as "priorities" or "prerequisites" of normal gait (35). This functional subdivision of the gait cycle encompasses the three tasks described previously, but adds swing period elements necessary to ensure appropriate weight acceptance and the global task of energy conservation. In the order of functional priority, these are stability of the weight bearing foot throughout stance, clearance of the non-weight bearing foot during swing, appropriate prepositioning of the swinging foot in preparation for initial contact, adequate step length, and energy conservation. This prioritization is influenced by Dr. Gage's interest in the gait of children with cerebral palsy, and includes a gait efficiency task (energy conservation) to address the reduced functional capacity or endurance of many individuals with pathologic gait. He also identifies several physiologic and biomechanical mechanisms common to normal gait that can improve energy conservation. These are eccentric muscle contraction, return of "stretch energy" from prestretched muscles immediately prior to concentric contraction, bi-articulate muscles functioning as energy transfer straps, and joint passive stability from the effects of ground reaction forces whenever possible to spare muscle activation (36). While other investigators have addressed the functional prerequisites of gait (15,25,37), the contributions described previously form the basis of the strategy described in this chapter.

Phases of the Gait Cycle

Since the tasks of weight acceptance, single limb support, and swing limb advancement can only be accomplished successfully if appropriate limb movement patterns occur sequentially and with correct timing, Dr. Perry developed a systematic method of subdividing the gait cycle to simplify pattern identification and facilitate observational gait analysis (24). Now known as the *Rancho* classification in honor of Rancho Los Amigos Medical Center where Dr. Perry and colleagues of the Pathokinesiology Service developed this method, it relies on eight subdivisions of the gait cycle, referred to as *phases* of gait. While in general, both phases and periods refer to specific time slices around the gait-cycle unit circle, Perry prefers to use the term "phase" for intervals that have specific functional significance and have a clear relationship to the three identified gait tasks described in the previous section. The Rancho classification provides a framework for functionally organizing the gait cycle harmoniously with the three fundamental gait tasks, and after 30 years of refinement, this approach has proven to be a powerful tool for identifying specific functional deficits or gait impairments during each phase of gait. Instrumented gait analysis can be used to quantify the magnitude of a functional deficit at a joint by reviewing the kinematic and kinetic data, or abnormal muscle timing by reviewing the EMG. This provides evidence and helps pinpoint the specific region or system most responsible for the overall gait abnormality, and suggests interventions to directly correct the identified functional deficit or gait impairment in each phase.

The eight phases described by Dr. Perry are identified in Table 16.2. Notice that all but the first phase (initial contact) represent separate time intervals between 0% and 100% of the gait cycle. Figure 16.12 illustrates the phases of gait in sequence around the unit circle, with stick figures signifying the temporal

16.2 The Eight Phases of Gait as Described by Dr. Jacquelin Perry

PHASE OF GAIT	DESCRIPTION
Initial contact	The moment when the foot strikes the ground
Loading response	Initial double support period when the limb is accepting weight.
Mid-stance	First phase of single support when the body advances over the stance limb ending ahead of the stance limb as weight is transferred to the forefoot
Terminal stance	Last phase of single support ending with opposite initial contact
Pre-swing	Final double support period when the knee rapidly flexes in preparation for swing and weight is shifted to the opposite limb
Initial swing	1st third of swing period where maximum knee flexion occurs
Mid-swing	Middle third of swing period where maximum hip flexion occurs, ending with a vertical tibia
Terminal swing	Last third of swing period where final knee extension achieves maximum step length and the limb is properly positioned for weight acceptance

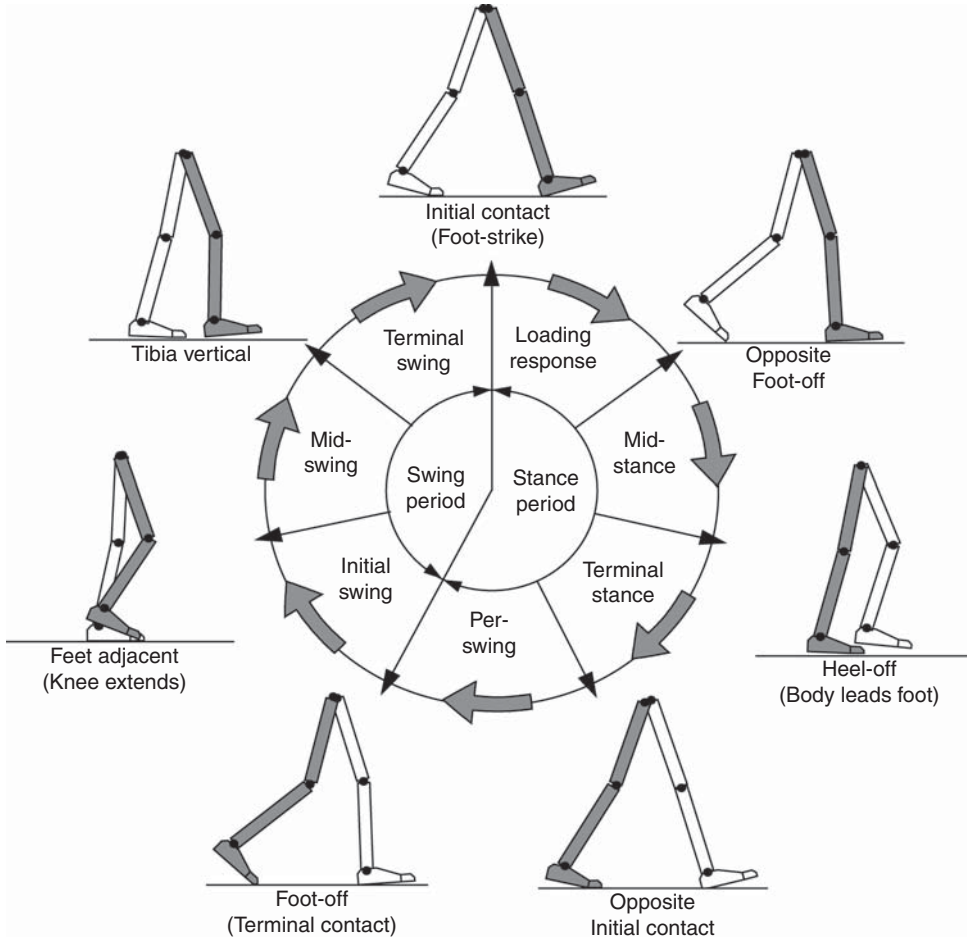

Figure 16.12 Typical gait cycle wrapped around a unit circle and subdivided into eight gait phases that have functional significance. The first phase is initial contact, and is equivalent to the temporal event by the same name. The phases are drawn in sequence and are equally spaced for clarity, but do not represent their usual duration. Refer to the text for a complete description of the phases and their functional significance.

event delineating the beginning and end of each phase. Phases are shown equally spaced in this figure for clarity; typically, each phase will not be of the same time duration. Initial contact is considered a phase of gait since it marks an important transitional point between swing limb advancement and the challenging task of weight acceptance, although unlike the other phases, it is a single instant in time. The other important transitional period (from stance to swing) occurs during final double support and is known as the phase of *pre-swing*. In terms of temporal events, final double support (and therefore pre-swing) is considered part of stance period because this interval ends with foot-off. However, in terms of gait phases, this interval also marks the first phase of the swing limb advancement task, highlighting the fact that from a functional standpoint, pre-swing has more to do with preparing the limb for moving forward than supporting the body during stance (24). Also notice that three new temporal events not associated with foot/floor contact have been introduced: heel-off, feet-adjacent, and tibia vertical. These terms have been used by Whittle (5) and others (4,38) and are useful to delineate the phases in normal gait; however, there is not yet a consensus among gait investigators if these are the undisputed event markers. For example, Perry acknowledges that a rising heel usually marks the beginning of terminal stance in normal subjects, but in patients with weak ankle plantar flexors, this heel-off may be delayed into pre-swing, which would technically eliminate the terminal stance phase. Dr. Perry prefers to identify the beginning of terminal stance as the point where the body moves ahead of the limb and weight is transferred onto the forefoot (39). Similarly, the event marking the transition between initial swing and mid-swing has been identified as the point where the feet are adjacent (5), when the swing limb is directly under the body (4,38), when swing limb acceleration changes to deceleration in normal gait (36), where the knee begins to extend and the foot clears the ground (39), and mid-swing (4). Fortunately, most investigators agree that the temporal

event marking the transition between mid-swing and terminal swing is the point where the tibia is directly vertical. These slight differences in the definition of the exact transition between phases are why some investigators report different phase durations for normal gait. This shouldn't be a concern, however, because determining the exact transition point between phases and comparing phase durations to normal are less important than recognizing that there are distinct phases in gait that can be identified and that certain functional accomplishments must occur in each phase.

CRITICAL EVENTS LINK GAIT IMPAIRMENTS TO POSSIBLE INTERVENTIONS

With the gait cycle now subdivided both temporally and functionally into discrete phases, all that is left is to identify specific joint positions or motions in each phase that directly contribute to the accomplishment of the three functional tasks of weight acceptance, single limb support, and swing limb advancement. Dr. Perry and her colleagues at Rancho Los Amigos Medical Center refer to these specific joint positions or motions as *critical events* (39). They have identified 13 critical events over the entire cycle, with one or more critical events in each of the 8 phases. Critical events occur at the foot, ankle, knee, or hip, and are largely focused on angular displacements in the sagittal plane. While there are other, more subtle motions occurring in all three anatomical planes, these 13 critical events are considered the most essential to producing a normal walking pattern, typically have the largest displacements, and are most easily observed from the walking subject, with or without the help of recording instruments. The significance of this approach is that once the critical events that are essential to producing a bipedal gait pattern are known, one can use the measures from instrumented gait analysis to determine functional reasons for why a particular critical event is absent, altered, or delayed. Interventions can then be focused on restoring critical events, leading to improved walking performance. The critical events for each phase of gait are shown in Table 16.3, including their relationship to stance and swing periods and each gait task. Notice that critical events and temporal events are quite different. As has been discussed throughout this chapter, temporal events are moments or instants in time used to delineate periods in the gait cycle, and critical events are important functional components that can be used to identify gait impairments.

16.3 Relationship Between Periods, Tasks, Phases, Temporal Events, and Critical Events During the Gait Cycle

	STANCE PERIOD				SWING PERIOD			
TASKS	**WEIGHT ACCEPTANCE**		**SINGLE LIMB SUPPORT**		**SWING LIMB ADVANCEMENT**			
Phases	Initial contact (0%)	Loading response (0%–12%)	Mid-stance (12%–30%)	Terminal stance (30%–50%)	Pre-swing (50%–62%)	Initial swing (62%–75%)	Mid-swing (75%-87%)	Terminal swing (87%–100%)
Temporal events	Initial contact	B: Initial contact E: Opposite foot-off	B: Opposite foot-off E: Heel-off (body leads foot)	B: Heel-off (body leads foot) E: Opposite initial contact	B: Opposite initial contact E: Foot-off	B: Foot-off E: Feet adjacent (knee extends)	B: Feet adjacent (knee extends) E: Tibia vertical	B: Tibia vertical E: Initial contact
Critical events	• Heel first initial contact	• Hip stability • Controlled knee flexion for shock absorption • Controlled ankle PF	• Controlled tibial advancement	• Controlled ankle DF with heel rise • Trailing limb posture	• Passive knee flexion to 40° • Rapid ankle PF	• Max knee flexion (>60°)	• Max hip flexion (25°) • DF to neutral	• Knee extension to neutral

B, beginning; DF, dorsiflexion; E, end; PF, plantarflexion.

Critical Events During the Weight Acceptance Task

The two phases associated with weight acceptance, initial contact and loading response, coincide with the period of initial double support, and include four critical events. The first critical event, a heel-first initial contact, must be present if forward momentum is to be preserved (24) and the energy from the falling body COM is to be redirected in the direction of progression (32). It is also necessary to prepare the new support limb for the demands of the next phase. The next three critical events of hip stability, controlled knee flexion for shock absorption, and controlled ankle plantar flexion, must occur during the loading response phase. Hip stability requires dynamic joint stiffness in the sagittal and frontal planes at the hip to prevent unnecessary forward pelvic tilt or increased pelvic obliquity, respectively. This places a high demand on the torque (moment) production ability of the hip extensors and hip abductors of the forward load-bearing limb. This demand is reflected in increased muscle activation (recorded using dynamic EMG) in the gluteus maximus, gluteus medius, and hamstrings. If a patient has weak hip extensors and fails to compensate for this weakness, there will be an increased anterior pelvic tilt and/or forward trunk lean directly associated with the inability of the hip extensors to meet the demand of this critical event. Alternatively, if the subject is successfully compensating for the weakness, they will exhibit a posterior trunk lean to position the whole body COM behind the hip joint center, thus reducing the torque production demand on the hip extensors. Evidence of either strategy is reflected in recordings of the hip sagittal plane angles (kinematics), the weight line (ground reaction force vector), dynamic EMG of the hip extensors, or by calculating the hip extensor moments and powers (kinetics). Similar strategies are used when there is weakness in the hip abductors, leading to either uncompensated or compensated Trendelenburg's gait patterns in the frontal plane. All can be related to the loss of the critical event of hip stability during the loading response phase.

Controlled knee flexion for shock absorption must occur to prevent unnecessary knee flexion during loading response, which wastes energy and places a higher demand on the quadriceps. Similar to the hip, quadriceps weakness can also be either compensated or uncompensated. Uncompensated quadriceps weakness will present as abnormal or increased knee joint angular displacements in the sagittal plane (increased knee flexion) with possible collapse at the knee. Compensated patterns will display body postures that shift the COM forward (forward trunk lean) so that the ground reaction force vector is closer to or in front of the knee joint, thereby reducing the quadriceps demand (quadriceps avoidance gait). Again, these patterns are reflected in kinematic, kinetic, and dynamic EMG measurements, within the context of meeting the needs of this critical event. Correctly controlled knee flexion during loading response is reflected in the slope and maximum knee flexion value during the first peak of the sagittal plane knee joint angular displacement curve, in the magnitude of the knee extensor moment and power absorption curve, and the EMG activity of different heads of the quadriceps.

The final critical event during loading response is controlled ankle plantar flexion. In this context, "controlled" is referring to the ability of the ankle dorsiflexors to eccentrically contract and lower the initially neutral foot carefully to the ground to provide a more stable base of support than can be provided by the calcaneus alone. This action is referred to as the heel or first rocker (40), the first of three important mechanisms that occur at the foot and ankle and facilitate progression of the entire stance limb (31). These three rockers are illustrated in Figure 16.13. If the pretibial muscles have sufficient strength to restrain the rate of foot drop, the action of the first rocker pulls the tibia forward, which in turn is transferred to the femur by the active quadriceps that is attempting to restrain the rate of knee flexion. This is how the energy of the falling body COM is redirected to provide forward progression, an important energy-conserving mechanism often lost when the heel rocker is absent. If the pretibial muscles did not have sufficient strength but the subject was able to achieve a heel-first initial contact, a noticeable foot-slap would occur as the unstable lever at the ankle allows the foot to plantar-flex uncontrollably. This is reflected in a steep descending initial slope on the sagittal plane ankle kinematic curve and absence of either a dorsiflexor moment or dorsiflexor power absorption on the corresponding sagittal plane kinetic curve (22). Experienced clinicians can also detect this event without all the modern conveniences by simply listening for the sound of the foot-slap! The effects of uncontrolled ankle plantar flexion at loading response are not as easy to detect when the subject fails to achieve a heel-first initial contact, as is the case with foot-flat, forefoot, or equinus initial contact. In this case, kinetic data are helpful, because as the point of foot/floor load bearing moves further in front of the ankle with progressively increasing plantar flexion at initial contact, there is a proportional increase in the magnitude of the incorrect plantar flexor moment during loading response. This plantar flexor moment (via the triceps surae) has opposite the desired effect on the knee, as occurs with eccentric contraction of the pretibial muscles when a true heel rocker is present; the knee extends when it should be flexing (41). This reduces the effectiveness of knee shock absorption and may eliminate it

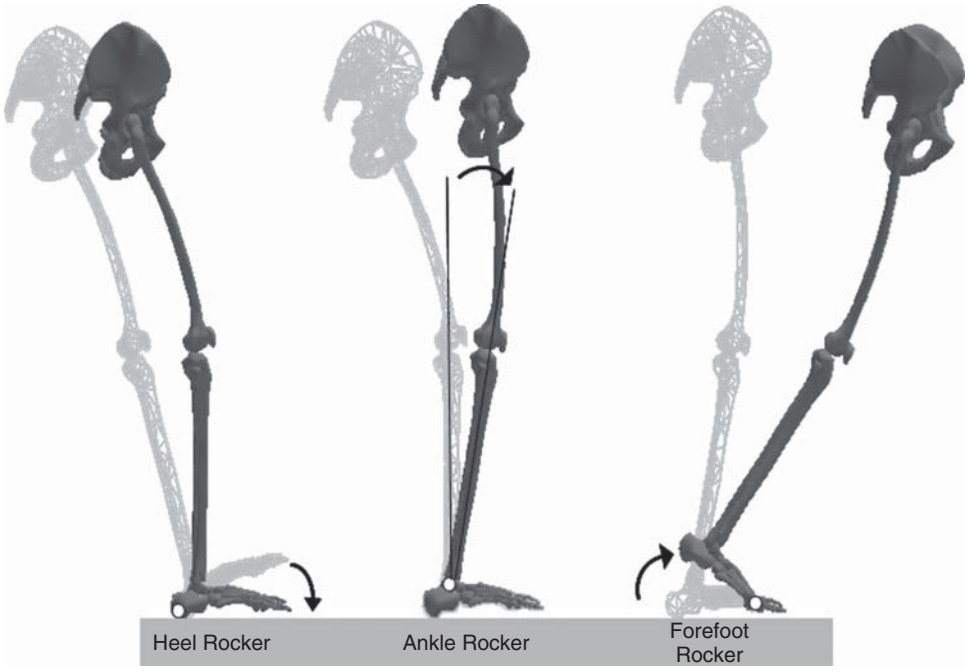

Figure 16.13 The three rockers representing normal ankle function in gait: These are the heel or first rocker, the ankle or second rocker, and the forefoot or third rocker. The lighter gray skeleton represents the beginning of each rocker, and the arrows signify the movement that is associated with each. Refer to the text for a full description of these important critical events.

completely, increasing bone-on-bone forces at the knee. The amount of shock-absorbing energy transferred to the hip and ankle is reflected in the hip, knee, and ankle powers during this phase, and is useful for describing the potential degree of impairment associated with incorrect ankle function at loading response. Note that the magnitude of the EMG activity of the pretibial and posterior compartment muscles by themselves do not explain the moments at the ankle, since major force contributors arise from the inertial and gravitational forces that occur during the first rocker. The EMG activity does help sort out if the pattern of motion is due to weakness (pretibial muscles with first rocker present, triceps surae without it), neglect (often seen in traumatic brain injury [TBI]), or poor motor control (seen in cerebral palsy or cerebrovascular accident [CVA]). This information can assist in determining whether a solid, leaf-spring, hinged, or floor-reaction AFO, Botox injections into the calf musculature, or tendon lengthening or transfer surgery would be the most appropriate intervention to use when the critical event of controlled ankle plantar flexion is abnormal or absent. Table 16.4 summarizes many of the gait measurements that are useful for identifying functional causes for absent or abnormal critical events during the weight acceptance task, and while not exhaustive, can help organize the array of measures available for assessing impairments during this task.

Critical Events During the Single Limb Support Task

As shown in Table 16.3, there are three critical events during the single limb support task. These are controlled tibial advancement during mid-stance, controlled ankle dorsiflexion with heel rise (heel-off) during terminal stance, and a trailing limb posture during terminal stance (39). During the two phases of this task (mid-stance and terminal stance), the responsibility of the stance limb is to simultaneously provide support against gravity without losing balance and contain the forward momentum built up by the contralateral swinging limb. Both of these objectives can be accomplished by controlling tibial advancement in the first half of single support and controlling ankle dorsiflexion in the second half. This will lead to the trailing limb posture (body COM forward of the base of support) necessary to permit a sufficient step length on the opposite side. If, at the end of loading response, the foot has achieved foot-flat, then during mid-stance, the ankle becomes the axis of rotation for the body's forward progression. This is referred to as the ankle or second rocker, and this mechanism continues until maximum dorsiflexion is achieved in terminal stance (Fig. 16.13). With the heel and forefoot firmly planted, the tibia can rotate over the talus smoothly under the selective control of the soleus, later assisted by both heads of the gastrocnemius, which simultaneously

16.4 Gait Measurements Useful for Identifying the Cause of an Absent or Abnormal Critical Event During the Weight Acceptance Task

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/DIST. MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Initial contact	Heel first initial contact: if absent, also consider mid-swing and terminal swing critical events	Strength • weak ankle DF ROM • tight hamstrings or triceps surae Neurologic triceps surae tone	• Reduced swing period, or reduced single support time on opposite side • Reduced step length	• Hip flexion > or < normal max of 30° • Knee flexion >4° • Ankle not at neutral	Refer to phases terminal swing or loading response	• Excessive hip extensor or hamstring activity • Reduced or absent ankle DF activity • Premature ankle PF activity
Loading response	Hip stability	Strength • weak hip extensors • weak hip abductors ROM • tight hip flexors, hip flexion contracture • femoral anteversion	• Prolonged initial double support time	• Hip flexion >30° • Increased pelvic tilt and/or obliquity • Increased hip internal rotation • Pelvic retraction on side of weakness	• Large hip extensor moment with possible initial power absorption	• Excessive hip flexor and hip adductor activity • Decreased hip extensor and abductor activity
	Controlled knee flexion for shock absorption	Strength • weak quadriceps ROM • tight hamstrings or knee flexion contracture	• Prolonged initial double support time	• Knee flexion < 4° or > 20° • Abnormal knee flexion wave	• Initial knee flexor moment with no phase reversal (quad avoidance) • Large knee extensor moment with excessive power absorption	• Excessive knee extensor activity • Increased co-contraction at the knee with prolonged knee flexor activity
	Controlled ankle plantarflexion (PF)	Strength • weak ankle DF ROM • tight triceps surae or reduced DF • tight hamstrings or knee flexion contracture	• Prolonged initial double support time • Reduced time to foot-flat, with possible foot-slap	• Tibia forward of vertical with ankle in DF • Abnormal 1st rocker (heel rocker) • Incorrect foot alignment with incorrect foot progression angle	• Large PF moment with high power absorption	• Reduced or absent ankle DF activity • Premature ankle PF activity • Premature tibialis posterior activity

DF, dorsiflexion; PF, plantarflexion; ROM, range of motion.

limits knee extension. The slow-twitch, fatigue-resistant muscle fibers of the soleus are usually well suited to the sustained eccentric contractions required to control tibial advancement. Weakness in the triceps surae, however, results in the tibia advancing too quickly, which prematurely allows the tibia to move past vertical and leads to sustained knee flexion during mid-stance, and premature or excessive dorsiflexion and lack of knee extension at terminal stance. In this circumstance, a rigid AFO or, in extreme cases of weakness, a floor-reaction AFO can effectively supplement the weak plantar flexors, restore a more normal plantar flexor moment, and control tibial advancement during mid-stance and dorsiflexion during terminal stance. Gage also suggests using a rear-entry, hinged, floor-reaction AFO in these circumstances (12), which permits ankle plantar flexion but resists dorsiflexion in mid-stance and terminal stance.

In normal adults and typically developing children, the forward progression of the body causes the origin of the ground reaction force vector (center of pressure or COP) to move forward to the metatarsal heads, causing the heel to rise at the beginning of terminal stance. Now the axis of rotation for the body's forward progression is the metatarsophalangeal (MTP) joint, giving rise to the forefoot or third rocker (31) (see Fig. 16.13). While the first two rockers were constraining forward progression using eccentric plantar flexor contractions, the forefoot rocker is an accelerating rocker, as evidenced by the large ankle plantar flexor moment and transition from power absorption to power generation (36). With the help of strong concentric contraction of the fast-twitch fibers of the gastrocnemius, the ankle is stabilized and continued dorsiflexion in terminal stance is halted. By the end of terminal stance, the ankle is plantar flexing in preparation for initial contact on the other side, which yields the trailing limb posture necessary for maximum step length. When there is plantar flexor weakness, the third rocker is ineffective, which fails to control continued dorsiflexion, allows the knee to prematurely drop into flexion, reduces trailing limb posture, and shortens the opposite side step length. All of these factors reduce overall walking performance. AFOs that store energy in the structure of the orthosis as the ankle dorsiflexes (rigid, leaf-spring, floor-reaction) can provide a plantar flexion assist as the foot is unweighted in early pre-swing, depending on the amount of stiffness and energy storage built into the custom orthotic. This assist can return some of the reduced plantar flexor moment that would occur without orthotic use, and evidence of this can be found in the plantar flexor moment curve comparing orthotic and barefoot conditions.

If, at the beginning of mid-stance, the foot has either not achieved or is past foot-flat (equinus, early heel-off, spring-foot), the normal ankle and forefoot rocker mechanisms may not be effective, and the three critical events of single limb support will not be achieved. In toe-toe gait (equinus) or jump knee gait (forefoot initial contact and excessive knee flexion at loading response, followed by rapid knee extension and ankle plantar flexion in mid stance), plantar flexors that are tight or have increased tone overly constrain forward tibial advancement in mid-stance and dorsiflexion in terminal stance, leading to excess knee extension and early heel-rise. While the mechanism is different from the case of weak plantar flexors, the end result is the same; reduced effectiveness of second and third rockers and inability to achieve the three critical events. In these cases, Botox injections into the triceps surae, tendoachilles lengthening, or intramuscular triceps surae lengthening (Strayer procedure) can be effective in restoring second and third rockers, depending on severity. Ankle plantar flexion moments and powers, and dynamic EMG recordings are quite useful in selecting which procedure is most appropriate (22). Another example is crouch gait deformity, where hip and knee contractures combined with weak or overlengthened plantar flexors lead to early heel-rise and premature forward advancement of the tibia in mid-stance, and premature and excessive dorsiflexion in terminal stance. In this case, the same impact on the second and third rockers described previously for weak plantar flexors will often occur. Dr. Gage has long been a proponent of performing single-event, multilevel (SEML) soft tissue and bony surgery for this deformity to restore the proper rocker mechanisms and, with the proper orthotics, the plantar flexion/knee extension couple that allows the patient to stand more erect and walk more effectively (12). Other centers have taken a more conservative approach of staging the procedures, which has the advantage of reducing the surgical impact at the time of the procedure, but may cause muscle imbalances at other joints, leading to additional surgeries down the road. In either case, or when nonsurgical interventions are warranted, the goal should be to restore the rocker mechanisms so that the three critical events of single limb support can be realized. Table 16.5 summarizes many of the gait measurements that are useful in identifying causes for absent or abnormal critical events during the single limb support task.

Critical Events During the Swing Limb Advancement Task

Swing limb advancement is the last task that must be accomplished to complete the gait cycle, and, as shown in Table 16.3, this task contains four phases and six critical events. There are two critical events in pre-swing: passive knee flexion to 40 degrees and

16.5 Gait Measurements Useful for Identifying the Cause of an Absent or Abnormal Critical Event During the Single Limb Support Task

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/DIST. MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Mid-stance	Controlled tibial advancement	Strength • weak ankle PF (advance too fast) ROM • with equinus, or excessive DF without equinus (advance too fast) • tight triceps surae or hamstrings (advance too slow) Neurologic • triceps surae tone	• With equinus (early heel-off) • excessive DF without equinus (delayed heel-off) • Reduced single support time • Reduced opposite step length	• Tibia forward of vertical with ankle in excessive DF • Abnormal 2nd rocker (ankle rocker)	• Abnormal slope (too flat) of the PF moment curve	• Prolonged ankle DF activity • Reduced or absent PF activity • Prolonged hip and knee extensor activity
Terminal stance	Controlled ankle dorsiflexion (DF) with heel-off	Strength • weak ankle PF (DF too fast) ROM • excessive DF or reduced PF (DF too fast) • tight or increased tone in ankle PF (slow DF, early heel-off)	• With equinus (early heel-off) • excessive DF without equinus (delayed heel-off) • Reduced single support time • Reduced opposite step length	• Abnormal 2nd rocker (ankle rocker) • Excessive DF • Premature knee flexion	• Abnormal slope (too flat) of the PF moment curve • Reduced or absent PF power absorption	• Prolonged ankle DF activity • Reduced or absent PF, inverter, and everter activity
	Trailing limb posture	Strength • weak ankle PF • weak opposite hip abductor ROM • excessive DF or reduced PF • tight hamstrings or tight hip flexors	• Reduced single support time • Reduced opposite step length	• Excessive hip flexion • Excessive knee flexion	• Prolonged hip ext. moment or delayed/absent hip flexor moment • Prolonged knee ext. moment or delayed/absent knee flexor moment	• Any activity in the hip or knee flexors, and hip or knee extensors (lack of passive stability)

DF, dorsiflexion; PF, plantarflexion; ROM, range of motion

rapid ankle plantar flexion. As previously discussed, pre-swing is an important transitional phase that, while still a component of stance period, is functionally more associated with preparing the trailing limb for the swing period to come. Achieving 40 degrees of knee flexion before the foot leaves the ground is essential. This is because once the foot is airborne, the leg acts as a compound pendulum, so further knee flexion is completely dependent on concentric contraction of the hip flexors, including the adductor longus, and the inertia of the lower leg and foot (36). At normal walking speeds, knee flexion during pre-swing requires no active muscle contractions around the knee (passive). It occurs by a complex mechanism that involves continuation of tibial advancement as the forefoot rocker continues from terminal stance; unloading of the limb as weight is transferred to the new stance limb; continued concentric contraction of the triceps surae, which produces rapid plantar flexion that propels the knee joint in front of the ground reaction force vector, and concentric contraction of the adductor longus to initially accelerate the thigh forward (31).

All of these actions push the ground reaction force vector so far behind the knee that it collapses in the absence of an equalizing knee extension moment produced by the quadriceps that normally are silent during pre-swing. So weakness in the plantarflexors, hip flexors, or adductor longus all have an adverse effect on achieving the necessary knee flexion. Since a trailing limb posture with hip extension to 10 degrees past neutral amplifies the effect of the third rocker to shift the tibia forward, hip flexion contracture can also reduce the ability of the knee to passively flex to 40 degrees, despite the fact that such a contracture often prevents the knee from fully extending at terminal stance. Furthermore, it is interesting that because of the hip extensor component of the biarticulate hamstrings, inappropriate activation or tightness of these open-chain knee flexors can inhibit passive knee flexion in pre-swing by resisting the hip flexors and adductor longus as they attempt to accelerate the thigh. Problems with this critical event can be identified from the sagittal plane knee kinematics, the hip moments and powers, and the hip knee and ankle dynamic EMG. Problems with rapid plantar flexion, which is necessary to produce sufficient knee flexion in pre-swing, are also evident from the ankle kinematics, kinetics, and dynamic EMG. Since the critical events in this phase are so dependent upon concentric contraction and power generation at the hip and ankle, interventions to replace hip flexor and ankle plantar flexor strength are somewhat limited to AFOs that can return plantar flexion moment in pre-swing or enhance the third rocker, or stretching, lengthening, or weakening muscles that may be inhibiting hip flexion using neurolytic agents or surgical procedures (42).

In initial swing, the only critical event is to achieve maximum knee flexion of at least 60 degrees. If 40 degrees of knee flexion has been achieved at the end of pre-swing and the hip flexors and adductor longus stop firing before the end of initial swing, then in the absence of inappropriate quadriceps or hamstring activity, sufficient knee flexion should occur naturally during this phase. The point of maximum knee flexion must occur before the end of initial swing (not during mid-swing or terminal swing), since this is the point where the swinging limb must be at its shortest functional length to successfully clear the ground. The ankle dorsiflexors (pretibials) are firing concentrically at this time to bring the foot from its point of maximum plantar flexion at the end of pre-swing to at least neutral by the end of initial swing so that toe clearance can be assured in mid-swing. In this phase, kinematics can be used to quantify the progress of the swinging limb, joint moments in the sagittal plane should be near zero, and dynamic EMG can be used to identify inappropriate muscle firing. Of particular interest in this phase is the activity of the rectus femoris. This biarticulate muscle initially is active in late pre-swing to assist with accelerating the thigh forward. At the moment the foot leaves the ground, continued activity of the rectus femoris may assist with hip flexion, but may have the negative consequence of providing open-chain knee extension through the patellar ligament. Since it has been shown that the brain uses the rectus femoris to accelerate the thigh to selectively control step length and cadence during swing (36), if hip flexor angular velocity is initially slow and 40 degrees of knee flexion was not achieved at the moment of foot-off, the rectus femoris may increase its activation in initial swing to serve as an auxiliary hip flexor. This abnormal compensatory activity of the rectus femoris is in an effort to produce increased thigh acceleration, but because of its biarticular structure, it yields the negative effect of producing a larger knee extension moment, exacerbating the problem of insufficient knee flexion in swing. Whether the rectus femoris is firing as a compensatory mechanism or because of incorrect motor control associated with upper motor neuron injury, if it continues to be active at the end of initial swing, it may contribute to the abnormality known as stiff-knee gait, the common name given to the gait abnormality of insufficient knee flexion in swing period. If kinematic and electromyographic evidence (reduced knee flexion peak and/or slope and prolonged activation) exist, then a rectus femoris transfer to the semimembranosus or sartorius may harness this inappropriate activity, or more likely, prevent concentric knee extension from limiting peak knee flexion in swing and thereby disrupt swing limb advancement. The rectus femoris transfer for the treatment of stiff-knee gait is a surgical procedure that was conceived

as a direct result of using IGA techniques (43) and has been supported by a series of laboratory investigations and long-term follow-up (44,45,46,47,48,49). It is now considered the standard of care for the treatment of stiff-knee gait when evidence from IGA confirms that the rectus femoris is responsible for failure to achieve the critical event of obtaining maximum knee flexion of 60 degrees during initial swing.

The two critical events during mid-swing are both related to achieving toe clearance as the limb swings through the lowest point in its arc of motion and it is at greatest risk to inadvertently make contact with the ground. These critical events are maximum hip flexion to 30 degrees and neutral ankle dorsiflexion. The hamstrings may fire near the end of mid-swing to begin decelerating the forward movement of the thigh or to slow down the cadence, but generally these muscles should be silent until terminal swing. Note that after the swinging limb clears the floor, there is typically no further need for hip flexion, and additional hip flexion will only decrease the likelihood of achieving the final critical event during terminal swing: knee extension to neutral. Children with cerebral palsy and other patients with upper motor neuron disease have a difficult time motor programming the previous two phases of motor activity, and often display excessive knee and hip flexion with peak values later than normal during mid-swing. Kinematics and dynamic EMG can help identify these incorrect patterns, and the usual procedures of stretching, lengthening, or injecting the offending muscles may be useful if they can permit the critical events in pre-swing and initial swing to occur. The critical event of neutral dorsiflexion is usually the responsibility of the ankle dorsiflexors, which typically initiate concentric activity in pre-swing. The pretibial muscles generally reduce their activity in this phase since they no longer need to concentrically contract from the plantar flexed position and are only needed to hold the foot against gravity. If they are weak or if there is an upper motor neuron injury preventing normal motor control, foot drop will result, which will adversely affect toe clearance and necessitate compensatory mechanisms of circumduction at the hip, increased ipsilateral pelvic obliquity (hip hiking) or contralateral early heel-off (vaulting). The most common solution to this problem is to prescribe an AFO with a plantar flexion stop to hold the foot in the correct position throughout swing period. In children and adults with TBI, dynamic EMG can be used to determine if the lack of dorsiflexion during swing is related to incorrect cortical control or an inability to correctly motor-plan the dorsiflexion activity. In the latter, training with biofeedback of muscle contraction may improve foot clearance during swing and eliminate the requirement of using an AFO.

The final critical event in the gait cycle is knee extension to neutral during terminal swing. This represents the last opportunity of the swinging limb to reposition the foot prior to weight acceptance, and if this critical event is achieved, a sufficiently long step length will result. In typically developing children and normal adults, hamstring activity will begin during this phase to decelerate the swinging lower limb so that a small amount of knee flexion (<4 degrees) is present at initial contact. In some cases during slow speed walking, the quadriceps will contract concentrically to assist with final knee extension, but in general, this is unnecessary if the subject displays proper motor control. In addition to kinematic recordings to confirm final position and dynamic EMG recordings to determine if there is excessive hamstring activity, large hip extensor and knee flexor moments with power absorption just prior to initial contact may be indicators that the hamstrings are tight or display increased tone. Lack of full knee flexion in terminal swing is one of the most common gait abnormalities in cerebral palsy (50), and may require neurolytic injections or surgical lengthening of the hamstrings and adductors when the physical exam and gait measurements provide appropriate evidence. As in the previous sections, a table has been prepared to summarize the gait measurements that are useful in detecting abnormalities in the six critical events associated with swing limb advancement (Table 16.6). If all six of these critical events are performed successfully, the limb will be properly prepared for initial contact and ready to begin the cycle of gait events again.

The 8 phases and 13 critical events described in the previous sections and summarized in Table 16.3 complete the functional decomposition of the gait cycle. Armed with this analysis framework, the pediatric physiatrist can utilize IGA measurements, radiographs, and a comprehensive physical exam to better understand the complex interactions of body structure and function that produce abnormal gait patterns in the pediatric patient. We will conclude this chapter with a case study illustrating the use of this framework in a subject with a common pediatric diagnosis but a unique and challenging gait abnormality.

MOVEMENT ANALYSIS CASE STUDY—DIPLEGIA

Cerebral palsy is the most common cause of motor disability in children, with an incidence of approximately 2 to 3 per 1,000 live births in the United States. It is most often associated with low birth weight, preterm infants or with multiple births, and despite improved prenatal care, the incidence has not decreased in several decades. Of the four main types of cerebral palsy

16.6 Gait Measurements Useful for Identifying the Cause of an Absent or Abnormal Critical Event During the Swing Limb Advancement Task

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/DIST. MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Pre-swing	Passive knee flexion to 40°	Strength • Weak hip flexors ROM • Tight hip flexors (limit hip extension)	• Delayed foot-off • Prolonged stance period	• Insufficient hip extension at beginning of Pre-Swing (<10°) • Incorrect slope, 2nd knee flexion peak	• Reduced peak hip flexor moment and power generation • Reduced knee extensor power absorption	• Reduced or absent adductor longus or hip flexor activity • Abnormal hamstring activity
	Rapid ankle PF	Strength • Weak ankle PF ROM • Excessive DF or reduced PF	• Delayed foot-off • Prolonged stance period	• Abnormal 3rd rocker (heel rocker) • Excessive DF early or reduced PF late in pre-swing	• Reduced peak PF moment at start of pre-swing • Reduced peak PF power generation	• Prolonged ankle PF activity into late Pre-Swing • Inappropriate co-contraction of ankle PF, DF
Initial swing	Maximum knee flexion (>60°)	Strength • Weak hip flexors ROM • Tight rectus femoris	• Asymmetric stance/swing ratio • High variability in swing period or step length	• Reduced or delayed peak knee flexion in swing • Incorrect slope in knee flexion wave • Slow ankle DF	• Non-zero moments and powers at hip, knee and ankle by end of Initial Swing	• Reduced or absent adductor longus and hip flexor activity • Reduced or absent ankle DF activity
Mid-swing	Maximum hip flexion to 30°	Strength • Weak hip flexors ROM • Tight hamstrings	• Asymmetric stance/swing ratio • High variability in swing period or step length	• Max hip flexion late or in next phase • Abnormal pelvic obliquity (hip hike) or hip abduction (circumduction)	• Non-zero moments and powers at the hip • Premature knee flexor power absorption	• Prolonged rectus femoris activity • Premature hamstring activity
	Neutral DF	Strength • Weak ankle DF ROM • Tight ankle PF	• Asymmetric stance/swing ratio • High variability in swing period	• Sagittal plane ankle curve with excess DF or PF	• Non-zero moments and powers at the ankle	• Reduced or absent ankle DF activity with foot drop • Any PF activity
Terminal swing	Knee extension to neutral	Strength • Weak hip flexors or knee extensors ROM • Tight hamstrings Neurologic • Hamstrings tone	• Asymmetric stance/swing ratio • High variability in swing period or step length	• Knee flexion >4° before initial contact • Compensatory and excess hip flexion for limb clearance	• Large hip extensor and knee flexor moment with power absorption just before initial contact	• Excessive knee flexor activity • Reduced or absent ankle DF activity

DF, dorsiflexion; PF, plantarflexion; ROM, range of motion.

(spastic, athetoid, ataxic, and mixed), more than 70% are classified as having spastic cerebral palsy, as is the subject of this case study.

"LD" is a nonverbal 13.5-year-old male with spastic diplegia and developmental delays. His mother had an uncomplicated pregnancy, and he was born full-term, but in his first year of life he demonstrated delayed developmental milestones and did not start walking until age 6. He is also hearing-impaired with cognitive, behavioral, and oral motor dysfunction. Previous treatments included oral baclofen, which had little effect on gait performance, and bilateral hinged AFOs. He had no neurologic or orthopedic surgical procedures performed prior to his visit to the cerebral palsy clinic at our institution, after which LD was referred to our motion laboratory for instrumented gait analysis. His family reported an increased incidence of tripping and falling over the previous 15 months with fast walking and a perceived reduction in overall gait performance. They also reported that the left leg was now turning out more than in the past.

The physical examination performed on the day of the gait analysis measured LD's height as 165 cm, his weight as 63 kg, and he had equal leg lengths. No fixed joint contractures were found, but he showed a popliteal angle of −65 degrees bilaterally, consistent with hamstrings tightness. We measured a thigh-foot angle of 35 degrees external on the left, 25 degrees external on the right, slight hindfoot valgus, moderate forefoot abduction, and moderate pes planus bilaterally. Ely and Thomas tests were normal, and there was no appreciable spasticity (1 on the Ashworth scale) in the hamstrings, quadriceps, peroneals, tibialis posterior, toe flexors, or triceps surae bilaterally. Strength information from manual muscle test of the major muscle groups was inconclusive due to the inability of the subject to perform an isolated muscle contraction and his difficulty understanding instructions due to cognitive limitations. However, the therapist performing the physical examination reported that most muscle groups should be at least in the range of 3–4 by observing other functional activities and by noting that the subject is an independent, limited community ambulator.

Radiographs taken at the time of the analysis showed slight adduction of the proximal femurs but no sign of femoral head uncovering and otherwise normal hip joints bilaterally. Standing anterior/posterior (A/P) and lateral radiographs of the foot showed forefoot abduction, uncovering of the talus, midfoot collapse, and a reduced height of the medial longitudinal arch (see Fig. 16.14). This was consistent with evidence of increased pressure at the navicular during the physical exam and redness caused by the orthotics in the same area.

Observational gait analysis using slow-motion video recordings while the subject walked barefoot in the laboratory showed a stiff-knee gait pattern during initial swing phase, reduced peak knee flexion in mid-swing, insufficient knee extension during terminal swing, and a reduced dynamic knee range-of-motion throughout the gait cycle, all observed bilaterally. LD shows a foot-flat initial contact bilaterally, with no sign of a heel or first rocker (see Fig. 16.13). At the ankle, LD showed premature tibial advancement during loading response and mid-stance with delayed heel-off bilaterally. In the frontal plane, LD demonstrated moderate lateral trunk lean during loading response on both sides consistent with a compensated Trendelenburg's gait pattern, suggesting weakness of the hip abductors.

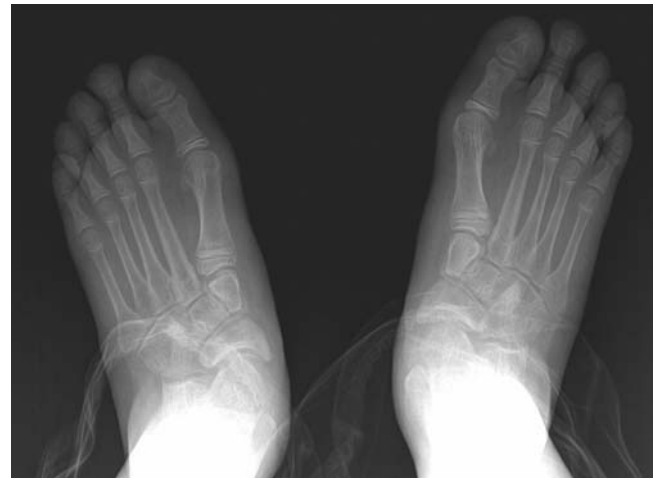

Figure 16.14 Bilateral A/P radiograph of the feet of the subject described in case study. This radiograph is commonly required when the subject presents with pes planus, to better understand the structural alignment of the foot.

The instrumented gait analysis included temporal-spatial measures, 3D kinematics, 3D kinetics, and dynamic EMG recorded from six muscles bilaterally using bipolar surface electrodes. Because of the report of pes planus, a plantar pressure recording was included to document the existence of excessive pressure in any area of the foot. The temporal-spatial recordings showed an average cadence of 103 steps/minute (88% normal) and an average walking speed of 51 meters/minute (65% normal). The left side average step length was 0.54 meters (80% normal) and the right side was slightly less at 0.47 meters (70% normal). There were no appreciable differences in gait symmetry or timing of gait events between the left and right sides, with the exception of a slightly longer single limb support time on the left that was not considered clinically significant (95% normal on the left, 89% normal on the right).

Kinematic, kinetic, and dynamic EMG data from both legs for the barefoot trial are shown in Figures 16.15, 16.16, and 16.17, respectively. On the

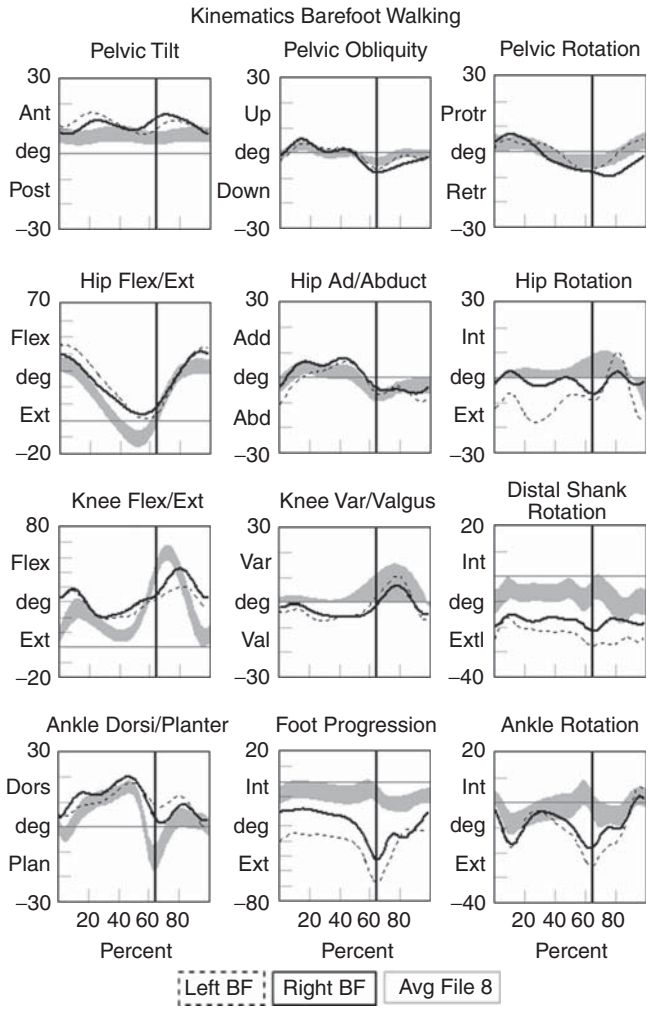

Figure 16.15 Three-dimensional kinematic graphs constructed from a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject, "LD." The solid line describes the right side, the dashed line shows the left side, and the gray band is from the age-matched normal database collected in the laboratory and used as a reference.

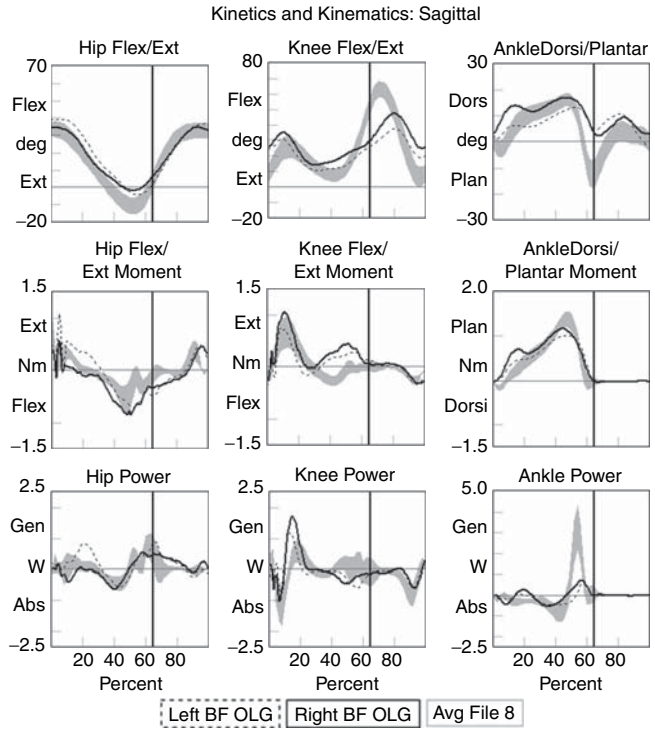

Figure 16.16 Three-dimensional graphs of sagittal plane kinematics, sagittal plane joint moments, and total joint power for the hip, knee and ankle constructed from a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject, "LD." The solid line describes the right side, the dashed line shows the left side, and the gray band is from the age matched normal database collected in the laboratory and used as a reference.

kinematic and kinetic curves, the right side is represented by a solid line and the left side uses a dashed line, and for comparison, a gray band is included on each graph representing the ensemble averages from our typically developing child database for this age group. The gray bands correspond to +/– 1 standard deviation across the ensemble average for that graph. For the EMG data shown in Figure 16.17, the right side is darker, the left side is lighter, and the black bar at the bottom of each graph is a normal timing reference.

There are a variety of ways to review these data systematically, including evaluating each joint or segment in sequence starting either proximally or distally, evaluating all graphs for a particular data type first and then moving on to the other categories, or reviewing all data for a particular phase of gait and then advancing to the next phase until the cycle is completed. Which of these procedures to follow is a matter of personal preference, and is sometimes dictated by the complexity of the case, but for the novice, it is a good idea to consistently follow the same procedure or review sequence until you are comfortable recognizing the significance of each graph individually. We typically start with the kinematic graphs and work distally from the pelvis, scanning the graphs across all phases of the gait cycle to identify deviations from the normal reference. We focus first on the portions of the curve that have the largest deviation from the reference data and then attempt to describe these deviations in the context of the 8 gait phases and 13 critical events described previously and summarized in Table 16.3. As needed, we jump to the subject's kinetic and EMG data for additional evidence to explain the absence of a critical event at a specific phase of the gait cycle, and using all of the evidence gathered in the analysis, develop a logical rationale for the subject's unique gait pattern or abnormality.

In the case of LD, we see evidence of slightly increased anterior pelvic tilt starting during loading

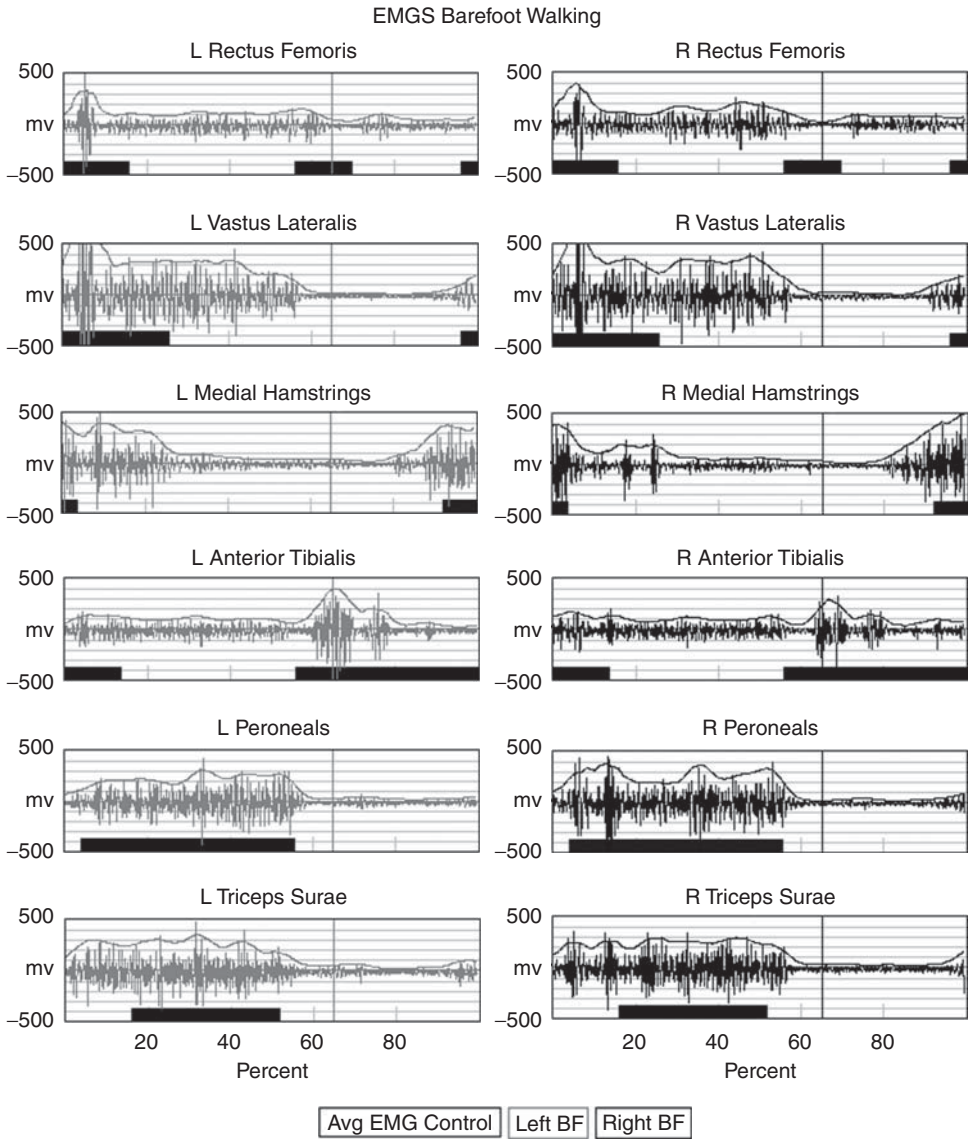

Figure 16.17 Filtered and time normalized EMG for 12 muscles of the lower extremity for a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject, "LD." The black bars at the bottom of each graph are constructed from published normal EMG activations and are used as reference values. The smooth curve above the EMG activation is a processed EMG signal obtained by rectifying and integrating the raw EMG.

response and reaching a peak of approximately 18 degrees at mid-stance on the left and 14 degrees on the right (Fig. 16.15, first row). This gives rise to a pattern often seen in diplegia called a "double bump" as the pelvis tilts slightly forward during weight acceptance on each side. It is often associated with weakness of the hip extensors and lack of shock absorption more distally, and can be attributed to reduced performance in the critical event of hip stability during loading response. Another cause could be tight hip flexors, but this is unlikely given that the Thomas test from the physical examination was negative. The existence of the second bump in the pattern during pre-swing and initial swing comes from the same mechanism occurring at loading response on the contralateral side. It is reflected in the ipsilateral pelvic tilt because the pelvis is, of course, a single segment and the graphs of each hemipelvis section are 180 degrees out of phase. Pelvic obliquity and pelvic rotation are near normal bilaterally until foot-off and the beginning of initial swing, when the right hemipelvis drops and retracts slightly compared to the normal reference and the left side. This suggests that the compensated Trendelenburg's gait pattern observed is not completely effective at maintaining appropriate pelvic position on the right side during initial swing, possibly due to weaker hip

abductors on the left side during loading response. All of these compensations can be attributed to difficulty achieving the critical event of hip stability during loading response and are evidence of proximal weakness during the task of weight acceptance.

Moving to the hip joint, LD shows increased hip flexion during loading response, decreased hip extension during terminal stance and pre-swing, and increased hip flexion at terminal swing bilaterally. Notice that the shape and range-of-motion for the hip flexion curve is virtually the same as the average normal curve, except that it is shifted up toward increased flexion by about 10 degrees. This is approximately the same amount that the corresponding anterior pelvic tilt curve is offset from its normal value. These two graphs are often coupled since hip joint angles are calculated relative to the pelvis, the more proximal segment. The lack of hip extension at terminal stance is the most significant limitation here, since it negatively affects the ability to achieve a trailing limb posture during terminal stance, which is essential to achieving maximum stride length on the contralateral side. Moving to Figure 16.16 and the sagittal plane kinetics at the hip joint, we see no significant deficits in the hip moment curve bilaterally, but a reduced hip power generation at pre-swing, approximately 70% normal on the left and 50% normal on the right. Since sufficient power generation at the hip is necessary to achieve the critical event of passive knee flexion to 40 degrees during pre-swing, and is also a necessary precursor to accomplish the task of swing limb advancement (50), reduced power generation at the hip may contribute to LD's increased incidence of tripping when trying to walk at higher speeds.

Returning to Figure 16.15, the transverse plane motion at the hip shows near normal hip rotation on the right side, but increased hip external rotation of approximately 10 to 15 degrees on the left side. This suggests that some of the reported external foot position on the left can be attributed to the hip. Looking distally down the kinematic chain, we see additional contribution to the final foot progression angle occurring at the knee (distal shank rotation, left approximately 20–25 degrees external, right approximately 10–15 degrees) and to a much lesser extent at the ankle bilaterally, yielding a final foot progression angle of approximately 40 degrees on the left and 20 degrees on the right during mid-stance and terminal stance. We use the term "distal shank rotation" here rather than knee rotation to highlight that the recording includes the external "twist" of the tibia or tibial torsion in the graph rather than just the amount of rotation occurring between the thigh and shank segment. The modified Helen Hayes marker set used to produce these curves assumes that the ankle joint axes and knee joint axes are offset in the transverse plane by the amount of the tibial torsion and normally wouldn't include this offset. We prefer to include the tibial torsion in this curve to better understand the contribution of tibial torsion to the overall foot progression angle, and therefore, call it the distal shank rotation to avoid confusion. It is good practice to have a clear understanding of how the link-segment model is calculating a particular quantity before utilizing it for clinical decision-making, and this curve in particular is frequently affected by vague or unstated model assumptions. To conclude the rotational assessment, we see a large peak in the external foot progression angle (left approximately 70 degrees external, right 50 degrees external) that corresponds to a lateral whip of the foot at foot-off, most likely as a compensatory mechanism to help with limb advancement. This large external foot progression angle with the left about 20 degrees greater than right, is consistent with the parents' description, and from the kinematic analysis, can be attributed to both the thigh and shank on the left, and from compensatory mechanisms at the foot and ankle bilaterally.

The analysis now moves distally to the knee, where some of the most significant gait deviations exist. In Figure 16.15, the bilateral knee flexion/extension curves show increased knee flexion (relative to the normal reference) during loading response, decreased knee extension during terminal stance, decreased and delayed peak knee flexion during initial swing (left more severe), and increased knee flexion during terminal swing and initial contact. This has the appearance of compressing the knee sagittal plane curve into the middle range of the normal reference, with a shallow rising slope from mid-stance through initial swing (left = 52 degrees/second, right = 65 degrees/sec, normal = 240 degrees/sec), and decreased dynamic range at the knee over the entire gait cycle (left = 24 degrees, right = 31 degrees, normal = 60–70 degrees). The existence of swing period gait deviations at the knee prevents the most important critical event in swing from being accomplished: achieving maximum knee flexion of at least 60 degrees. Their presence also provides evidence of a bilateral stiff-knee gait pattern that, as previously described, adversely affects the task of swing limb advancement. But when taken together, this combination of excessive knee flexion in stance and insufficient knee flexion in swing has the effect of disrupting all other critical events associated with normal knee function, including controlled knee flexion during loading response, achieving trailing limb posture during terminal stance, passive knee flexion to 40 degrees during pre-swing, and finally reaching full knee extension during terminal swing. With this many critical events absent, altered, or delayed, all three fundamental gait tasks are compromised. Therefore, in order to see any significant improvement in walking ability, these critical events need to be restored,

which by necessity prioritizes any intervention directly affecting knee range of motion during the gait cycle. To find support for specific interventions, we return to the kinetic and EMG recordings shown in Figures 16.16 and 16.16, respectively. In the top row of Figure 16.17, we see that both the left and right rectus femoris EMG recordings show muscle activation beginning late in initial swing and continuing until terminal swing, with a small peak in initial swing slightly before peak knee flexion. This abnormal EMG activity in combination with insufficient peak knee flexion in initial swing and a shallow slope of the knee curve during pre-swing provide strong evidence to support the use of a rectus femoris transfer procedure bilaterally 46,49). When successful, this procedure can improve both the peak knee flexion in swing and the slope of the knee flexion wave starting in pre-swing, addressing two missing critical events at the knee. To address the other affected critical events, we must review the kinetics and EMG recordings during initial contact, loading response, mid-stance, and terminal swing phases. With greater than 20 degrees of knee flexion throughout stance period, there is a significant force demand on the knee extensors during weight acceptance and single limb support. Evidence of this can be found in the large knee extensor moments during loading response and terminal stance, shown in the middle graph of Figure 16.16, and the prolonged stance phase EMG activity of the vastus lateralis and rectus femoris shown in Figure 16.17. These findings are consistent with a mild "crouch gait" deformity, set up by the limitation in knee extension at terminal swing.

While LD is able to overcome this biomechanically disadvantaged position and maintain an upright posture at this time, as he matures and grows heavier, any increase in knee flexion during stance may increase the demand to a level greater than he can withstand, which would severely limit his overall gait performance. It is reasonable here to consider the more aggressive surgical procedures that have been shown to improve knee function in cases of persistent crouch gait, namely a knee extension osteotomy to reduce knee flexion contracture and patellar advancement to treat patella alta and improve the function of the quadriceps (51). Since LD showed no significant knee flexion contracture on physical examination or radiographic signs of patella alta, and the crouch deformity was considered mild since he could achieve 20 degrees of knee flexion at terminal stance, these surgical procedures were deemed unnecessary at this time. However, since there was evidence of tight hamstrings on physical examination (popliteal angles of –65 degrees) and the EMG recording of the medial hamstrings (third row, Figure 16.17) showed premature onset in mid-swing, the team felt hamstring lengthening procedures would be appropriate. Nonsurgical techniques such as phenol injections to the hamstrings could be considered here, but with strong evidence for rectus femoris transfer and the ease of performing a hamstring lengthening at the same time as the rectus procedure, the surgical path seemed most appropriate for this patient. Furthermore, the combination of these two procedures has the best chance of restoring all missing critical events at the knee in the shortest amount of time to prevent continued progression of the crouch gait deformity as LD grows larger through adolescence.

To complete the instrumented gait analysis, we move distally once more to the remaining graphs describing the ankle and foot. The sagittal plane ankle kinematics graph in the lower-left corner of Figure 16.15 provides evidence of what was seen during the observational analysis: increased dorsiflexion at initial contact and no sign of a first or heel rocker during loading response. This is consistent with the foot-flat initial contact observed, and is shown on the kinematic graph as an increasing slope in the first 10% of the gait cycle starting at 5 degrees of dorsiflexion, rather than a decreasing slope starting from a near-neutral ankle position for the normal reference. The right side shows increased dorsiflexion continuing into mid-stance, with a peak at about 15% of the gait cycle, after which the dorsiflexion stabilizes and then increases at a more normal rate (slope of the ankle curve) near the upper extreme of the normal reference until the beginning of terminal stance. After beginning in a dorsiflexed position at initial contact, the left side dorsiflexion increases at a normal rate, tracking closely the slope of the reference value and providing evidence of a near-normal second or ankle rocker. Following peak dorsiflexion in terminal stance, the period of rapid ankle plantar flexion during pre-swing begins, which is associated with the third or forefoot rocker. Unfortunately, maximum plantar flexion stops at a joint angle of approximately 8 degrees dorsiflexion on the left and 2 degrees dorsiflexion on the right—clearly insufficient compared to the normal reference. The ankle then maintains a dorsiflexed position throughout swing period bilaterally. Considering these elements together and describing them in terms of fundamental gait tasks and critical events, we begin to see a clear picture of the impact of these gait deviations at the ankle. First, we have evidence that during weight acceptance LD is missing a heel-first initial contact and controlled ankle plantar flexion (first rocker) bilaterally. Second, during single limb support, controlled tibial advancement (second rocker) is altered on the right and controlled ankle dorsiflexion (DF) with heel rise is delayed bilaterally. Finally, while starting the task of swing limb advancement, rapid ankle plantar flexion (third rocker) in pre-swing is reduced bilaterally. Fortunately, LD does maintain sufficient dorsiflexion in mid-swing to clear his foot so as not to compound the lack of knee flexion and stiff-knee

pattern already affecting swing limb advancement. As with the analysis at the knee, failure to achieve these critical events at the ankle represents significant gait dysfunction and must be addressed. Additional insight can be obtained from the kinetic graphs on the right-most column of Figure 16.16 and the EMG recordings in the lower three rows of Figure 16.17. As is seen on the right side of Figure 16.16, the combination of foot-flat initial contact, increased knee flexion, and increased ankle dorsiflexion during loading response places a large demand (external moment) on the ankle plantar flexors, and they respond by increasing the net ankle plantar flexion moment (internal moment) during loading response and the early portion of mid-stance. This is most likely a compensatory response to the external demand, and is accomplished by prematurely activating the peroneals and triceps surae during terminal swing, initial contact, and loading response. Notice that the peak in right ankle dorsiflexion at approximately 15% of the gait cycle is accompanied by a peak in the plantar flexion moment and just preceded by a small peak of ankle plantar flexor power absorption, shown in the lower-right graph of Figure 16.16. The ankle power curve then reverses to produce a small amount of power generation at the point in the cycle (mid-stance) when continued ankle dorsiflexion is briefly reversed and the ankle plantar flexion moment returns to normal levels. This suggests that although biomechanically disadvantaged by foot position and excessive knee flexion, the ankle plantar flexors initially absorb energy during loading response as the tibia falls forward, but then limit excess dorsiflexion with a brief burst of power generation at the ankle at the beginning of mid-stance. Since the physical examination was inconclusive, it isn't clear whether this is due to true ankle plantar flexor strength or simply the resistance or viscoelastic behavior of the musculotendon unit. Regardless, it does explain the early dorsiflexion peak in the ankle sagittal plane graph and suggests there is some eccentric control over tibial advancement during loading response and mid-stance.

However, ankle function is not as good during terminal stance and pre-swing, when the powerful concentric contraction of the triceps surae is needed to produce rapid ankle plantar flexion. The strongest evidence of this is shown in the reduced ankle power generation during pre-swing in Figure 16.16, where the power generation is approximately 25% normal on both sides. Since power generation is normally larger at the ankle than at any other joint, and substantial power generation from both the hip flexors and ankle plantar flexors is necessary to produce passive knee flexion during pre-swing, this is a profound deficit that affects both the knee and the hip, and is the strongest evidence of plantar flexor weakness in the analysis.

The experienced gait analyst might cite the delayed heel-off and short step length of this "calcaneal gait" pattern as obvious indicators of calf weakness. While this may be true, the ankle power data provides a strong quantitative justification for such a claim, and has the added benefit of gauging the degree of dynamic plantar flexor weakness that occurs at this critical point in the gait cycle. This evidence, along with the excess dorsiflexion during stance period and our concerns about more severe crouch gait deformity as LD matured, eliminated any thoughts of a tendo-achilles lengthening or intramuscular lengthening of the gastrocnemius for this subject.

Based on the results of the instrumented gait analysis and the other physical examination and radiographic evidence, and following consultation with the patient and his family, our clinical team recommended that LD undergo bilateral rectus femoris transfers to the semitendinosis, bilateral hamstring lengthenings, bilateral Evans calcaneal lengthenings, and a left tibial osteotomy of approximately 20 degrees internal. The rectus femoris transfers were clearly indicated from both kinematic and dynamic EMG evidence and the presence of a stiff-knee gait pattern. The hamstring lengthenings were supported by physical examination and IGA data, and could be efficiently performed in conjunction with the rectus transfers. Since there were no previous surgical procedures performed on the hamstrings, we prefer to transfer the rectus femoris to the semitendinosis, although we have found no evidence to rule out the other potential transfer sites of sartorius or gracilis (49). The Evans calcaneal lengthenings were primarily supported by the radiographic evidence that showed significant uncovering of the talus and forefoot abduction with mid-foot collapse. While not always a part of our IGA procedure, for this case, a foot plantar pressure measurement from each limb was recorded using a two-meter pressure plate mounted in the motion laboratory walkway after the force platform array. These recordings confirmed the existence of pes planus and showed an increased pressure under the first metatarsal heads and medial border bilaterally, with the pressures higher under the left foot. This information, combined with concerns expressed by the family regarding LD's flat feet, as well as the clinical team's hope that a more rigid and properly aligned foot could improve the power generation capability of the ankle plantarflexors during pre-swing, convinced us to add this procedure to the list. The left tibial osteotomy was warranted based on the rotational kinematic findings that showed a distal shank rotation of approximately 15–20 degrees greater than normal, a foot progression angle approximately 25–30 degrees greater than normal (including the contribution from the external hip rotation that was believed to be compensatory), and the family's concerns about the limb asymmetry and increasing external foot position.

Finally, bilateral leaf-spring ankle foot orthoses were prescribed to provide some plantar flexor assist and to help control tibial advancement in the presence of the weak plantar flexors that would most likely persist after LD recovered from his surgical procedures. However, we are hopeful that the improved biomechanical position, increased knee flexion in swing phase, increased knee extension at initial contact, and more rigid foot that we expect will result from these surgical interventions will reduce the physical demands of walking sufficiently for the AFOs to be discontinued once LD fully recovers. While a follow-up gait analysis to confirm our recommendations could not been included here since it had not been completed at the time of this writing, it really isn't the purpose of this case study to demonstrate our gait analysis prowess using a single sample. Rather, we hope this clinical example serves to illustrate how instrumented gait analysis and a systematic analysis procedure based on functional decomposition of the gait cycle can be used to make complex clinical decisions for the pediatric patient with gait dysfunction.

SUMMARY

In this chapter we have attempted to provide an overview of the methods, procedures, and strategy for utilizing instrumented movement analysis to assist with the clinical interpretation of gait deformity in children. Focusing on the functional subdivisions that naturally occur during normal walking, and identifying the specific critical events that must be accomplished in each phase of gait, we have developed a framework that can be used for both instrumented and observational gait analysis and that can be applied to children and adults. By providing a brief description of modern computerized systems for movement analysis and linking measurements from these systems to functional tasks and critical events, we hope that instrumented gait analysis will be less intimidating and more clinically relevant to the pediatric physiatrist. Controversy remains regarding the value of IGA and its place on the modern rehabilitation service, with staunch advocates (8,10,35,52,53) and ardent detractors alike (54,55). It is our hope that armed with a solid background in the principles of gait analysis and an objective and impartial understanding of the benefits and limitations of current methodologies (56), every pediatric physiatrist can make the best clinical choices for the complex neuromuscular patients who rely on their decisions.

PEARLS, PERILS, AND RESOURCES

- Instrumented gait analysis (IGA) supports decision making for the child with walking problems, but doesn't replace a sound clinical and technical understanding of normal gait.
- Normal gait is naturally cyclical and symmetric, so any movement asymmetry should be investigated.
- There are 13 critical events that must occur during 8 distinct phases of the gait cycle to produce a normal gait pattern. Each critical event has functional significance, and so provides a link between observed gait abnormalities and possible interventions.
- IGA provides evidence of absent, altered, or delayed critical events, and provides the framework for identifying treatments to directly address these functional limitations.
- While skill, experience, and practice are required to fully utilize IGA results for clinical interpretation, by following the strategy outlined in this chapter, the process can be less intimidating and more clinically relevant to the pediatric physiatrist.

Resources

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Psychosocial Aspects of Pediatric Rehabilitation

Lee Renee Lucas

While many clinicians have heard the iconographic illustration of childhood disability in the piece "Welcome to Holland" (1), there persists a tendency to ignore the vital lessons embedded there and go on to "preach" based on anecdote and stereotypes. This chapter will seek to explore both the literature on the family experience with childhood disability and clinical experience of the author after almost decades in the field. The clinical experience enlivens and enlarges on the existing body of literature, giving attention to the vital aspect of the actual experience embodied in work such as "Welcome to Holland."

OVERVIEW

The focus of this chapter must be the family. Any credible work in this field is based on the premise that the family's adjustment underlies that of the child/adolescent. This is consistent with the developmental understanding of the "typical" child, and the added issue of disability only magnifies this relationship between child and parent or caregiver. Issues of dependence beyond more typical childhood limits often exist in addition to the added management and stress of involvement with medical and social systems not encountered to any similar degree with a more typical child.

The scope of this chapter will encompass those children with disabilities and their families within a medical rehabilitation context. This work will cover those with congenital and acquired disabilities, with some relative focus on those with acquired disability due to injury or illness. Many children now survive what in times past would have been lethal assaults by illness or injury and now mature into adulthood. So now the issue is quality of life for the 1 to 2 million children who have a severe chronic physical condition in the United States (2).

Quality of life means several things in this context. Good quality of life depends on the family's ability to maintain its own integrity by developing an idiosyncratic "normal" from which to preserve its primary job of guiding the child with a disability (or any child) to independence. In real terms, this means performance of tasks unique to children with disabilities. These tasks include, but are not limited to, mastery of the medical system, needed accommodations from the educational setting, navigation of the "rough waters" of social acceptance, and integrating the reality of the disability into the family structure. Specific to this is empowering the family by providing the necessary educational materials as well as mentoring to facilitate adept handling of the two behemoths of the medical and educational/vocational bureaucracies. It is essential that the family remain the center of all

training and education of the child with a disability. The family is the expert on its own functioning, and without deliberately tapping that expertise, inappropriate goals and agendas are developed. Without doing this, in trying to "help," the family is left feeling isolated as they travel their journey of childhood disability and injury.

FAMILY-CENTERED CARE

Across the country, hospitals and other medical institutions are acknowledging the importance of including the family in critical medical and mental health decisions. Gone are the days where the clinician makes decisions without the patient's and family's input. In essence, there has been a paradigm shift, where the cultures of many health care organizations are not only inclusive of families, but also are actively recruiting their involvement. The goal is to empower families to ask direct and courageous questions by giving them access to medical information and placing more emphasis on the importance of human interactions among all health care providers (3).

"Family-centered care within the nursing profession is not a new trend, with roots dating back as early as the 1950s" (4). However, it did not receive national recognition until 1987, when former Surgeon General Koop made it a primary initiative (5). These initiatives broadened the definition of family and acknowledged the diverse cultural backgrounds that make up families in our nation.

Smith, Terrel, and Conant (6) state in their article "Making family-centered care a reality," that "Children get better faster when their emotional and social needs are met along with their medical needs—a hospitalized child is still first and foremost a child."

While this statement may ring true for most children and families, the literature also suggests that providing family-centered care may be daunting for some practitioners. For example, Newton (7) states, "There is no consensus as to how much and what form of parental involvement should exist and how far that participation should extend." Barriers such as balancing parental involvement and participation need to be addressed honestly and openly with the health care team. Keeping the child's medical condition in the forefront of decision making will allow a positive experience for all participants.

In an effort to gain a better understanding of family-centered care, Eichner and Johnson and The American Academy of Pediatrics have defined seven core principles (8) for the practitioner to incorporate into their interactions with families during each encounter to improve outcomes. Each principle is based on a *collaborative* relationship between family and health care practitioner.

1. Respect
2. Honoring racial, ethnic, cultural, and socioeconomic diversity and its effect on the families' experience and perception of care
3. Recognizing and building strengths of each child and family, even in difficult and challenging situations
4. Supporting and facilitating choices for the child and family about approaches to care
5. Ensuring flexibility in organizational policies and procedures so services can be tailored to meet the needs, beliefs, and cultural values of the family
6. Sharing honest and unbiased information with families on an ongoing basis in ways that are useful and affirming
7. Providing and/or ensuring formal and informal support for the child and parent/caregiver during each developmental phase

While patient satisfaction is the primary goal, the author also describes several benefits for the pediatrician (and other health care providers) as well (8). These include but are not limited to improved clinical decision-making from better information and a collaborative process; improved follow-through, as the family has been consulted; improved communication among health care team members; and greater child and family satisfaction with the health care team.

Family-centered care also recognizes that institutional leadership and policies must promote the family's best interests and support the activity of the direct care clinician. Although family satisfaction has increased from such initiatives as specialty meals, access to technology, and more inclusive visitor policies (eg, younger siblings), economic considerations exist. "Health care decision makers, providers, and third-party payers require evidence that family-centered care is not only effective but cost-effective" (9).

The Family and Health Care Team Partnership

"You are so strong"…"I don't know how you do it"…"I could never take care of a child with…."

In well-meaning attempts at support or solidarity, friends, family, and health care providers offer such words to a family who is absorbing the impact of a severe trauma or diagnosis of severe illness. Though such sentiments are heartfelt, the net result is distancing, with families feeling more isolated.

Families of children with disabilities or illness need ongoing knowledge, guidance, education, and training at each developmental milestone to prepare

them for the road ahead. It is the responsibility of the clinician to assist the family in integrating their child with special needs into a world that may be unprepared to meet them. Acknowledgement of the disability or illness in a respective and professional manner, while at the same time maintaining the family's perspective, is paramount to all involved with the child's care.

Resilience

In ecology, "resilience" has been defined as "two competing fashions that emphasize two different aspects of stability" (10). It may be said that when a child is diagnosed with a serious illness or injury, the two "competing forces" are the family system and the medical setting. The family system is trying to maintain their current homeostasis while allowing a new organism into their system, and the medical setting is trying to maintain its current state while integrating the child into its system.

In this section, the author explores the literature pertaining to resilience—changes that occur in the family when their child has been diagnosed with a serious illness, disability, or injury, as well as the health care team—and suggest interventions for the family to once again achieve homeostasis. There will be further exploration of a framework that will not only engage practitioners in using these models when a child or adolesencent is diagnosed with a serious illness or injury, but also assist families to anticipate and plan for the future (11).

The challenge of keeping consistent schedules and balancing the predictability or unpredictability of the child with illness or disability is not new. The family's life is often set between constancy and change, or between predictability and unpredictability. Roland and Walsh imply that "a family resilience framework is grounded in the recognition that crises and persistent challenges [affect] the whole family and, in turn, key family processes mediate the adaptation of all members and their relationships" (11). Therefore, the family requires a new road map along this journey. A fluid road map, where there are resources along the way to provide information, guidance, and support, is necessary.

The Family System

"After hearing that my child was injured, I felt as if I were slapped in the face; I felt the burning sensation for several minutes. There are some days when I can still feel that sensation on my cheek." –Mrs. S

It is not uncommon for a parent to have such a visceral reaction after hearing that their child has been diagnosed with a serious illness or disability. The way in which the parent or caregiver views the world is immediately changed. They lose their naivety and are forced to recognize the unfairness of the world. The process of this change is at the same time very fast and in slow motion. A parent or caregiver must immediately begin to make sense of the medical information presented to them, while at the same time devise a plan for siblings, spouses, or other family members. The family system as they knew it has been permanetly altered, and caregivers must now face the challenge of learning new tools to facilitate their understanding of the child's illness or disability.

Depending upon the diagnosis, the period of understanding the child's illness or disability may unfold within days, months, or years. Physical, emotional, and spiritual development will continue to take place, and the caregiver's role as parent will also continue to evolve as they learn new facets of their child's life. Extended family members who may not have been as involved in the past may now join the new family system in order to care for the siblings of a child with an injury or disability.

Traditional family roles and finances may be affected as the family prepares to accommodate the special needs of their child. Parents who in the past never had to rely on social services agencies may now require the tools to apply for public assistance. Applications for Social Security disability; Women, Infants, and Children (WIC); and food stamps may now need to be made in order for the family to survive. This process is not easy, and parents will not only require specific information about local and national agencies, but also emotional support so as not to feel "guilty or ashamed" about applying for these services.

Days before the diagnosis, the family may have been maintaining an organized schedule of work, school, church, recreation, and minimal physical- or health-related issues. Now, the caregiver system is engaged in learning about the special needs of their child, and at the same time, needing to rely on a "health care team" of individuals, none of whom know their child or family history. One complaint that parents have during these intial meetings with the health care team is that it is difficult for the parent to establish themselves as primary caregiver when other members of the team have so much influence upon how the child will be cared for. Parents may have the sense of being "steamrolled" over by the health care team and will need to maintain their role as primary caregiver (8).

In order to prevent these feelings, a parent should be supported by the health care team to continue to be the "expert" about their child and also be encouraged to partcipate as a member of the health care team. A parent or caregiver will be the most successful in caring for their child with an injury or disability if

they are given the opportunity to partner with the health care team. The parent of a child with special needs begins to develop new roles and learns how to be the child's advocate, broker, educator, and project manager.

Advocate

In the article "How to advocate for your child," Shekerjian (12) offers 10 tips to advocate for your child as follows:

1) Define and examine your concerns.
2) Develop possible solutions.
3) Prepare a written document.
4) Meet with the teacher (case manager, staff member).
5) Approach the meeting with a positive attitude.
6) Define the next step.
7) Document events.
8) Follow the chain of command.
9) Consider all educational options.
10) Never forget that you are responsible for the education (treatment, success) of your child.

Further, Faust (13) believes advocacy to be "a vital element because systems are not always responsive to the individual client." Parents, as advocates, take on the role of negotiating home care services, educational plans, and other social systems when their child has been diagnosed with a serious illness or injury. Parents have become more and more influential as social change agents, and advocacy is one way in which they make change. For example, a parent may need to call the administrator for their health care policy or legislator in order for their child to receive specialized equipment that may not otherwise be covered. The parent's opinion and understanding of her child's illness or disability may have more influence than the medical team has on the insurance system. One caveat to this example, however, is that parents may not have the energy or expertise to navigate this system without supports.

Broker. To manage, or to "broker," is the process in which a parent acts as a link or bridge to services (14). Parents with children with special needs are often the primary bridge between the medical staff and ancillary staff in the hospital, community resources, and the educational system. This role is necessary to ensure that services are appropriate and accessible to their child. This role also involves the parent or caregiver being concerned with the "quality and quantity of services." In other words, a parent will not rely on only the services that are offered, but will challenge the system to communicate in order to create change. This change can occur within or without the hospital system. On the rehabilitation team, for example, the parent communicates with team members from one hospital to another in order to prepare for discharge and ancillary services.

Educator. Parents begin their child's educational process even before the child is born, as soon as they begin to read the book *What to Expect When You Are Expecting* (15), and they continue the process as long they live. Parents of children with special needs are no different from those parents without children with special needs, but from the time they hear that their child has been injured, diagnosed with a disability, or become seriously ill, these parents become experts regarding the child's health care. At the initial team meeting, parents may have completed their own research on the diagnosis and may have found information unknown to medical staff. Members of the rehabilitation team rely on parents to inform and educate them about a patient in order to give the child the best care. Parents' knowledge of the child's likes, dislikes, and temperament are invaluable to the staff.

Project Manager. "Parents of children with disabilities do not see their children's needs dividing neatly along program lines" (16). However, the roles that parents take on should be on their own terms and should be evaluated periodically with staff. This evaluation should include the successes as well as the challenges faced by the family members. A careful and honest negotiation is necessary to clarify the needs of the patient and their caregivers. A parent who becomes a skillful advocate, broker, and educator is first and foremost a parent and should be supported in that role, not evaluated on their effectiveness in other roles (17).

The Health Care Team System

"Every wise physician knows that the best he can do for a patient [family] is to assist nature in healing" (18).

It is important to keep in mind that the best health care is given when both the parent and the health care team work in conjuction with one another. As in any relationship, communcation and mutual respect are essential. Fallowfield (19) reminds us that parents may have difficulty hearing the information regarding their child and that information from the caregiver may often need to be repeated. Furthermore, when giving critical information regarding the patient's illness or disability, it is practical for the practioner to have a "plan" to reinforce that the family has heard/understood the information that is being presented (19).

Davis (17) offers suggestions to organizing your "plan" when giving critical information to a family member or caregiver:

1. Begin with what the parent/family knows; ask specific questions, such as:

   "When we spoke last, you asked . . ."
   "What is your understanding of your child's disability?"
   "What did the emergency department physician tell you about the accident?"

2. Present the information. Facts and data are important in this segment of the conversation, but in small, digestible parts that parents can integrate into their knowledge base.

3. Check the result. Observe the family and their reaction to the information that is currently being presented. Invite the family at this time to ask questions or receive clarification.

4. Ensure retention.

It is also wise to offer written information about the specific topic, and refer to appropriate members of health care team (ie, case manager, social worker, psychologist) who can continue to process this information with the family. Many families may request an audiotape of the interview with the physician to share with other family members. The authors suggest that this process be established before the interview begins to ensure the privacy of the people involved.

When an audiotape is not appropriate, Cunningham and Newton (20) reported that using a written consultation questionnaire was also highly effective in confirming that families understood the medical information being presented. This tool became valuable to the parents as an effective means to communicate with the medical team and offered a voice to parents who were not confident to prepare individual questions themselves.

Stille and Antonelli (21) summarize in their article, "Coordination of care for children with special health care needs," that coordination is highly dependent upon communication." They go on to explain that "a team approach involving nonphysican staff and families as primary partcipants to be the best option in health care." Furthermore, it is the responsibilty of all members of the health care team to assist patients and families during all aspects of the child's illness or disability. The hope for all members of the health care team is that we view children and their families within a holistic approach, acknowledging that as a team we are separate and at the same time equal.

TASKS OF THE FAMILY

After either the initial diagnostic period or initial conversations about the child's injury or disability, the family's goal must be to return to "life as it was." Often, one parent must return to work, siblings must return to school, and the diagnosis or disability begins to integrate into the family's world. It is suggested here that a parent will have a better opportunity to achieve a new sense of "normalcy" and get their needs met when they are able to approach the child's illness or disability with a sense of confidence and inherent understanding that they will adapt and cope with an uncertainty that life may bring. Sandler (22), in her book *Living with Spina Bifida*, describes nine tasks that resilient parents of special-needs children learn. They are as follows:

- Balance the disability with other family needs.
- Maintain clear family boundaries.
- Become competent at communication.
- See situations in a positive light.
- Maintain family flexibility.
- Be committed to the family unit.
- Engage actively in coping strategies.
- Be well-integrated socially.
- Develop cooperative relationships with professionals.
- Obtain information/education.
- Learn new parenting skills.
- Achieve equilibrium or homeostasis.

Working Toward a New "Normal"

While loss is pivotal in a person's experience, it is not the loss of a child that these families are experiencing. Rather, it is the "loss" of the way in which their child experienced the world. It is the loss of what was "normal" and the comfort of what used to be.

After her child was injured and began to use a wheelchair, a parent spoke about missing walking with her child through the leaves in the autumn. The mother felt sad that she could no longer hear her son's footsteps crunch underfoot, but after a period of adjustment, was able to feel a new kind of happiness watching her son role himself through the leaves. Instead of giving up on a favorite pastime, the family learned that pushing the child in the wheelchair was fulfilling, just different from their previous experience.

Family members and caregivers are subjected to multiple tasks at the time of diagnosis, and one of these tasks is how to reframe their child's experience and create an experience that not only meets the child's needs, but also that of the family. The following are some examples of how families have reframed those initial negative feelings into feelings that are more productive or healthy:

Helplessness → Empowerment
Fear → Cautious optimism
Sadness → Openness of feelings
Anger → Advocacy

TASKS OF THE PRACTITIONER

Families with children with disabilities face the similar, mundane, everyday life struggles of families with typical children, yet there are volumes of research material investigating the differences between coping styles and functions in the family of a child with a disability, illness, or injury.

A study in *Pediatrics* (23) "provided a compare and contrast" of children with chronic illness to those children without. This study revealed that while all children and families should receive a psychosocial assessment, practitioners should not "assume that dysfunction" exists because of the disability in the family. It appears from this study that assessment of the family's needs should come first and then a look at the family's strengths and weaknesses in order to devise a support plan.

Similar research conducted by Press and Nolan (24) found that disease or disability does not predict adaptation to disability and that the psychosocial adjustment of family life before the illness or injury was the same as the psychosocial adjustment after the illness or injury. The predictors for positive outcome included good communication between family members, low conflict in the home setting, and the positive expression of emotion.

With that being stated, the role of the practitioner and the interventions offered to the family must always "start where the family is." In other words, the practioner must listen to the family's needs and offer emotional support at all developmental milestones or turning points in the family's life. The practitioner can act as a positive role model for the family by acknowledging changes in the family dynamics or community systems and reframe these changes so that the family can maintain function.

However, in order for a family to function at an optimal level, one must provide the family with the information and resources necessary to succeed. The practitioner or medical staff member should investigate information and resources in the family's community. Information on local community support groups, as well as resource information on funding opportunities, will be helpful to the family. Concrete assistance, such as filling out a Medicaid application or arranging a Social Security interview, will relieve the family's stress greatly.

Communication is also an essential intervention when working with any family, regardless of their child's diagnosis. Most families may not remember the exact phrases or words that the practitioner uses to disseminate information, but the family will often remember the tone, setting, and approachability of the practitioner during the interview. The physician's ability to restore competence in the family by acknowledging and validating their fears or concerns will ensure that the family feels as if they are a member of the health care team.

SELECTIVE TIMELINE OF THE HISTORY OF DISABILITY RIGHTS

A great deal of change has occurred within the last 40 years with regard to people with disabilities, and "some of the factors influencing the success are the nature and severity of the disabilities, accommodations available in society, and attitudes toward the disabled" (25). What follows is a selective timeline of the disability rights movement and how key pieces of legislation broke down the barriers of society in order to allow all individuals the right to education, accommodations, and freedom (26,27).

1964: Civil Rights Act passed outlawing discrimination on the basis of race in public accommodations and employment, as well as in federally assisisted programs. This law became the model for subsequent disability rights laws.

1965: Autism Society of America is founded by parents of children with autism in response to the lack of services. Parents found their children were being discriminated against by the medical "experts" who believed autism was the result of poor parenting as opposed to a neurological disability.

1968: The Architectural Barrier Act was passed, mandating that federally constructed buildings and facilities be accessible to people with physical disabilities. This act is generally considered to be the first ever federal disability legislative law.

1970: The Physically Disabled Students Program (PDSP) was founded by Ed Roberts, John Hessler, Hale Zukas, and others at the University of California in Berkley. With its provisions for community living, political advocacy, and personal assisted services, it became the nucleus for the first Center for Independent Living, founded two years later.

1973: Passage of the Rehabilitation Act of 1973 marked the greatest achievement of the disability rights movement. In particular, Title V and especially Section 504, with the first line confronting discrimination against people with disabilities. Litigation arising out of Section 504 generated such concepts as "reasonable modification," "reasonable accommodations," and "undue burden." This act became the framework for federal law (ie, Americans with Disability Act of 1990).

The Education for All Handicapped Children Act (Pub. Law 94–142) was passed establishing the rights for children with disabilities, including a public education in an integrated environment. The act is a cornerstone of federal disability legislation. Over the next 20 years, millions of children with disabilities were educated under its provisions, radically changing the lives of people in the disability community.

1975: The first Parent and Training Information Center is founded to help parents of disabled children exercise their rights under the Education for Handicapped Children Act of 1975.

1976: Passage of an amendment to the Higher Education Act of 1972 to provide services to physically disabled students entering college.

1980–1983: The parents of "Baby Doe" in Bloomington, Indiana, are advised by their physicians to forego a surgical procedure to unblock the baby's esophagus due to the fact that the baby had Down's syndrome. "Baby Doe" starved to death before legal action could be taken. However, this case prompted the Reagan administration to issue legislation calling for "Baby Doe squads" to safeguard the civil rights of newborns.

1984: The "Baby Jane Doe" case, like the previous baby in Bloomington, Indiana, involved an infant being denied medical care because of the infant's disability. This case resulted in litigation argued before the U.S. Supreme Court in *Bowen v. American Hospital Association* and, in turn, led to the passage of The Child Abuse Prevention and Treatment Act Amendments of 1984.

The U.S. Supreme Court rules in *Irving Independent School District v. Tatro* that those school districts are required under the Education for All Handicapped Children Act of 1975 to allow a school nurse or an aide to perform intermittent catheterization as a "related service to a disabled student." School districts can no longer refuse to educate a disabled child because they might need such a service.

1985: The U.S. Supreme Court rules in *Burlington School Committee v. Department of Education* that public schools must pay expenses of disabled children enrolled in private programs during litigation under the Education for All Handicapped Children Act of 1975 if the court rules such placement is needed for the child to receive education in the least restrictive environment.

1988: The U.S. Supreme Court in *Honing v. Doe* affirms the "stay-put rule" established under the Education for All Handicapped Children Act of 1975, under which school authorities cannot expel, suspend, or otherwise move disabled children from the setting agreed upon in the child's Individual Education Program (IEP) without a due-process hearing.

The National Parent Network on Disabilities was established as an umbrella organization for the Parent and Training Information Centers.

1990–present: The Americans with Disabilities Act is signed by President George H.W. Bush and witnessed by thousands of disability rights activists. The law is the most sweeping disability rights legislation in United States history, giving people with disabilities full legal privileges. This law mandates that local, state, and federal programs become accessible; that businesses with more than 15 employees make "reasonable accommodations" for disabled workers; and that public areas such as restaurants and stores make "reasonable modifications" to ensure access for all disabled citizens. Finally, this act also mandated access to public transportation and communication.

The Education for All Handicapped Children Act is amended and renamed the Individuals with Disabilities Act (IDEA).

The final federal appeals court ruling in *Holland v. Sacramento City Unified School District* affirms the right of disabled children to attend public school classes with nondisabled children. The ruling is a major victory in the ongoing effort to ensure enforcement of the IDEA.

PEARLS AND PERILS

- Start where the family is. Allow them time and space to integrate the diagnosis or situation.
- Look to the family's strengths. Allow them to show you what works well.
- Never say "I know how you feel." Each individual experience is different.
- Set limits and be consistent. Families need to know that someone is in charge and is an expert. They rely on that when they are confused or overwhelmed.
- There is nothing that kindness and compassion can't help. Even in the worst situation, a family will appreciate it if kindness was demonstrated to their child.

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