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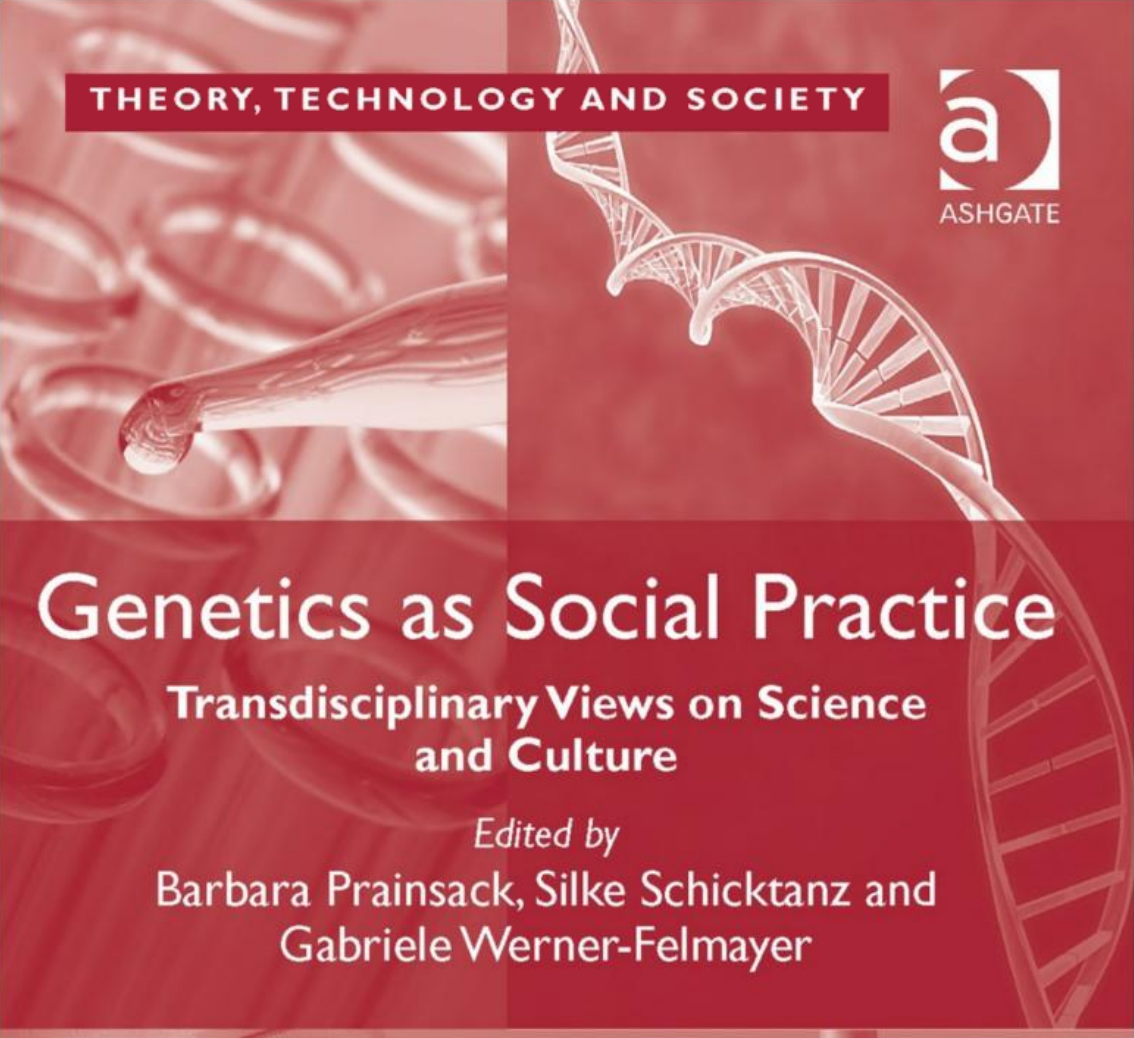

Genetics as Social Practice

Transdisciplinary Views on Science and Culture

Edited by

Barbara Prainsack, Silke Schicktanz and Gabriele Werner-Felmayer

GENETICS AS SOCIAL PRACTICE

Theory, Technology and Society

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Published by
Ashgate Publishing Limited
Wey Court East
Union Road
Farnham
Surrey, GU9 7PT
England

Ashgate Publishing Company
110 Cherry Street
Suite 3-1
Burlington, VT 05401-3818
USA

www.ashgate.com

British Library Cataloguing in Publication Data
A catalogue record for this book is available from the British Library

The Library of Congress has cataloged the printed edition as follows:
Prainsack, Barbara.
Genetics as Social Practice: Transdisciplinary Views on Science and Culture / by Barbara Prainsack, Silke Schicktanz, Gabriele Werner-Felmayer.
pages cm.—(Theory, Technology and Society)
Includes bibliographical references and index.
ISBN 978-1-4094-5548-6 (hardback: alk. paper)—ISBN 978-1-4094-5549-3 (ebook)—ISBN 978-1-4724-0718-4 (epub)
1. Genetics—Social aspects. 2. Genomics—Social aspects. 3. Sociobiology. 4. Science and civilization. I. Title.
QH438.7.P73 2014
304.5—dc23

2013033634

ISBN 9781409455486 (hbk)
ISBN 9781409455493 (ebk – PDF)
ISBN 9781472407184 (ebk – ePUB)

Contents

List of Tables and Figures

Tables

Figures

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Friederike Kogel received her MD at University Medical Center Göttingen, Germany, in 2013. Since 2011 she has been working on her PhD research in the field of medical genetics under the supervision of Silke Schicktanz. Her major research focus is on ethical issues of genetic susceptibility tests for Alzheimer's Disease.

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Barbara Prainsack is Reader (Associate Professor) in the Department of Social Science, Health and Medicine at King's College London. Her work focuses on the societal, regulatory and ethical dimensions of bioscience. In particular, she is interested in the ways in which science, politics and religion mutually constitute each other, and how they interact with understandings of humanness, corporality, personhood, and citizenship.

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Silke Schicktanz is Professor in the Department of Medical Ethics and History of Medicine at the University Medical Center Göttingen, Germany. Her research is located at the intersection of normative bioethics and socio-empirical studies of medical practice, with a strong emphasis on cross-cultural comparative work. Recent research topics include ethical and social studies of ageing medicine and predictive biomarkers as well as the question of how concepts of responsibility and autonomy can provide a link between lay people's morality and professional bioethics.

Sigrid Sterckx is Professor of Ethics at Ghent University and at the Vrije Universiteit Brussel (VUB) in Belgium. She gives courses in Theoretical Ethics, Methods in Ethics, Global Ethics, and Environmental Ethics. Her current research focuses on ethical aspects of biobanking, organ transplantation and patenting of genes and living organisms; human enhancement; medical decision-making at the end of life; and environmental justice. Her most recent book on patents, with Julian Cockbain, is *Exclusions from Patentability – How far has the European Patent Office eroded Boundaries?* (Cambridge University Press, 2012).

Paul Vanouse is an artist who has been working in emerging technological media forms since 1990. He is a Professor of Visual Studies at the University at Buffalo. His recent projects, 'Latent Figure Protocol', 'Ocular Revision' and 'Suspect Inversion Center', use molecular biology techniques to challenge genome-hype and to confront issues surrounding DNA fingerprinting.

Gabriele Werner-Felmayer is Associate Professor for Medical Biochemistry in the Division of Biological Chemistry, Biocentre, at Medical University Innsbruck in Austria. Her work focuses on concepts of hope and promise in modern biomedicine and how these affect notions of consciousness, personhood and humanness. She also initiated the bioethics network *Ethucation*, the Austrian partner in the UNESCO Chair's in Bioethics (IL) International Network, which aims to improve bioethics education.

Johannes Zschocke is head of the Division of Human Genetics and of the Medical Center of Genetics at Innsbruck Medical University, Austria. He specialises in inherited metabolic disease and the genetic basis of developmental disorders.

Foreword

The reflection about what and who we are is part of what makes us human. At present, science-based scholarship plays an important role in this endeavour. Throughout history, the scientific exploration of human diseases has provided important theoretical insights into the dynamics and the material bases of life. Genetics emerged during the second half of the nineteenth century as a field of expertise that played an increasingly important role for understanding these processes, but also for shaping notions of individuality and identity outside the realm of science and medicine. By now, we live in an era where most aspects of personality, physiology and pathology seem to have a genetic dimension.

A brief look at how understanding diseases has changed over time illustrates the extent to which genetics has altered our perspectives. In the nineteenth century, cellular pathology led to an understanding of human bodies in terms of specialised organ functions. The idea that traits have a physical endogenous basis, which may play a role in pathogenic mechanisms, had not yet emerged. It was only in the first decade of the twentieth century that William Bateson rediscovered the Mendelian laws, and Theodor Boveri and Walter Sutton developed the chromosomal theory of the cell. When Archibald Garrod finally introduced the concepts of chemical individuality and inborn errors of metabolism, the concept of hereditary factors entered medical science. Even then, it remained a mystery – for many more decades to come – how (and to what extent) genetics influenced bodily functions. For a long time, known links between genetic alterations and disease were largely descriptive and restricted to large microscopically visible chromosomal aberrations. Only few monogenic diseases correlated clearly with aberrations at the level of DNA. For most complex diseases, as well as personal traits and other phenotypic features, a molecular basis remained totally enigmatic.

Over the last 25 years, this has changed fundamentally. The emergence of genomics and bioinformatics along with a rapid progress of technology has transformed medicine as well as biology at remarkable speed. Consequently, the focus of understanding human biology in the context of health and disease has shifted from a few hundred different cell types to many ten thousand different molecules and their complex interactions. We have started to recognise the huge spectrum of genetic variants and their possible impact. Moreover, studying the human genome at its molecular level has challenged numerous traditional notions related to the definition of genes, and to mechanisms of gene expression and regulatory circuits. It underlined the non-linear complexity of the genotype-phenotype relationship. We have now entered a phase in which we are starting to appreciate this complexity. While on the one hand we may identify associations of certain genetic variations with certain phenotypes (mostly in so-called monogenic diseases), we had to learn that they are of limited significance for diagnosis and

prediction regarding complex conditions. Problems with the characterisation of phenotypes are one reason for this; insufficiently characterised phenotypes lead to inflations of genotype-phenotype correlations.

It is plausible to assume, however, that predictive genetic testing in the context of multi-factorial conditions will become more robust along with an improved understanding of the aforementioned complexity and the integration of genomic with other data. This will also affect practices of generating, communicating, and receiving genetic risk information.

In the context of some well-studied monogenic diseases, we already have considerable experience with predictive genetic testing. The most dramatic example of this is Huntington's disease, an autosomal dominant neurodegenerative disease inherited with a 50 per cent probability from an affected parent, for which no curative treatment is available. Predictive genetic tests have also become routine for familial diseases such as particular hereditary cancer syndromes, cardiac disorders, and others. For most conditions, the prognosis is not as bleak as in Huntington's disease, and often there are effective treatment options. Many of these tests are effective tools of preventive medicine, and health insurers cover their costs. However, there are also an increasing number of tests for variants of little predictive power and clinical utility, and new sequencing technologies will lead to the identification of many novel genetic variants with very unclear meaning. Furthermore, it is now possible to test for presence of recessive disease mutations that have no clinical relevance for the person but indicate an increased risk of monogenic diseases in the person's offspring.

The increasing availability of genetics for personal use comes along with a number of challenging questions: How can we learn to integrate these types of information from genetic tests into our own lives? How does genetic knowledge affect our 'naïve' self-image of who we are and what we can achieve? Does it increase or decrease our personal freedom to know genetic factors that may influence our future? Moreover, how would we deal with the possibility to select for children who were 'free' from any known genetic risk factors? Will this affect the attitude of our society to persons with disabilities and special needs? Indeed, are we intelligent and knowledgeable enough to really understand what we are doing?

One of the two maxims on the Apollon temple in Delphi was γνῶθι σεαυτόν – know thyself: the art of prediction is not to tell the future, but to find ways how to deal with it. Oedipus had to learn this the hard way. The oracle correctly predicted the future twice. The wrong conclusions were reached twice, with dramatic consequences. Genetic tests have the potential of resembling an oracle in that they are difficult to understand and may influence how we think, feel and act in unexpected and maybe irrational ways. Truly helpful guidance in the rapidly developing field of human genetics, both for individuals and the society, can only be provided through interdisciplinary discourse.

The multi- and interdisciplinary dialogue represented in this book began with a Symposium entitled 'Genetics as Culture in a Consumerist Age' organised by

the editors of this volume in Innsbruck, Austria, in October 2011. Since the initial meeting in Innsbruck, the social, regulatory and ethical dimensions of genetics and genomics have become even more salient also in public debates. This edited volume is thus a highly timely contribution to both the scholarly and the public debate. The breadth of information and diverse standpoints collated in this volume will be of interest and use to a large number of people in various disciplines and contexts of practice.

Johannes Zschocke
Innsbruck, May 2013

Acknowledgements

This book is the result of a longer dialogue on genomics in the twenty-first century between a group of colleagues from different disciplines, including social sciences, philosophy, biology and bioethics. The concrete beginnings of the book date back to October 2011, when the three editors of this book organised a symposium on 'Genetics as Culture in a Consumerist Age' in Innsbruck, Austria. Participants from 12 countries and from a diverse range of backgrounds – including anthropology, philosophy, ethics, genetics, epidemiology, sociology, political sciences, law, and visual and performing arts – contributed to this meeting. We would like to thank all of them for their enthusiastic, timely, and at the same time critical contributions and for sharing their work with us. We are also indebted to numerous colleagues, institutions and sponsors who supported the meeting financially: Lukas Huber at the Biocenter, Medical University of Innsbruck (and the SFB021 as well as Oncotyrol); Barbara Tasser at the Center for Italian Studies, and Ursula Moser, Center for Canadian Studies, both from the University of Innsbruck; the cultural departments of the municipalities of Innsbruck, Imst, and of the federal state government of Tyrol; Anna Ifkovits Horner at the Swiss Embassy in Austria; as well as our two cooperation partners Josef Quitterer at the University of Innsbruck, and Siegfried Walch at the Management Center Innsbruck. Without their great help the symposium could not have taken place.

We would like to express our sincere gratitude to all contributors to this book; we hope to continue our mutual exchange in the future. Sincere thanks go also to Claire Jarvis and Kirsten Giebutowski, our editors at Ashgate, for their support and guidance; it has been wonderful to work with them.

Barbara Prainsack, Silke Schicktanz and Gabriele Werner-Felmayer
London, Göttingen and Innsbruck, May 2013

Chapter 1
Geneticising Life: A Collective Endeavour and its Challenges

Barbara Prainsack, Silke Schicktanz and Gabriele Werner-Felmayer

Since biology has started to become molecular in the 1950s, two scientific icons have kept catching the collective imagination: the DNA double helix, and the gene (Anker and Nelkin 2003; Nelkin and Lindee 2004). No other scientific project has ever received as much attention from mass media as the Human Genome Project (Rödder 2009; Schäfer 2009). More recently, with the emergence of high-throughput technologies and the commercialisation of sequencing services, genes and genomes have moved even further into the centre of public debates – and with them, the promises of individualised medicine. Virtually any scientific and cultural concept pertaining to health and disease, or to family, ancestry, identity and personality, now has a genetic dimension. In addition, the extension of genetics beyond the clinical realm into other scientific disciplines and into the public arena goes hand-in-hand with changing ways of sharing and participating in research. These changes challenge some of the core categories and dichotomies of disease research and health care (see below). They also foster the convergence of different traditions and technologies, and they raise new ethical and regulatory challenges in the context of privacy, consent and ownership, just to name a few.

The aim of this book is to explore how genetics and society are shaped by technology, cultural beliefs, and current social practices. Voices from various disciplines such as sociology, ethics, philosophy, biomedicine, anthropology and the arts, aim at a multi-faceted reflection on promises and pitfalls, hopes and fears, as well as categories of personal freedom, autonomy and responsibility in times of personal genetics. Contributions to this volume speak to three main themes: creating identities (Part I), sharing knowledge (Part II), and participating in the social laboratory (Part III).

Part I is opened by Jennifer Fishman and Michelle McGowan's insightful discussion of whether and how the relatively new field of personal genome testing has been altering individual identities (Chapter 2). While much of the bioethical and social scientific literature assumes that, for better or worse, these tests will be transformative for those who use them, the authors argue that test users' perspectives do not as yet corroborate these claims. Josef Quitterer (Chapter 3) addresses a similar question, yet from a philosophical perspective: he argues that a distinction between 'weak' and 'strong' concepts of self enables us to give a more nuanced answer to the question of how concepts of self and individual identity

have been affected by the shift from clinical to post-clinical genetics. Troy Duster, in his chapter on ancestry testing (Chapter 4), subsequently sheds light on the social dimensions of a form of genetic testing that is not only located entirely outside of the clinic, but also has no immediate medical relevance. Via the claims that genetic ancestry testing entails about the test-takers' genetic heritage, however, it joins the ranks of genetic technologies that are involved in the articulation and production of identities. The latter is a role that Priska Gisler, Mo Diener and Luzia Hürzeler challenge in the final contribution to Part I (Chapter 5): by reflecting on narratives from visual imagery and other representations of kinship and relatedness, they look for answers to questions about ancestry in a manner that deliberately bypasses the help of genetic technologies.

Part II of our book, entitled 'Sharing Knowledge', opens with Paul Vanouse's 'hack' of DNA technologies (Chapter 6). Vanouse's is a thought-provoking statement on the many different ways in which DNA can speak. Gabriele Werner-Felmayer (Chapter 7) then presents Con'Sequences, a reflection on multi-layered artwork relating to DNA as a carrier of information. She analyses how such an artistic engagement reveals the genetic codification of language against the backdrop of an ideology of hope and hype shaping scientific and social practices. Werner-Felmayer's chapter is followed by Sigrid Sterckx' and Julian Cockbain's discussion of intellectual property rights in the context of personal genetics (Chapter 8). The authors convincingly show how – despite wide agreement across many disciplines that genetics is, among other things, also a social practice – the implications of this have not reached patent law. Patent law has been virtually unaffected by the social and ethical debates on post-clinical genetics.

Part III of our book looks at different practices of social, moral, and political participation in the wider field of genetics. Barbara Prainsack's contribution discusses instances of 'citizen science' in genetics, and unpacks the notion of participation in this context (Chapter 9). Gisli Palsson then takes a closer look at the notion of labour specifically; he argues that the labour carried out by subscribers to personal genome testing services has remained largely unrecognised, and not sufficiently conceptualised also in the scholarly debate (Chapter 10). Aviad Raz, Nitzan Rimon-Zarfaty, Julia Inthorn and Silke Schicktanz then turn to yet another kind of 'work', namely the moral management of and dealing with genetic testing for late-onset diseases. Drawing on a German-Israeli comparison, they show how such labour of responsibilisation can take different forms depending on socio-cultural contexts (Chapter 11). This moral participation takes different forms, depending on socio-cultural contexts, as the comparison between data from Germany and Israel reveals. Our volume is concluded by Silke Schicktanz and Friederike Kogel's discussion of how the wish to know one's genetic risk for Alzheimer's disease must be seen in conjunction with considerations about kinship, belonging, and the notion of future-oriented responsibility. The authors argue that people must be protected against blame for 'bad health' in later years (Chapter 12).

As an introduction into this transdisciplinary collection, this first chapter will start with describing important recent developments in the field of genetics

and genomics. We will then turn to the role of complexity within the nature v. nurture debate as well as to relevant aspects of public perception of genetics and genomics. In the final section of this chapter we will discuss new challenges of socially and culturally embedded genetics and analyse limitations of existing ethical paradigms.

Setting the Scene: Biosciences in the Twenty-First Century

'Biology today is being transformed by an explosive growth of data emerging from laboratories worldwide' (NCBI at a Glance 2004a). This statement, prominently placed at the US National Center of Biotechnology Information (NCBI) website, echoes a common notion within the life sciences today. But how exactly does this alleged 'transformation' of biology affect professional and every day practices? And does it refer 'only' to the struggle of turning 'data tsunamis' (Lareau 2012) into meaningful information, or does the possibility of large-scale data generation and data-mining spur a larger transformation of how scientific knowledge is produced? Moreover, are common concepts of genetics and inheritance moving away from a reductionist variant of gene-centredness towards an embrace of the complexities of biology in a way that will empower us to live happily and healthy ever after, as was the visionary promise of genomic medicine[1] (Green et al. 2011)?

Let us go back in history a little bit: in 1988, the NCBI was founded as a division of the National Library of Medicine (NLM) at the US National Institutes of Health (NIH). The aim was, and still is, to apply computerised information processing methods to biomedical research (NCBI at a Glance 2004b). What began some 25 years ago as a repository for DNA sequences in GenBank, and for scientific publications in PubMed, soon became an essential tool for bioscientists and a major platform for making their work visible to the broader community (by providing data and publications to such collections). Also other institutions, such as the Wellcome Trust Sanger Institute in England, or the European Molecular Biology Laboratory (EMBL), provide open access to curated data of high quality. Several of these institutions work together, and they all share important features in common: first, the data in these systems and repositories come from the research community; second, the curation and interpretation of data takes place at these institutions but relies on user review and contribution; and third, these databases link genomic sequence data with additional information (like protein sequences and structures, sequence patterns, gene expression profiles in various tissues and cell types, or diseases). The Human Genome Project and the rapid progress in sequencing technologies (see Box 1.1) enabled the establishment of these essential tools and platforms.

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1 The term genomic medicine is often used as a synonym for molecular medicine. Personalised (or individualised) medicine is a more comprehensive endeavour aiming at the increasing consideration of individual differences in prevention, diagnosis, and treatment, in which genomics plays an important role (European Science Foundation 2012).

These developments have paved the way for what is now often referred to as 'big biology' and seen to hold the potential to change our understanding of genetics, inheritance, health and disease as well as evolution and taxonomy of species from persistent reductionist to more complex versions (see Boxes 1.2 and 1.3). 'Big biology' is characterised by joint efforts of large research consortia such as the Encyclopedia of DNA Elements (ENCODE) and several programmes devoted to technology development, bioinformatics, genome function and genome variations (see also Boxes 1.2 and 1.3).[2] One recent addition to this list is the Human Microbiome Project (HMP; hmpdacc.org). The concept of the superorganism, i.e., the human organism in combination with our microflora (our 'microbiome', also termed 'metagenome'), captures the new focus on interactions between the host genome and the microbiome in thinking about disease etiology (Genome Reference Consortium 2012; Human Microbiome Project Consortium 2012; Murdoch and Detsky 2012).

Numerous challenges still lie ahead. Some of them are technology-related. For example, the human reference genome produced at the beginning of the new millennium contains numerous sequencing errors (despite still being the highest quality assembly of a mammalian genome ever produced) (see Box 1.1). In addition, personal human genome sequences that may soon be available from personal genomics services for a reasonable price will require cost-intensive re-analysis for the purpose of finding out which variations are rare variants of the genome and which are sequencing errors (Mardis 2010).

Even more difficult to address is the challenge of translating advances in genomics research into clinical applications. One particular difficulty in this process is that the tools to study, and the concepts to understand the significance of genomic variants, rare or common (see e.g., in Offit 2011), are still under development (see also Boxes 1.2 and 1.3 for an overview of current perspectives on genome function and variation). Moreover, the need to negotiate increasing amounts of uncertainty about the clinical relevance of observed genomic variations is likely to complicate interactions between patients and physicians (Hessling and Schicktanz 2012).

Getting Together: Contexts of a More Integrated Science

A popular assumption is that 'big biology' with its data-drivenness and its inherent technology-pull is a novel phenomenon. There is an ongoing debate about whether data and machines really do change the scientific method (e.g., Leonelli 2012). This debate is underpinned by an assumed opposition between hypothesis-driven and data-driven research, again with the assumption that they are mutually exclusive (see e.g., Golub 2010; Weinberg 2010). There is antagonism between 'large-scale

2 A comprehensive overview of large-scale projects in genomics and how they shape the research landscape is provided by Kaye 2012, for example.

genomic studies' run by research consortia that 'will not be sufficient for gaining a fundamental understanding of biology' on the one hand, and research carried out by 'individual investigators' who are empowered by data catalogues 'to pursue more effective hypothesis-driven research' in order to achieve innovation on the other (Green et al. 2011: 207). Such portrayals of researchers as either data-hungry number crunchers or smart innovators, however, also reflect the bitter struggle of different camps for funding and priority. This may be interpreted as the 'noise' of academic life, but it also puts in perspective more idealistic characterisations of genome research as a process of unlimited, altruistic data generation and sharing, one of genomics' heroic narratives. Data sharing and pre-publication data release were hence identified as keys to success in terms of scientific productivity and public benefit (Toronto International Data Release Workshop Authors 2009). Moreover, data sharing sits uncomfortably with other public goods and individual rights such as intellectual property, data privacy and confidentiality (see Sterckx and Cockbain in this volume). This presents an ongoing challenge for governance systems aiming to protect research participants (e.g., Kaye 2012).

Despite this, much of the attractiveness of 'big biology' projects lies in the promise of a democratisation of the scientific endeavour itself, as science entails new possibilities for interdisciplinarity, multi-national collaboration, and also participation by non-professionals. The research community has been extended from professional experts to potentially everyone, and genetics is a field where this trend is particularly visible. Online genomics services enable everyone who is willing to contribute their genomic and lifestyle data to research to do so. OpenSNP is one such platform (Plagnol 2012) that offers consumers of online genetic and genomic testing services ways to 'publish their test results, find others with similar genetic variations, learn more about their results, find the latest primary literature on their variations and help scientists to find new associations'.[3]

Public contributions to science often entail that participants waive privacy and ownership rights for the sake of a joint public effort to increase genomic knowledge. The emerging concept of 'citizen science' (Prainsack, in this volume) is an instance where, in rich countries, the main obstacle to contribute to research is no longer seen in professional authority and financial resources, but in motivation and passion. Ethical and legal instruments to facilitate these collective efforts are under development; what is needed now is a critical assessment of what values and political or economic imperatives underpin these efforts (see Palsson; Raz et al.; and Schicktanz and Kogel, in this volume). Such a 'participatory turn' in the context of biomedicine, and instruments and concepts such as 'collective consent' (i.e., presumed consent by the public, as long as there is no open resistance), open or broad consent (e.g., Sheehan 2011, or solidarity (Prainsack and Buyx 2011) fit well

3 Quoted from *Welcome to* openSNP. Available at: http://opensnp.org [accessed 6 August 2012].

into the spirit of an era in which a hard-to-define common good[4] is increasingly expected to accommodate the needs of science, and the actual or alleged 'interests' of individuals (Rommetveit 2011).

Debates about the 'participatory turn' in science and medicine also resonate with intensifying debates about the need for higher levels of sharing and participating in collective actions in the realm of science (see below). The distinction between science and society is becoming even blurrier than before, as social scientists, lawyers, ethicists, 'lay' people, and representatives of media, popular culture and art, contribute to science in increasing numbers (see Gisler et al.; Vanouse; and Werner-Felmayer, in this volume). A closer look at attempts to render human genome research more transparent shows that this process has not been entirely 'voluntary' but it represents, at least partly, a strategic move to calm the public debate over DNA research (Koski 2005). One consequence has been an increasing popularisation (Schäfer 2009) of genomic and other biomedical large-scale research in print media and on television, and an increasing coverage of scientific news more generally. 'Big biology' projects are typically presented with powerful metaphors (see e.g., Marshall 2011), and the latter have, in turn, influenced scientific research: For example, the human genome was often framed as a code, a blueprint, or a scripture, and the ability to 'read' it was allegorised as enabling humanity to understand the 'book of life' (Kay 2000; Nerlich and Kidd 2005). More recently, metaphors like these have been complemented by a language that accommodates and highlights dynamic interactions. The Human Microbiome Project, and the embrace of the notion of the superorganism by both scientists and journalists, illustrate these dynamics (Nerlich and Hellsten 2009).[5]

Regardless of what metaphors are used, they always bring with them connotations from the respective fields where they originate. Ironically, it is eroding trust in institutions within these other fields that can then, via the metaphor, bleed into public debates about science.[6] The popularisation of science also entails that there are incentives for scientists to overstate the benefits of their research in order to obtain funding, a situation that has attracted critical attention from social scientists and ethicists (Holm 2007). As the case of embryonic stem cell research illustrates, this can create a vicious cycle where unrealistic promises almost inevitably lead to disappointment among politicians and the public. Also in human

4 However, the discussion about 'open consent' in the context of direct-to-consumer genetic tests also revealed that consumers often did not intend to agree that their personal data can lead to patentable innovations for test providers even though they signed a document of 'the kind one might quickly click past while installing software' (see Check Hayden 2012: 312).

5 One report in the New York Times for example referred to the 'self' as being a 'community property' of the body's microflora (Nerlich and Hellsten 2009: 29).

6 For example, the decreasing trust in banks amidst the ongoing economic crisis may well influence our understanding and trust in biobanks. The mixing of different dimensions of meaning, a by-product of the popularisation of science, can also affect core concepts and categories in scientific agendas and practices.

genome research, inflated hopes have been stirred, particularly in the context of genomics-based personalised medicine (Khoury et al. 2010). One issue of concern here is that the communication tends to simplify new insights. For example, new research on the numerous regulatory circuits of gene expression and hence the labyrinthine ways of inferring a phenotype from a known genotype (see Boxes 1.2 and 1.3) are rarely addressed in public media. A time-lag between gaining insights and introducing these new insights into general knowledge is a common epistemic phenomenon (Fleck 1980: 150). However, in genomics the issue is complicated by the apparent reluctance of molecular geneticists to discuss their own results if they challenge conventional theory. This situation has led to an 'unacknowledged crisis in biology' and 'a serious problem of public misinformation' (Commoner 2009: 29).

Genetics as Culture: Beyond the Nature v. Nurture Debate

'Big biology' is taking place against the backdrop of the increasing synthesis of some previously distinct categories: Oppositions such as nurture v. nature, professional v. lay, and individualist v. communitarian, are no longer active dichotomies in many areas of biology. The increasing ease with which categories, ontologies, and methods from previously distinct areas and disciplines are now travelling into new domains provides an important condition of possibility for further advances, and an important challenge for scientists, funders, and publics. For example, the nature v. nurture debate has been one of the main ideological and ontological dividing lines in life sciences and humanities/social sciences. Where one stands in the nature v. nurture debate used to have profound implications for one's perspective on the hierarchy between different scientific disciplines, and one's view of what role of public policy could and should play in reacting to, or otherwise shaping, the 'facts' that nature or society respectively was seen to create. Such deterministic world views bore the danger of becoming blind to anything that did not fit the dominant narrative. Behaviourism in psychology, neuro-determinism in neuroscience, and genetic determinism, are examples of this problem. Genetic determinism implies that genes are the immutable 'hard ware' of human beings that ultimately determine the boundaries of what is changeable by individual and social practice (for an overview of critical social science work see Lemke 2013). Although radical genetic determinism in its crude form has never been fully adopted in the modern life sciences (Plomin et al. 2003; Prainsack 2012), it continues to influence public media coverage and public discourse. This is perhaps also due to the important role that genetic determinism plays as a straw man to heat up media controversies. Related to this, but different in its emphasis, is the concept of genetic essentialism. Dar-Nimrod and Heine's (2011) definition of genetic essentialism is underpinned by the understanding that humans aim to see the essential characteristics of entities in order to give sense to a complex world. In other words, essentialism is a way to reduce complexity. This process,

however, is shaped by a variety of different factors including personal experience, essential needs, and cultural context. In the case of genetics, essentialism can lead to often unintentionally reductionist views about personal identity, traits or the aetiology of diseases. Such reductionist views are reinforced by the way genes and genetics appear in public discourses, and, while motivations and rationales that underlie genetic determinism and genetic essentialism might differ (Dar-Nimrod and Heine 2011), the outcome is often comparable. Overcoming such reductionist assumptions is one of the major challenges in the communication of human genomics research, and a serious educational issue. In the public realm, a rather black-and-white-picture still prevails: It is typically *either* the biological inheritance of a person (her genes, her 'blood') that is seen to cause a particular behaviour, *or* her social upbringing (see Duster, and Quitterer, in this volume, discussing aspects of this pertaining to ancestry and individual identity respectively).

More recently, however, a new approach to explaining both biological and social phenomena has emerged out of the relatively young field of epigenetics and system-based approaches. This new approach can best be described as *normative complexity*. It is normative because it disadvantages approaches that do not accommodate non-linearity; it rejects the application of linear causalities to living systems or organisms. For example, the growing popular science literature on epigenetics (e.g., see Carey 2012; Francis 2011; Spector 2012) promotes the idea that neither genetic nor extra-genetic factors can sufficiently explain (almost) any phenomenon, but that it is a complex interplay of the two that we have only begun to explore (see also Boxes 1.2 and 1.3). This commitment to normative complexity is not restricted to scientific discourses but it is entering public discourses as well. What these endorsements of normative complexity often fail to do, however, is to spell out how the new insights from epigenetics or systems biology change the very categories we still use to describe phenomena of nature and nurture. For example, what happens to the notion of the gene when, via epigenetic mechanisms, social environments and practices can become literally inscribed in the gene and the genetic is folded into the social (see Keller 2010)? The logic of popularisation in public media and popular science discourse, however, leads again to the simplification of explanations that were designed to accommodate complexity.

Your Spitting Image? Genetics Goes Online (and Personal)

The engagement of lay people with genetic and genomic data online provides a good example of how the question of genetic determinism vs. normative complexity creates new battlefields (see Fishman and McGowan in this volume). Internet companies offering genetic tests to consumers online have been subject of considerable criticism not only because the test results were seen as potentially harmful for test-takers, but also because they were seen to promote – or at least imply – a genetic determinist worldview. Moreover, as some studies on the level

of 'genetic literacy' suggest, many people are ill prepared to integrate genomic information into their health-related decision making (Kung and Gelbart 2012). The idea that genes play an important role in almost every behaviour, trait, or human phenomenon (i.e., the gene-centric stance) should, however, not be conflated with the assumption that genes are *the* most important, or sometimes even *the only* important element in explaining these phenomena.[7] Particularly the websites of genetic testing companies in the health domain embed DNA analysis as their central selling point in an enlarged space of discussing non-genetic and non-genomic factors, such as natural and social environments, and particularly lifestyle factors such as physical exercise and nutrition (see also Schicktanz and Kogel in this volume). By saying this, we do not mean to deny that services such as 23andMe can be seen as promoting a gene-centric approach to understanding health (Bartol 2012); here, we need more socio-empirical research on the attitudes and understandings of users and non-users of these services.

Members of health internet platforms that do not offer DNA testing but instead focus on patient support, like Patients Like Me (patientslikeme.com) often also share parts of their genetic and genomic information, either in discussion forums, or in order to integrate genetic information with other health-related data (e.g., curetogether.com). These platforms do not treat these genetic and genomic data as an isolated phenomenon, but they aim to integrate them with other kinds of information. Yet what is easily and often overlooked in the discussion about the risks and benefits of people's sharing of their genomic information is that it also contributes to a new way of modern social life. In personal genome testing, for example, playfulness and entertainment are important elements of the user interface (Vayena et al. 2012). The reliance on clinical utility in evaluating the usefulness of personal genetics thus neglects the point that the motivations of people engaging with these platforms may be broader than what clinical utility is able to capture, or different from clinical concerns altogether. This, hopefully, will be explored in wider empirical research with users of these services in the near future (see also Fishman and McGowan, in this volume). Apart from exploring the motivations of users it will be also important to look at some of the unintended consequences of their engagement. Companies like 23andMe, but also not-for-profit platforms revolving around patient-support and patient-self-help, engage their members in creating value for somebody else – for the common good, for perhaps problematic public health policies, and/or for the financial profit of enterprises and individuals. This needs to be kept in mind in light of the rhetoric about 'empowerment' and the 'democratisation of genomics' (see also Palsson, in this volume).

What we need to ask is whether there is a genuine devolution of power from elites to non-elites in this assumed process of increasing openness and democratisation. Of course, the rhetoric of consumer empowerment often serves as a tool to channel the *a priori* unaligned, divergent, and partly subversive objectives and health

7 See Commoner 2009 for a critique of popularizing this view through the Watson/Crick theory.

goals of individuals into ordered activities which render consumer participation valuable for a company. At the same time, the 'creation of one's self', offline and online, has become a pleasurable and entertaining activity for many people in late-modernity (see Giddens 1991), and it is interwoven with dominant patterns of consumption. Such patterns of consumption play an increasingly important role in the production and reception of health information, and in its incorporation in the self-conception of citizens. The relevance of this arguably extends to the larger field of interactive and participatory health services on the Internet, as the notion of consumer value co-creation has been an intrinsic feature of the Web 2.0 era.

Looking Ahead: Moving to a New Research Agenda

The new paradigm of a more complex and a more socially embedded genetics leads us to a critical reflection of existing values, norms and ethical standards. Are we sufficiently equipped, conceptually and methodologically, to deal with these new challenges of scientific complexity, context sensitivity and participatory ideals? The development of new paradigms and ethical approaches in genomics that interact with society and culture at so many, scientific as well socio-political, levels, requires an understanding of the limitations of existing scientific, political, social, and ethical paradigms. With 'ethics' we refer to the discursive formation of guiding principles and critical reflections upon the mutual relationship between genetics and life science on the one hand, and individual and social practices on the other. Ethics should serve as a tool to help us deal with scientific and moral uncertainties in a complex and reflexive modernity (see also Nowotny et al. 2001). Such an understanding of – applied – ethics favours a combination of scholarly work on moral philosophy including adjacent areas *and* work on publicly shared values or moral opinions. Hence, the way that people adopt, interpret or criticise genetics is an important source for a new ethical agenda that acknowledges their participation not only as consumers or research participants, but also as a normative authority in a democratic system.[8]

Academic ethics in particular has, in a joint endeavour with Western and international law, predominantly focused on *one* ethical principle to guide and govern genetics so far, namely autonomy (e.g., Chadwick et al. 1999; Fox and Swazey 2008; Sherwin 2011; Widdows 2011). In reference to autonomy, the 'right

8 Critics of 'bioethics' have rightly pointed to areas where it has been reduced to a tokenistic thetoric to foster and promote life science instead of providing critical input (Rabinow 2003). This concern should be taken seriously, but it is important to keep in mind that bioethics exists in many forms and guises, and that especially also applied bioethics offers much more. Another main criticism of bioethics has been its expertocratic practice and its formalistic, abstract approach (Evans 2012). Because of this, so the argument continues, bioethics ignores the everyday way in which people are affected. To overcome such an expertocratic stance, various approaches in ethics argue for the systematic integration of lay and affected people's perspectives (Schicktanz et al. 2011).

to know' as well as 'the right not to know' articulate corresponding professional and social duties in dealing with genetic information. The right to know includes the professional ethos to inform patients of the purpose, alternatives, results and possible consequences of genetic diagnosis (e.g., in the case of prenatal testing, of the arising question about abortion), while the right not to know refers to professional and societal duties to protect the privacy of patients, relatives, or particular vulnerable groups, such as children. According to this right, people should not be confronted with information on their genetic risk if they do not want to know. None of these rights were a matter of course, apart from a very basic understanding of informed consent, until 1997, when the European Convention on Human Rights and Biomedicine and the UNESCO Universal Declaration on the Human Genome and Human Rights adopted both (see Andorno 2004). This late development of even a right not to know might mirror societal and political difficulties in many industrial countries in acknowledging their history of explicit eugenics (such as forced sterilisation) or the recent practice of so called neo-eugenics (by means of implicit social pressures on women in prenatal testing). While the right to know can be seen as biopolitical form of self-governance, the later addition of a right not to know opened spaces for resistance against eugenic and biopolitically motivated self-governance. This is not ethically unproblematic either. Researchers sometimes decide unilaterally not to share new information with patients, referring to the latter's right not to know, yet without actually asking patients whether they wanted to exercise this right in that instance.

Whether an ethics that remains focused on autonomy is a suitable tool to harness the complexity of our personal and societal engagements with genetics, and to critically scrutinise the assumed societal benefits of genetics in the twenty-first century, is questionable. The shortcomings of autonomy-, choice- and interest-driven discourses can be summarised as follows: The 'geneticisation' (Árnason and Hjorleifsson 2007; Lippmann 1991) of our understanding of health and illness, even of the conception of and relation between members of family (Featherstone et al. 2006) or society have created a web of internalised or anticipated responsibilities towards oneself, towards our loved ones, towards society as a whole, and towards future generations. The term 'responsibility' challenges traditional meanings of ascribing guilt and blaming by adding more forward-oriented forms of care, solidarity and acknowledging power asymmetries. It addresses health professionals, individual patients, as well as whole social groups. And it cannot be done justice by a solely legalistic or patient-centred concept of autonomy and individual professional duties. Moreover, genetics and medicine nowadays take place outside of the clinic as well as within. It takes place in families, patient groups, state organisations, on the Internet, and on the international market (see Schicktanz and Kogel, in this volume).

In a 'traditional' understanding of geneticisation, people use genetic explanations to make sense of personal misfortunes, difficulties, identities, based on the idea of 'genetic' relationships. As empirical studies suggest, however, genetic explanations do not redefine kinship relations. Instead, existential

phenomena such as shame, love or responsibility that are already embedded in these relationships are 'enriched' by genetic meaning (see also Featherstone et al. 2006). This explains many cases where parents feel guilty when they are told that they carry genetic mutations that caused clinical symptoms in their children. The genetic dimension here does not introduce the category of guilt into the relationship between the parent and the child; most parents feel responsible for their children in many ways, and they feel guilt when they sense that their responsibility as a parent is compromised in any way. What genetic information does, here, is to provide a vehicle for the expression of a particular feeling rather than introduce the feeling. It renders the relation between the parent and the child even more complex, as it adds assumptions of 'causality' (i.e., the genetic mutation has caused the suffering of the child). The result can be that parents are blamed for having children with disabilities, insinuating that 'there is no need for such a thing to happen in this day and age'.

Another manifestation of geneticisation can be found in new partnerships between patient advocacy groups and genetic researchers (Rabeharisoa 2003). Parents with children suffering from severe rare genetic disorders have created new partnerships with life science research by sharing genetic material, personal data and participating in research (e.g., Genetic Alliance, geneticalliance.org; or EURODIS, eurordis.org). Some of these partnerships are not only motivated to develop future therapies, but also to eradicate those diseases altogether. An example is the recent market introduction of a gene chip for pre-conceptional genetic testing. In 2011, it covered over 450 rare diseases, but it is constantly further developed and aims to cover already 1200 rare diseases by 2013. Its development is heavily supported by some patient advocacy groups (Bell et al. 2011). These new forms of partnerships differ from those social movements that regard themselves as emancipatory from science or more constituency-based health care movements (Brown et al. 2004). The new collective endeavours as well as new categories of aims (such as pre-conception genetic testing or pre-implantation diagnosis), mark a new area of the geneticisation of individual and social life, perhaps a new form of socially induced eugenics, not yet sufficiently explored or even recognised.

But it is also important to understand the new dimension of lay and patients' involvement in such projects. The case of rare disease research shows new possibilities for patients and affected persons to take part in the governance of the academic-industry-complex, especially in those areas often neglected by politics and pharmaceutical industry (Wehling 2011). Hence, patients and citizens can obtain more agency, beyond merely deciding whether to know or not to know. Any ethical approach suitable to address the needs of genetics in the twenty-first century must empower patients, as well as their families, friends, and care givers, to take active parts in decision making beyond merely choosing between different options presented to them. They need to be able to participate in formulating the options themselves. Reflections of the opportunities and their risks and social implications, however, are still very rare (see Schicktanz and Kogel, in this volume).

Ethical reflections concerning the social implications, beyond the individual and her family, can be subsumed under the term of 'social responsibility'. In this broad understanding, social responsibility considerations entail forward-oriented normative claims based upon duties, rights but also care and commitment (for details see Schicktanz and Schweda 2012). Different agents and subjects are involved, e.g., the state and public actors, as well as the objects, e.g., social groups or professionals, of a responsible behaviour. In the particular context of genetic testing in order to possibly reduce disease risk the question arises how the above mentioned normative claims could be dealt with by these different parties.

Considerations of social responsibility are accompanied by a revived debate of the role of solidarity instead of the former domination of individual autonomy and informed consent. This trend might be seen as a broader turn or reconsideration of communitarian values and the politicisation of ethics, and it goes hand in hand with a shift from individual ethics to collective ethics in the field of genetics more generally (Raz 2009, 2010).

The concept of solidarity, while less at home in Anglo-American discourse, still plays a crucial role in European political traditions, and assumes an important place also in Latin and African socio-political movements. It can be defined as shared costs and duties between (two or more) individuals or members of a community, depending on a recognition of a similarity in a relevant respect based upon a common choice or fate (see Prainsack and Buyx 2011, 2012). It underpins many social welfare and public health care systems. Especially the socio-political meaning of the term adds an important dimension to the recent neoliberal discourses which tend to blend out social cohesion and a public awareness of the vulnerability of others. Solidarity, however, should not be understood as ethical principle that always trumps autonomy and self-determination. Similar to the Musketeer[9] slogan, 'one for all, all for one' it articulates a basic commitment of the members of a group towards the wellbeing of each other (see also Wildt 1995), often based upon a collective identity. In this sense, as an ethical principle it may be helpful to distinguish it from altruism on the one hand (as the bearing of costs or motivation to act in a supererogatory way, and also because solidarity, in contrast to altruism, entails both self- and other-directedness) and from justice on the other (see Bayertz 1998). However, solidarity can be understood as a notion that links the abstract principle of justice or fairness to reciprocal recognition of needs (or interests and rights) and thus helps to render concrete the just allocation of goods in a particular group (e.g., between and within generations).

A solidarity-based discussion requires a critical and reflective perspective on ongoing debates about values in health care and research practice. The concept of solidarity, on the one hand, helps to understand why many patients and citizens share and participate in genetic research, even when they are fully aware that they may not personally benefit in an immediate or direct manner. The renewed

9 After the French novelist Alexandre Dumas and his world famous book *The Three Musketeers*.

currency of the term, however, also calls for a critical perspective on how solidarity is 'motivated' in specific concrete cases, and whether the underlying conditions are fair, transparent, efficient etc. For instance, one might ask what kind of information, or even what kind of hope-and-hype-rhetorics, were used to motivate patients to contribute samples and data to biobanks. Solidarity can also help us in framing particular concerns regarding patenting and commercialisation of genes and biobanks, if these developments are likely to restrict and limit their use for vulnerable groups. Finally, it is also crucial to scrutinise problematic rhetoric in which, in the name of solidarity, citizen's rights for bodily integrity or self-determination are undermined. Such a critical case would be where donations of tissue or organs are justified by presumed consent, against all empirical evidence that the majority does not agree, but in the name of 'solidarity'.

Conclusion

'Big biology', genetic and genomic research is currently transforming into a collective endeavour that challenges some of our social, cultural and ethical equipment developed particularly in the last three decades of the twentieth century. Its most distinguishing characteristics are complexity, multi-data-driven technologies, close collaborations between professional scientists, patients, and 'lay' people, and an attention by media and art that has challenged the self-understanding of the research community itself. Genetics in the twenty-first century would benefit from an inspired debate that accommodates, and provides ways to understand and harness the complexities inherent in health and disease and the dynamics of underlying research results and concepts. Such a debate must be open to contributions from different professional and practical contexts. BioArt, for example, provides a fruitful arena to address questions, concerns and alternative understandings beyond the analytical constraints of science, humanities and social science. Empirically informed ethics as well as social sciences that are engaged with societal and political developments will equip us well to investigate interactions between the moral, social and political dimensions of genetics and genomics, still too often neglected.

Box 1.1 The Human Genome and Sequencing Technology

Human Genome Project: The official duration of this multi-national endeavor, which is sometimes referred to as archetypical large-scale project in biomedicine, was from 1990 to 2003 (more information at: http://www.genome.gov/10001772). 'Draft' sequences were published in 2000, a first 'finished' version, i.e., a near-complete sequence of 2,851,330,913 (or 2.85 billion) base pairs, corresponding to ~ 99 per cent of the haploid nuclear genome was published in 2004 (International Human Genome Sequencing Consortium 2004). The sequence is a consensus sequence and does not represent an individual person's genome as DNA from 'about 13' people was used (Nature Methods 2010). Due to high structural variability of individuals in certain regions such as deletions, insertions, repetitions or inversions, there are still gaps, assembly errors and other uncertainties in the sequence (Dolgin 2009).

Human Reference Genome: Besides further polishing the human genome sequence, a major aim of the Genome Reference Consortium (CRG) is to include alternative assemblies from regions of high variability in order to represent genomic variation and to provide a more robust reference for genome analyses. The currently available reference assembly GRCh37.p8 consists of 3,190 (XX sex chromosomes) and 3,094 (XY sex chromosomes) billion base pairs[1] and contains 357 gaps.[2] Comparison of the reference sequence with individual genome sequences from the 1000 Genomes Project identified some 27,000 divergent bases awaiting clarification whether they are 'erroneous' or 'rare' bases (Genome Reference Consortium 2012).

Personal Genomes: The first personal genomes to be published were that of Craig Venter in 2007 using first generation technology (see below) and that of James Watson in 2008 using second generation technology (Wadman 2008). Unlike in the Human Genome Project these personal analyses used diploid genomes, i.e., DNA from both sets (one inherited from each parent) of the 23 chromosome pairs were sequenced giving important clues on genome variability. Genome variability (see Box 1.3) is an increasing field of investigation. Also worth mentioning in this context is the Personal Genome Project (PGP) at Harvard Medical School (http://www.personalgenomes.org), which uses the genomic, lifestyle-related, and other relevant information of volunteers to advance personal genomics.

First Generation Sequencing: Methods are based on a preparatory and analytical procedure termed Sanger method as used in the Human Genome Project. As of 2001, fragments of 500 to 600 bases could be sequenced per run, yielding about 115,000 base pairs per day. Sequence production, and not analysis, was rate-limiting (Mardis 2011).

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1 Assembly statistics for the current human reference genome GRCh37.p8 is available online at: http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/human/data/index.shtml [accessed: 28 June 2012].

2 http://www.ncbi.nlm.nih.gov/projects/genome/assembly/grc/data.shtml [accessed: 29 June 2012].

Next Generation Sequencing (also referred to as second or third generation sequencing): Since 2005, the capacity of sequencing technology has increased by several orders of magnitude. This was achieved by developing so-called massively parallel devices and various preparatory and analytical methods essentially different from the Sanger-based instruments. Data analysis became much more complex as compared to first generation sequencing due to the amount and different quality of data generated by novel sequencing platforms (Mardis 2011).

Sequencing Costs: Sequencing costs per whole human genome declined steadily from estimated 100 million USD in 2001 to about five million USD by the end of 2007. This development accelerated rapidly during the past five years down to less than 10,000 USD in 2012 and was mainly caused by switching from first to second generation sequencing technology. The costs for Craig Venter's genome were 100 million USD whereas those of James Watson's genome were less than 1.5 million USD (Wetterstrand 2012).

Sequencing Coverage and Depth: Coverage and depth refer to the likelihood that a certain base on the genome was identified in the sequencing process, and thus to the quality of the sequence. Deep sequencing coverage is a prerequisite for detecting genomic variants, for correcting sequencing errors and for including both chromosomes of a person's genome. With current platforms, a person's DNA has to be sequenced about 28 times (or 28X) in order to enable the assessment of the entire genome (Mardis 2011).

Box 1.2 Genes, Genomes and Others

Gene: The term 'gene' was coined by Wilhelm Johannsen in 1909 for the 'special conditions, foundations and determiners […] [by which] many characteristics of the organism are specified' (cited in Gerstein et al. 2007: 669).[1] The common perception of genes is that they are entities containing hereditary information, or more specifically, that they are DNA stretches that code for proteins. This notion is challenged by current research as being too simplistic (Gerstein et al. 2007, see also below).

Genetics: Genetics can be defined as the science of protein-coding genes in the context of heredity. However, in a genomic era this definition is increasingly inadequate, as the concept of gene has to be broadened (see below).

Genomics: Genomics may be seen as a subfield of genetics studying the genome in its entirety instead of single genes. Genomics is not confined to what we define as genetic information encoded by an iconic double helix, but it also deals essentially with structure and hence function of the three-dimensional macromolecular DNA packed in our chromosomes as the genome is a highly dynamic molecule.

1 A timeline on the gene as evolving concept is given in Gerstein et al. 2007.

A Changing Definition of Gene in the Era of Genomics: Less than 1.5 per cent of the human genome sequence encode for proteins (about 20000). Finding out functions of the rest of the sequence was and still is a major focus of genomics research. In 2003 the ENCyclopedia of DNA Elements (ENCODE) project was launched, aiming to 'identify all functional elements in the human genome sequence' (ENCODE Project Consortium 2004: 636). The project initially focused on 1 per cent of the sequence scattered over 44 discrete regions. Since then, the concept of the gene has turned 'into something complex and elusive', and in 2007, at the end of ENCODE's pilot phase, it was concluded that a 'gene is a union of genomic sequences encoding a coherent set of potentially overlapping functional products' (Gerstein et al. 2007: 669). These functional products do not only comprise protein-coding genes but a large number of RNA transcripts with various regulatory functions spread throughout the genome sequence (ENCODE Project Consortium 2011). Some of them are defined as non-coding genes which amounts to a substantial change in the definition of gene. A major challenge of covering all functional elements of the human genome is that many of these functions are expressed in a cell-type-specific and/or developmentally differential way thus increasing the diversity of individual gene expression. Recent advances by ENCODE, however, allowed to assign at least one biochemical function for about 80 per cent of the genome sequence (ENCODE Consortium 2012).[2]

Exons, Exome and Genomic 'Dark Matter': Exons are genomic sequences which are transcribed into messenger RNA (mRNA). The exome comprises the entirety of exons. If applying the term only to protein-coding genes the human exome consists of about 180,000 exons covering about 1 per cent of the genome (S.B. Ng et al. 2009). Various large-scale projects have recently suggested, however, that not only such a small fraction, but most of the genome sequence can be transcribed and thus contributes to the RNA pool of a cell type or tissue. Transcribed genomic sequence that cannot be annotated to known exons is sometimes termed genomic 'dark matter'. The respective RNA transcripts are called 'dark matter' RNA. Increasing evidence indicates that aberrant expression of 'dark matter' transcripts is associated with certain cancers and neurological disorders (reviewed by Kapranov and St. Laurent 2012). The authors conclude that it would therefore 'not be surprising if 'dark matter' transcripts would eventually occupy a central place in our conceptual understanding of the molecular events underlying human development and disease' (Kapranov and St. Laurent 2012: 6).

Transcriptome: The term refers to RNA transcripts as a whole. As gene expression varies among cell types and at different stages of development, an individual's transcriptomes are much more diverse than her underlying genome. In addition to coding RNA transcripts, long non-coding RNAs (lncRNAs) were identified that regulate and fine-tune gene structure and expression, a process called 'RNA-directed epigenetic control'. It was shown that such processes are particularly important in the brain suggesting that 'long-held ideas of gene regulation in development and cognition will have to be reassessed' (Mattick 2012: 516).

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2 Since 6 September 2012, *Nature* has provided an interactive information tool for ENCODE in order to help readers make sense of the 30 papers ENCODE published so far. Available at: www.nature.com/encode/#/threads.

Epigenetics and Epigenome: Epigenetic modifications, like e.g., methylation of DNA or histone modification, influence gene expression and hence phenotypes. One prominent example is genomic imprinting, i.e., the silencing of either the maternal or the paternal gene copy (allele) as is found in a small proportion (< 1 per cent) of genes. Another example is X chromosome inactivation by which it is warranted that only one X chromosome is active in female mammals thus excluding that females express twice the amount of X chromosomal gene products. Recent research indicates that random epigenetically driven monoallelic expression is a more common phenomenon than previously assumed, impacting, among others, neurodevelopment and brain function as well as immunoglobulin formation. Monoallelic gene expression accounts for the unique identity of cell types within a given tissue as well as between individuals (Chess 2012). It also causes different disease susceptibility of monozygotic twins and plays a vital role in ageing. (Milosavljevic 2011) Like the genome itself also the epigenome, i.e., the sum of epigenetic modifications, is highly dynamic and regulated by interactions of genetic and environmental factors. Intriguingly, epigenetic information can also be passed on to future generations, most likely through RNA (Daxinger and Whitelaw 2012). Both genome and epigenome are crucial for a phenotype's constitution.

Microbiome, Metagenome and Supergenome: The complexity of genomics and epigenomics is further increased by the versatile and individual microflora inhabiting our bodies. The concept of the superorganism was developed to take interactions between microorganisms and host into account. Importantly, the microbiome contributes the majority of genomic information of the human meta- or supergenome. The aim of the Human Microbiome Project (http://www.hmpdacc.org) is to analyse the genomes of microbial communities at different human body sites and to correlate these findings to health and disease (Human Microbiome Project Consortium 2012). For example, important insights about resistance and susceptibility to common inflammatory diseases such as type 1 diabetes, ulcerative colitis and Crohn's disease were obtained from microbiome studies recently (Virgin and Todd 2011).

Genotype and Phenotype: The genotype comprises the genome of a cell type or individual. It is sometimes referred to as a person's genetic makeup. As numerous regulatory circuits influence gene expression (see above) the genotype is distinct from the phenotype. Humans are diploid, i.e., they have one maternal and one paternal set of chromosomes and hence they have two copies (alleles) for any gene (an exception are genes of the X and Y chromosomes in males). Two identical alleles of a given gene are termed homozygous whereas different alleles are heterozygous. If an allele is dominant it masks the phenotype of the other allele which is then recessive. These relationships were first described by Gregor Mendel and can be observed in so-called Mendelian traits.[3] Mendelian inheritance is also termed monogenetic inheritance as usually mutations in a certain gene are responsible for the observed phenotype, e.g., mutations in phenylalanine hydroxylase cause phenylketonuria. Most common diseases, however, like e.g., heart disease, diabetes and others are

3 A comprehensive catalogue of Mendelian traits and disorders is found in the Online Medelian Inheritance in Man (OMIM) database. Available at: http://omim.org.

defined as multifactorial or polygenic and follow complex non-Mendelian patterns of inheritance. In fact, the differentiation into monogenic and polygenic inheritance is somewhat misleading as it suggests that the first is simpler than the latter. However, also Mendelian traits are complex and influenced in not yet fully understood ways by other genes. For example, many monogenetic diseases, including phenylketonuria, show an inconsistent genotype-phenotype relationship. This means that the genotype is not a sufficient predictor for outcome (e.g., not all individuals with mutations causing phenylketonuria develop the impaired cognition usually observed (Scriver 2007)). The practical lesson from these findings is that 'the actual phenotype not the one predicted from genotype at a major locus' should be treated (Scriver 2007: 840).

Box 1.3 Human Genetic Variation and Medical Applications

Human Genetic Variation: Sites of genome variation currently studied in detail comprise single nucleotides – stretches of DNA – which show variations of copy number (CNVs) caused by deletion, insertion, inversion, and duplication. Only a small proportion (about 0.1 per cent) of the whole genome shows genetic polymorphisms. These variations can be used for genome-wide association studies (GWAS, see below), correlating certain variants with the expression of common multigenic diseases or other common traits. Genetic variation also underpins ancestral testing. Genetic ancestry tests use either Y-chromosome specific variations for the paternal lineage, or variations in the mitochondrial genome for the maternal lineage (for a recent review on gene variation and phenotypes see Marian 2012). The HapMap Project launched in 2002 (http://hapmap.ncbi.nlm.nih.gov) catalogued several single nucleotide polymorphisms (SNPs) and defined so-called haplotypes (in this context, haplotypes are associated SNPs which are transmitted together). Launched in 2008, the 1000 Genomes Project (http://www.1000genomes.org) aims to map all human DNA polymorphisms using DNA from 2,500 individuals from five large regions of the world. It aims to find all genetic variants that have frequencies of at least 1 per cent in the populations studied. Results obtained so far indicate that, on average, each person carries 250 to 300 putative loss of function variants in annotated genes and 50 to 100 variants associated with inherited disorders (The 1000 Genomes Project Consortium 2010). Genetic variation of individuals is analysed by so-called genotyping, a collective term for numerous methods assessing the occurrence of the above mentioned variants in fractions of the genome. By using whole genomes for analysis it is expected to obtain more comprehensive information for association studies.

GWAS and EWAS: GWAS have identified a large number of candidate variants (mostly SNPs and CNVs) being associated with numerous common diseases or traits. However, to identify disease-causal genome variants is still a major challenge and requires to determine whether a given SNP or CNV has a functional effect on the molecular level and if so, whether this effect is deleterious for the organism (recently reviewed by Cooper and Shendure 2011). In order to minimise this gap, also

epigenome-wide association studies (EWAS), investigating particularly variations of DNA methylation, are performed. Although promising, they also create new challenges, because methylation patterns can be the cause or consequence for a given phenotype (Rakyan et al. 2011). When translating results from this kind of research into medical practice or any kind of personal genomic testing, it should be kept in mind that risk assessments on the basis of genetic variations struggle with at least two sorts of bias: ascertainment bias and publication bias. Ascertainment bias means an overestimation of effect for genetic variants as a result of the statistical procedures applied (Xiao and Boehnke 2011). It contributes to the observed discrepancies of disease-risk predictions for individuals by different online genetic testing suppliers (e.g., Ng et al. 2009). Publication bias refers to over-reporting positive results, a general phenomenon which also applies to genetic association studies (Valachis et al. 2011), particularly when only a small number of polymorphisms are evaluated. Thus, the relevance of a certain genetic variant for disease causation might be easily overrated and the provisional nature of such results could be overlooked. Moreover, next generation sequencing has lower sequence coverage and poorer SNP-detection capability in the regulatory regions of the genome (Wang et al. 2011) which could add to the biases affecting risk assessment of genetic polymorphisms. That the use of genetic information for disease risk assessment is still in its infancy and is not a frequently addressed topic within research on attitudes of test-takers.

Biomedical Cloud: In order to improve the quality of predictions one of the current goals of genomic medicine is to build up a 'biomedical cloud' that integrates data from genomics, systems biology and biomedical data mining (Grossmann and White 2012). As such efforts require maximum computing capacity they rely on cloud computing. However, the meaningful introduction of genomics into clinical practice in this manner is not only a linear function of computing capacity but it requires the input from diverse disciplines.

Acknowledgements

We are grateful to Priska Gisler, Gisli Palsson and Aviad Raz for very helpful comments on earlier versions of this chapter.

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PART I
Creating Identities

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Chapter 2
Will Personal Genomic Information Transform One's Self?

Jennifer R. Fishman and Michelle L. McGowan

Introduction

Direct-to-consumer (DTC) personal genome testing has provoked controversy since its inception. In the past five years, there has been much speculation within the medical, scientific, bioethical and social science literature regarding the potential impact of genomic information on individual identity. In both promotional and critical assessments of DTC genome services, there is the presumption that obtaining personal genomic information will have a significant impact on the ways in which recipients of this information understand themselves, and, further that 'knowing oneself' in this way will have lasting effects on one's conception of self, relationships with others, and approaches to personal health care going forward.

Claims have been made by those within the commercial sector about the transformative possibilities of acquiring new genetic knowledge. For instance, recent analyses of the marketing rhetoric used to advertise DTC personal genome testing (PGT) show that commercial entities characterise genetic information as empowering, creating individuals who are capable of using their own personal genomic risk susceptibility information to promote their own health and well-being (Harvey 2010; Saukko et al. 2010). The companies' mission statements and promotional materials that we accessed online reflect these analyses. 23andMe (2012), for example, promotes the following as two of its 'core values': 'having the means to access one's genetic information is good' and 'your genetic information should be controlled by you' – invoking a neoliberal, rights-based approach to direct access to personal genomic information. deCODEme's website suggests that 'getting to know your personal genome will *empower* you and provide you with a road map to improve your health' (deCODEme 2012, emphasis added). These companies have also emphasised the ideology of individual responsibility for health management to catalyse consumers: deCODEme (2012) again says, 'armed with knowledge from your unique genetic risk profile, you can start making the right lifestyle choices.' 23andMe (2012) similarly directs users to 'take a more active role in managing your health.'

On the more sceptical end of the spectrum, scholars publishing in the medical and bioethical literature have raised concerns that imagined consumers or 'virtual users' (Saetnan 2000) may lack the 'genetic literacy' required to interpret the complex medico-scientific information contained in personal genome test reports

which could result in unnecessarily dramatic responses to or conversely hollow reassurances and complacency regarding reported health risk susceptibilities (Cho 2009; Hunter et al. 2008; McGowan and Fishman 2008). McGuire and colleagues (2009) have hypothesised that consumers of personal genome testing may be likely to interpret the results of these tests as informative for healthcare decision-making and possibly even diagnostic, even though these services are currently marketed with the caveat of being informational and recreational rather than medical products. These cautionary conceptualizations of genomics and identity fit into Brian Wynne's (2005) argument that expert discourses of public science impose meanings on genomics by casting them in reductionist terms of discourses of risk.

Yet, the meanings and uses consumers are ascribing to DTC genomic risk assessment are still emerging and these claims have been made in the absence of empirical data on the subject. To date utopic and dystopic claims about genomic information and identity have largely been made in the absence of examining the users themselves to understand how they understand the impact of genomic information on their self-conceptualisation.

We acknowledge the complexity and intellectual ambiguity inherent in the term 'identity.' As we hope will become clear over the course of this chapter, we employ the term 'identity' as a general one that references the social scientific literature on the creation of the self within society, specifically from symbolic interactionist traditions. In the American tradition of George Herbert Mead (1934) and Herbert Blumer (1969) we think of the making of the 'self' as an ongoing social process that is enacted through one's interactions with other individuals, institutions, and social objects, including new technologies. We use the term identity to signal the ways in which the 'self' is in fact a social entity where one also categorises and characterises oneself into pre-existing social categories and groups. Our interest here lies in personalised genomics' claims that genetic information gives different categories of and differential access to self-knowledge and might therefore allow us to characterise and know our 'selves' in a new and different way.

Our aim in this chapter is to describe the landscape of viewpoints on the emergent relationships between personal genomics and individual identity, and to test the theoretical spectrum of utopic and dystopic perspectives against the findings of empirical research with early users of these services. We argue that many of the speculative claims about how new genetic knowledge will alter individual identity have not been borne out in the empirical studies of personalised genomic services thus far. We will conclude by looking towards the future to suggest what new research questions need to be addressed in order to analyse how personal genomic information may impact individual identity.

Dystopic and Utopic Views of Genetic Information and Individual Identity

In the early 1990s Abby Lippman (1991: 19) coined the term 'geneticization,' which she defined as 'an ongoing process by which differences between individuals are reduced to their DNA codes, with most disorders, behaviours,

and physiological variation defined, at least in part, as genetic in origin.' She argued that the danger in this trend of viewing genes as the key to understanding humanity is that individuals are reduced to their genes in a range of social spheres. Ideas about geneticisation have expanded to include the increasing use of genetic interventions in Western medicine.

Others have continued to develop this line of analysis to explore the dangers of genetic reductionism and essentialism. Evelyn Fox Keller (1992: 281) claimed that the popularity of the idea that 'our genes are what make us "what we are"' depends on an essentialist notion of genetic makeup as destiny, which contributes to a 'eugenics of normalcy' (299). In their analysis of genetics in American popular culture, Dorothy Nelkin and M. Susan Lindee (1995) argued that as ideas about genes and the implementation of genetic technologies have proliferated in the public sphere, biomedical explanations of the benefits of the gene and genetic knowledges have come to dominate popular attitudes in the United States, a phenomenon they characterise as genetic essentialism. These scholars have expressed the fear that the increasing genetic explanation of social problems raises the potential for attributing social problems to individuals' genes. They also raised a concern about the possibility of a (re)emergence of wide-scale population control tactics that are meant to curb social problems (Nelkin and Lindee 1995). In this context, Nelkin and Lindee (1995: 16) characterise genes as a 'convenient way to define personhood, identity and relationships in socially meaningful ways' that are relevant in medical contexts but one that also conveys 'guilt and responsibility, power and privilege, intellectual or emotional status.'

Critics writing in the 1990s also pointed out that genetic diagnosis far outstripped the capabilities of gene-based therapies, and raised the concern that 'prevention' simply meant preventing the birth of children with known genetic mutations and that parents of children with disabilities may be seen as irresponsible if they could have prevented the birth of a child with needs that require social support beyond the structures of the immediate family (Asch and Geller 1996; Bérubé 1996; Keller 1992; Nelkin and Lindee 1995). Beyond reproductive decision-making, geneticisation theorists have feared that equating one's genes with individual identity would exacerbate existing social stigmas and lead to further discrimination towards people with disabilities and specific genetic susceptibilities, including genetic discrimination in hiring practices, health insurance coverage, and education (Nelkin and Lindee 1995; Saxton 2000; Stempsey 2006). It has also been suggested that geneticisation would lead to the eventual development of a biological underclass, which could be defined as a marginalised social group whose social disadvantages and expectations could be linked to being labeled as genetically flawed (Duster 1990; Nelkin and Tancredi 1989). In geneticisation theory, being reduced to one's genetic makeup is projected to have profound negative implications for self-identity. For, as William Stempsey (2006: 198) has put it: 'if one's identity is to have a particular genetic constitution, and one's particular genetic constitution is in fact a genetic disease, then one's very identity is disease. We no longer have a disease; we *are* a disease [emphasis added].'

In contrast to the dystopic view of the effects of genetic information for our everyday lives, there is a group of scholars who focus on the potential liberatory benefits of personalised genetic information on one's sense of self and in particular the relationships that will form with others with whom they might find kinship. These theorists tend to celebrate the inherent potential in 'knowing more' about one's self, characterising such self-knowledge as perhaps ultimately liberating and opening up new opportunities to affect self-change and sociality.

Much of the social theory used to think about genetic information and identity in this way stems from the later work of Michel Foucault on the role of governmentality in shaping our understanding of the self. Governmentality is Foucault's (1991) concept of how power operates in modernity, which begins with the premise that there are no natural rules of governance; rather principles of governance are cultivated based on the need to provide rational explanations for modes of rule. Governmentality scholars have argued that in the contemporary Western context, neoliberalism is a prominent governing rationality (Lemke 2001; Rose 1999), and that 'the key feature of the neo-liberal rationality is the congruence it endeavours to achieve between a responsible and moral individual and an economic-rational actor' (Lemke 2001: 201). For example, the biomedical governmentality to 'know thyself' often relies on a neo-liberal and ultimately consumerist discourse to be 'proactive,' and 'take charge' of one's health, in order to become a self that is actualised, enlightened, and a socially and personally responsible citizen (Rose 1999).

Scholars portraying the largely utopic potential of genetics often rely on neoliberal governing rationalities. For instance, Novas and Rose (2000) have argued that the complexity of individual subjectivities results in a plurality of approaches to genetics and what constitutes individual health. They argue that the discursive shift towards creating a population of individuals who are 'genetically at risk' is not necessarily deterministic, but rather raises the potential for individuals to actively manage their genetic health and futures. Through the development of a genetically responsible citizenry, Novas and Rose claim that who counts as an 'expert' on genetic health is in flux. Now individuals can become self-experts in the pursuit of personal genetic health who work together with traditional experts in science and medicine. Paul Rabinow (1992) has characterised this move towards genetic citizenship as 'biosociality', the idea that as genetic discourses become dominant in the medical and popular spheres, individuals and groups will take up new definitions of themselves using the rhetoric of genetics which in turn will allow for new forms of identity politics along genetic lines. Used as a foil to 'sociobiology,' biosociality focuses on the potential development of new communities formed around genetic identities which also disrupts the individualism of geneticisation. Hence, genetic networks can be an important new way for people to share information and camaraderie that also facilitates genetic responsibility.

Heath, Rapp and Taussig (2004) have asserted that biosociality opens up the space for new forms of democratic practice, knowledge production, and the distribution of power. In their analysis of genetic citizenship in the contemporary

US context, they claim that new forms of genetic expertise among lay people are opening up spaces for rights claims and social recognition in the realm of genetic health, difference, and what constitutes a Foucauldian ethic of care (Foucault 1988): a component of genetic citizenship articulated around specific genetic conditions and political interests.

In the most positive sense, theorists of biosociality and genetic citizenship celebrate the liberatory potential of contemporary governing rationalities of genetics and the potential social distribution of power, but the proponents of these theories also recognise some pitfalls in these new social configurations. Through the somatisation of genetics and incorporation of biomedical language and technologies into everyday life, what once may have been the discourse of science is now also within the purview of lay experts (Novas and Rose 2000). Instead of being solely democratising, this type of unfettered access also becomes a moral responsibility and is a facet of the increasing biomedicalisation of conceptualisations of health, illness and self-care (Clarke et al. 2003). Lemke (2002) also argues that while neoliberal political rationalities celebrate individual choice and responsibility, there is also the potential for individual choice to slide into obligation. Thus while 'genetic citizens' can be seen as actively constructing knowledges and management of genetic risks, the constraints of what constitutes 'genetic responsibility' and of responsible genetic citizenship should not be downplayed.

Furthermore, Heath and colleagues (2004) have argued that a neoliberal political rationality, ironically, constrains free choices and restricts the range of possible approaches to taking active responsibility for genetic health by mobilising genetic research with therapeutic ends and invoking genetic citizenship claims. Thus, they argue that the rhetoric of individual rights and responsibilities operates within governing rationalities where 'the discipline and health of the body … both objectify and subjectify modern peoples' (Heath et al. 2004: 154). In other words, although this rationality is rooted in ideas about the free and modern liberal actor to act in ways that maximise his self-interest, this mode of thinking actually inverts 'free choices,' compelling individuals to behave in ways to become 'good' citizens thereby acting in the best interest of the state for devolving the responsibility to 'take care' of oneself to individuals alone. And individuals do so, not due to state coercion or threat, but because to the individual himself, it seems like the most rational, and even empowering, course of action. In the contemporary neoliberal context, 'the good subject…thus becomes the individual who will modify their lifestyle responsibly in relation to their genetic risk' (Novas and Rose 2000: 495).

Other scholars have argued that when coupled with genetic technologies, the Internet further serves as a potential catalyst for new forms of genetic subjectivities, knowledge building, and sociality by allowing individuals 'translocal engagements' (Heath et al. 2004: 155) around shared genetic and biomedical experiences (Novas and Rose 2000; Saukko 2004). Clarke et al. (2003: 182) argue that genomic-based technologies and the Internet are two (of many) developments that share the potential for constructing new 'technoscientific identities' – whether

those are identities inscribed upon us by others or are a product of the new subjectivities that arise through a biomedical governmentality that encourages such desire, demand, and need to inscribe ourselves with these identities. Given that the Internet mediates many interactions with DTC personal genomic testing and how consumers currently access genomic information, we wonder about the potential synergies and compatibilities between these two types of technologies for transforming identities. We should also consider the ways broader inequalities may be compounded in terms of access when the digital divide meets the health care divide, or perhaps rectified given the widespread (albeit still incomplete) access to web-based technologies, which give rise to yet other types of identities.

Perspectives on Individual Identities and Genomics

Much of the literature cited above references genetic and genomic technologies of the 1990s and early 2000s. Even as these technologies were emerging, Adam Hedgecoe (1998, 1999) and others (e.g., Condit 1999) called into question the lack of empirical evidence to support claims that geneticisation and, moreover, genetic essentialism were on the rise and whether these developments necessarily needed to be conceptualised in negative terms. However, research and technologies have changed considerably since then. Christine Hauskeller (2004) has argued the shift from genetic to genomic understandings of the world entails moving from acquiring 'certain' knowledge (i.e., of carrying a mutation or not) to 'uncertain' knowledge, now conceptualised in terms of susceptibilities and risk. And these susceptibilities and our understandings of what they mean are likely to change over time with 'new' research. Risk calculations and assessments are likely to be in need of constant re-evaluation. Claims about the highly 'personalised' nature of genomic information acquired through personal genomic testing raises the stakes about its ability to transform identities. Yet, again there is little empirical evidence to demonstrate whether or how these shifts play out in terms of understandings of identities. Thus, it is timely to revisit the claims made about the impact of genetic and genomic information on personal identity to examine if any of these fears have been borne out.

Recent conceptual work in social theory has wrestled with the notion of whether and how the advent of personal genomic information will inform understandings of personal identity. Nordgren and Juengst (2009) argue that consumer genomics companies utilise the rhetoric of personalised genomic medicine in advertising their services, suggesting that they can fulfil the promises of PGM by providing individualised risk assessments and allow consumers to take personal responsibility for their unique health risks. In so doing, personal genomics capitalises early on the potential of bioinformatics for encouraging users to embrace self-directed learning, which Nordgren and Juengst (2009: 164) characterise as an illustrative enactment of 'the populist "open-source" ethos of the Internet and postmodern suspicion of authority and paternalistic expertise.' Their analysis suggests that the availability

of personal genomic information invokes a do-it-yourself mentality towards knowledge of the self and one's genomic identity. Similarly, Harvey (2010: 371) has argued that 'genetic susceptibility testing provides information that the 'genetic entrepreneur' can use not just to protect but to generate 'vital capital.'

From a different angle, Zwart (2009) argues that individual identity formation will be greatly influenced by information produced by the field of behavioural genomics. Drawing from Goffman's (1959) work on presentation of self in everyday life, Zwart (2009: 125) projects that 'genomic bioinformation will increasingly be built into our self-images and used in order to tailor and adapt our practices of Self to our "personalised" genome. We will keep working on ourselves, no doubt, not by modifying our genomes, but rather by fine-tuning our behaviour.' With the advent of behavioural genomics, Zwart cautions that little is known yet about whether knowledge of behavioural genomic traits will be empowering or disempowering for individuals.

Piggybacking on Rose's (2006) work, Lee and Crawley (2009: 35) characterise the personal genomic information that 23andMe provides as a 'locus of biosociality' which 'forges social relationships based on beliefs of common genetic susceptibility that links risk, disease, and group identity.' However, they caution that the mechanisms through which 23andMe promotes biosociality around personal genomic information provides little guidance as to how to make found commonalities in genomic makeup meaningful in terms of shared social identity. Lee and Crawley (2009) contend that using personal genomic information for social networking has potential to realise Rabinow's (1992) concept of biosocial groups, yet little is known yet about whether genomic biosociality may give rise to relationships between genomic researchers and participants that could be characterised as 'democratic genomics' (Lee and Crawley 2009: 39).

Less optimistic about the potential for genomic information to manifest in biosociality and biological citizenship, Raman and Tutton (2010) have critiqued Rabinow and Rose's (2006) concept of 'molecularisation' for being overly reliant on a progress narrative of genomic science and its transformative potential to influence the ways in which individuals understand and govern themselves in molecular terms. They argue that new forms of knowledge produced through science may have the potential to change processes of identification. However, to presume that biological citizenship is the only logical endpoint for enacting identity in light of genetic information gives short shrift to the importance of population-level biopolitics and has the potential to be exclusionary for members of marginalised populations ill-equipped to engage in pursuit of their individual vitality. Perhaps, they argue, Rabinow and Rose's position that individuals are now obligated to govern their own health in light of genetic risk is a privilege for those whose resources allow them to effectively participate in biosociality and biological citizenship. Being a rational economic actor or a genetic citizen is not a subject position that may be available (or deemed warranted) for all, and the privileges embedded within these discursive and material constructions are worthy of analysis.

Unintentionally lending himself as example of this privilege, Francis Collins, arguably PGT's most prominent advocate, describes his interpretation of his personalised genome test as revealing 'specific threats' and responded by being 'more attentive … I had resolved to go ahead with a long-postponed plan to contact a personal trainer and work harder at a diet and exercise program, knowing that this was the best prevention for whatever diabetes risk that still remained' (Collins 2010: xx). Collins' ability to respond to 'threats' to his own health through enacting a highly regimented and costly exercise plan serves to exemplify the privilege of enacting a genomic identity in a neoliberal context.

Foster and colleagues (2009) also argue for a qualified approach to the impact of personal genomic information, stressing that personal utility and impact could be highly variable across individuals and groups. For instance, Juengst and colleagues (2012: 433) speculate that by marketing the potential for genomic risk information to reveal aspects of one's identity (e.g., family relationships, ancestral origins, or future potentials) 'consumer genomics can powerfully reinforce the importance of these socially ascribed identities for both the consumer's self-identification and identification by others.' There is, therefore, the potential for the re-inscription of social categories that replicate the current social order rather than provide liberation from it.

There is a small but growing body of empirical literature assessing the uptake and use of personal genomic information, and to date has primarily focused on prospective and actual consumers' attitudes regarding personal genomic risk assessment, their interest in using these services, and whether and how personal genome test results inform health behaviours and healthcare decisions.

Studies assessing *prospective users*' assessments of genomic risk assessment demonstrate relatively high levels of interest in PGT for health risk assessment, but low uptake of testing (Cherkas et al. 2010; Kaphingst et al. 2010; McGuire et al. 2009; Ortiz et al. 2009). McGuire and colleagues (2009) reported that a minority of prospective users expressed concern about getting 'unwanted information' and that the majority of prospective users of personal genome testing anticipated that they would want a physician to help interpret the results of the test, despite the fact that these services are offered directly to consumers without medical mediation. Another study of individuals who anticipated obtaining personalised genomic risk assessment revealed some concern that individual results would be worrying, but the majority of prospective users believed that obtaining genomic risk information in the context of participation in a clinical research study would provoke health-promoting behavioural changes (Gollust et al. 2011). Additionally, Bloss and colleagues (2010) suggest that interest, uptake, and concerns about DTC genomic risk assessment may vary across gender, racial, ethnic and socioeconomic lines.

In 2009, we conducted an empirical study of early users of DTC personal genome testing to assess their motivations, experiences, and reflections upon personal impact of receiving genomic risk susceptibility information (McGowan, Fishman and Lambrix 2010). Our study assessed the potential benefits and hazards of this technology and its mode of delivery, and whether and how personal genome

testing generates new forms of technoscientific identities (Clarke et al. 2003; Novas and Rose 2000). Counter to medical, scientific, and bioethical speculation that consumers may not be equipped to interpret results and may understand personal genome test results as health informative or diagnostic (Cho 2009; Hunter et al. 2008; McGuire et al. 2009), our findings suggest that early users approach personal genome testing technologies with both optimism and personal interest in the burgeoning field of genomic research yet scepticism about the genomic technology's current capabilities. So while Harvey (2010: 371) has argued that 'genetic susceptibility testing provides information that the "genetic entrepreneur" can use. . . to generate 'vital capital', attaining a state of optimal wellness specific to their genetic constitution,' our respondents typically acknowledged 'health' as an important value, but most did not go so far as to argue that new information about themselves had inspired them into health-seeking action. While a small number of our respondents took targeted preventive health-related measures upon receiving their genomic risk susceptibility reports, we argue that these individuals may be more accurately characterised as armed with knowledge of familial medical history and disease manifestation that prompted their health-related actions (including seeking out PGT in the first place). While our respondents may have initially had an interest in health-related information that PGT could provide, they certainly acknowledged specific limitations of the genome test itself, as well as the field of genomics, regarding its contemporary capabilities to impact personal health outcomes and behaviour. Nevertheless, they held overwhelmingly positive and optimistic views for the future of genomics to actualise its promissory potential.

Other recent studies of PGT users report similar findings. One study reported that some respondents believe that genomic risk information would empower personal health improvement, longevity and health decision-making for themselves and their families (Su et al. 2011), and other respondents 'expected that the genetic information could provide insights into their identities and what makes them unique' (Su et al. 2011: 141). Yet, other studies also report that PGT results neither provoked anxiety nor prompted changes to health behaviours and screening or post-test genetic counselling, even when freely available (Bloss et al. 2011; Gordon et al. 2012; Lee and Vernez 2012). Kaufman and colleagues (2012) have suggested that consumers' propensity to engage in post-test health information seeking, follow-up with healthcare providers and make changes to health regimens may be dependent on one's personal circumstances, including perceptions of personal health, family history of disease, and personal interpretations of genetic risk rather than just as a result of knowledge acquisition itself. Taken together, these studies suggest that recipients of PGT do not interpret their results as deterministic, and most appear to understand that PGT results are not going to provide them with any certainty but rather that it can provide additional information that can be used in conjunction with other health-related information in order to make better informed lifestyle decisions (Gordon et al. 2012).

Therefore, at this juncture, we cannot make a generalisable claim that this new technology enacts a uniform moral imperative or unique ethic of self-care for users

of the technology, nor can we argue that it fails to do so for at least some users. Further, given the small number of empirical studies assessing user perspectives of PGT and the small number of users seeking out this testing, additional research is needed to assess whether those who receive results conveying elevated risk factors for specific diseases would characterise themselves as 'genetically at risk', 'the worried well' or 'genetic entrepreneurs' more than users who did not receive eye-catching risk susceptibility results.

Conclusion

Given the rapid expansion of marketing genomic services directly to consumers, we strongly believe that it is inadequate to rely on the promotional rhetoric of the companies marketing these services or on the speculative social theory arguing that new identity formations are emerging. Neither the utopic nor dystopic view of genetic technologies has been able to capture the ways in which the information gleaned from these still emergent technologies are integrated into one's sense of self and identity. Yet, they can be used as launching pads for understanding the nuances and complexities of how new uncertain, ever changing, and value-laden information must be considered in a world with particular governing rationalities.

The perspectives of users of PGT are imperative for assessing the social and cultural construction of genomic information technology and whether and how they might be creating, inscribing, or enacting new technoscientific identities, new forms of genetic citizenship, and/or biosocial relationships. Furthermore, it will be important to differentiate users to understand what set of unique circumstances, structural arrangements, personal risk susceptibilities, prior health (care) experiences, and other factors contribute to an emergent subjectivity in the face of new genomic information. Beyond the broad brushstrokes of grand theories are the analyses that can only be achieved with empirical data to draw out the complex interrelationships between identities, technologies, self-knowledge, and the future. If we are interested in assessing emergent identities and how users are incorporating this information into their sense of self, we may need methods other than interviews, surveys, and narrative analysis to assess this phenomenon. One can imagine studying emergent user communities and interactions amongst members, or studying how one communicates this 'new' genomic information to others – family members, health care providers, or prospective partners/spouses. Perhaps the most important dimension to capture is change over time: as the technologies and risk profiles evolve and change as each user ages (with age itself as a risk factor for disease), health risk information may take on new meanings and need to be constantly reintegrated into one's identity. How can we study that process as identities shift and alter over time and with new experiences (Blumer 1969; Mead 1934)? And genomic technologies are hardly the only ones at our disposal in the twenty-first century that we use to access 'personal' information (about ourselves and others). The desire to know oneself extends to numerous other (non-medical)

realms. Where and how do these other acquisitions of personal information get woven into our emergent identities and ways of knowing? As analysts, it will likely be important to compare these other types of information seeking with genomic information in order to better understand our contemporary socio-historical-technical context. We believe that these future studies are instrumental for understanding the implications of conceptualizing the relationships between health status, healthcare and personal identity in genomic terms, and provide new insight into the largely informational rather than medical value that early users attribute to personal genome testing technology in its current state.

Acknowledgements

Support for the preparation of this chapter was provided by the US National Human Genome Research Institute, Grant NIH R01 HG005277. The authors wish to thank the other members of our research team for their integral contributions to our larger project, 'Anticipating Personalized Genomic Medicine': Eric Juengst, Richard Settersten, Jr., Marcie Lambrix, Michael Flatt, Marcie Lambrix, Timothy Ottusch and Roselle Ponsaran. They are also very appreciative of the helpful comments from the editors on an earlier version of this chapter.

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Chapter 3
The Changing Self: Philosophical Concepts of Self and Personal Identity in a Post-clinical Age of Genetics

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The concept of the 'self' plays an essential role in discussions of personal identity. In these discussions, the human genome can be seen as a science-based key to personal identity (Klitzman 2009). With the availability of personal genomics services, the possibility of obtaining 'personalised' calculations of one's genetic risk mostly through commercial providers, it appears that more and more people integrate their personal genetic data into the so-called 'autobiographical self' (Klitzman 2009: 887). This concept refers to how an individual understands and defines her identity in the light of her values, convictions and aims. It includes also ideas and interpretations of the body and their relationship to concepts of human identity (Schicktanz 2007). Genomic information undergoes a specification as soon as it becomes part of the autobiographical self. The answer to the question about the relevance of 'personalised genomics'[1] (Klitzman 2009; Zwart 2009) for human identity therefore depends not only on genetic testing or clinical evidence but also on the meaning which we ascribe to concepts like 'personal identity' and 'self'. This chapter will explore the role which direct-to-consumer (DTC) genetic tests play in this quest for identity.

Identity and Self

Before discussing the relevance of DTC genetic tests to the question of personal identity, the concept of identity which is involved here has to be clarified. In the field of genetic research the concept 'identity' is mainly used to express 'qualitative identity', which means the coincidence of genetic properties: The term 'genetic identity' is used to express the fact that certain genes are 'exactly alike' in one or more individuals (Zeiler 2007: 28). With the term 'personal identity', on the other hand, we express the numerical identity of persons; that means we refer to the intuition that we are one and the same entity. Concerning 'numerical identity', a

1 Both terms, 'personalised genomics' and 'personal genomics', can be found in the specialist literature but 'personal genomics' seems to be more commonly used. Unless citing from other authors, I will use the term 'personal genomics' here.

synchronic meaning must be distinguished from a *diachronic* one: Synchronically, 'identity' designates the *unity* of a person. In this case the identity-question 'who am I?' is understood as 'with what in the world am I identical?' In the diachronic sense, with 'numerical identity' we express our basic intuition that we remain the same person over time: On this second meaning, the concept 'identity' is employed in questions like: 'Am I the same person as I was thirty years ago?' (Noonan 1989: 104–105).

At first glance there seems to be no intrinsic connection between qualitative genetic or genomic identity and numerical personal identity. It is obvious that genomic identity does not imply the numerical identity of persons: Two twins with identical genomes are two numerically different persons. The twin example shows that qualitative genomic identity is no sufficient condition for the numerical identity of living beings; it can be asked, though, whether qualitative genomic identity might be a necessary condition for their diachronic identity. The relevance of qualitative identity for diachronic identity is more obvious when we consider the problem of qualitative change: The diachronic identity of animals and persons is affirmed even if many of their properties change over time. For example, I assume that my cat is the same as she was one year ago even if she is much bigger now than she was one year ago. A complete qualitative change, however, poses a serious problem for the numerical diachronic identity of living beings. If nothing remains the same, then it is hard to answer the question of the diachronic identity of persons and living beings affirmatively. For this reason, many rely upon a minimal qualitative identity – the sameness of some essential properties – to justify the claim that persons and other living beings are diachronically identical. The solution to the problem of diachronic identity, namely, 'what makes me the same person over time?', thus depends on the solution to the problem of qualitative identity – are there essential qualities which remain the same during my lifetime.

Questions about the essential qualities of living beings have traditionally been answered by appeal to concepts like 'soul', 'substantial form', or 'essence'. With such concepts philosophers designated essential properties which could guarantee the diachronic identity of persons and other living beings. The metaphysical assumption of a soul, though, is nowadays regarded as unscientific because there is no empirical evidence for the existence of essential qualities which together would constitute a 'substantial form'. From this perspective, the concept of genomic identity seems to be a good candidate for a scientific answer in the quest for essential properties which remain the same during an organism's lifetime. Even if there is no absolute genetic identity during an organism's lifetime,[2] the

2 Recent studies indicate that the genetic profile is subject to changes within one individual during lifetime (Forsberg et al. 2013). As Zeiler (2007: 28–29), however mentions, the fact that there is no complete genomic identity from birth to death does not exclude the more general assumption of genomic identity.

genome seems to constitute a set of stable properties which could guarantee the identity-over-time of human beings.[3]

The rise of human genomics, however, coincides with a period of growing scepticism towards the assumption of diachronic identity itself. Concepts like 'essence' and 'nature', which were previously used to account for personal identity, have been abandoned and replaced with ontologically weaker principles of identity. One example of this shift is the replacement of the concept of soul with the modern concept of *self*. Whereas the concept of soul or substantial form originally designated an identity-preserving principle whose essential properties remain the same during the lifetime of a person, the concept of self designates the self-representing activities of a person. Without doubt it was the pioneer of English empiricism, John Locke, who made the concept of self prominent in modern philosophy. Rejecting the older notion of the soul as unclear and inaccessible to human experience, he deemed the concept of the self to be sufficient for guaranteeing personal identity throughout life. What is new in Locke's argument is that the identity of persons is detached from their organic identity: Locke distinguishes the concept of 'self' from the concept of 'man' [sic!]. 'Self' is a psychological term denoting human persons. 'Man' is a biological term denoting the human organism and her bodily existence. For Locke, the diachronic identity of an organism depends on the organic life of that animal:

> An animal is a living organised body; and consequently the same animal … is the same continued life communicated to different particles of matter as they happen successively to be united to that organised living body. (Locke: Ch. 27, § 8)

The diachronic identity of persons, by contrast, is constituted by the cognitive function of self-reflection or self-consciousness:

> in this alone consists personal identity, i.e., the sameness of a rational being: and as far as this consciousness can be extended backwards to any past action or thought, so far reaches the identity of that person; it is the same self now it was then … (Locke: Ch. 27, § 9)

In this conception of the diachronic identity of human persons, the concept of the self plays a crucial role. The 'self' is based on consecutive acts of consciousness bound together because of a person's ability to remember. X is the same person through time as Y if and only if X possesses the same consciousness as Y. To possess the same consciousness is to remember previous states of that consciousness:

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3 According to Dupré (2010: 26), the idea of a 'stable [genetic] core' has 'provided extremely useful applications […] ranging from phylogenetic analysis to forensic DNA fingerprinting.' In the same article, however, Dupré expresses his concerns that an over-emphasis of this stable core 'can be one of the most fundamental sources of misunderstanding in theoretical biology'.

> So far as any intelligent being can repeat the idea of any past action with the same consciousness it had of it at first, so far it is the same personal self. … [it] will be the same self, as far as the same consciousness can extend to actions past or to come. (Locke: Ch. 27, § 10)[4]

The introduction of two different principles of identity – one for the organic and another for the personal life – is a clear deviation from the tradition, where the notion of the soul as the person's substantial form designates one and the same principle of identity for the human being's organic and cognitive life. Locke's notion of the self and his distinction between biological and what we may call higher-order principles of identity are well received in contemporary philosophy of mind, social science, and bioethics (Singer 1979: 76). It has prompted psychologists, social scientists, and philosophers to create a classification of different principles of identity, of which the following three types can be distinguished:[5]

a) The biological principle of identity (biological self) is based on an organism's *fundamental self-regulatory processes*.
b) The autobiographical/personal self is the principle of personal identity and is based on consecutive acts of consciousness bound together by *memory.*
c) The social/interpersonal self is a principle of social identity and is produced by *intersubjective activities* like communication and social interaction.

The relevance of personal genomics for personal identity must be seen in the light of the modern discussion of different forms of self and identity. If personal identity is a product of the self-representing activities of the individual, there are two distinct ways in which genetic information provided in DTC genetic testing is relevant for personal identity. On the one hand, genomic data provide information about qualitative genetic identity, giving rise to the question of how the genome shapes the identity of the individual on the biological, cognitive and social levels. On the other hand, genetic information has an impact on personal identity as soon as it becomes an object of the self-representing activities of the individual, when she considers how it might be integrated into the different kinds of selves – especially into her autobiographical and social selves. The following analysis is dedicated mainly to this second way of identity-building in which genetic information is an object of a person's self-representing activities. In this way genetic information provides knowledge about oneself. The schematic categorisation of different

4 cfr. also § 17: 'Self is that conscious thinking thing […] which is sensible, or conscious of pleasure and pain, capable of happiness or misery, and so is concerned for itself, as far as that consciousness extends.'

5 This classification can be understood as the common denominator of the different classifications of the 'self' which can be found in James (1890), Neisser (1988), Dennett (1991) and Flanagan (1992).

selves given above can be used to give a more detailed analysis of the specific character of genomic self-knowledge. What kind of self-knowledge does personal genomic information provide?

The Genomic Principle of Identity

At first glance, genomic self-knowledge seems to provide knowledge about the biological self. What distinguishes genomic information from other biological data such as blood pressure, pulse, or blood glucose level? Genomic information pertains to the basic dispositions of our organisms. It is commonly seen as information about our biological or molecular 'blueprint' (Zwart 2007): as such it concerns not only the probability of our developing certain diseases; in addition, the most essential traits of our phenotype like hair colour, size or bodily appearance seem to be anchored in the structure of our DNA. The meaning of these data, though, reaches far beyond biology. Some people associate genomic data more or less directly with the autobiographical and social selves. A prominent example of such an enlarged functional role ascribed to genomic information is provided by Craig Venter, a leading figure in genomic research. In the publication of his own DNA sequence Venter and his group relate genetic information not only to specific organic dispositions, like the 'higher risk of acute myocardial infarction' (Levy et al. 2007: 2131), but also to behavioural characteristics, including socially relevant ones like tobacco addiction, alcoholism, antisocial behaviour, and conduct disorder (Levy et al. 2007: 2134–2135). The Dutch philosopher Hub Zwart observes that, in his autobiography, Venter considers links between genomic data and specific behavioural dispositions like risk-seeking and the ability to tolerate stress as relevant for some of his own past decisions and experiences (Zwart 2009: 128). Craig Venter presents genomic information as having explanatory value for the autobiographical and social self. This case can be seen as a paradigm example in the field of so-called personalised genomics (Levy et al. 2007: 2131), where genomic data are regarded not only as providing additional biological information, but also as being part of a principle of identity which provides an explanatory framework for organic processes as well as for one's autobiography and social relations. In this way genomic information constitutes a principle of identity which comprises the biological, the autobiographical, and the social self. In the following I will call this principle 'genomic principle of identity' (GPI).

The GPI seems a perfect successor to traditional identity-providing concepts like the soul or the substantial form. In the Aristotelian tradition the soul is the formal principle which guarantees the identity of all living beings – it is the substantial principle in virtue of which a body is a living body (Aristotle: 412^{a}, 7–9). Aristotle classifies the form of reality possessed by the soul as *dispositional* rather than manifest (Aristotle: 412^{a}, 24f.). The soul, as the basic capacity of all cognitive and non-cognitive activities, is supposed to guarantee identity even through the most dramatic changes which living beings might undergo. In the Aristotelian

tradition the soul plays a decisive role in explaining mental phenomena. But its explanatory scope does not end here: its *explanandum* also includes biological phenomena. Aristotle, who assumes that even plants have souls, argues against a strict separation of the mental from the biological. Aristotle's tripartite distinction of the soul into vegetative, sensible and rational 'parts' shows that the basic capacities of nutrition, growth or sense-perception are necessary pre-requisites for the rational part to function well (Aristotle: 413^{a}, 25 – 413^{b}, 10). For Aristotle there is just one subject – the animate organism (plant, animal or human being) – which in virtue of its nature is able to do all the things that a living being of a specific kind typically does. Therefore, the Aristotelian soul accounts for the causal connection between biological and cognitive functions. The GPI is analogous to the soul in these respects: Like the soul, the genomic principle of identity refers to a dispositional reality; the genomic principle is not constituted so much by the actual existing molecular structures of the DNA, but more by dispositions (such as the susceptibility to specific diseases) or behavioural characteristics (like risk-averseness and stress tolerance) which can be connected (with a higher or lower probability) to a certain genome. Another way in which the genomic principle of identity resembles the soul is in the range of its integrative function: Like the soul, the genomic principle accounts for an individual's organic and cognitive capacities.

Despite these similarities, there is one striking difference between the two principles of identity. Unlike the soul, which entails the top-down explanatory model of the hylomorphic (form-matter) account, the GPI is a bottom-up explanatory model. The top-down model begins its explanation with the entire organism on the macro-level and deduces the constituents and processes of the micro-level. Individual micro-level events and phenomena can be causally explained only as components of larger functional units at the macro-level. In the case of the human soul, the physical and mental activities of a person can be explained only by locating them in the organisational principle of the entire human organism. The bottom-up explanatory model, on the other hand, is based on the assumption that every macro-phenomenon can be deduced from micro-physical causal processes. This deterministic model is presupposed by most research strategies in the natural sciences. In the case of the genomic principle, all macro-phenomena, such as the organism's biological structure, activities, and so forth could be explained in terms of the processes and structures of specific molecular patterns on the micro-level. The GPI seems to present a bottom-up approach which allegedly integrates the biological, autobiographical, and the social selves into a coherent scientific picture. Is personal genomics the scientific answer to the age-old question of human identity and self-knowledge, as the 'essentialist rhetoric' of DTC DNA testing maintains (Nordgren and Juengst 2009)? Does the sequencing of my personal DNA reveal my individual uniqueness,[6] and can it determine who I am?

6 The 23andMe research homepage sketches a very optimistic picture on the potential of genomics for determining my personal uniqueness: 'Participate in research

Conceptions of the Self and Their Influence on the Interpretation of Underdetermined Genomic Information

The use of genomic information for questions about self and identity points to obvious limits of the GPI: On the one hand, the GPI seems to require some form of genetic determinism. One could say that personal genomics would provide a satisfying answer to the question 'who am I?' only if my identity could somehow be derived from my genome. On the other hand, experts agree that there is no straight connection between genome and phenotype. The initial optimism concerning the existence of 'mono-causal relationships between genes and traits' (Zwart 2007: 181) has been replaced by the more nuanced view that higher order properties are not genetically predictable. One of the important discoveries of the Human Genome Project is that there is no one-to-one correlation 'between organismal complexity and gene number', and that the complexity of who we are cannot be derived from the underlying genome (Hauskeller 2004: 294; Zwart 2007: 191). Research has shifted from assuming a strict correspondence between geno- and phenotype to taking human genes to have a 'functional plasticity' (Zwart 2007): One and the same genetic type can yield multiple phenotypes, not least due to the effect of epigenetic processes; this multiple realisability between geno- and phenotype is confirmed also by twin studies which have shown that 'the same genome does not always produce the same phenotype' (Hauskeller 2004: 296; a recent review on the genotype/phenotype discordance of common diseases in monozygotic twins is found in Bell and Spector (2011)).

Genetic functional plasticity has important consequences for the causal relationship between the genome and the biological, autobiographical and social selves. One is that what I am on these levels cannot be derived from my genome. There are other factors which co-determine my personal traits. In the case of the biological self, epigenetic mechanisms and environmental influences play important roles in further developing what I am on the biological level. By referring to these epigenetic mechanisms, we may compensate in a certain way for the indeterminacy resulting from the functional plasticity of the genome. Do we have similar 'epigenetic' factors for the autobiographical and social selves, or is it up to the consumer of personal genomics to provide a more detailed account of their relevance to genetic data? There are different levels on which the subject herself interprets her own genetic information.

As we saw above, the autobiographical self depends on a person's capacity of self-representation. A first interpretation occurs already in this self-representing process. When I represent myself I (normally) do so not from an objective third-person perspective, but from a subjective first-person perspective. 'Having a first-person perspective' can be defined as having the 'ability to conceive of oneself as

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while exploring your own genetics: […] Learn new things about yourself – and what your genes may have to do with them. Find out which traits make you stand out from the crowd', https://www.23andme.com/research [accessed: 25 January 2013].

oneself' (Rudder Baker 2000: 66) or as having 'a perspective from which one thinks of oneself as an individual facing a world, as a subject distinct from everything else' (Rudder Baker 2000: 60). Here it means having the ability to know that specific genetic data are from my own genome: 'I am the person who has this genetic profile which can be associated with my own phenotype.' Genetic information is interpreted as soon as this genetic evidence is discovered to be about oneself. For example, the same information about a specific probability for hypertension is interpreted differently by a person who finds out that the information is about her own genome than by someone for whom the information is just one genetic sample among others. Self-attributed genomic information might be connected with emotions, expectations, and preferences, which not only have a specific influence on one's behaviour but also on the interpretation of the information itself.

A second level of interpretation of genomic information arises when the subject integrates her personal genomic data into her existing autobiographical and social selves. As we have seen above, companies are offering DTC DNA tests which provide information about probabilistic causal relations between one's genome and one's ancestry, character traits, and other features of the autobiographical and social selves. This has the effect that personal genomic information is not only regarded as additional biological evidence by test-takers, but also as providing information for the autobiographical and social selves. Personal information concerning one's own genetic code is not only filled into the narrative of the autobiographical and social selves; its meaning is in turn shaped by those very selves. Concerning the autobiographical self, for example, the genomic information provided by modern medical techniques undergoes such a specification as soon as it becomes part of the subject's belief system. The autobiographical self can be considered a 'centre of gravity' (Dennett 1991; James 1890) for a set of beliefs, forming an implicit theory for the person in question. This implicit theory provides a pattern by which she can then interpret genetic information. In particular it entails a very selective reading and interpretation of genomic data; for example, a person whose belief system contains the belief that intelligence is highly valuable will have a tendency to ascribe greater importance to those data which in some way can be connected to intelligence; in ancestry testing they might pick out those ancestors who – in some way – fit with their own ideal of intelligence.

A third level of interpretation, which has to do with genomic self-knowledge, arises when subjects use the information provided in DTC DNA-tests for self-reform or self-improvement. In this case, genetic self-knowledge is not an end in itself; it is supposed to enable the consumers of genomic information to improve or reshape themselves. According to Zwart (2007: 182), 'the shift from knowledge to power is important' for the attempt to obtain genetic self-knowledge: 'The basic goal of biotechnology was clear – the evidence-based amelioration of human nature: (self-) knowledge is power' (Zwart 2007: 185). The 'shift from knowledge to power', though, requires a subject who autonomously decides whether to use DTC DNA testing services and – even more importantly – it is he or she who sets the standards for self-reform. These standards cannot be derived from genomic

evidence itself because they depend on a subject's beliefs, expectations and desires. A person who intends to change her drinking and smoking habits might interpret a genetic disposition to cardiac diseases in a different way than a person who wants to become a professional tennis player.

This process of interpreting genetic information on the level of the autobiographical and social selves by the consumers themselves can result in contradicting interpretations of one and the same data. Sometimes personal interpretations of genetic data stand in direct opposition to their scientific/clinical counterpart. In the following I present an example of such contradicting interpretations of the same genetic material. Even if the case is taken from traditional genetic testing and not from personal genomics, it illustrates how the dichotomy of personal[7] and clinical interpretations of one and the same genetic data can be a source of ethical problems.

Many deaf people consider deafness not as a disability but as a valuable component of their identity. Dennis (2004) reports a case in which a deaf couple 'desperately wanted a deaf baby'. Genetic testing could be used to achieve this goal: 'They could use prenatal genetic testing, and abort the foetus if it can hear. Or they could consider *in vitro* fertilisation (IVF) combined with preimplantation genetic diagnosis to select deaf embryos for transfer to the womb' (Dennis 2004: 895). Even if it turns out that the genetic probability of 'success' would be extremely low, the case illustrates, in the following way, the possibility of giving incompatible interpretations of the same genetic data: Deaf parents who desire a deaf child argue that deafness is not a disability, but rather that it is 'constitutive of a unique and valuable cultural identity' (Harrosh 2011: 3). According to the Royal Association for Deaf People, 'Deaf people are only 'disabled' by the effects of discrimination and exclusion' (Harrosh 2011: 3). As a matter of fact, the definition of deafness given by the Royal Association for Deaf People is incompatible with a standard medical evaluation. In clinical practice, genetic pre-implantation testing for deafness has been allowed only for the sake of avoiding the implantation of an embryo carrying deafness. The supervising authorities of the same clinic which allowed genetic tests for deafness (Monash IVF, Melbourne) made it clear that they 'would not allow a couple hoping for a deaf child to use the test': "Our policy states that the procedure should be used to avoid a genetic abnormality," says Helen Szoke, the authority's chief executive' (Dennis 2004: 895).

The two conflicting interpretations of the genetic disposition for deafness cannot be resolved on the level of biology. It seems clear that deafness as a biological phenomenon is often correlated with a specific genetic structure. The incompatibility between the personal and the professional medical view of the genetic disposition for deafness, however, depends on the way in which deafness is

7 'Personal' here is understood as belonging to the belief-system of a group of individuals which share a specific attitude towards deafness.

integrated in the autobiographical and social selves of the persons involved.[8] In the case described above, genetic information about deafness is evaluated positively by the majority of those who have integrated deafness as an essential component of their autobiographical and social selves. For them deafness is 'constitutive of a unique and valuable cultural identity'. This evaluation stands in direct opposition to the clinical or scientific interpretation of genetic information regarding deafness. On this view, deafness is seen as an abnormality or even as harmful. To be deaf, on this view, is to be 'made worse off than we could have been relative to the potential of our species to fully realize its nature' (Harrosh 2011: 4).

Is there a way to resolve this discrepancy in the interpretation of genetic information? Who has the authority to decide what the 'potential of our species' is? As we saw above, specific concepts of the autobiographical and social selves have a direct impact on the interpretations of genetic data made by different persons, groups or institutions. Therefore, a solution to the problem of incommensurable interpretations of genomic information depends on the existence or non-existence of objective standards for the integration of genomic information into the higher-order selves. In the following I compare a conventional (ontologically weaker) understanding of the self with a biology-based (ontologically stronger) concept of the self. I will show that only the ontologically stronger concept provides a theoretical framework on which scientific and private interpretations of genetic data can be compatible. The following discussion refers mainly to conceptions of self and personal identity as they are presented in the field of philosophy of mind, cognitive science and neuroscience.

A Weak Concept of Self and Personal Identity

An ontologically weak conception of self and personal identity is presupposed by Daniel Dennett, Owen Flanagan and Thomas Metzinger. Dennett, for example, regards the self as a fiction of self-representing biological systems: '[…] selves are not independently existing soul pearls, but artefacts of social processes that create us, and, like other such artefacts, subject to sudden shifts in status' (Dennett 1991: 423). Dennett draws a clear distinction between a biological and a personal principle of identity – between a biological and a personal (autobiographical) self. For Dennett, the biological self is a relatively stable reality because it is anchored in an organism's fundamental self-regulatory processes. Its limits coincide with those of the body. Whilst the biological self is a relatively stable entity, the conscious self of adult human beings is an explanatory fiction generated by the self-representing system itself. In contrast to the biological self, the limits of the conscious self are not confined by the human organism's biological structure. The physical correlate

8 It is clear that the child's autobiographical self plays no role here. After all, she is not even born yet and may never be. It is the members of two different communities who are disagreeing over the value of a genetic variant on the basis of their own autobiographical and social selves.

of the conscious self consists in parallel distributed neurological processes in a highly plastic brain (Dennett 1991: 187). This plasticity of the brain is for Dennett the main reason to assume that the biological self is not the ontological basis of the personal self which is constituted through various forms of self-representation.[9] Far from it: Dennett categorically rejects such an interpretation: The autobiographical and social selves do not have the biological self as their ontological basis; rather, it is through self-representation that these selves are constituted in the first place. Our self-representations are structured in such a way that a self can be ascribed to them only retrospectively. For this reason Dennett calls the personal self a 'Centre of Narrative Gravity' (Dennett 1991: 410): This self is not a real entity but rather an explanatory fiction or a useful theoretical construct.

The philosophical notion of a personal self of Dennett and others suggests a very weak concept of personal identity. This weak concept of identity is not only characteristic of contemporary philosophy but is also commonly employed in the social realm. As Hauskeller puts it, the stable patterns of personal identity provided by the Judeo-Christian tradition are no longer accepted in a society where 'acquired positions need regular reaffirmation and societal recognition' and 'the abandonment of an 'innate' social status goes hand in hand with the need to form oneself into a subject, to develop an identity'. One consequence is that 'the resulting identity is unavoidably weak and fragile, because of its incalculable dependence upon others and upon societal recognition patterns. The stability of social recognition is dependent upon the success of an identity 'performance' (Hauskeller 2006: 1–2).

The self as the centre of narrative gravity is the philosophical version of the weak and fragile identity described by Hauskeller in the social realm. But it is not just philosophers and social scientists who subscribe to this view of identity; rather, it is shared by many scientifically minded 'common people'. The divergent interpretations mentioned above result from an ontologically weak (narrative) understanding of the self, made explicit by philosophers of mind and social scientists. It is these weak concepts of the autobiographical and social selves which typically frame the answers to questions about how to use genetic information. When we assume this narrative concept of the self, we can hardly avoid incommensurable interpretations of the same genetic data. An unbridgeable gap between scientific and private interpretations of genomic information will always remain. Since the autobiographical and social selves are not continuous with the biological self, there are, on the genomic view, no objective criteria for evaluating the adequacy of the different interpretations of genetic information. Scientifically important aspects might be irrelevant to a person's autobiographical

9 As a materialist Dennett assumes that there is a physical correlate for the self in a similar way as there is one for illusions or false beliefs; but there is no *proper physical correlate* for the 'self-illusion' – in the brain there is nothing which corresponds in the proper sense to that what we have in mind when we refer to our self. For Metzinger, e.g., the self is the product of the self-misunderstanding of a system which self-represents itself (Metzinger 1993: 157), for Dennett the self is an explanatory fiction (Dennett 1991: Chapter 13).

self, and other aspects of genetic information which are less important from a clinical point of view might take a central position in the autobiographical self.

If the autobiographical and social selves are useful fictions, there are no criteria to correct the interpretations made on the basis of these fictions. Criteria which appeal to the biological nature of the individual or to the 'potential of our species' – as proposed by the above quoted case on deafness – are unavailable, since there is a discontinuity between the autobiographical and social self on the one hand and the biological self on the other hand. In this case, the only criteria for the best reading and use of genomic information are pragmatic. If it should turn out that the deaf couple's interpretation of the genetic disposition for deafness is based on very deep layers of their personal belief-systems, then the clinical personnel might have a strong ethical motive to accommodate the deaf couple's wish for a deaf baby.

If we assume an ontologically weak narrative conception of the self, it is hard to avoid the consequence that one and the same genomic material is subject to incommensurable interpretations. However, the problem of finding objective standards of interpretation for genomic information can be seen in a new light when an ontologically stronger concept of the self, as for example in the works of Antonio Damasio, George Butterworth, and others, is assumed. It can be shown that an ontologically stronger (biology-based) concept of the self renders the first- and third-person interpretations of genetic data compatible, because it sets constraints on the interpretation of underdetermined genomic information.

What a Strong Concept of Self Implies for Interpreting Genomic Information

George Butterworth extensively explored the origins of self-perception in infancy (Butterworth 1992); his studies seem to back the thesis that there is no discontinuity between the biological self as the basis of an organisms' self-regulatory processes, on the one hand, and the autobiographical and social selves as the basis of higher forms of self-conception, on the other hand. These different kinds of selves are not incompatible with each other. The higher forms of self-conception presuppose the more basic ones. There is a continuum beginning with primitive ways of self-regulation and bodily self-perception and terminating with a mature concept of an autobiographical and social self in adult human beings:

> The point is that movement synergies reveal properties of the material self as an *organized totality*; species-typical-developmental processes will determine the extent to which such aspects of the categorical self become elaborated within higher order cognitive processes. (Butterworth 1992: 108)

According to this view, a human being's biological self is the precursor of the autobiographical and social selves. The assumption that there is continuity between the biological and the higher order selves can also be found in the work of the neurobiologist A. R. Damasio. He states that consciousness and self-

consciousness depend on structures that belong to older phylogenic areas of the brain which are closely interconnected with basic biological functions. Damage to parts of the diencephalon, of the brainstem, or the upper part of the *formatio reticularis* leads to various forms of loss of consciousness. These structures are responsible for the regulation of basic living functions of the organism – the so-called 'inner milieu'. From the fact that these brain areas are involved in the control and representation of bodily processes, Damasio concludes that there is a direct connection between conscious experience, neuronal representation and the control of bodily processes (Damasio 1999: 236). A central condition for the development of human subjectivity and self-consciousness – according to Damasio – is the representation of the organism's basic regulatory mechanisms, or its dynamic equilibrium (homeostasis). Since these regulatory mechanisms are relatively stable, they provide an optimal foundation for referring to an identical subject, such as the one presupposed by self-consciousness. Damasio explicitly rejects the relativistic suggestion that the personal self is merely a fiction. The intuition that we remain the same person over time is not a fiction but refers to a biological reality – the organisational principle of our bodily functions, which remains the same over time. Even if we undergo a permanent change throughout life, the structural and functional principle of our organism remains largely unchanged. Bodily processes are grounded in a unifying principle, which persists from the beginning to the end of our life. Self-representation generates the impression of the identity and immutability of a stable self, because this invariant organisational principle of our organism is constantly represented as well:

> The reason why representations of the body are well suited to signify stability comes from the remarkable invariance of the structures and operations of the body. Throughout development, adulthood, and even senescence, the design of the body remains largely unchanged. To be sure, bodies grow in size during development, but the fundamental systems and organs are the same throughout the life span and the operations that most components perform change little or not at all. (Damasio 1999: 141)

This organisational principle is no fiction but rather is a real aspect of our organism, and moreover it controls fundamental bodily functions. According to Damasio, without this principle, neither consciousness nor self-consciousness could arise. This organisational principle of our organism constitutes personal identity on a fundamental level. In this view, the biological self is not discontinuous with higher-order principles of identity such as the autobiographical and social selves.

In deciding on the adequate interpretation and legitimate use of personal genomic information, the following constraints are implied by an ontologically strong self: The autobiographical and social selves are unfolding the potential of an underlying biological self. According to Damasio these higher-order selves 'manifest the same goal as the form of automated homeostasis' which can be found in the biological self. 'They respond to a detection of imbalance in the life process, and they seek to correct it within the constraints of human biology and of

the physical and social environment.' (Damasio 2010: 292). In this way important standards for the determination and interpretation of genomic information are set by the bodily system as a whole. The standards for an adequate interpretation of genomic information are not found in the personal approach of the autobiographical self but in the entire organism as a functioning unit.

The assumption that there is continuity between the biological, autobiographical, and social selves puts us in a position to develop standards for integrating genomic information into the autobiographical and social selves in a way that is analogous to the way in which genetic information is integrated into the biological self through the means of epigenetics. If the relevance of genomic information for the autobiographical and social selves cannot be fully determined on the genetic level, it is up to scientific disciplines like neuroscience, cognitive psychology, and social sciences to discover the quasi-'epigenetic mechanisms' for these more complex forms of identity. Information about my genome provided by DTC DNA tests improves the quality of my self-knowledge only in combination with the results of other scientific disciplines. Initial studies of consumer responses to DTC genetic tests seem to confirm that consumers themselves share this view about the interpretation of genomic information (McGowan et al. 2010): they seem less impressed by the essentialist rhetoric of genomic companies than one might expect and they are generally very sceptical about the explanatory power of personal genomic information. Moreover, since many are aware of the limitations of such tests, they have a strong motivation to discuss these results with a clinical expert (such as a personal physician) and to integrate them into a larger picture of their own identity.[10]

Conclusion

Some promoters of DTC genetic tests assume that genomic information provides knowledge about not only one's biological but also one's autobiographical and social selves. Even if it is taken for granted that DTC genetic information matters for questions about self and identity, it remains unclear to what extent the genome causally influences these higher-order properties of human beings (Heinemann 2006); genetic information is underdetermined as a principle of identity. The indeterminacy of the genetic information concerning the biological self can be compensated for in a certain way by epigenetic mechanisms. But whether there are objective standards for interpreting the relevance of genomic information for the autobiographical and social selves depends on which notion of 'self' is at issue. If selfhood is ontologically weak (such as the narrative conceptions discussed earlier) then the only constraints on the interpretation of genomic information are

10 These findings are more or less confirmed by Gollust et al. (2012): 91.7 per cent of a selected group of 'early adopters' of personal genomics 'stated that they were likely to share their results with their physicians' (Gollust et al. 2012: 27). According to this study, though, there seems to be a discrepancy between the declared intention to share the results with medical professionals and the actual practice of doing so; only 15–29 per cent of the respondents actually shared the results with their physicians (Gollust et al. 2012: 28).

pragmatic. Following this view, there can thus be divergent interpretations of the same genetic information. Things are different when an ontologically stronger – biology based – concept of the self is presupposed. In this case, scientific disciplines like neuroscience, cognitive psychology, and social sciences would not only set standards of adequacy for interpreting how and whether genomic information relates to the autobiographical and social selves; these disciplines could also cast light on the way in which complex epigenetic mechanisms fill the gap between the genomic information and the higher-order structures of the autobiographical and social selves.

Acknowledgements

I would like to thank Katherine Dormandy, Barbara Prainsack, Silke Schicktanz and Gabriele Werner-Felmayer for their helpful comments and critical remarks.

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Chapter 4
Ancestry Testing and DNA: Uses, Limits – and Caveat Emptor[1]

Troy Duster

Direct consumer use of DNA tests for ancestry tracing has taken off in recent years, and we are not just talking about probes for first-generation genetic lineage as in *Who's Your Daddy*?, popularised on daytime 'reality' television. Between 2002 and 2006, nearly a half-million people have purchased tests from at least two dozen companies marketing direct-to-consumer kits (Bolnick et al. 2007) and since then the DNA ancestry industry continuously proliferated (Royal et al. 2010; Wagner et al. 2012). The motives for testing range from the desire for ancestral links to those who lived on other continents five-hundred plus years ago – to a more modest interest in reconstructing family histories (reviewed in Bolnick et al. 2007; Royal et al. 2010). For many African- Americans, the quest to find a link to regions and peoples of sub-Saharan Africa can take on a spiritual or even messianic quest, at least partially explained by the fact that the Middle Passage across the Atlantic during the slave trade explicitly and purposefully obliterated linguistic, cultural, religious, political and kinship ties. The 2006 PBS[2] television series, *African American Lives*, brought this quest into sharp relief. First celebrity and later ordinary Blacks were mesmerised by stories of DNA matches that claimed to reveal or refute specific ancestral links to Africa, to Native American heritage, and surprising to some, East Asian or European populations.

In sharp contrast, CBS'[3] *60 Minutes* aired a dramatic segment in the fall of 2007 (October 7) that portrayed a direct and sharp challenge to the claims-making about such ancestry testing. The segment began with Vy Higgensen, an African-American woman from New York's Harlem triumphantly affirming her connection to 'new kin' (one of whom was a white male cattle rancher from Missouri). But as the program unfolds, we see a disturbing cloud of doubt drift over the last part of the segment that ends with a less than subtle hint at specious claims. A first test from the company *African Ancestry* claims that Higgensen is linked to ancestors in the Sierra Leone, the Mende people. She rejoices. 'I am thrilled! It puts a name,

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1 An earlier version of this chapter appeared in *Race and the Genetic Revolution*, edited by S. Krimsky and K.Sloan. New York: Columbia University Press, 2010.

2 PBS (Public Broadcasting Services) is the US network of publicly funded TV stations (http://www.pbs.org).

3 CBS (Columbia Broadcasting System) is a US network of private TV and broadcasting stations (http://www.cbs.com).

a place, a location, a people!' But then she is shown the result of a second test from another company, *Relative Genetics*, which claims that she instead has a genetic match to the Wobe tribe of the Ivory Coast. She seems philosophical. Yet a third test, from still another company, *Trace Genetics*, claims that her ancestors are from Senegal, the Mendenka. Now she seems agitated, visibly concerned, confused – and most certainly disappointed that what began as a definitive match to a particular group or region of Africa has now turned into a 'you pick which one you want to believe' game.

The very next month, serious questions about the tests were revisited when Henry Louis Gates, who had hosted the aforementioned *African American Lives*, said that the same thing had happened to him. Here is how the *New York Times* (Nixon 2007) cast the story:

> HENRY LOUIS GATES JR., whose PBS special "African American Lives" explores the ancestry of famous African-Americans using DNA testing, has done more than anyone to help popularize such tests and companies that offer them. But recently this Harvard professor has become one of the industry's critics.
>
> Mr. Gates says his concerns date back to 2000, when a company told him his maternal ancestry could most likely be traced back to Egypt, probably to the Nubian ethnic group. Five years later, however, a test by a second company startled him. It concluded that his maternal ancestors were not Nubian or even African, but most likely European.
>
> Why the completely different results? Mr. Gates said the first company never told him he had multiple genetic matches, most of them in Europe. "*They told me what they thought I wanted to hear*," Mr. Gates said [my emphasis].

Here we have the first sally into a combined definitional and epistemological conundrum – beginning with the meaning of 'ancestry'. While this is typically used to refer to geographic areas where one's biological ancestors lived, with just a few minutes of reflection, we can see an enormous problem to which even common sense will alert us: *Which ancestors*? Easy enough if we are only dealing with mom and dad, or four grandparents – or we can even handle three generations back with eight great-grandparents. But if we go back six generations, that means we all have 64 direct biological ancestors. Since each of these 64 could be said to have made an equal biological contribution to our makeup, why would we choose to represent any one or two as our 'real' biological lineage? (Eight generations gives us 256 such ancestors, and twenty generations places the figure at 1,048,576.)

The Capacities and Limits of Using DNA to Test for Ancestry

What can DNA tell us about our genetic lineage, and where does it fall short? What explains Vy Higgensen's multiple results from different testing sites? Flawed methodology? Partial truths hyped as definitive findings? Did the testing companies use different methods, or deploy different reference populations – or both?

Let us begin with what DNA testing *can* tell us about biological ancestry. There are two different tests – one for males and another for females, and each can provide relatively definitive results along one particular line of our genetic ancestry.

Males inherit the Y chromosome from their biological fathers. The markers are sufficiently distinctive so that the test can not only identify the father, but also the father's father, and if the data were available, the father's father's father. This path to ancestry identification can go on for as many generations as data are available – which is how Thomas Jefferson (or one of his brothers) was linked to Sally Hemings' offspring. For more than 150 years, historians argued and debated as to whether Jefferson had children with one of his slaves, Sally Hemings. Only in the last decade has Y chromosome analysis settled the debate in favour of those who have claimed that the historical record pointed to Thomas Jefferson.

The test for female ancestry has an interesting parallel. We can definitively answer 'Who's your mommy?'! Mitochondria, the cell's energy producers located outside the cell nucleus, have their own genomes. All of a mother's children inherit her mitochondrial DNA (mtDNA) but only the daughters pass it on as, in general, only the mitochondria of the egg cell but not of sperm survive in the early embryo. Thus, for a female, it is possible to trace and identify her mother, her mother's mother, etc. (along the same line as just noted for males using Y chromosome analysis). This was the way that granddaughters were linked to their grandmothers in the aftermath of Argentina's 'Dirty War' (1976–83). Thousands of young fathers and mothers 'disappeared' by acts of the ruling junta, and their orphaned small children were given to couples who wished to adopt (Penchaszadeh 1992). It was through mitochondrial DNA testing that the grandmothers were reunited with the children of their (murdered/disappeared) daughters. These two tales reveal not only the power of DNA ancestry testing, but their significant and consequential social and political uses as well.

But it is also vital to re-state the limitations – that these two tests can identify, for example, only two of the 64 great great great great grandparents. Indeed, only two of the next generation further back, of 128, can be so identified, only two of 256, and so on. Yet each of the other (62 or 126 or 254) contributed equally to our genetic makeup as the two we can trace by the sex-linked paternal or maternal lines. The Genographic Project of the *National Geographic Magazine* (https://genographic.nationalgeographic.com/) uses these two tests, supplemented by a selection of 22 additional markers. The researchers correctly inform participants who send in their DNA that there are limitations to what can be claimed. Nonetheless, people who receive the results are often led to believe that if their test does *not* match the archival sample of a particular Native American or Eskimo group, then they are *not* genetically linked to that group. Several years ago, when Genographic Project scientists sampled people in the Arctic North, Lorianne Rawson, a 42 year-old woman who had strong social ties to, and who believed that she was descended from, the Aleuts of Alaska, submitted her DNA to the Genographic Project. She was informed by the testers that results linked her instead to the Yup'ik Eskimos,

the enemies of the Aleuts (Harmon 2006). Personal and political trauma can understandably ensue from such seemingly authoritative reassignments. This kind of 'result', however problematic in terms of disclaimers or caveats, happens when the technology inevitably limits the analysis to particular corridors or silos of the ancestry tree, and locks in on that limited corridor. While the results are presented as an authoritative claim, the laity is not provided with the tools to understand how the many other ancestral links noted above are excluded by the limits of ancestry tracing through DNA analysis.

Sometimes these putative links (or lack of same) have significant financial repercussions. The Black Seminoles have been struggling with this very question – of whether to use DNA analysis to 'authenticate' their relationship to the Seminole Indian Tribe. The reason is straightforward and serious: money. The federal government, pursuant to a land-settlement claim, made an award to Seminole Indians in 1976, poised to distribute upward of $60 million. In 2000, the Seminole Nation of Oklahoma amended its constitution so that members needed to show 'one-eighth Seminole blood' (Johnston 2003: 262). The Black Seminoles could use either Y chromosome analysis or mtDNA to link themselves through very thin chains back on two edges of the genealogical axis (mother's mother's mother, etc.; or father's father's father, etc.), but that would miss all other grandparents (14 of 16, 30 of 32, 62 of 64). The stakes are even higher for the Florida Seminoles. In 2006, the tribe purchased the entire Hard Rock Café chain for approximately one billion dollars. If you were offered a genetic ancestry test of either Y chromosome or mtDNA analysis, would you really want to engage the probabilistic Russian-roulette type gamble?

To supplement the limitation of Y chromosome and mtDNA testing, a group of researchers has come up with a procedure to discern the frequency of certain markers that are hypothesised as belonging, selectively, to our ancestors (see below). However, there are several blind assumptions that have to be accepted in order to have confidence in the links to ancestral populations so defined.

Ancestral Informative Markers (AIMs) – The New Proxy for Race

Unlike Y chromosome DNA or mtDNA tests, this technology examines a group's relative share of genetic markers found on the autosomes – the non-sex chromosomes inherited from both parents.

Since ancestral informative markers (AIMs) are overwhelmingly shared across all human groups, it is therefore not their absolute presence or absence, but their rate of incidence, or frequency, that is usually being analysed, and this is especially true when it comes to claims about continental populations. How did these markers come to represent ancestral populations of Africa, Europe, and Native America? The vast majority of these markers are *not* 'population specific', as the inventor of AIMs originally claimed (Shriver et al. 1997). Because the companies marketing ancestry tests hold proprietary interests in their techniques, most do not make

them available for possible scientific replication, and their modelling constructs are therefore undisclosed. Thus, we are left to speculate about the threshold level of frequency that is used to determine the grounds for inclusion or exclusion, as well as what counts as a 'pure' referent population.

In one lab that permitted its procedures to be studied by a medical anthropologist, ancestry percentages were generated by formulas that compare the relative frequency of markers (44 in total) between selected populations of recent European, African, and Native American descent (Fullwiley 2008a, 2008b). All those in the defined group were tested for the frequency of markers that the researchers hoped would provide *relative* distinguishability. Recall that the frequency at which each marker appears in each group is noted – and whole continents are never sampled. Finally, the researchers compare marker frequencies between the three groups to come up with values which, when taken together, yield a probability result about ancestral percentages. This procedure generates the baseline for the statistically-based notion of a 100 per cent pure European (or African, etc.), so that when you send in your DNA from the saliva swab, and it turns out that you have one-third of the markers that have been designated as 'European' – you are told that you are 33 per cent European. It is by this statistical legerdemain that we have come to the molecular re-inscription of race in contemporary human genetics (Duster 2006; Fullwiley 2007).

There are a number of deeply problematic, even flawed assumptions behind that percentage claim. What is this 'reference population' that has become the measuring stick by which we inform people of their 'per cent ancestry to a putatively pure continental population' (read 'race' here) (Duster 2006: B13). Let us re-examine such a result if reported back to someone of recent African descent. First, more than 700 million people currently inhabit the African Continent – and human geneticists have known for decades that this is the continent with the greatest amount of genetic variation on the globe. The reason for this variation was noted by Pilar Ossorio (2009: 4):

> For many regions of the human genome, there are more variants found among people of Africa (and the recent African diaspora) than found among people in the rest of the world. This is probably because humans have resided in Africa for much longer than we have resided any place else in the world, so our species had time to accumulate genetic changes within the people in Africa.

A scientifically valid random sampling of even one per cent of this population would require a prohibitively expensive research program – a database of seven million. So instead researchers have settled for 'opportunity samples' – namely, a few hundred here or there, or even thousands that have been collected for a variety of reasons. No attempt has ever been made to take theoretically driven or random samples from African tribes such as the Lua, Kikiyu, Ibo, Hauser, Bantu, Zulu (with all the linguistic, cultural and political complexities of defining the boundaries of such groups), not to mention the thousands of language groups spread across the continent. How then, can we have any sense of reliability or

validity for a claim that says someone is 80 per cent African – when the baseline for that claim is based upon the transparent scaffolding of chance – not purposive sampling?

Yet, when taken together, we are told that these markers appear to yield sufficiently distinctive patterns in those continental populations tested. So now we see how a specific pattern of genetic markers on each of a set of chromosomes that have a *higher frequency* in the 'Native Americans' sampled becomes established as a 'Native American' ancestry reference. (The fact that there are more than 480 different populations of the Tribal Council – the vast majority of which have never been sampled – is no small matter here, but that is not the focus of the critique I am about to make.) The problem is that millions of people around the globe will have a similar pattern that is, they'll share similar base-pair changes at the genomic points under scrutiny. This means that someone from Bulgaria whose ancestors go back to the fifteenth century could (and sometime does) map as partly 'Native American', although no direct ancestry is responsible for the shared genetic material. There is an overwhelming tendency for those who do AIMs analysis with the purpose of claims about ancestry to arbitrarily reduce all such possibilities of shared genotypes to 'inherited direct ancestry'. In so doing, the process relies excessively on the idea of 100-per cent purity, a condition that could never have existed in human populations.

While this is a huge problem, yet another issue looms even larger. If a computer program produces an outcome indicating that 35 per cent or more of a particular genetic marker exists in population A (let's call them East Asian), while 35 per cent or less occur in population B (let's call them European), the researcher may use that marker to say that someone is from East Asian ancestry. To make matters even more complicated, claims about how a test subject's patterns of genetic variation map to continents of origin and to populations where particular genetic variants arose, require that the researchers have 'reference populations'. The public needs to understand that these reference populations comprise relatively small groups of *contemporary* people. Those groups sampled may have migrated over several centuries, and thus these researchers must make many untested assumptions in using these contemporary groups to stand as proxies for populations from centuries ago, whether putatively representing a continent, a region, or a linguistic, ethnic or tribal group. To construct tractable mathematical models and computer programs, researchers bracket these assumptions about ancient migrations, reproductive practices, and the demographic effects of historical events such as plagues and famines. Given these intractable barriers to even low-level probabilistic reliability, geneticists are on thin ice telling people that they do or don't have ancestors from a particular people.

Thus, instead of asserting that someone has no Native American ancestry, the most truthful statement would be: *It is possible that while the Native American groups we sampled did not share your pattern of markers, others might since these markers do not exclusively belong to any one group of our existing racial, ethnic, linguistic, or tribal typologies.* But computer-generated data provide an appearance

of precision that is dangerously seductive and equally misleading. Now we come to one part of the answer as to why different companies come to different results. We cannot conclude that an individual has a close affinity to a particular ethnic or racial group or local geographical population simply because their DNA markers match that population. 'Such a conclusion would require demonstrating that the DNA sequence is not present in other places, it would require demonstrating that the gene pool of that ethnic group or local population had been close and immobile for centuries and millennia…' (Weiss and Long 2009: 709).

Be Especially Wary of Applications of These Claims

There is a yet more ominous and troubling element of the reliance upon DNA analysis to determine who we are in terms of lineage, identity, and identification. The very technology that tells us what proportion of our ancestry can be linked, proportionately, to sub-Saharan Africa (ancestry-informative markers) is the same being offered to police stations around the country to 'predict' or 'estimate' whether the DNA left at a crime scene belongs to a white or black person. This 'ethnic estimation' using DNA relies on a social definition of the phenotype (phenotype being the observable physical or biochemical characteristics of an organism, determined by both genetic makeup and environmental influences). That is, in order to say that someone is 85 per cent African, we must know who is hundred per cent African. *Any molecular, population, or behavioural geneticist who uses the term 'per cent European' or 'per cent Native American' is obliged to disclose that the measuring point of this 'purity' (100 per cent) is a statistical artifact that begins not with the DNA, but with a researcher's adopting the folk categories of race and ethnicity.*

The Segue to Forensics and Criminal Justice and 'Molecular Race'

It is possible to make arbitrary groupings of populations (geographic, linguistic, self-identified by faith, identified by others by physiognomy, etc.) and still find statistically significant genetic markers shared between those groupings. For example, we could simply pick all of the people in Chicago, and in Los Angeles, and find statistically significant differences in DNA marker frequency at *some* loci. Of course, at many loci, even most loci, we would not find statistically significant differences. When researchers claim to be able to assign people to groups based on marker frequency at a certain number of loci, they have chosen loci that show differences between the groups they are trying to distinguish.

The work of Evett et al. (1993, 1996), Lowe et al. (2001) and others suggests that there are only about ten per cent of sites in the DNA that are 'useful' for making distinctions. This means that at the other ninety per cent of the sites, the allele (one member of a pair or series of genes that occupy a specific position on a specific chromosome) frequencies do not vary between groups such as 'Afro-

Caribbean people in England' and 'Scottish people in England'. But it does not follow that because we cannot find a single site where allele frequency matches some phenotype that we are trying to identify (for forensic purposes, we should be reminded), that there are not several (four, six, seven) that will not be effective, for the purposes of aiding the FBI, Scotland Yard, or the criminal justice systems around the globe in highly probabilistic statements about suspects, and the likely ethnic, racial, or cultural populations from which they can be identified – statistically.

So when it comes to molecular biologists asserting that 'race has no validity as a scientific concept', there is an apparent contradiction with the practical applicability of research on allele frequencies in specific populations. It is possible to sort out and make sense of this, and even to explain and resolve the apparent contradiction – but only if we keep in mind the difference between using a taxonomic system with sharp, discrete, definitively bounded categories, and those which show patterns (with some overlap), but which may prove to be empirically or practically useful.

When representative spokespersons from the biological sciences say that 'there is no such thing as race' – they mean, correctly, that there are no discrete categories that come to a discrete beginning or end, that there is nothing mutually exclusive about our current (or past) categories of 'race', and that there is more genetic variation within categories of 'race' than between. All this is true. However, when Scotland Yard or the Birmingham, England police force, or the New York City police force, wants to narrow the list of suspects in a crime, they are not primarily concerned with tight taxonomic systems of classification with no overlapping categories. That is the stuff of theoretical physics and logic in philosophy, not the practical stuff of helping to solve crime or the practical application of molecular genetics to health delivery via genetic screening – and all the messy overlapping categories that will inevitably be involved with such enterprises. That is, some African- Americans have cystic fibrosis even though the likelihood is far greater among Americans of North European descent, and in a parallel if not symmetrical way some American whites have sickle cell anaemia even though the likelihood is far greater among Americans of West African descent. But in the world of cost-effective decision-making, genetic screening for these disorders is routinely done based on common-sense versions of the phenotype. The same is true for the quite practical matter of naming suspects.

Searching for Racial and Ethnic Markers in Forensic DNA

In the July 8, 1995 issue of the *New Scientist* entitled, 'Genes in Black and White', some extraordinary claims were made about what it is possible to learn about socially defined categories of race from reviewing information gathered using new molecular genetic technology. In 1993, a British forensic scientist published what is perhaps the first DNA test explicitly acknowledged to provide 'intelligence information' along 'ethnic' lines for 'investigators of unsolved crimes'.

Ian Evett, of the Home Office's forensic science laboratory in Birmingham, and his colleagues in the Metropolitan Police, claimed that their DNA test can distinguish between 'Caucasians' and 'Afro-Caribbeans' in nearly 85 per cent of cases.

Evett's work (1993), published in the *Journal of Forensic Science Society*, draws on apparent genetic differences in three sections of human DNA. Like most stretches of human DNA used for forensic typing, each of these three regions differs widely from person to person, irrespective of race. But by looking at all three, the researchers claimed that under select circumstances it is possible to estimate the probability that someone belongs to a particular racial group. The implications of this for determining, for practical purposes, who is and who is not 'officially' a member of some racial or ethnic category are profound.

The legal and social uses of these technologies are already considerable by the *cognoscenti*, and they are poised to 'take off'. Here are some examples:

More than a decade ago, several states began keeping DNA database files for sexual offenders. Three factors converged to make this a popular decision by criminal justice officials that would be backed by politicians and the public: 1) sex offenders are those most likely to leave body tissue and fluids at the crime scene, 2) they rank among the most likely repeat offenders, and 3) their crimes are often particularly reprehensible in that they violate persons, from rape to molestation and abuse of the young and most vulnerable. Today, all fifty states store DNA samples of sex offenders, and most states do the same for convicted murderers. Moreover, now thirty-four states store DNA samples of all felons (Simoncelli 2006).

While 39 states permit expungement of samples if charges are dropped, almost all of those states place the burden on the individual to initiate expungement. Thus, civil privacy protection, which in the default mode would place the burden on the state, is reversed. In other words, instead of 'innocent until proven guilty' it has become 'criminally suspect until proven innocent' so to speak. Twenty states now authorise the use of databanks for research to develop new forensic techniques. Based on the statutory language in several of those states, this could easily mean assaying genes or loci that contain predictive information – even though current usage is supposed to be restricted to analyzing portions of the DNA which are only useful as identifying markers. Since most states retain the full DNA (and every cell contains all the DNA information), it is a small step to using these DNA banks for other purposes. The original purpose has long been pushed to the background, and the 'creep' expands not only to other crimes besides sexual offenses, but to misdemeanours and even those merely arrested as well, California being a case in point. Following the passage of a state proposition in 2004, it is now legally permissible for authorities to collect DNA from those merely arrested for certain crimes.

On January 5, 2006, the president of the United States signed into law HR 3402, the Department of Justice Reauthorization bill of the Violence Against Women Act of 2005. This legislation for the first time permits state and federal law enforcement officials the right to transfer DNA profiles of those merely arrested for federal crimes into the federal Combined DNA Index System (CODIS) database.

Previously, only convicted felons could be included. Those DNA profiles will remain in the database unless and until those who are exonerated or never charged with the crime request that their DNA be expunged. Thus the default will be to store these profiles, and expunging requires the proactive agency (and resources) of those who have been arrested.

This announcement was the source of celebration by one of the leading providers of DNA testing services, Orchid Cellmak Inc., of Princeton, New Jersey. The president and chief executive officer of Cellmak, Paul J. Kelly, immediately issued a statement applauding this development (Orchid Press Release 2006: 1), stating:

> This is landmark legislation that we believe has the potential to greatly expand the utility of DNA testing to help prevent as well as solve crime… It has been shown that many perpetrators of minor offenses graduate to more violent crimes, and we believe that this new legislation is a critical step in further harnessing the power of DNA to apprehend criminals much sooner and far more effectively than is possible today.

But there is yet another reason why we must be much more wary of these developments. Criminologists and statisticians have provided enough convincing evidence that reliability may be a systemic issue with regard to 'exact matches', leading to false 'hits' with traditional short tandem repeat (STR) approaches (Thompson 2008). As for the possibility of using full DNA samples for forensic research, attempts to determine physical features, such as skin colour, hair texture, and eye pigment, have already been made (Fullwiley 2008b). These techniques, as they rely on 'admixture estimates' discussed earlier, are also rife with reliability issues despite their veneer of exact precision with regard to continental genetic affinity, or, put bluntly, racial diagnosis. This kind of categorizing of subjects and patients is occurring in medical and health journals, often with the idea that pharmaceuticals could be tailored to patients based on putative notions of their ancestral genetic 'admixture'. Researchers are also finding new ways to identify genetic variants related to 'admixed' populations that they believe may be 'linked' to variable complex disease conditions, such as end-stage renal disease (Kao et al. 2008). Here whole areas of the genome are assumed to be ancestrally 'African' or 'European' with very little discussion of how such prior determinations of purity are – or are not – relevant for all self-identified Africans and Europeans.

An Unregulated No-Man's Land – No Oversight, No Guidelines

Much like the industry of assisted reproduction in the United States, there is a complete absence of regulation or quality control with genetic ancestry testing. There is no requirement for transparency in the construction and use of reference populations. Any company can claim that their laboratories can analyse your DNA to provide accurate information about your ancestry. If three different companies

provide three different answers (as in the *60 Minutes* report noted at the outset), what is a consumer to do? Which company is correct, or more to the point, which one is more likely to be correct? There is no way of knowing, since we have no 'gold standard' for excellence or professional self-policing. This was pointed out in *Science* six years ago (Bolnick et al. 2007), and in November 2008, the American Society of Human Genetics (ASHG) issued a statement on ancestry testing that included five recommendations emphasizing the need for greater responsibility, research, explanatory clarity, collaboration and accountability by these direct-to-consumer companies (ASHG 2008). The statement also pointedly warned of several important limitations to the scientific approaches used to infer genetic ancestry, including the false assumption that contemporary groups are reliable substitutes for ancestral populations, and most significantly, the lack of transparency regarding the statistical methods that companies use to determine test results (Lee et al. 2009).

But while the ASHG statement calls for greater transparency, we have seen that private sector providers of ancestry testing have proprietary reasons for keeping secret their own particular combinations of key technology, software and population sampling procedures. Most are unwilling to disclose the size and composition of their reference populations. Without mechanisms to enforce transparency, there is no way of assessing the scientific basis for specific assertions of 'per cent ancestry'. For example, until and unless there is a publicly available version of what constitutes a 10 per cent European or a 100 per cent African, etc., claims about 80 per cent ancestry cannot be fully understood or tested, much less replicated.

Building on the ASHG recommendations for transparency, there is a need for specific policies enforced by federal agencies. For example, the Federal Trade Commission and the Centers for Disease Prevention and Control can and should play pivotal roles in setting industry standards for what constitutes responsible and accountable practices. These agencies can promote the research necessary to identify minimal guidelines for presenting the fair uses and clear limitations of current genomic technologies. Guidelines for transparency would also include clear statements spelling out the risks associated with over-extrapolating or misinterpreting genetic ancestry results. The active involvement of regulatory agencies would provide infrastructure for the interdisciplinary dialogue necessary to create effective policies and for maintaining industry standards (Lee et al. 2009). While supporting such measures, we should not be naïve about their effectiveness, since the demands on these companies to generate profits are strong and insistent. It is difficult to exaggerate the role that money plays in this whole process, whether for ancestry testing companies trying to stay in business or members of groups seeking to cash in on casino gambling by being designated an Indian tribe by the US Interior Department's Bureau of Indian Affairs (BIA).

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Chapter 5
Other Stories: Artistic Explorations of Genealogy and Identity

Priska Gisler, Mo Diener and Luzia Hürzeler

Preamble

For many years, one of the authors of this chapter, Mo Diener as a performance artist was involved in a longsome search for her ancestors. She was, and still is, undertaking research in order to eventually confirm (or prove wrong) the rumour that her mother's ancestors were travellers. Mo Diener developed her work through a series of photographic images, which she collected from family members, through interviews she made with them, and also through information she dug up in archives. It is surprising to see, in a series of images, how her research led her to condense this broad body of work in a photographic installation with exemplars of her mother's clothes and then to recognise how her longing to know more about the past might be tied to an intense mother-daughter relationship.

Luzia Hürzeler's artistic work approaches questions of ancestry from a somewhat different angle; she enquires into animal husbandry and taxonomy. Luzia Hürzeler explored the different institutions and possible bonds that connect living and dead animals. For a video-installation, which has been shown in a series of art exhibitions but was presented in scientific contexts only exceptionally, she worked with two lions who were biological relatives. Through this work, Luzia Hürzeler asked in-depth questions about representations of animals and also about the meaning of humans' encounters with animals, finally, in what way they could, actually and authentically, be regarded as based on a kind of mutual understanding.

Based on these artistic works of two of us, we seek to offer two alternative ways of genetic story-telling on different aspects of genetics as a science dealing, among other things, with the topics of heredity and ancestry. Preceding the two narratives that will be outlined in the following, were a series of conversations between us, the two artists and a sociologist of science, about family and kinship, about humans and animals, about the living and the dead; in other words, about how living beings relate to each other. The conference 'Genetics as Culture in a Consumerist Age',[1] held in Innsbruck, Austria, in 2011, offered the perfect

1 More information at: https://www.i-med.ac.at/ethucation/Veranstaltungen/Folder_end.pdf.

opportunity to pool ideas and reflections and to further develop our own thoughts. At the conference, Luzia Hürzeler and Mo Diener each gave a lecture performance and showed an artistic video, and Priska Gisler brought them into interaction with each other and the audience. It soon became clear that the different stories we were telling and showing to each other were all, in one way or another, responses to questions of genealogy, social order and identity. The broader context within which our conversations were situated extends to recent developments in biotechnology and medicine. In this chapter, we consider how the many socio-technical strands which are opened up by genetics – as well as the analytical tools, objects, and institutions that accompany and form the assemblage upon which we draw here – influence the ways narratives generate orders and identities. From both a science studies as well as from an artistic perspective, we seek to explore how we are part of such a socio-technical assemblage, and also consider how this ensemble of practices and objects could eventually be made visible.

Beyond merely exploring what one can gain from genetics – considered as much a tool for testing (of ancestries and disease-related phenotypes) as it is for classification (of species), our contribution seeks to reveal – while closely intertwining sociological and artistic thinking – how ideas and understandings about where we come from or how we belong are made. It will consider some of the institutions, practices and discourses that order meaning and tie bridges between the living and the dead, the 'here' and the 'there', the 'us' and the 'others'. Our chapter, therefore, seeks to add some new stories, or 'other stories,' to the scientific narratives that are so widely distributed and heard.

Artistic work is certainly part of, but can also play with and reflect some of the social assumptions about current biomedical technologies. Artists explore a range of technologies in order to find out more about their wider impacts. They also question the roles played by institutions in assembling, ordering and using these technologies. An artistic perspective does not intrinsically stand in opposition to other methods of knowledge production or story-telling, but it might contribute unexpected knowledge about how knowledge is produced and constantly transformed.

Artistic exploration often aspires to be a form of condensation. Hypotheses, observations, and reflections are combined or compared in an unexpected way and exposed to the public – and by doing so, artists try to make certain yet unarticulated aspects of a theme obvious, they highlight insights that have remained unspoken or unthought, insights that nobody has dared to articulate or explicate. They therefore take from the invisible, the unspoken, the unheard, in order to make something epistemologically accessible.

Knowledge Practices in Worldly Webs: Artistic Genealogy in Times of Biotechnology

> […] at every level of the onion, scientific knowledge, like all other kinds, remains constitutively historical and non-innocent. All of the actors, human and non-human alike, in these knowledge practices are situated in dense, worldly

> webs. Primate sciences–both what is known and how knowledge gets crafted – are naturalcultural all the way down. No more than any other kind of knowing, progressive knowledge does not ever precipitate out of the viscid brew of worldly configurations. (Haraway 2004: 4)

In the second half of the 20th century, the possibilities for accessing and assembling data about a person's history and family, for enriching knowledge about genetic ancestors or individual dispositions, and also for finding ways to connect with formerly unknown relatives all attained new dimensions with the emergence of novel biomedical technologies (Egloff et al. 2011; Jasanoff 2007; Ong and Collier 2005). These social-biological technologies increasingly seem to allow deep introspection in the bodies of human and non-human beings. They are prone to feeding age-old longings to know more about where we come from and how closely we are related to others, both humans and other animals.

Without a doubt, and accordingly, the biomedical sciences and their technologies can be understood as being embedded in an assemblage of heterogeneous practices and discourses (Michael 2011). They dispose of a complex potential to create new (human/human as well as human/non-human) orders, and they exert influence on how social (power) relations are enacted and also how needs and desires of belonging are inspired and experienced. More importantly though, these biotechnologies and the accompanying scientific knowledge are both informed by, and constitutive of, changing narratives about kinship, family and genealogy, which allow multiple fantasies about free as well as enforced memberships and affiliations.

When we, a temporary community of three scientific and artistic researchers, started our conversations about ancestry and identity, we came across a certain range of institutions and specific technologies which seemed important: Among them were art and natural history museums and zoological gardens as well as communal archives, family picture albums and ancestry tests. They are all full of images and narratives as a means of arranging arguments and logics, which contribute towards constructing the world in a certain order (Haraway 1989). But of course we also aimed to find out how far re-arrangements of these narratives could go. Maybe we should state even more clearly, as a kind of heuristics to this chapter, that we tried to find a way to undo what Irvin and Michael have called 'ethno-epistemic assemblages', that is, 'knowledge claims that are established and circulating locally; knowledge claims that embrace certain representations of the future as well as scientific knowledge, general knowledge, juridical knowledge, journalistic knowledge etc.' (Michael 2011: 59; or also Irwin and Michael 2003: 113). In other words, we set out to undo a specific, seemingly stabilised, kind of knowledge about genealogy and belonging by operating with artistic means.

In contemporary western societies, genetic insight is very often expressed in the form of scientific knowledge and expertise. Yet it is also entangled and wrapped in stories that tell of the human longing to access the past, to find personal, individual history. The internet in particular serves as an ideal platform to host the stories of

people who are looking for common ground or trying to bridge perceived gaps between themselves and those who lived in the past, or those who are presently and seemingly too far away.[2] Genetic storytelling is undoubtedly closely connected to questions of identity. Or, as Jerome Bruner puts it: 'Telling oneself about oneself is like making up a story about who and what we are, what's happened, and why we're doing what we're doing' (Bruner 2002: 64).

The stories are complicated by the fact that a dense network of actors and practices is contributing its version to the meaning and positioning of genetic knowledge as a biomedical technology. They all add sociological, medical, philosophical, ethical, artistic viewpoints and understandings in order to provoke, as well as produce and contribute, to the assemblages and their inherent representations. The way in which scientific knowledge, the behaviours of animals, personal emotions, poetic insight, as well as 'naturalcultural' imagination are translated into each other continues to intrigue us, and will form the focal point of the remainder of this chapter when we present the two artistic works more closely.

Disguise and Network: A History of Descent

For the last few years, the performance artist Mo Diener has been pursuing an artistic research project on the genealogy of her own family. By repeatedly seeking to interrogate her mother and other relatives, by looking through family albums, researching in national and communal archives and finally by producing her own performances, photos and films, she nourishes her suspicions about the gaps in her family history, of illegitimate children, vagrant ancestors and failed existences, and thereby reflects on how these ideas might shape and make-up her own personality. Mo's work that will be discussed here mainly consists of two parts. We will discuss certain aspects of a lecture-performance that drew on her archival work and which was combined with pictures of an artistic installation made some months earlier.

The starting point of Mo Diener's lecture-performance was her mother, Doris Diener Naef. An early photographic portrait shows young Doris, a healthy looking girl with two thick plaits, in a portrait style typical of the 1940s. Doris' father was station master of the Swiss National Railway and the family had to move many times. But what made Mo Diener's mother difficult and interesting, both to her daughter and possibly to others, is a strange recurring (family) reputation of having vagrant ancestors. This tale was not based on documents or any other material evidence, but on mere hearsay. Moreover, Mo Diener's mother was fond of colourful clothes, never got on very well with the neighbours, was resistant to settling down when her husband bought her a house, and was very happy when the family bought a caravan for holidays. Her mother Doris' restlessness and the memory of some family rumours about her mother's ancestors followed Mo Diener into her own adult life and stayed with her long after she had become an artist. It

2 For example: https://www.23andme.com/stories [accessed: 17 September 2012].

eventually led Mo to consider taking a DNA test as an artistic research project. She saw this as a step towards realising her quest to gain more clarity about the family history and to follow some of her – more suspected than evidence-based – personal-historical biographical traces. Yet, instead of undergoing genetic testing, Mo started to combine her artistic approach with an ethnographic research method.

Over the course of time, Mo Diener visited a range of archives, searched through relevant literature, got to know some travellers and repeatedly talked to her mother and other relatives. She soon realised how difficult it was for her to access her family's past. There were not many records documenting the life of her ancestors. Perhaps the most precious souvenirs she managed to get hold of were a couple of photo albums. Amongst them, she found a photo of her grandmother Rosa Josephine Brunner and two other pictures of her great-grandmother, Anna Luise Peter, which – during Mo Diener's lecture performance – the public came to see (Figure 5.1, p. 88).

One of the pictures shows her great-grandmother at a market (Figure 5.1, p. 88). The story behind it, which was passed on to Mo by her mother, is as follows: The woman had lost her first husband early in life and was therefore left with four children to feed. Subsequently, and in order to make a living, she worked as a stallholder in the years that followed. Mo Diener remembers well that her mother and her aunts were not talking too nicely of this woman. To work on a market had not been respectable enough for the rest of the family and the young aunts had been told not to greet their grandmother in public on the market.

The picture does not convey much of Anna Luise Peter's daily work. But we see her in action, posing in a humorous shot with someone in a bear costume. Yet the picture contains a question mark. Who was behind the mask? The absence of an answer to this question makes the picture well suited to encourage all possible fantasies about what happened, there and then, to this great-grandmother or her relatives: was this her lover, a traveller, might he or she be connected to the myths prevalent in the family?

Interestingly, the picture finds its echo in a second one. This time, it shows the same great-grandmother amongst other family members (Figure 5.1, p. 88). In the absence of reliable information, we estimate that the photo was taken sometime in the 1920s or 1930s. The sitters in the picture are arranged in a traditional way: the head of the family – we suppose – is positioned in the centre, next to him his wife, six children in the front row and some relatives to the side. Yet the only known person in this picture is Mo Diener's great-grandmother with her (unnamed) second husband. The adults, men and women, may have put on their best clothes for the photograph, while most of the children were allowed to remain barefoot. Mo could not say much about this photo and the empty space left her, as well as us, guessing again. Maybe a wandering photographer had passed by and taken the picture? Was it the birthday of the head of the family?

The pictures convey to us in what we see – the arrangement, the centering of the *pater familias*, etc – some of the social uses of photography (Bourdieu 1989), but more importantly here, they also point to the fact that it does not take long for

intimate knowledge to become lost. Apart from the great-grandmother, neither Mo Diener nor we know whom or what we are seeing, and who is looking out at us in the pictures.[3]

Neither of the two pictures gives us any hints about travellers in the family; both vagrants and other such identities are left to oral tradition and the imagination. Such 'lay sociograms', of which we have two examples here, 'provide an extant visual record of social roles and relations' (Bourdieu and Bourdieu 2004: 601). But – as family secrets – they are 'typically stored away in a box as it would be indecent or ostentatious to display them in one's home' (Bourdieu and Bourdieu 2004: 601). Indeed, it took Mo Diener some power of persuasion to get hold of the family albums in the first place and to come to some minimal information about some of the photos, in the second.

While the information she could withdraw from these photos was only so much, Mo was continuing her research in public archives in order to find out more about her origins. This was knitty-gritty work, but it allowed her to reconstruct the maternal family tree for the last five generations. It was in the archive of St. Gallen that she found the only factual gap. For Mo Diener's great-great-grandfather Heinrich Albert Peter, born the 9th of June 1855, only a mother, Magdalena Sophie A. Peter, was registered in the document of civil status. A father seemed missing and could – therefore – be anybody, a wandering tradesman, even a traveller, a Jenisch, as they are called in Switzerland. Where to go from this information?

Still we have no clue about which story might be more accurate and whether all of this is completely exaggerated. Mo Diener herself explicitly did not want to think in such concrete terms of lovers and secrets. The pictures did not challenge her fantasy and this was one reason why she later searched for a new access through an artistic operation. We realise that, together with the photos, puzzles of such narratives often remain fragmentary or hidden. Indeed, Mo told us that she had to overcome the sceptical looks, the incredulous questions, also the silences, regarding her interest in these long forgotten pictures, the archival documents and the stories behind these materials when she approached her mother, her aunt or other relatives. Her questions functioned as a kind of accentuation of the imprint of something that occurred in the past, of stories that had been told long ago but that were now returning as fragments, when the re-presentations were brought out of the dark, bringing with them the emotions of happy days, fears or loss.

Mo's search for her relatives and the knowledge about her family's ancestry seemed to face dead-ends and silences, the documents she had found brought more questions than answers. It was at that time that she decided to do a photographic

3 A family tree is inexistent, which leaves both Mo Diener and us with not much more than the uncertain assumption that the photograph does indeed display family ancestors. We can presume that the kind of photographic postures, the mystical market situation as well as the more classical family portrait are directly connected to the (hierarchical?) social relations fostered by – we guess – a rural or semi-rural society, 'in which the lineage and the 'house' have more reality than the particular individuals who compose them' (Bourdieu and Bourdieu 2004: 602).

work using her mother's clothes. Again, it was not easy for Mo to convince her mother to hand out some of her most personal belongings, to take those clothes out of the boxes in her wardrobe. Like an echo to the fragmentary imprints of her great-grandmother, the installation 'Daily routine. An artistic workout' (Swiss Jura 2010) contained some clothes her mother herself had made. In this way, the installation represented a tangible, condensed form of Mo's search. Her mother had always loved sewing and knitting, in fact the movements of her hands had always been an important driving force for her desires. Over the course of her life she had produced a series of colourful clothes – jackets, shirts, skirts, pullovers, dresses. They – according Mo – mirrored her inner world while they also allowed as a kind of second skin her appearance in public. For her installation work, Mo hung them up between some trees in a rural landscape, and also on a hanger on the wall of the house, and took the photographs while they were floating in the wind (Figure 5.2, p. 89).

The result was astonishing. The clothes were able to open up the space, they formed nets, they made personal structures visible. The photos allow insights into the contours of a person; something is absent but seemingly imprinted in the clothes. Their colours build stark contrasts with the environment around them. They form traces of memories. They reflect the shapes of the human inside them. They reveal a form, an aroma, a body.

In some ways we – and amongst us, Mo Diener, the daughter – are now given a representation of this woman, whoever she was and whatever fantasies and wishes she might have had. And the ideas of past lives, whether glorious, adventurous or tragic, of innumerable grand- and great-grandmothers can be projected onto this representation. The pictures of her mother's clothes allow a new perspective onto something Mo must have seen many times; they cast new light onto her mother and her relations to the world.

They even allow two different views of her, for Mo differentiated between the clothes that were sewn and those that were knitted. While the latter were produced and used in an informal, family context, the mother made the sewn clothes for more conventional, formal reunions. The photographic installation – again a kind of Bourdieu's 'lay-sociogram' – therefore, not only brought to the fore something of this lived life, but also the social roles taken on by the mother, not only for her daughter but for others as well. It also offers insight into the contingency of Mo's understanding of her, and also of her relationship with her mother. Identity, according to Deleuze, is never fixed. 'The experience of self, of 'I' comes into being through an assemblage of relations' (Fox 2012: 77).

It is this unstable experience which we encounter again in a small story told by Mo Diener, and one which was connected with the transformation necessary for this artistic work. Before being able to work with these clothes, Mo felt the urge to wash them thoroughly. The stabilisation of the representation, the installation – which was also necessary, in a similar way, for the photographs to be developed, fixed and made visible – was in need of a procedure where some characteristics had to go in order to be replaced by others. The analogies to the production of

photographic images were still present in the exhibited work. This aspect came to the fore in an email she wrote while we were drafting this chapter:

> My daily routine. An artistic workout (installation, Jura, 2010) with these materials shows the potential of their material flexibility in a moment of conflict with myself and my perception. It was easy to stretch them between branches, and they found new existence in the intermediate spaces. (The sewn clothes were at their best hung up; their form played in the wind). They could be hung and dried both conventionally as well as upside-down … the momentary appearance, not the form of the objects, became the subject (…) I carried this work out with intense concentration – it was, in this sense, a creation akin to that of a sculptor: I 'let the material speak' … so perhaps the clothes speak of something very specific.

The traces that were left behind Mo Diener for her to grasp hold of and start telling stories with were few. A couple of photos, some civil registry data, a series of conversations with her mother and her aunt, and a photographic performance of her mother's clothes drew attention to the huge gaps in knowledge. These gaps – however – became much more important: they became telling, speaking. They functioned as triggers, provoking a response. Instead of being blanked out – as is usually the case when genetic material is tested – the gaps became meaningful. In between the pictures, data, and words, life had been going on, life was experienced. Maybe a traveller came along and enabled an intense romance, maybe poverty was suffered or a life on the margins could not be avoided. But as life had to go on, other times came, and there was a mother who loved travelling, and when she couldn't go on a journey, she was knitting and sewing. And it is all these ideas about what might have happened that become readable and thinkable when the laundry is transformed into a piece of art. And not only this; the meaning of the dresses themselves has also changed, what they bear – the lives of at least two women, Mo and her mother's, will be understood differently from now on.

The photographic installation, however, also gives some hints into the ways family histories are told. What would a genetic test – a slightly different representation – have made with Mo Diener and her artistic work or with her relation to her mother? She would have found out, via the genetic information, something about her genetic proximity (or distance) to Roma, Sinti or Jenisch, i.e., travelling ancestors. Thus, it might have enabled her to tell another story than the one she came across via photo-albums and historical archives. But what would she have found out about her relationship with her mother? She would have had to make sense with the statistics in the first place and have to decide how much the numbers mattered. It is hard to judge since a test had not been taken, but we may guess that despite an apparent certainty, the space for fantasies would have remained widely open.

Surely, the in/stability of narrative information through oral tradition or through scientific measurement is entirely different. But the artistic photographs, the representation of a mother-daughter-relation, bear something more than just some

old clothes or raw numbers. They tell of a childhood, they speak of emotions, but they also show the entangledness and networks of relatives, finally they disclose how difficult it is to talk about family matters even within close generations and therefore, sometimes, as simple as that, images reveal more than words.

Living and Dying in an Exhibition

A second work of art that sparked lively debate was a video installation by Luzia Hürzeler, created in 2009/10. In this work, questions such as what constitutes humans or non-humans play a central role. Beyond this, it is also dedicated to the relationships between humans and animals, how they construct each other, and how these relationships are articulated or can be made visible artistically.

In her work 'Il Nonno',[4] of which we have included stills in this chapter, a lion encounters his taxidermic grandfather. While we don't know whether *he* knows this, we know that the lion is facing his dead relative. We watch him watching, yet the human viewer ultimately has no clue what he/she sees the animal seeing. The pictures (Figure 5.3, pp. 90–91) show a kind of 'neutral' enclosure. An animal gazes quietly towards the lens of the camera. Only on very close reading does it become clear that the lion is dead, that it is in fact a stuffed animal. When watching the video, we are left with the illusion that this is a still-photograph, until the calm is broken and a living animal enters the scene.

When the living lion enters the set (Figure 5.3b, p. 90), first of all he circles the stuffed animal, then he also snuffles and gazes around (Figure 5.3c, p. 91). But there comes a moment where he poses stock-still, directly in front of his taxidermic companion (Figure 5.3d, p. 91). Once he has eclipsed him fully, he gazes directly into the camera. What a happy coincidence: The young lion enacts the perfect scene!

In her filmic installation, Luzia Hürzeler enables the audience to watch a lion at close distance, and this can be seen again in the stills she chose for this chapter. In addition, she also aimed at bringing together two institutions which are important and topical: the natural history museum and the zoo. In one of her video stills, we see a stuffed animal that could be placed in a collection, and we also encounter a lion, which she has borrowed from a family of lion tamers. The gaze in each plays an important role.

One has to remember that the eyes of the stuffed animals in natural history museums are always artificial. They constitute the most difficult part of the taxidermic process. Yet, Donna Haraway writes about the animals in the famous dioramas of the American Museum of Natural History: 'Each diorama has at least one animal that catches the viewer's gaze and holds it in communion.' But the taxidermist is not playfully arranging this setting. Haraway continues: 'The animal is vigilant, ready to sound an alarm at the intrusion of man, but ready also to hold forever the gaze of meeting, the moment of truth, the original encounter. The

4 The Grandfather.

moment seems fragile, the animals about to disappear, the communion about to break … ' (Donna Haraway 1989: 30). The very thing which is enacted in these famous dioramas and constantly searched for by the visitor during a trip to the zoo enriches Luzia Hürzeler's video: The lion's gaze into the camera is key to this work. The lion performs exactly what the viewer is looking for.

At first sight, Luzia Hürzeler's story about the two lions seems to be quickly told. But it also offers a series of complex themes if we look closer and listen more carefully to what the artist tells us about how it was made. The title 'Il Nonno' does not reveal the place or time, let alone names. In the video installation, as well as in the stills, an unnamed lion encounters his taxidermic counterpart. But they meet in a place which is neither the arena nor the museum. The location for the encounter is a kind of third space. A concrete floor covered partially in mulch, with white linen attached to the cage. It represents a White Cube, not a pure one, but one which would need some cleaning.

Luzia Hürzeler, who created this work during an artist residency in Rome, will never forget how difficult it was to find an institution which would allow the meeting she was planning. Neither the Museo Civico di Zoologia di Roma nor the Giardino Zoologico di Roma granted her access for this purpose. She eventually came across a circus on the outskirts of the city which enabled her experiment to take place. Interestingly, this family of animal-holders handed down their business from one generation to the next and lived in close proximity to their lions, while the lions were bred and raised again from one generation to another. The stuffed lion, the grandfather, actually, had been a kind of family hero since he had starred in a film and roared in a commercial. After his death he had been stuffed and later been put in a storage. When he was brought out to the circus for Luzia Hürzeler's artwork, the family was seemingly touched by the reunion.

The aspect of belonging is important here, the lion ranged as a kind of family member to the circus owners. By doing so an important paradox comes to the fore that very often beleaguers exotic animals in zoological gardens. Usually, zoo animals are assigned with a place of origin to provide background information for the visitors. This observation is clearly underlined in zoological gardens and also perpetuated by the statements on the labelling plates in front of animal enclosures. However most of these animals have never seen their 'origin'; they were born and brought up in the zoo. This offers up an analogy to genetic identity testing. Even in the context of the zoo – or the animal taming family as in Luzia Hürzeler's work – the desire to know the *one* place of origin remains crucial. No matter how many generations have been raised in zoological gardens, their original 'lebensraum' is stabilised, even constantly reiterated by the labels on the plate – as they are on the information sheets of genetic identity kits, and narratives. The Roman circus-family did it their way: When the lion was brought from the storage, first of all, they placed their youngest, a girl, on top of the taxidermy and took a family snapshot to remember this event.

Luzia Hürzeler's work thematises these questions of belonging. By setting the encounter in a 'neutral' territory, she allows the audience to speculate as to the

place and origin. As an artist, she can mobilise and unite objects that would usually have remained separate institutionally. This equals a kind of gesture; she follows an obsession, she strives to see something which she – and others – have not seen in this way so far.

The terrain proved to be significant in a second way, too. During her stay in Rome, Luzia began to visit the Museo Civico di Zoologia di Roma regularly and became fascinated by the corpses of dead animals. She started to talk to the taxidermist who was responsible for the preparation of many of the animal exhibits. She soon learned that a large number of the objects were not representing animals that had been caught in the wild, but that they were brought there from the nearby zoo. The taxidermist perceived himself as a sculptor, breathing new life into the dead animals. He had often known the animals that he worked on personally, and also the reason why they had died. His work was carried out in a little house that was placed between the two institutions, the zoo and the museum. It was not by chance, however, that this place was not located in the main building. The preparation of the animals, as Luzia was told by the taxidermist, was considered to be 'dirty' work. Dirt – according to Mary Douglas (1966) – is symbolic for disorder. The preparatorium, the house where the animals are stuffed once they come in from the zoo and before they leave for the museum, is key in working towards the restoration of order.

The preparation of animals, the dirty work, as Luzia learned during her encounters with the specialist, is a laborious process. The preserved parts, such as the fur and the bones, are carefully combined with other materials, like plastic mass and glass eyes. Parts of the dead animals are then completed and arranged in such a way that they correspond with humans' perceptions or images of a living animal. We recognise the animal from its outer form and the bodily traits. The stuffed preparations are, in a way, similar to the shapes of the mother's clothes in Mo Diener's installation, and surely also to the imprints of photographs.

What differentiates between living animals and the dead? Through the preparation process, the dead animals are transformed. In the process of being prepared for the exhibition, they lose their individuality and start to represent their species as a whole. The system of representation is specific to the institution, and at the same time recognisable in the represented object. (It does not necessarily follow, of course, that the represented animal is ever aware of the categories it is representing.) It is just for the short journey from the zoo to the Natural History Museum that we might become aware of the (visual, oral, literary, sculptural, genetic etc.) construction of identities. The institutions mobilise these forms of articulations and keep them going in order to generate their objects, but also to recreate their orders (Rose 2001). Yet it is precisely this generative work that has been repeated by the artist here, and it is in this moment that we come full circle. The young lion, which would be tied to his origin in the zoo, is confronted with his dead grandfather, a representative of his species in the Natural History Museum. The representation, in this case, deep-freezes the origin of the lion in Africa.

The following is key: We realise that while the animal is looking, we still do not know what he sees, who he is looking at and what this means to him. Does he remember his next of kin and what does he recognise in it, does he want to know where his relative has come from? And does this matter to him or us?

We cannot really answer these questions, but it becomes clear that the representation remains decisive. It allows us – humans and animals – to see from the outside, to grasp some glimpses of what might hold us together, and at the same time, what makes us companion species: Institutions, relations, presentations are constituted of practices of doing and making genealogy at the same time.

So, which novel story can we tell in relation to genetics here? Maybe not so much more than that: this artistic work refers to the fact that we, humans, are able to recognize a lion in a stuffed animal. But the piece furthermore reveals something about the status of representations. The taxidermy represents an animal that once was alive, we can refer to him as an animal even. However, what we really see are parts of his dead body that has been emptied and arranged together with parts that have been made. Thus, we mainly see his skin and look into his artificial eyes. By not knowing whether the young lion recognizes the representation of his dead grandfather, it becomes visible that a representation is made and related through conventions to the human cognition. Genetic information is mainly based on statistics, similarities, commonalities, probabilities etc. By the artistic work the stuffed animal as well as the genetic information becomes visible as crafted objects. As every other representation the taxidermy as well as the genetic knowledge are limited and never can represent a being as a whole.

Conclusion

In this chapter, we sought to gain a better understanding of ancestry tracing and how visual, oral, sculptural narratives are used to generate identities. The topic of belonging, of being part of an assemblage of objects, discourses and institutions in times of biotechnology consistently intrigued and challenged us. We approached these questions from an ethnographic-artistic perspective. Artists can be experts in visualising identity constructions and also questioning them. They explore the means to do so and, in some cases, enable current perceptions and understandings to be called into question. Artists as much as scientists are influenced by discourses of identity – why else would they come up with the idea to develop or take a genetic test in order to find out more about one's mother or other ancestors? And: only profound curiosity in human and non-human relations might bring an artist to confront a living lion with his dead grandfather.

Both works reveal the desire to learn more about where living beings come from and – perhaps – about where they are going. This is what they are both dealing with or trying to find: Only the representation, the video of the lion seeing his dead relative, as well as the one showing a mother's clothes, allow a glimpse of oneself from the outside. Almost as difficult as seeing oneself from the outside –

and mirrors do not really do a good job here – is seeing one's own mother who gave birth to us, who has known us from when we were little, from when we were first able to think. Just as a photographic self-portrait or one's voice on a recorder is hard to bear, the representations force a kind of 'distanciation'. A representation may perhaps even allow us to see some decisive traits for the first time.

So what kind of stories does the artistic evidence tell in contrast to those which a genetic test would have revealed? After all, biotechnological procedures also generate images and therefore representations. These images require no less interpretation than the photos of a great-grandmother at a market-stand. The conclusions from genetic tests come as basic statistical information and as probabilities. Both are intertwined with the desire to pose new questions, to find other answers, to integrate personal (genetic) data into the autobiographical and narrative self (Featherstone et al. 2006).

The two artistic examples shade two aspects of alternative versions of genetic story telling. As in Mo Diener's case, the genetic test that was not taken, remains in the end as another possibility, as an imaginary window into the past that has not yet been closed. A genetic test still bears the hope to be able to tell another story, one that might be differing from the more typical dramas that haunt the average Swiss family, similar to those that Mo Diener came across with her historical search. Luzia Hürzeler's work points to a second aspect of story-telling, one which all representations – be they genetic or artistic etc. – have in common. A representation is crafted and made. Worse, it will never be able to be the object it tries to represent. Genetic tests generate fantasies and they open up the imagination. They are propositions in need of interpretation.

The two artistic pieces made visible that a representation allows us to see something unrecognisable from another side. A kind of imprint, a cast, a cover is produced in the examples we have discussed in this chapter. The dead lion, cleaned and carefully prepared by the taxidermist, is represented by a stuffed animal that does not emanate pheromones and therefore is perhaps not easily recognisable to the younger, living one anymore. We had photos of relatives that were developed through chemical baths before the pictures were able to emerge. We had clothes on a washing-line that had to be washed many times before the daughter could start working with them artistically.

Scientific knowledge opens up space for the imagination – about origin, relatives, migration, and uncontaminated by problems, conflicts, fears. Old photos seem alike; they are imprints of past lives, not the life itself. But they may also indicate what the search is about: They are accessible only through narrative (for example, one has to be told who is in the picture). Clothes floating in the wind are reminders of the mother who crafted them, but who – significantly – is now absent and does not reveal all that could be known about her. The stuffed lion refers to all that we want to know about his living counterpart.

Maybe the pictures are contemplative: Since when do lions in the zoo have grandfathers? Why should animals in the Natural History Museum be 'related' to animals in the zoo, and what sort of kinship would that be? Is the term 'grandfather'

not already a concept that presupposes a very anthropomorphic understanding of social relations, relations we cannot assume animals to have? How are clothes on a washing-line related to other people? What do they show if not that they are, first and foremost, reminders of an absent, lived life? Do they help to speculate that alongside missing husbands, migration, (animal) captivity and death perhaps there has been joy, colours, friendship and freedom?

Finally, while we seek to analyse how artistic work contributes to the production of the cultural significance of genetics, we also wonder whether the stories themselves flow into a discourse that might even and already be our own, and also whether despite doing so, these articulations are able to shed some new light onto all too familiar knowledge?

Acknowledgements

This publication was supported by the Swiss National Foundation (Project 137991, 'Wir sind im Winterschlaf: Eine künstlerische und sozialwissenschaftliche Untersuchung des Mensch-Tier-Grenzen im Zoo' ['We are hibernating: an artistic and social science exploration of human-animal boundaries in the zoo']. We would also like to thank Jamie Lee Searle for her careful proofreading, and helpful comments in completing this chapter.

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Left: Anna Luise Peter, my great grandmother. Born the 1st of June 1882 in Elsau, Zurich.

(Market booth, St. Gall, Switzerland, approx. 1925.)

Rosa Josephine Brunner, my grandmother. Born the 10th of August 1909 in Stein, Appenzell A.R.H. Switzerland.

Doris Naef, my mother. Born the 25th of October 1930 in Dietikon, Zurich, Switzerland.

Anna Luise Peter (2nd from left) with her second husband (left).

(Names, place and date of the photograph unknown.)

Fig. 5.1 Mo Diener – Family portraits

installation view. swiss jura. 2010

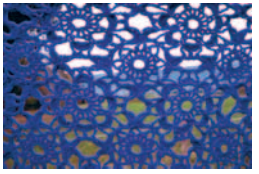

Fig. 5.2 **Mo Diener – Daily routine, an artistic workout**

a

b

Fig. 5.3 **Il Nonno by Luzia Hürzeler – video stills**

c

d

Fig. 5.3 (continued)

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PART II
Sharing Knowledge

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Chapter 6
The Latent Figure Protocol – A Photo-essay[1]

Paul Vanouse

1 Portions of this essay were originally published in Czas Kultury (6, 2010).

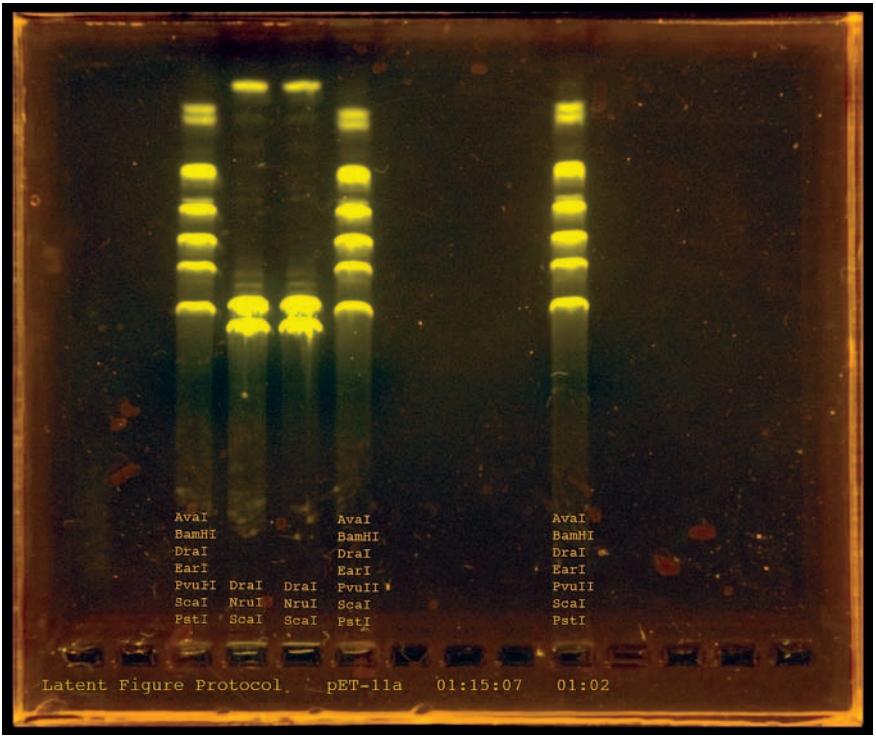
I am an artist who works in Emerging Media forms. My artwork addresses complex issues raised by new techno-sciences (such as how they enter into existing social structures) using these very techno-sciences as a medium. I utilise emerging technologies that have been isolated in their communicative potential and through creative re-use, mis-use and/or even ab-use turn them into communicative media forms.

Recently, I have sought to force the arcane codes of scientific communication into a broader cultural language. The 'Latent Figure Protocol' utilises DNA sequencing technologies to create recognisable images and icons – not simply images of a sequence of DNA in a gel (like a standard DNA fingerprint), but rather highly controlled images that are created with actual DNA in an imaging gel. Each of the following images was created using the same source of DNA, but varying which 'enzymes' were used to process it. The project uses DNA technologies differently than in typical lab work as I believe that artists working in emerging technologies should go beyond use of only pre-existing techniques and creatively 'hack' in this domain.

Information, transcription, translation, code, redundancy, synonymous, messenger, editing, and proofreading are all appropriate terms in biology. They take their meaning from information theory (Shannon, 1948) and are not synonyms, metaphors, or analogies.

—Hubert P. Yockey, *Information Theory, Evolution, and the Origin of Life*, Cambridge University Press, 2005

DNA is a long-stranded, organic, material substance found in every



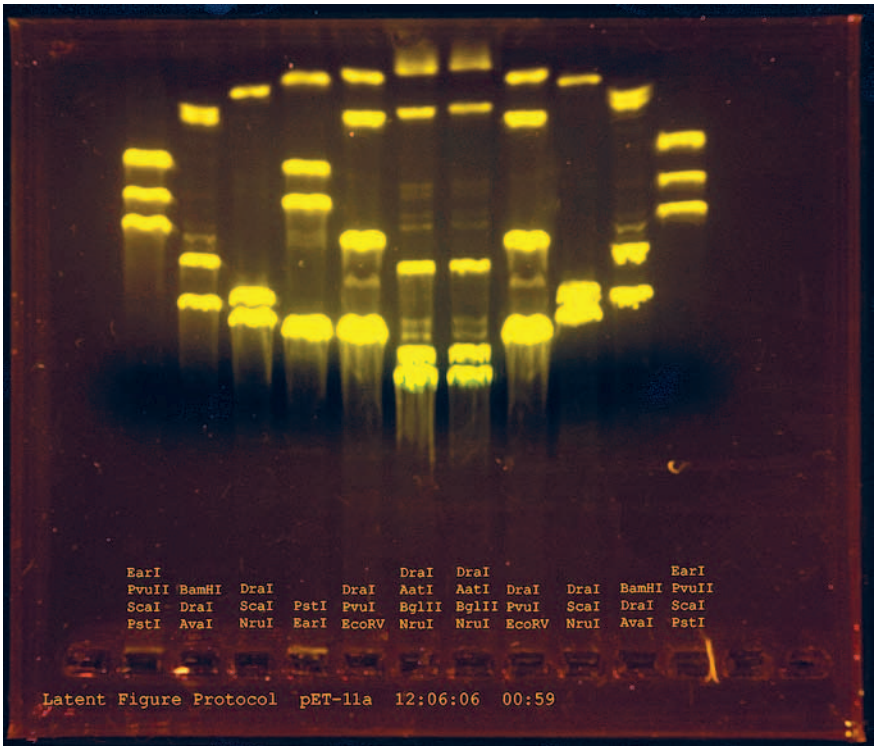
cell of every living organism. However, it is deeply intertwined …

Any company that wants to be in the business of using genes, proteins or antibodies as drugs has a very high probability of running afoul of our patents. From a commercial point of view, they are severely constrained – and far more than they realise.

... Two years from now, we have a big opportunity to remonetise our asset and still have plenty of intellectual property to ourselves ...

—Dr. William A. Haseltine, CEO Human Genomic Sciences in Lawrence M. Fisher, 'The Race to Cash In On the Genetic Code', *New York Times*, August 29, 1999

with slippery analogies and operational metaphors that conveniently



de-materialise it into pure information – particularly when this …

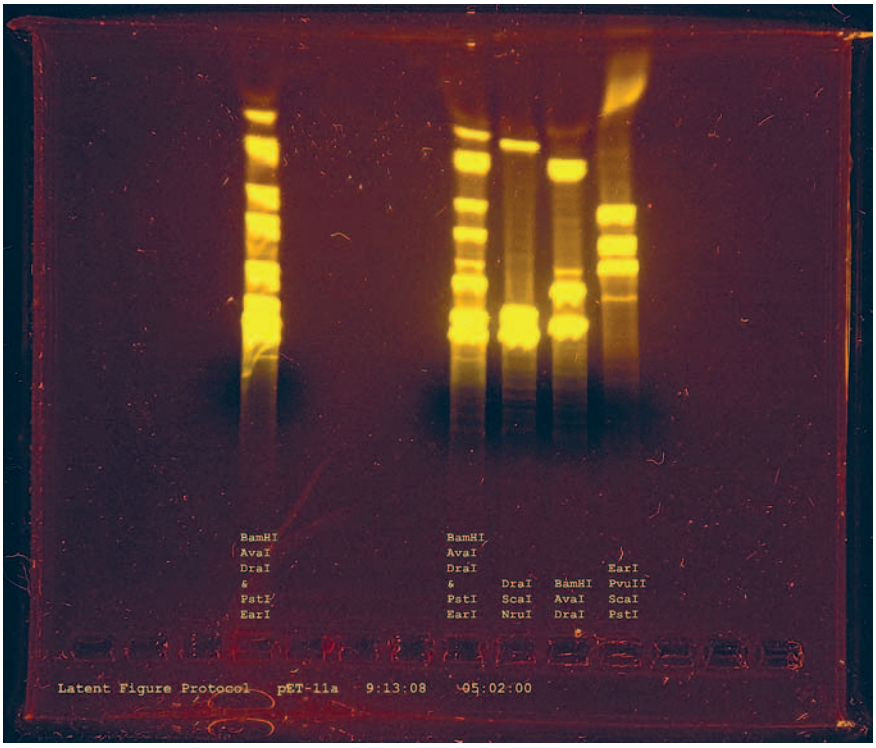
DNA analysis can now rightly be called 'DNA fingerprinting'. The term 'invokes in the mind of the jury that we are identifying one individual to the exclusion of all others'.

—Dwight Adams, chief of the scientific analysis section at the FBI laboratory in Washington, DC

If we had called this 'idiosyncratic Southern blot profiling', nobody would have taken a bit of notice. Call it 'DNA Fingerprinting', and the penny dropped.

—Dr. Alec Jeffreys, genetics researcher and 'inventor' of 'DNA Fingerprinting', University of Leicester, UK

de-materialisation enables new economies of identity, property



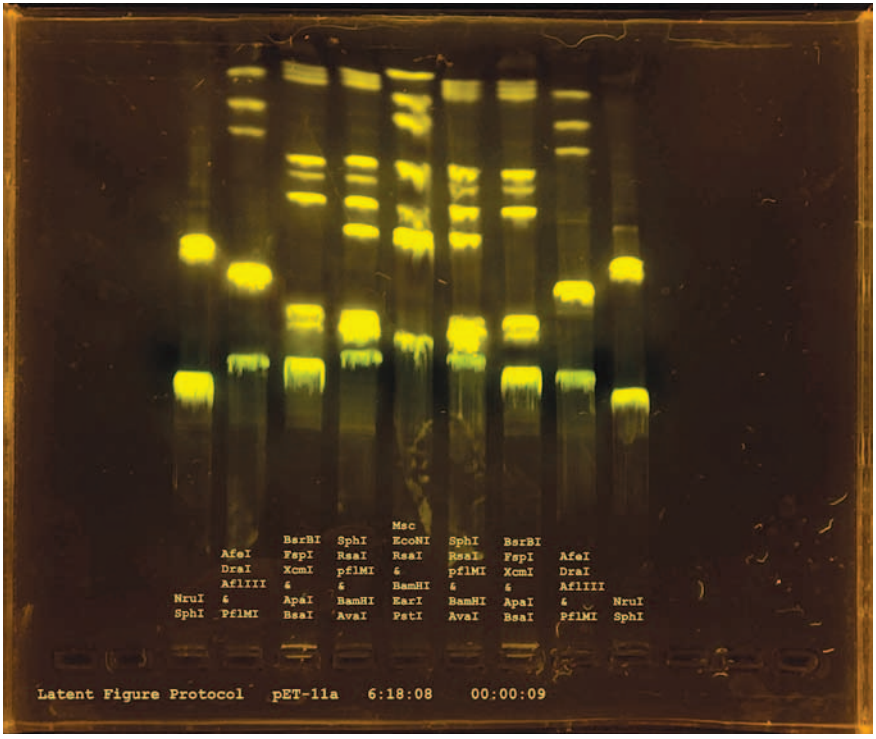
and communication. Likewise, determinist notions and wild …

In health-conscious, sports-oriented Boulder, Atlas Sports Genetics is playing into the obsessions of parents by offering a $149 test that aims to predict a child's natural athletic strengths. The process is simple. Swab inside the child's cheek and along the gums to collect DNA and return it to a lab for analysis of ACTN3, one gene among more than 20,000 in the human genome …

The analysis takes two to three weeks, and the results arrive in the form of a certificate announcing Your Genetic Advantage, whether it is in sprint, power and strength sports; endurance sports; or activity sports …

—Juliet Macur, 'Born to Run? Little Ones Get Test for Sports Gene', *New York Times*, November 29, 2008

hyperbole such as 'DNA as destiny' permeate popular media



and fuel reductivist fantasies. —Paul Vanouse, 2012.

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Chapter 7

Consequences of Sequences, Codes and Messages: Artistic and Scientific Readings of Chromosomes in an Era of Consumerism

Gabriele Werner-Felmayer

> What exactly distinguishes nature from artifact? Are genes the essence of an individual and a sacred part of human inheritance? Or are they […] a form of currency? (Anker and Nelkin 2003: 181)

Capturing the physical basis of inheritance was, and still is, a stimulus for creating ideas about our nature, in a literal as well as in a figurative sense. Along with the development of technical equipment over the past centuries – from the microscope to the DNA sequencing device – the human body was dissected into smaller and

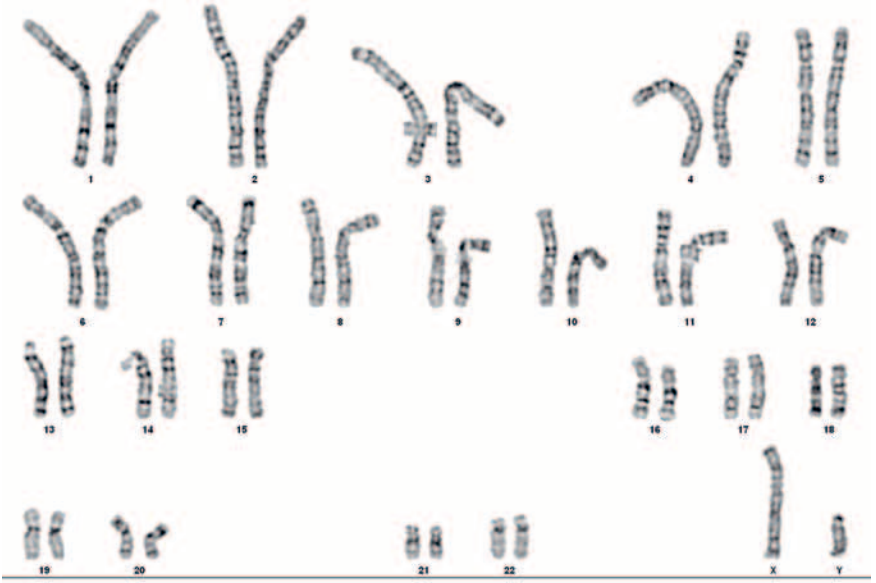

Fig. 7.1 **Male human diploid chromosome set (karyotype) (courtesy of Dr. Christine Fauth, Division of Human Genetics, Medical University of Innsbruck, Austria)**

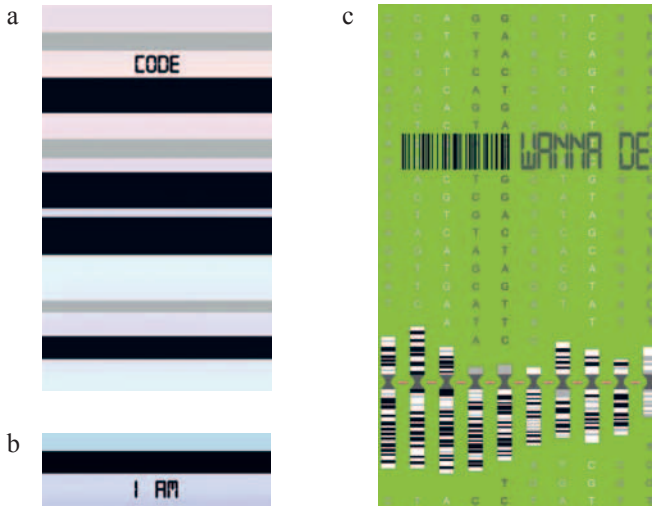

Fig. 7.2 **Design for banding patterns for p-arms of chromosome 9 (a) and of the Y chromosome (b), and for a fragment of the wall hanging (c)**

Fig. 7.3 **Federica Esposito during the performance of Con'Sequences**

smaller entities. Fragmenting the body into its molecular structures undoubtedly improved our scientific understanding of biological processes. Beyond science, it created what Suzanne Anker and Dorothy Nelkin called 'The Molecular Gaze' (2003). (See above quote.) This molecular perspective focuses on DNA, genes and genomes, and their codes and messages. It has influenced Western culture since several decades. Shaped by such an angle of vision, mere chemical structures and molecular models turned into ideograms of the genomic and post-genomic era: The double helix, for example, is no longer just a molecule. It has become a sign of wealth, growth and innovation and in some cases serves as eponym for investment funds as well as skin care products.[1] Moreover, the three-letter acronym DNA has entered business jargon.[2]

A large number of social and cultural science studies deals with this transformation from molecule to idea, and with its consequences on science itself (e.g., Keller 2002; Kay 2000) and on concepts of life and the human constitution (e.g., Nelkin 2001; Nelkin and Lindee 2004; Nerlich, Dingwall and Clarke 2002; Nerlich, Elliot and Larson 2009; Nerlich and Hellsten 2009; Rothman 2001). The visual arts have always played an important role in this transformation. They have functioned as a medium to popularise scientific achievements or as a sensuous, often critical way to uncover broader meanings and cultural interpretations of science. As Anker and Nelkin (2003: 194) observed, 'there remains little doubt that the vision of artists can contribute to our understanding of the real ambiguities of a powerful science'.

This chapter presents and discusses the multi-layered artwork Con'Sequences, developed by the Austrian artist Helene Keller in cooperation with other artists (see below) for the interdisciplinary symposium on 'Genetics as Culture in a Consumerist Age' in 2011.[3] Con'Sequences refers to the iconography of well-known depictions of the chromosome set and its typical banding patterns and explores meanings of humanness and individuality. Its core is a sculpture of human chromosomes that can be integrated into a multimedia performance. Before investigating some of the scientific and cultural coherences regarding genetics in a consumerist age that the artwork takes up, I will provide backdrop information regarding my own role as well as the development of Con'Sequences.

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1 See logos at: http://www.helixpartners.com or http://www.helixskin.com. A skin care product review blog with a double helix model and other images inspired by molecular biology textbooks on its homepage is even called The Triple Helexian (http://thetriplehelixian.com) [all sites accessed: 27 October 2012].

2 In a recent newspaper column, for example, the chief editor of the career section wrote that 'controlling mechanisms run deep in the DNA of businesses' ('Kontrollmechanismen sitzen tief in der DNA von Unternehmen') (Bauer 2012: K1).

3 The symposium, held in October 2011 in Innsbruck, was organised by the editors of this volume. More information at: https://www.i-med.ac.at/ethucation/Events/conference_2011.html.

Viewpoints

The perspective of this analysis bears on a particular line-up: On the one hand, Helene Keller and I have been engaging in a dialogue on form, patterns and shapes of the living already for a long time. This ongoing dialogue occasionally results in my contribution of texts to her objects,[4] as it was also the case for Con'Sequences. On the other hand, however, I perceive the artwork in my role as a scientist and I do this in a dual way: first, I participate in experimental work that requires expertise in genetics and genomics; secondly, I critically explore cultural and ethical dimensions of these research fields (Werner-Felmayer 2007, 2012; Introduction to this volume). My analysis thus reflects this position of being part of a process while at the same time being an observer and commentator. As a result, this chapter does not fit within usual categories such as cultural or science studies, science, or ethics, but it is a systematic reflection of my own interdisciplinary connections with the theme.

At this point, I would like to add some remarks on interpretation: Any literary 'text is a lazy machine that asks the reader to do part of its work […] a machine for the production of interpretations' (Eco 2011: 38). Like such a text also any other work of art allows – or even calls for – numerous interpretations. Which coherences become tangible for a beholder depends on a complex, partly unconscious, and indirect interaction between the one who creates and then releases a piece of art with the one who views, reads, and interprets it. The outcome of this process is hardly predictable, as will also be illustrated below. With regard to interpreting literary writings, Eco defines 'empirical readers' who 'often use a text as vehicle for their own passions, which they project onto the text or which the text coincidentally provokes in them' (2011: 46).[5] Comparable to those 'empirical readers', I allowed myself to use Con'Sequences as a vehicle for my own passions when writing this chapter. Therefore, this text meanders between biological facts and science narratives with special emphasis on the meaning of words. The artist did not intend all of the readings and connotations on which I elaborate here. I also may not have grasped all of the threads she has laid out in her work. Nevertheless, the coherences presented here are the result of our many conversations on Con'Sequences during and after its making.[6]

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4 Helene Keller uses ceramics and different other materials. For her work and some text examples (in German), see: http://www.helenekeller.com/index.php.

5 I translated the quotations from the German edition of Eco (2011): 'Ein Text ist eine faule Maschine, die vom Leser verlangt, einen Teil ihrer Arbeit zu machen […] eine Maschine zur Erzeugung von Interpretationen' (38); 'Empirische Leser', die 'oft den Text als Vehikel für ihre eigenen Passionen [benutzen], die sie von außen an den Text herantragen oder die der Text zufällig in ihnen auslöst' (46).

6 Despite her essential input to this analysis and interpretation, Helene Keller refused to be a co-author of this chapter as she consistently avoids writing about her own artistic work.

Evolving the Artwork

Helene Keller obtained a PhD in microbiology before she turned to visual arts in the late 1980s. In her artistic explorations, she has concentrated on interpretations of sphere, lens and female archetypes. More recently, she has started to engage with expansive installations of different materials referring to the theme of changeability. In her work, the former biologist usually takes a back seat. An affinity to structures and materials found in nature as well as a reference to forms and meanings of life, however, is always tangible. When I told her about the topic of our planned symposium back in 2010 she was instantly interested. She felt intrigued both by the recent progress of genomics and by the marketing and consumer dynamics coming along with it. As she told me recently, the first thing that had come to her mind while we were talking had been pastel colours. She had felt that the shrill advertisement world of consumer genomics with its platitudinous slogans calls for some soft, ethereal counterpart reflecting the beauty of the genome and poetic facets of life. When leafing through biology textbooks, it occurred to her that the banding patterns of stained chromosomes resemble barcodes. This gave her a clue of how she could visually connect the genome with consumerism.

Having found colours, patterns and form, Helene Keller asked me to contribute texts to her work. Words and their meanings as well as signs, codes, scriptures, messages and their apocryphal contents have long been major topics of our mutual interest. The genome as a text and *we* as its *product* are powerful ideas inspiring literary engagement. Moreover, language contains melody. Helene Keller therefore invited Nadia Braito[7] to develop a performance for the artwork's exhibition, musically improvising these short text messages. At this time, Nadia had just found old letters from which she learnt about her grandmother's life in a small village in the Trentino region during the era of Italian fascism. Helene Keller's enquiry therefore coincided with Nadia's dealings with ancestry, kinship, and the quest for identity. During one of our conversations about the project, it occurred to us that in addition to English versions of the text messages, also Italian and German versions should be set to music. In so doing, we wanted to acknowledge several issues: first, that native language is an important part of a person's identity; secondly, that Italian is a highly melodic language important for various music genres; and third, that Trentino is close to the Italian-German language boundary. This boundary is an essential ingredient for the rich regional culture. For setting the texts, Nadia developed her music in a joint project with the sound designer and music programmer Saverio Monti;[8] the project was carried out at the Conservatorio di Como. This allowed for integrating the artwork in a performance with sound and movement as described in more detail below. She

7 Nadia Braito graduated in vocal jazz at the conservatoire of Como, Italy. She sings in various jazz formations and teaches pop and jazz singing in Italy and Switzerland.

8 Saverio Monti (trained at the Accademia delle Belle Arti in Milan and the conservatoire of Como) explores new forms of interaction of various sensory perceptions. More information available at: http://saveriomonti.it.

also got the dancer Federica Esposito[9] on board of this artistic project that had been developing throughout almost the whole year before the symposium.

Because of this extensive interdisciplinary cooperation,[10] the artwork, particularly when showing its full potential by the performance, transforms letter, sequence, pattern and form into the manifold expressions of humans. It refers to a life that is much richer than anything is we can buy or than any DNA sequence may tell us. Additionally, an unintended consequence became obvious during writing this chapter: The artwork communicates science and could even serve educational purposes, as Helene Keller's object represents a systematic conversion of a scientific image (more details below). This is, of course, no straightforward process, as it requires scientific knowledge about genomes, genetics and other fields of biology. In the following, I will outline this background to some extent before referring more specifically to Con'Sequences and possible readings of it.

Chromosomes, Karyotypes, Idiograms and Ideograms

Looking at pictures of chromosomes (Figure 7.1, p. 107), we would certainly not think that they *represent* us, or part of us, even if the picture had been taken of chromosomes from our own cells. Of course, if we remember anything from school biology lessons, we know that Figure 7.1 represents chromosomes of a human somatic cell. In this case, it shows a karyotype, i.e., the entirety of chromosomes of a male individual without any obvious chromosome aberrations (see Box 7.1 for details). If we then consider that these structures contain the roughly three billion base pairs of our genome, they may become more intriguing at least for some of us.

Identifying the sequence of the DNA that is contained in the chromosomes may seem like a key to identifying ourselves. In some respects, this is indeed the case. There is no doubt that our genome is an essential factor for 'making' us the way we are. However, there is also no doubt that the genome is not sufficient to constitute all dimensions of us and our individuality (see also Keller 2002, 2010, and the Introduction to this volume). On the path from DNA to individual, images of chromosomes serve as symbols of individual identity. Companies marketing direct-to-consumer (DTC) personal genetic tests frequently use images of chromosomes. The website of 23andMe, for example, shows chromosome-shaped bi-coloured Xs (23andMe 2012). Another provider, *Genelex*, uses a banded chromosome symbol instead of the x in its name (Genelex 2012). This is notable for

9 Federica Esposito is a graduate from Scuola Professionale Italiana Danza in Milan and obtained training in Simons technique in New York. Her work focuses on dance theatre (she participated, for example, at the festival Grec in Barcelona and the Biennale in Venice). She teaches contemporary dance in Italy and Switzerland.

10 In addition, Werner and Martin Dobler, Karlheinz Ehart, Kajetan Fuisz, Martin Konrad, Adalbert Kathrein and Christian Raffeiner contributed essentially to the technical realisation of Con'Sequences.

Box 7.1 Some Facts about Chromosomes

Karyotype: Human somatic cells contain two sets of 23 chromosomes of maternal and paternal origin, respectively. The cell therefore contains a diploid genome organised in 46 chromosomes. Forty-four of these chromosomes are so-called autosomes and form pairs but the X and Y chromosome do not as they are sex determining and highly different from each other (see Box 7.2). Egg and sperm cells contain only one chromosome of each pair and are haploid (23 chromosomes). Unless showing chromosome aberrations characteristic in certain conditions, schematic karyotype representations or chromosome preparations from cells therefore show 44 chromosome pairs as well as either a pair of X chromosomes (female) or an X and a Y chromosome (male). Karyotype analysis is an important diagnostic and taxonomic tool.

G-banding: Staining chromosomes with a blue dye (Giemsa) yields characteristic G-banding patterns (Bickmore 2001). Giemsa stains phosphate groups of DNA and attaches to regions with adenine-thymine bonding. Areas with a high content of adenine and thymine (AT-rich) therefore stain darker as compared to those with a high content of cytosine and guanine bases (CG-rich).

Centromere and Sister Chromatids: The centromere is the constricted region of chromosomes that separates the shorter chromosome arm (p, from petite) from the longer arm (q, from queue) (O'Connor 2008). During cell division, the replicated sister chromatids are connected at their centromers (symbolised as X) before separation. Sister chromatids are copies considered identical but may be slightly different due to mutations.

Chromosome Maps: Cytogenetic chromosome maps refer to the International System for Cytogenetic Nomenclature (ISCN), a numbering system of chromosomal positions based on banding patterns (Bickmore 2001; O'Connor 2008). For example, the gene for haemoglobin beta is found at position 11p15.4 which means that it is located on the short (or p) arm of chromosome 11 in subregion 4 of band 15 (National Center for Biotechnology Information 1998–).

Condensed Chromosomes: Microscopically observable chromosome stages are 'condensed'. Such chromosomes have already replicated. Condensed chromosomes get compact and transportable and their sequence is no longer accessible for the usual processes of gene transcription. In most cell types, they align along an equatorial plane in order to prepare for segregation of the two sister chromatids and distribution to the two future daughter cells. The molecular processes underlying chromosome condensation remain unclear but the observed irregular folding patterns suggest that chromosome condensation is a self-organising process (Thadani 2012).

several reasons: First, chromosomes are relatively 'old-fashioned' representations of genetics. As compared to the double helix structure of DNA, they are observable by light microscopy and scientists identified them already in the second half of the nineteenth century. Although chromosomes contain the whole genome of a cell, their shapes do not refer to the high-tech sequencing technology and bioinformatics expertise that is necessary for genome analysis. Moreover, this simple X refers to a stage of chromosomes that is visible only at early mitoses, immediately after replication and before separation of sister chromatids. The banding patterns are the result of a staining procedure (see Box 7.1). Chromosomes therefore represent only a transient phase of genome organisation during cell division but not the genome itself or its sophisticated three-dimensional structure.

It seems unlikely that clients of personal genomics services that use schematised chromosomes in their logos know much about the biology of such coloured or banded Xs. It is also doubtful that they are aware of the ambiguity of concepts of identity in the context of genomes and genetics (Dupré 2010; Quitterer in this volume; Werner-Felmayer 2012). Nevertheless, chromosome symbols, apparently, are useful messengers for something that is often called 'information' (see below), that is personal and can be purchased in order to 'know' and 'manage' oneself in a way that is considered to be informed and timely. Hence, abstracted chromosomes, like the double helix, have become true ideograms, graphic symbols representing ideas about who we are as a species as well as individuals. In addition, X-shaped chromosomes have become advertisement vehicles, as they are easily recognisable. They can therefore address a pluralist audience irrespective of the degree of factual knowledge about biology. In analogy to postmodern architecture and art, such symbols of chromosomes seem to integrate the 'slow-moving codes of the past' with 'the ephemeral tastes of fashion' (Jencks 2010: 22).

In this context, reading the scientific literature carefully reveals an interesting inconsistency: scientists use both terms, idiogram and ideogram, synonymously when referring to chromosome schematics (O'Connor 2008). However, the prefixes *idio* and *ideo* have different linguistic roots and meanings. *Idio* – derived from the Greek term *idios* – ('individual') means 'specific to a particular individual' (Prefix Dictionary 2012), whereas *ideo* refers to the Greek *idea*, signifying 'idea', 'form', 'model' or 'general principle' (Word Information 2012). Here, I use the term id*i*ogram for karyotype schematics and chromosome maps and id*e*ogram when referring to chromosomes (or other entities such as the double helix) as symbols of broader ideas and concepts.

Traditional cytogenetics (Figure 7.1, p. 107) shows condensed chromosomes stained with dyes yielding typical banding patterns (see Box 7.1). There are numerous methods available, but G-banding is one of the standard techniques for identifying chromosomes and finding chromosomal aberrations (summarised by Bickmore 2001). Combining various banding techniques, chromosome maps (see Box 7.1) can be generated which inform about location of genes as well as domain organisation of the genome, including patterns of DNA replication as genome regions replicate at different times and this affects the banding patterns (Blickmore

2001). Such an image was the basis for the design of Con'Sequences (see below and Figure 7.4, p. 116).

In an era of genomics, where chromosome maps at single nucleotide resolution are available,[11] it is possible to generate idiograms of any sort by computational methods. Idiograms therefore do not necessarily show the classical banding patterns of cytogenetic analysis but visualise highly complex connections of sequence, location and genetic relationship. Currently, circular idiograms generated by the Circos software (http://circos.ca) are popular and appear to be state of the art when presenting complex relationships of genomic data in scientific journals and other media (Krzywinski et al. 2009). Another software, Idiographica (Kin and Ono 2007), generates less fancy but more familiar karyotype idiograms with banding patterns. It is possible, for example, to generate a karyotype showing the recombination pattern of paternal and maternal genome portions from personal genomic data, thus turning abstract figures into a telling image (Bisignano 2009). Such idiograms also turn to ideograms in the true sense as they allow viewers to develop ideas about how their individual genome and its peculiarities may constitute them, e.g., because of parental recombination patterns.

Con'Sequences

Helene Keller started her artistic exploration for Con'Sequences from idiograms of G-banded chromosomes as originally published by Francke (1981). Figure 7.4 shows the design for Con'Sequences based on a haploid human karyotype representation commonly found in textbooks as well as on websites of genomic databases.[12] This karyotype displays the 22 somatic chromosomes as well as both sex chromosomes (X and Y). Therefore, it shows all 24 chromosomes of the human genome sequence rather than the karyotype of an individual's somatic cell (46 chromosomes) or gamete (23 chromosomes). The line connects the centromeres, the short p-arms pointing up and the long q-arms pointing down (see Box 7.1 for background and terminology).

11 Chromosome Map Viewer developed by the National Center for Biotechnology Information is available at: http://www.ncbi.nlm.nih.gov/mapview.

12 See for example the representation of the human genome organised in chromosomes on Map Viewer at: http://www.ncbi.nlm.nih.gov/projects/mapview/map_search.cgi?taxid=9606&build=104.0 [accessed: 16 May 2013]. This representation includes the mitochondrial genome as an additional entity. Such a design refers to the whole human genome. If it represented the genome of an individual, it was male as it includes the Y chromosome.

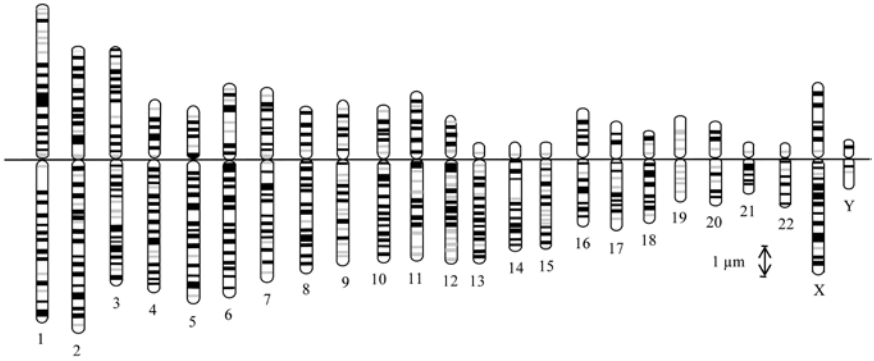

Fig. 7.4 **Design of Con'Sequences based on a haploid karyotype including both sex chromosomes of G-banded human chromosome idiograms**

In referring to banding patterns, the artist tied in with metaphors of language, script and code, that have influenced biological sciences (reviewed by Kay 2000) as well as different art movements for several decades now (for numerous examples see Anker and Nelkin 2003: 9–45). She also resumed some of her previous work where she had dealt with cryptographs. Biologists like Helene Keller and myself, who started their training in the late 1970s, grew up with the concepts of code or code words, information, letters, and messages of a DNA sequence that had entered the field of biology only during the second half of the twentieth century. The idea that deciphering a code could explain life itself or tell us what it means to be human, however, rests upon an excessive simplification of complexity. To go along with Erwin Chargaff,[13] an early critique of this notion, life appears to be an unfathomable mystery as 'a strictly scientific understanding of life is impossible'[14] (1989: 46–47). The basis of this stance reflects neither an anti-science position nor metaphysical considerations. Neither does it deny the usefulness of metaphors to understand some molecular mechanisms and biological processes. Rather, it is the result of many years of observing and marvelling at manifestations of life from various perspectives, and an 'affinity for philosophy', as Erwin Schrödinger (1964: 10–11) called the 'astonishment' about experiencing limitations of logic and science in view of a complex whole.

13 Erwin Chargaff (1905–2002) was an Austrian biochemist who emigrated to the US in 1935. He discovered base pairing of nucleic acids (Chargaff's rules, i.e., adenine pairs with thymine, cytosine with guanine). His essays and texts (including aphorisms and poems) about humanity, nature, history, language and the role and responsibility of scientists are an important critique of science in an age of consumerism and its reductionism and exaggerated aspirations.

14 Original text: 'ein strikt naturwissenschaftliches Verständnis des Lebens [kann es] nicht geben'.

In this sense, Helene Keller's sculpture is not only an aesthetic representation of human chromosomes and their banding patterns but also a playful and ironic interpretation of exaggerated scientific claims regarding its capacity to explain personal identity, humanness, and life itself. The name of the artwork is programmatic as it plays with various significations of 'con': the Italian preposition 'con' means 'with' as the artwork deals with sequences; the Latin prefix 'con' turns sequences into consequences which signifies effects, impacts, implications or results. Something can also be of no consequence, however, and the artwork takes up some of these connotations. Moreover, if sequences were 'con sequences', one better should not trust them at all.

The Sculpture and Its Patterns

Con'Sequences consists of acrylic glass rods illuminated by light-emitting diodes (LEDs).[15] The rods symbolise 'chromosomes', fixed at their 'centromeres', i.e., stainless steel connectors between the p- and q-arms, on a horizontal stainless steel moulding. The dimensions[16] of the rods are true to scale (100000:1) indicating that chromosomes 1 and 2 are the largest whereas the Y chromosome is among the smallest ones.[17] A human cell's nucleus, and hence its chromosomes, contain nearly two metres of DNA (McHugh and Heck 2003). In the scale of the sculpture, this approximates 200 km.

The banding patterns characteristic for each chromosome were printed in black and grey (referring to different densities of the G-banding patterns as indicated in Figure 7.4 based on Francke 1981) on transparent foil with different pastel colours between the bars. In some of these coloured sections, key words like 'code', 'message', 'decode' and short texts, e.g., 'I am', referring to text messages (see below) were printed. Figure 7.2 (a, b, p. 108) shows two examples of the design. The sculpture is supplemented by a wall hanging made of tarpaulin (5 x 1 m) with a printed 4 letter code sequence (referring to a DNA sequence) interrupted by text fragments translated into barcodes (see Figure 7.2, c, p. 108). In principle, it is possible to translate whole sentences into barcodes (see Figure 7.5).[18] However,

15 See: http://www.youtube.com/watch?v=uVBgekWKgSA [accessed: 18 December 2012] for a short video sequence of the illuminated sculpture.

16 The longest rod (left, 'chromosome 1') measures 109 cm, the shortest (right, 'Y chromosome') 17.5 cm. The sculpture spans over 284 cm and its width is 16 cm.

17 Total length is 249, 243 and 59 million base pairs, respectively, according to assembly statistics for the human reference genome (Human Genome Assembly Information 2012). Note that chromosome condensation is irregular (see Box 7.1) and therefore the length in base pairs does not exactly correlate with the length measured for condensed chromosomes. For example, chromosome 21 (48 million base pairs) is the shortest in terms of sequence length (Human Genome Assembly Information 2012) but in cytogenetic karyotype preparations the Y chromosome appears to be smaller (Francke 1981).

18 The text was barcoded using the free software TEC.IT available at: http://barcode.tec-it.com/barcode-generator.aspx?LANG=de [accessed: 18 December 2012].

Fig. 7.5 Barcoded sentence

the wall hanging displays only barcodes of text fragments that are accessible to mobile tagging.[19] This allows viewers to decode the messages encrypted in the barcodes using their own mobile phones.

The Performance

For the performance, Saverio Monti had woven Nadia Braito's vocalised interpretations of the English, Italian and German text messages (Table 7.1) into electronic sound figures. Equipping each 'chromosome' rod of Con'Sequences with a touch screen allowed Federica Esposito to call up the vocal improvisations according to her movements while performing.

Figure 7.3 (p. 108) shows Federica Esposito during the performance on 28 October 2011. The multi-media performance of Con'Sequences took place in the evening. Therefore, the only light sources were the illuminated 'chromosomes' of the sculpture in the otherwise dark baroque hallway of the Faculty of Theology at Innsbruck University.[20]

The performance started by the dancer lying motionless in front of the sculpture, nearly invisible to the audience, and draped in fabric. The only sign of life was the acoustic voice of the singer rising out of the dark. She slowly approached the audience from a distance of about 80 metres, her beautiful voice carried by the exquisite acoustics of the exhibition hallway. In this mystical atmosphere, the dancer started to move, freeing herself from the textile like a pupa from its cocoon. Upon having released herself, she started to touch individual 'chromosomes' calling up the electronic versions of the texts while the live voice of the singer faded away into the dark. By her choreography, the dancer expressed human complexity that goes far beyond being an encoded market product. She allegorised individuals as the mysterious and vulnerable creatures they are, searching for identity, fighting for individuality, freeing from bondages, codes and matrices.

19 Mobile tagging includes reading (e.g., by photography with a mobile phone); decoding by a mobile software application; connecting to internet sites and into virtual networks for, e.g., mobile marketing (information at: http://en.wikipedia.org/wiki/Mobile_tagging) [accessed: 18 December 2012]. The quality of the mobile phone camera limits the length of barcoded texts, as electronic reading requires a certain resolution of the barcode.

20 There are regular art exhibitions in this hallway, which is about 110 m long with large windows, arches and stuccoed ceiling.

Table 7.1 Text messages for Con'Sequences

I am a coded message.	Ich bin eine verschlüsselte Botschaft.	Sono un messaggio cifrato.
Wanna decode me?	Willst du mich entschlüsseln?	Vuoi decodificarmi?
Barcode is not a code at the bar.	Barcode ist kein Code an der Bar.	Barcode non è il codice del bar.
Cod the code and escape the codex.	Überliste den Code und entrinne dem Kodex.	Aggira il codice e salvati dal codice.
Decode the code and find the message, if there is any.	Entschlüssle die Verschlüsselung und finde die Botschaft, falls es eine gibt.	Decifra il codice e trova il messaggio, se c'è.
Follow the codex, find the code, bargain for life, and gain barcodes.	Folge dem Kodex, finde den Code, handle ums Leben und erwirb Barcodes.	Segui il codice, trova il codice, contratta la vita e vinci codici a barre.
Crack the code, decipher the message, find my bareness, and cherish its rareness with some caress.	Knacke den Code, entziffre die Botschaft, finde meine Nacktheit, schätze ihre Besonderheit mit etwas Zärtlichkeit.	Risolvi il codice, decifra il messaggio, trova la mia nudità e apprezza la sua unicità con un pò di tenerezza.

Note: Texts were authored by Gabriele Werner-Felmayer, interpreted by Nadia Braito and electronically adapted by Saverio Monti.

The Provocative Y Chromosome

The sculpture of chromosomes raised a revealing reaction when presented to the public: The clearly visible smallness of the Y chromosome irritated two viewers of the sculpture, both male experts of medicine and science, albeit not in the fields of genetics and genomics. Both wrapped their criticism in humour but evidently, they did not like seeing the unimpressive Y chromosome next to the substantial X chromosome. Independently from each other, they therefore claimed that a representation of 23 chromosomes was scientifically more correct and suggested removing the Y chromosome from the artwork. This was surprising for the artist, as bringing in a gender perspective had not been on her agenda in this case. Neither had she intended to ridicule stereotypes of maleness in this work. On the contrary, she included both sex chromosomes in their 'real' proportions to refer to the entire human genome and she related to current genome and chromosome representations aiming to meet the scientific knowledge of today.

Interestingly, however, such a representation apparently called up gender images based on simple equations of size and quality. In the case of the Y chromosome, the popular, yet recently disproved, notion of the 'vanishing' Y chromosome has perpetuated such clichés. Based on this notion, the idea of the male sex as being prone to extinction has been in vogue for a while in newspaper feature sections (see Box 7.2 for more details) and obviously also had impressed the two visitors. As if 'I' was Y, they felt concerned and, funnily enough, requested exactly what they might have been afraid of: the vanishing of Y.

Box 7.2 The Y Chromosome Debate

Y Chromosome Evolution: According to current understanding, the human X and Y chromosomes evolved from a common ancestor autosome. The Y chromosome lost most (97 per cent) of the genes from this common ancestor during the past 200–300 million years in favour of evolving and selecting male-specific genes. Hence, 95 per cent of the Y chromosome comprise a male-specific region called MSY (Hughes et al. 2010; Skaletsky et al. 2003). This selection process is the basis of mammalian sex differentiation and is termed 'purifying selection' (Hughes et al. 2012: 82). Due to current concepts, the MSY has evolved in five evolutionary selection events accompanied by a loss of non-male specific genes and the evolution of some new, male- and human-specific genes (Hughes et al. 2012).

The Misconception of the Vanishing Y Chromosome: The Y chromosome was long considered to be a 'genetic wasteland' and a 'profoundly degenerate X chromosome' (Skaletsky et al. 2003: 825) as its sex-determinative role started to emerge only in the 1950s. Moreover, scientists could identify and sequence the human MSY (see above) only recently (Hughes et al. 2010, 2012; Skaletsky et al. 2003). Therefore, 'Men, or at least male biologists, have long been alarmed' that the Y chromosome might vanish and anticipated that '[t]he male sex would then become extinct, […], leaving women to invent some virgin-birth method of reproduction and propagate a sexless species' (Wade 2012: online).

Moreover, the episode also illustrates gender hierarchies which are quite common in medicine and science: The (male) 'experts' did not consider the possibility that they may not know sufficient details of current concepts regarding the human genome. Instead, they asked the (female) artist to change her work and insinuated that she had made a mistake when designing her sculpture with 24 chromosomes. However, as already mentioned above, the representation of 24 chromosomes is perfectly correct from a human genomic sequence perspective. The gag is that the sculpture with its 24 chromosomes refers in fact to a *male* genome. Removing the Y chromosome would turn it into a female's genome.

As we can learn from genomics research, the devil is in the detail: When we speak of 'the human genome', we refer to the haploid human genome sequence. In case of females, this consists of the entire DNA packaged into 22 autosomes and the X chromosome (see Box 7.1). In the case of males, it consists of the entire DNA packaged into 22 autosomes, the X chromosome *and* the Y chromosome.[21] A male's genome, therefore, is slightly larger than a female's but that is not all: Phenotypes arise from the diploid genome, which functions as a complicatedly interacting entity of our maternal and paternal genome copies (Levi et al. 2007).[22] Therefore, all chromosomes contribute to the phenotype, a notion that 23andMe took up in its name referring to the 23 chromosome pairs. However, the expression rate of X-linked gene products is higher than that of other chromosomes, in order to maintain their dosage in males, as they have only one X chromosome. In female somatic cells, one of the two X chromosomes is virtually inactive. Otherwise, X-linked genes were overexpressed (Payer and Lee 2008).[23] Therefore, on both the haploid and the diploid levels, the male genome's size is slightly larger as compared to the female one. This fact may evoke further connotations of size-related gender discrimination but I rather prefer to get back to Con'Sequences and another topos it revisits.

Information Matters

In a world of commerce, barcodes are ubiquitous. Graphically, they resemble chromosome-banding patterns as represented in Con'Sequences. The invention of barcodes by Norman Joseph Woodland and Bernhard Silver in the 1950s[24] crucially changed shopping practices, as barcodes allow the use of tills equipped

21 In addition to the nuclear genome organised in chromosomes, the human genome of both also contains a mitochondrial genome.

22 Sequencing the diploid genome revealed the high degree of genomic variation, which plays an important role for phenotype expression. For more details, see Chapter 1 in this volume.

23 Only about 5 per cent of genes from the inactivated X chromosome escape silencing (see Payer and Lee 2008 for details).

24 More information at: http://en.wikipedia.org/wiki/Norman_Joseph_Woodland [accessed: 15 December 2012].

with electronic barcode readers. Barcodes facilitate the automated tracking of goods and controlling warehouse stocks.[25] There are numerous kinds of 1D and 2D barcode systems. Many of them have become familiar to us as e.g., ISBN numbers of books or 2D matrix codes on train and flight tickets.[26] For Con'Sequences, Helene Keller applied the frequently used Code128, a barcode symbology of very high information-density[27] that closely resembles the chromosomal G-banding patterns. It can be used to barcode words and whole sentences.

As I will show here, the analogy of the two patterns goes far beyond design: it refers to data, messages and words as well as to practices and a language of consumerism that have entered scientific perspectives and representations of the genome. Barcodes not only label goods but also, for example, the animals we eat as they have turned into 'industry products'. Carrying the analogy of barcodes and genomes to extremes, one may comprehend DNA sequences as a barcode (see below), identifying individuals unequivocally like a barcode identifies a product. In an era of DNA fingerprinting this notion no longer seems farfetched. Why should we not think of individuals as 'products' of their genomes, as we already speak of genes and their products in the laboratory?

A major attribute of barcodes as well as chromosomes and genomes is that they contain *information*.[28] In the case of barcodes, this information is encrypted in black and white bar patterns combined with various symbols (e.g., digits, letters, special and control characters) and these patterns and symbols are ordered in a way that allows unequivocal identification of a product. The information of barcodes is not dynamic, regulated or regulatory, it is simply informative, an identifier, particular for the one item for which it was designed. In the case of chromosomes and the genome sequence, however, the term 'information' is much more complex. Here, information is understood as either being a sort of language (quite in the sense of product barcodes, see also below) or as 'informed matter', information being inherently embodied in matter which is conceived being the basis of self-organising living systems. As Evelyn Fox Keller details in a recent essay on this issue, these debates are 'rooted in distinctions between form and matter inherited from classical traditions' (2011: 174). In fact, controversies in this context reflect the perspectives of Plato's immaterialism and dualism as well as Aristotle's materialism and naturalism. For Plato, 'knowledge required abstraction of the timeless forms out of, and away from, their material manifestations'

25 While not immediately appreciated as being useful, barcodes finally became the basis of the Universal Product Code (UPC), introduced in 1973 in the US and later on in many other countries as the main barcode system.

26 For examples refer to Wikipedia at: http://en.wikipedia.org/wiki/Barcode [accessed: 15 December 2012].

27 Information at: http://en.wikipedia.org/wiki/Code_128 [accessed: 15 December 2012].

28 The most general definition of the term information is knowledge that is communicable or conceivable. Depending on the context, the term may refer to a binary code used for computers or to more complex codes and syntaxes.

whereas Aristotle and later on Aquinas rejected the idea of 'unknowability of matter, arguing instead for intelligible principles of material things' (Keller 2011: 174). Echoes of 'ancient form-matter dualisms can also be found in information theory and computational science' (Keller 2011: 175) as well as in the opposing perspectives of classical physics (as a way to define the laws of nature from simplicity and abstraction instead of complexity) and modern biology (as a way to understand living systems in terms of complexity). With regard to genetics, phenotypic plasticity (see Keller 2009 for a discussion of heritability of traits) and the dynamics of the genome may be an expression of self-organisation of 'informed matter'. Consequently, a genetic programme is not the still popular, reductionist version of a developmental programme hard-wired into the DNA and expressed under centralised control. It is rather a regulatory circuitry of molecular networks. This circuitry is not fixed but dynamic (Keller 2002). The degree of complexity may be such – and many recent findings support this view (see Chapter 1 in this volume) – 'that this program is irreducible – in the sense, that is, that nothing less complex than the organism itself is able to do the job' (Keller 2002: 101).

The genetic code was first conceptualised in the late 1950s (reviewed by Kay 2000). Experiments verified then that so-called DNA codons, i.e., triplets of nucleotides on DNA are 'transcribed' into complementary messenger RNA codons and then 'translated' into the respective amino acids they are encoding for (Nirenberg et al. 1965). Here, the metaphor of a genetic code of information reaches its limits, as only protein-coding genes have this kind of 'informational or semantic property' (Godfrey-Smith 2007: 8). Nevertheless, the metaphor of *genetic information* being a sort of language proliferated throughout the fields of genetics, biology in general and even law, reiterating a simplistic and deterministic understanding of the issue (for an in-depth analysis of factors shaping and fertilizing this proliferation see Kay 2000). Although more meaningful concepts of biological *information* have recently been developed by different scientific fields like 'supramolecular chemistry, robotics, interactive computation, and embodied cognition' (Keller 2011: 179), the deterministic and reductionist approaches culminating in the 'book of life' metaphor are still powerful in the discourse on genetics and genomics.

The reason for this ambivalence may be that simple perspectives on life match much better than complex ones with current socio-economic trends such as rationing and commodifying all aspects of life. Framing 'genetic material as disembodied information' supports this practice since it turns the genome to a matter for engineering, patenting and marketing (Didur 2003: 104). In line with such trends, one can present the Human Genome Project as a new El Dorado that opens the genome as a good to the ubiquitous logic of maximizing profits of all sorts. Such a stance is found e.g., in the official presentation of the Human Genome Project as a project 'to identify all […] genes', 'to determine the sequence', 'to store this information in databases', 'to improve tools for data analysis', that is a catalyst for 'the multibillion-dollar US biotechnology industry' (Human Genome Project Information 2012: online). This statement summarises emergent coproductions

of life sciences and capitalism based on the increasing transformation of biology into information science against the background of changing values, thus creating 'biocapital' on a global scale. This era is characterised by a 'speculative capitalism' that 'contains its own future-oriented grammar' pertaining to 'a political economy of hype' (Sunder Rajan 2007: 14). In this realm of ideas, one can, for example, speculate that until 2020 'Science Will Pinpoint What Makes Us *Homo sapiens*' and that 'Personality will Move From Art to Science', suggesting that it will become possible to 'roughly deduce' from an individual's genome 'not only what she looked like, but, for example, how she acted' (Than 2010: online). From here to predicting and finally somehow managing personality traits, it seems only a small step, although most of these claims have no sound scientific basis whatsoever.

Language itself has generated new patterns in an era when science and capitalism have merged – the language of information, of sequences, of data, of codes and of messages has become widespread. It helped to prepare the ground for commercializing genomics and vice versa, the language of commerce has entered science like any other field of human endeavour.

Illustrative examples for this coupling are 'DNA barcodes' (Hebert et al. 2003): The term refers to DNA sequence profiles specific for a certain species.[29] Despite recent criticism of the methodology,[30] 'DNA barcoding' has become widely used in taxonomy for species identification. The ambitious International Barcode of Life Project (iBOL) aims at 'Bridging the Biodiversity Gap,' referring to the fact that only about two of the 10 to 100 million species inhabiting Earth have been described so far (iBOL 2013). iBOL is a typical large-scale genomics project (see Chapter 1 in this volume) run by a big international research consortium sharing data and knowledge using bioinformatics approaches.[31] One of the goals of this 'largest biodiversity genomics initiative ever undertaken' is to set up the Barcode of Life Database BOLD 'towards the ultimate goal of a barcode reference library of all life on Earth' (iBOL 2013).

iBOL uses the information-encoded language and is basically a bioinformatics approach integrating DNA sequence with other information, as it is typical for biocapital (Sunder Rajan 2007). iBOL and DNA barcoding do exactly what Con'Sequences refers to: a species-specific DNA sequence and additional information are integrated; a UPC barcode is generated; a comprehensive database of barcodes then creates 'a digital identification system for life' (iBOL 2013).[32]

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29 Species-characteristic sequence profiles of the mitochondrial cytochrome c oxidase gene are used for species identification in mixed organism populations, e.g., in soils.

30 It was recently argued that the 'barcoding movement' would have to recognise its limitations in order to avoid becoming 'irrelevant' in an era of next generation sequencing technology (Taylor and Harris 2012: 377).

31 iBOL is a publicly funded not-for-profit Corporation (launched by Genome Canada).

32 For an analysis of using DNA barcodes as ordering principle and its social context see Waterton (2010).

Coda

Am I then bits and bytes that are 'hard-wired' into a genome? Could I therefore be barcoded like a product – a product of the genome that is *my* genome after all, making me unique? Do I have to buy my genome information in order to be my owner? What would I do with all the information that I buy? Should I just have it, store it or could I sell it? Could I share it with someone? Could I turn it into a meaningful story?

The so-called 'genomic revolution' and the emergence of genetics as social practice in times of consumerism came along with a variety of ambiguities oscillating between simplistic and complex perspectives. The artwork Con'Sequences makes some of them perceivable by ironically examining the barcode, information and language metaphors. These metaphors appear to conserve the above-mentioned dualistic view of immaterial information that matters and some less defined matter, which does not really matter. They also imprint our perception of genomics and the results we may obtain with connotations of all sorts. In his commentary 'A metaphor too far', Philip Ball (2011: online) states:

> Books of life, junk DNA, DNA barcodes: all these images can and have distorted the picture, not least because scientists themselves sometimes forget that they are metaphors. And when the science moves on – when we discover that the genome is nothing like a book or blueprint – the metaphors tend, nonetheless, to stick. The more vivid the image, the more dangerously seductive and resistant to change it is.

In the flood of data, genomics and bioinformatics are generating and in the economy of hope and hype, that markets them, the main challenge is to use both science and metaphors wisely. Otherwise, all kinds of misunderstandings and flawed conceptions could make life terribly difficult. Dealing with Con'Sequences illustrates that art can 'help us to arrange the cultural assimilation of the genetic revolution' and 'can also form political conceptions' as the 'gaze of artists dwells on the values challenged by a rapidly growing science' (Anker and Nelkin 2003: 4). It also allows us to comprehend life and ourselves beyond the technical talk of science and challenges our own stereotypes regarding practices of genetics in a broader sense.

Acknowledgements

I thank Helene Keller for all her input and Ernst R. Werner for inspiring discussions. I am also grateful to Barbara Prainsack and Silke Schicktanz for their knowledgeable comments on a previous version of this text and Maya J. Pandya for language editing.

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Chapter 8
The Ethics of Patenting in Genetics: A Second Enclosure of the Commons?

Sigrid Sterckx and Julian Cockbain

Introduction

Genomics brings us valuable information for the identification of susceptibility to disease and for the development of drugs. This information was always present in our DNA – it was just hidden from us. The question we address in this chapter is whether those who reveal the existing but hidden information should be enabled by patent laws[1] to monopolise it, to turn it into private property.

Are we facing a second enclosure of the commons? European patent law states that discoveries are not patentable. Should we allow discoveries on our 'blueprint' to become privatised, or, in the words of the US Supreme Court, are some things 'free to all men and reserved exclusively to none' (*Funk v. Kalo* 1948: 130)?

The question is particularly important as it concerns our health, and possibly even our lives. Patents allow companies to charge high prices for life-saving drugs and for diagnostic tests that indicate whether drugs or surgery are necessary – prices that not all can afford. This is not a question of having to wait before being able to afford a new luxury item – by the time a drug has gone off patent, many of those who could have benefitted from it will be dead.

Patents are being granted in Europe, the US and elsewhere for human genes and their use in diagnostic (and other) methods. In this chapter we will illustrate various ethical problems resulting from this. We will not address the basic question as to whether patents are inherently objectionable on ethical grounds. However, patents are privileges, exceptions to free trade, and have no valid justification if their benefits to society do not outweigh their detrimental effects.

Three major strands of ethical concern arise with the patenting of human genes and their uses – access, consent, and inherent patentability.[2]

First, the effect of patents in restricting *access*, is similar to that encountered with any life-saving invention, in particular of a food or medicine. While deferring

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1 A patent generally provides a 20 year monopoly on the 'inventions' defined in the patent claims.

2 The question of inherent patentability is not the same as the justification of patenting as such; the former starts from the position that patents are justified and then asks whether the patent law permits the grant of patents for some particular form of subject-matter.

the possibility of enjoying a new type of bicycle because we cannot afford the patentee's inflated prices may be annoying, it is not life-endangering. The same cannot be said of medicines. Even the USA, during the anthrax attacks that followed 9/11, considered overruling patent rights to Cipro, the drug of first choice for treating anthrax infection (Sell 2003: 160). However, the ethical concerns relating to access to healthcare, including genetic testing, have been written about at length and will not be considered further here (see, for example, *AMP v. USPTO* 2010; Gold and Carbone 2008; Hestermeyer 2007; and Sterckx 2007).

Second, ethical concerns regarding *consent* arise from the fact that the genetic information on which many gene patents are based derives from patients' and research participants' body material. Two relevant questions arise. Firstly, was the consent, if any, given by the persons providing the body material, sufficient to support the grant of a patent to the researchers? Secondly, are the people from whom that body material was taken entitled to share in the benefits of such patents, e.g., access to drugs or tests developed or to the financial rewards? Again, much has been written on this question of benefit sharing, not least in relation to clinical trials, so we will refrain from further comment. On the question of the ethical validity of consent to justify the grant of a patent, we will examine the leading European case, T-272/95 *Relaxin/HOWARD FLOREY INSTITUTE*. Considering US law, it is important to note that consent by the body material sources appears not to have any bearing on the researcher's entitlement to a patent. In this regard, we will comment briefly upon the *Moore* case as well as on the grant in 2012 of a patent relating to a genetic test for susceptibility to Parkinson's disease.

Third, ethical issues arise around the question of the *inherent patentability* (or more precisely *patent-eligibility*) of human genes and their uses. Quite simply put: are human genes, which are products of nature, suitable subject-matter for patents? Different patent laws provide different bases on which patents for products of nature might be denied. For example, the patent-eligibility of the cancer-related genes BRCA1/2 has been challenged in Europe and the US. A second US Federal Appeals Court decision issued in 2012 and, at the time of writing, the case is under consideration by the US Supreme Court.[3] We will review and discuss the European and US cases (*T-1213/05 Breast and ovarian cancer/UNIVERSITY OF UTAH* and *AMP v. USPTO*), finding a similarity in the bases that could be found to deny patent-eligibility despite the differences in the wording of the laws. Finally, we note how ethical issues relating to genetics have received insufficient attention in patent law, and we conclude with some suggestions as to how this deficiency might be corrected.

3 Since the submission of this chapter, in June 2013 the US Supreme Court issued its decision in *AMP v. Myriad* (2013). The Court decided that isolated DNA molecules which had the same sequences as in endogenous DNA were not patent-eligible. cDNA, however, was found to be patent-eligible.

Does Consent to Research Equate to Consent to Patenting?

In 1983, Howard Florey Institute (HFI) filed a patent application relating *inter alia* to DNA fragments encoding the human protein H2-preprorelaxin. The contribution was said to be the production of recombinant human relaxin in a potentially therapeutically useful form, to alleviate birth complications.

The European patent was opposed[4] by the 'Green' political party, *inter alia* on the basis of Art. 53(a) EPC, the so-called 'morality provision' of the European Patent Convention (EPC): 'European patents shall not be granted in respect of … inventions the commercial exploitation of which would be contrary to "ordre public" or morality' (European Patent Office 2010: 107).

The opponents argued that genes are part of the common heritage of mankind. Converting that common heritage into private property was said to violate human dignity (Aglietta 1992: 6–8). HFI responded that:

> [T]he ovarian tissue [the material used as the basis of HFI's 'invention'] was obtained at the time of a [surgical intervention] for ectopic (tubal) pregnancy. Ectopic pregnancy is a life-threatening condition … The material was obtained with patient consent and appropriate Ethics Committee approval was obtained for the collection of this tissue. … The tissue … would otherwise have been discarded. There was nothing unethical in the obtaining of the [tissue]. (Howard Florey Institute 1993: 4)

In its decision maintaining the patent, the EPO's Opposition Division observed that, according to HFI, the women whose tissue was used, had consented to its removal. Further, it was noted that the use of human tissue as a source of useful products was accepted by the vast majority of the public (European Patent Office 1995: 13). The opponents appealed, and the EPO's Technical Board of Appeal 3.3.04 dismissed the appeal in 2002. In its decision, regarding the consent point, it merely referred to two EPC Rules which were introduced following the issue of an EU Directive on the legal protection of biotechnological inventions (EBD) (European Parliament 1998) and suggested that no further arguments were needed. However, as we have noted elsewhere, the introduction of the EBD-derived Rules was highly problematic[5] (Sterckx and Cockbain 2012: 49–56).

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4 A European patent application is examined by the Examining Division of the European Patent Office (EPO). If accepted, the European patent is granted. Within nine months of patent grant, interested parties may file oppositions which are heard by the EPO's Opposition Division. The Opposition Division's decision (to maintain or revoke the patent) may be appealed by the patentee or the opponent to the EPO's Technical Board of Appeal.

5 The EPC Rules are incapable of modifying the scope of the EPC Articles. However, the Rules introduced in response to the EBD did seek to modify the scope of the Articles that define the scope of patent-eligible subject-matter. This was a 'quick fix' – amending the Rules could be done by the EPO's Administrative Council whereas amending the Articles would require ratification by all EPC member States.

The question of consent was also raised before the same Board of Appeal in the later case *T-1213/05 Breast and ovarian cancer/UNIVERSITY OF UTAH* (see below). In that case, one opponent argued that no proof had been provided by the patent holder that the donors of the relevant genetic material had given informed consent for its use. A *valid* informed consent, the opponents argued, would have to have included explicit consent to the commercial exploitation of the research results via patents. Since such proof was lacking, it had to be assumed that severe breaches of ethics had occurred. The Board's response was that the EPC did not require evidence of informed consent to be submitted.

In this context, the Board quoted EBD Recital 26: 'if an invention is based on biological material of human origin or if it uses such material, where a patent application is filed, the person from whose body the material is taken must have had an opportunity of expressing free and informed consent thereto, in accordance with national law'. From this, the Board concluded that the: 'legislator [had] thus not provided for a procedure of verifying the informed consent in the framework of the grant of biotechnological patents' (*T-1213/05 Breast and ovarian cancer/ UNIVERSITY OF UTAH*: para. 49). Accordingly, the opponent's arguments regarding informed consent were rejected.

In *Moore v. University of California*, the Californian Supreme Court likewise held that consent (in this case by John Moore to the patenting of a cell line based on his cancerous tissue) was not relevant when assessing whether the patent had been validly granted to the University. (Stanton 2008) Unlike the EPC, however, the US Patent Law does not contain any provision to the effect that inventions the commercial exploitation of which is immoral are not patent-eligible.

Thus, in effect, consent is not an issue for patent grant. However, it should be. As has been argued by bioethicist Julian Savulescu:

> Each mature person should be the author of his or her own life. Each person has values, plans, aspirations, and feelings about how that life should go. People have values which may collide with research goals … To ask a person's permission to do something to that person is to involve her actively and to give her the opportunity to make the project a part of her plans. When we involve people in our projects without their consent we use them as a means to our own ends. (Savulescu 2000: 649)

Indeed, we consider that consent should include an authorisation for bodily material or information to be the subject of a patent. Patients and research participants may consent to donate biological materials and phenotypic data for research aiming to promote the development of clinical applications. However, if they are not aware that this development might happen through commercialisation involving patents, this renders their consent ethically invalid since they were not enabled to make their own informed decisions on whether to allow the material to be used for such purposes.[6]

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6 Even if the consent explicitly mentions that the material or data may be used in patent applications, questions might still be asked about the validity of the consent, for others besides the person consenting have the same genes. As argued by Widdows (2012),

This is important, not only in view of theoretical ethical arguments, but also because it is empirically supported that many people do care about this issue. For example, Cook and Hoas (2011) have conducted a qualitative interview study in the US, exploring attitudes of participants when deciding whether to participate in medical research. They found that most participants desired more information about the commercial purposes of the research and that the information they had been given was not sufficient to enable the participants to realise:

> that some studies might be designed for commercial purposes, such as extending a patent … Participants thought it was dishonest not to be transparent about … the full purpose of a study and said that hiding such information would not be acceptable. Most (90%) wanted to know whether a study had such a commercial purpose and the vast majority (80%) reported that disclosure of such information could influence their decisions about participating in research in the future. Said one participant: "[T]he person should know the purpose of the study … I think the study participant should be told exactly what is going on. It's coercion otherwise. … " Said another [participant]: "Patents. Sure. Absolutely, for sure. I absolutely want to know. …" … Among those who said it would not influence their decisions about participation, they still felt they should be informed about such issues. (Cook and Hoas 2011: 4–5)

The importance of transparency on these issues is also clearly illustrated by the outrage expressed by various customers of the direct-to-consumer genetic testing company 23andMe, when it announced in May 2012 that it was to be granted US Patent No. 8187811 (*Polymorphisms associated with Parkinson's Disease*) on the next day (Wojcicki 2012). Several clearly believed that, by providing their data in the context of a research project on Parkinson's, they were participating in an altruistic exercise to promote the development and accessibility of diagnostic tests and therapies. To quote a few reactions posted on the company's blog 'Spittoon' (Wojcicki 2012):

> [T]his is simply crowd-sourced greed. As a longtime 23andme customer, this patent is extremely disappointing and alarming. Our family is done with your service.
>
> It would seem that the ethics of one company profiting from the knowledge of others because it patented a gene variant could do with some scrutiny, especially if it turns out that patients, who provided samples for the original research, were not aware that the results would be patented.
>
> [E]veryone coming to [23andMe's] service, either by paying it or by funded invitation (e.g., Patient groups …) needs to know clearly what this is about and make their own informed decision to join or not.

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the genetic self is the connected self because genetic information always gives information about related individuals. However, this need not concern us too much here, since any 'connectedness-based' concerns regarding the sufficiency of consent would seem to be dealt with if the criterion of patent-eligibility were to be properly applied as suggested below.

Clearly, for many, what has undermined trust it is not the profit motive in itself but the fact that, until the day before the patent was granted, the company did not provide any indication that it was seeking patents on its discoveries. The key lesson to be learned from this is that any (private or public) organisation involved in research that relies on human participation, whether by providing information or body material or both, needs to be transparent (Sterckx et al. 2013). This transparency should be not only about research goals but also about strategies and policies regarding commercialisation, including patenting and licensing policies. This is crucial to enable participants to decide whether those goals and policies are in line with their moral values, and whether they want to contribute to those goals by providing information or body material.

What Is a Discovery?

The EPC provides that discoveries are not patent-eligible, but limits this exclusion to discoveries 'as such', i.e., *applications* of discoveries are not necessarily excluded. Accordingly, for European patent law, the question arises as to whether human genes and their use in diagnostic (and other) methods are 'discoveries' and thus unpatentable.

But what is a discovery? 'Discovery' normally means *to find something that was pre-existing* – one discovers a previously unknown plant or mineral, or that energy is proportional to mass squared, or that a human gene sequence codes for a protein. To the extent that native genes, their variants, and their correlation with disease states are *pre-existing*, identification of the gene and its variants surely represents a *discovery*.

Another meaning of discovery relates to situations where *something is identified which had not previously been in operation*. For (the hypothetical) example, where a product of a marine microorganism, if injected into the human brain, can slow the progress of Alzheimer's disease, then to use the product to treat Alzheimer's disease would involve the *application* of a discovery which had not previously been in effect.

In 1998, after long heated debates, the EBD entered into force. In a controversial attempt to make the EPC compliant with the EBD without requiring a diplomatic conference of the EPC member states,[7] various EPC Rules were introduced in 1999. Two of these Rules are particularly relevant. One provides that: 'Biotechnological inventions shall also be patentable if they concern: … biological material which is *isolated* from its natural environment or *produced by means of a technical process* even if it previously occurred in nature' (European Patent Office 2010: 330, emphasis added). The other states that: 'An element *isolated* from the human body

7 A rule change requires a majority vote in the EPO's Administrative Council, whereas an amendment of an EPC Article requires ratification by *all* the EPC Member States. The latter procedure would obviously have taken much longer and it is very likely that not all Member States would have agreed.

or otherwise produced by means of a technical process, including the sequence or partial sequence of a gene, may constitute a patentable invention, even if the structure of that element is identical to that of a natural element' (European Patent Office 2010: 332, emphasis added).

In 1995, Myriad Genetics (a spin-off from the University of Utah), the University of Utah Research Foundation and the United States of America[8] filed a patent application relating *inter alia* to isolated human DNA coding for BRCA1 (a breast cancer gene) or modified forms of it that are functionally equivalent or associated with a predisposition to breast or ovarian cancer, and methods for screening cancer drugs. The European patent was granted in 2001 and opposed by eight parties, including Greenpeace. At the opposition hearing, two opponents argued that genes are unpatentable discoveries and that the central concept underlying the patent was the discovery of a link between a gene and a disease (European Patent Office 2005a: 8–9). The EPO's Opposition Division disagreed, arguing that:

> The fact that a link between the claimed probes, BRCA1 and breast cancer exists does not preclude the claimed probes to be patentable. … Moreover, Rule [29(2)] EPC explicitly states that "elements isolated from the human body or otherwise produced by means of a technical process including the sequence or partial sequence of a gene may constitute patentable invention even is the structure of that element is identical to that of a natural element". Since, the nucleic acid probes were described … as having been obtained by technical processes, they fulfill the definition of patentable elements of the human body given in Rule [29(2)] EPC. The probes as claimed cannot therefore be considered as mere discoveries (see T 272/95 [*Relaxin/HOWARD FLOREY INSTITUTE*]). (European Patent Office 2005b: 26)

The Opposition Division's decision was appealed and Technical Board of Appeal 3.3.04, the Board responsible for the flawed *T-272/95 Relaxin/HOWARD FLOREY INSTITUTE* decision discussed above, simply applied its own earlier flawed reasoning to the 'discovery' issue:

> [The patent claims] relate to … probes comprising partial DNA sequences of the human BRCA1 gene, which are described … as having been obtained by technical processes … These probes are thus isolated elements of the human body … and thus patentable subject-matter. (*T-1213/05 Breast and ovarian cancer/UNIVERSITY OF UTAH:* paras 44–45)

Thus by being simply reduced to a 'novelty-type' test, the EPC's exclusion from patent-eligibility of 'discoveries' has been made substantially toothless. By a 'novelty-type' test, we mean a test for *identicality* with the excluded. Thus 'isolated DNA', being free from other cellular material, is not *identical* with DNA as found in the body (the 'discovery') and is therefore not excluded. However, since materials

8 The US National Institutes of Health had been involved in the research.

found in nature must in general be separated from their environment to be useful, and since 'discovery' of the endogenous gene is also the discovery of the gene as such (i.e., the 'isolated' gene), something more than a novelty test is required in order for the legislators' intention in excluding discoveries to be respected. Indeed, as we have argued elsewhere (Sterckx and Cockbain 2012), an inventiveness test is also required – what is claimed must neither be the discovery itself nor anything which is obvious in the light of that discovery.

The Myriad Case in the US – Patentability of 'Isolated' DNA

EPO case law on discoveries remains scarce. However, the exclusion of discoveries from patent-eligibility under European patent law corresponds closely to the exclusion of phenomena of nature from patent-eligibility under US case law, so looking into the meaning and basis of the latter exclusion will aid our investigation as to whether discoveries should be part of the commons.

As far as natural products and phenomena are concerned, the patent-eligibility test to be applied by the United States Patent and Trademark Office (USPTO) under Section 101 of the US Patent Law (i.e., 35 USC 101), is well illustrated by the 1980 US Supreme Court decision in *Diamond v. Chakrabarty*:

> The laws of nature, physical phenomena, and abstract ideas have been held not patentable. Thus, a new mineral discovered in the earth or a new plant found in the wild is not patentable subject matter [under Section 101]. Likewise, Einstein could not patent his celebrated law that $E = mc^2$; nor could Newton have patented the law of gravity. Such discoveries are "manifestations of … nature, free to all men and reserved exclusively to none". (*Diamond v. Chakrabarty* 1980, quoting from *Funk v. Kalo* 1948)

The language used by the Court, 'free to all men and reserved exclusively to none', calls to mind comments on the human genome being the common heritage of mankind. Both the United Nations Educational, Scientific and Cultural Organization (UNESCO) and the Council of Europe (COE) have declared that human DNA has a special nature that warrants extra protection, since the human genome should, in a symbolic sense, be considered as the 'heritage of humanity' (Universal Declaration on the Human Genome and Human Rights (UNESCO 1997: Article 1) and Protection of the Human Genome (Council of Europe (2001)). However, widely diverging opinions are expressed on whether this concept offers decisive arguments against the patenting of genes (see, for example, Ossorio 2007). Moreover, the UNESCO and COE recommendations are ambiguous. Even though Art. 4 of UNESCO's Universal Declaration on the Human Genome and Human Rights provides that '[t]he human genome in its natural state shall not give rise to financial gains', the Preamble of this Declaration mentions that the declaration is 'without prejudice to the international instruments which could have a bearing on the applications of genetics in the field of intellectual property'. Moreover, the

words 'in its natural state' in Art. 4 are unclear - do they merely refer to 'native' or 'wild type' DNA? A similar lack of clarity exists with regard to the words 'as such' in Art. 21 of the COE's *Convention on Human Rights and Biomedicine*, which provides that: 'The human body and its parts shall not, as such, give rise to financial gain' (Council of Europe 1997).

The claims under consideration in *Diamond v. Chakrabarty* related to a genetically engineered microorganism that was not naturally occurring. Finding those claims acceptable, the US Supreme Court distinguished over its 1948 decision in *Funk v. Kalo*:

> Concluding that the patentee had discovered "only some of the handiwork of nature," the [Funk] Court ruled the product [a combination of known bacterial species] nonpatentable: "No species acquires a different use. … The bacteria perform in their natural way. Their use in combination does not improve in any way their natural functioning. They serve the same ends nature originally provided and act quite independently of any effort by the patentee." … Here, by contrast, the patentee has produced a new bacterium with *markedly different characteristics* from any found in nature and one having the potential for significant utility. His discovery is not nature's handiwork, but his own; accordingly it is patentable subject matter under [Section 101]. (*Diamond v. Chakrabarty* 1980, emphasis added)

In the *AMP v. USPTO* case which will be analysed in this Section, one of the main questions at issue was whether claims to 'isolated' DNA failed the case law test as being claims to naturally occurring substances, i.e., discoveries which are not markedly different from 'products of nature'. Put simply, is 'isolated' DNA not simply the DNA which is a product of nature, or does 'isolation' sprinkle fairy dust over it, making it something markedly different?

Obviously, DNA does not exist in nature in isolated form. However, isolation or purification does not change the fact that the claimed DNA *itself* is a product of nature. As with the 'isolated DNA' claims considered by the EPO (discussed in the previous Section), the question arises as to *whether the wording of a patent claim (e.g., as an isolated, purified or synthetic version of a natural product) can make the claim patent-eligible*.

In *AMP v. USPTO*, the Association for Molecular Pathology and others challenged fifteen claims in seven patents licensed to Myriad Genetics. These claims were directed *inter alia* to 'isolated' DNA corresponding to the human genes BRCA1/2, to fragments of such DNA, and to cDNA. Also at issue were claims to diagnostic methods for screening for BRCA1/2 variants correlating with enhanced propensity to breast and ovarian cancer, and to methods of screening drugs for utility as anti-cancer agents. Myriad offers BRCA1/2 screening for several thousand dollars a time and, due to the patents, is the only commercial supplier of such screening in the US.

At the first instance, the US District Court for the Southern District of New York found these fifteen claims not to be patent-eligible in accordance with

Section 101 of the US Patent Law. Section 101 provides an initial hurdle that an 'invention' must pass, before being assessed for patentability using the more conventional tests of novelty, non-obviousness, and utility.

The US Supreme Court has for over a century considered that laws and phenomena of nature are not patent-eligible. This position is clearly expressed, for example, in *Diamond v. Chakrabarty*, a landmark decision from 1980 which threw open the door in the US to the patenting of life forms. However, for over a century the USPTO has been granting patents for things which, to a layperson, seem to be naturally occurring products. More to the point, it has been granting patents for human genes for decades. Typically, a gene may be claimed as 'an isolated DNA molecule coding for protein X and having the nucleic acid sequence Y'. The apparent reasoning is that (i) in nature, the DNA is associated with other materials, and (ii) the gene, in nature, is part of a much larger molecule and hence the chemical bonds at the start and end points of the 'isolated DNA molecule' will be different from those at the corresponding sequence positions in native DNA.

The central point at issue in *AMP v. USPTO* was therefore *the necessary difference* between what is claimed and what is natural for the claimed subject-matter to be patent-eligible. Is *any* difference sufficient, and, if not, then how *great* a difference is required? In *Diamond v. Chakrabarty*, for example, the US Supreme Court has made it clear that not just *any* difference is not enough, and that the characteristics of what is claimed must be 'markedly different' from what can be found in nature.

This, however, raises the further question of the *basis* on which the magnitude (i.e., the 'markedness') of the difference should be assessed. An example may help here – if we take your pink car and re-spray it red, there is clearly a visible difference, but no functional difference. If, instead, we apply a colourless speed radar absorbing coat, the car looks just the same but will function differently. US courts have struggled to find the best candidate answer for the question of deciding the sufficiency of the difference. A first possible candidate is that the claimed subject-matter is a new thing (relative to the natural). A second candidate is that it has properties that are not possessed by the natural. A third candidate is that the claimed subject-matter must be inventive over the natural. However, all three 'smuggle' into the Section 101 test, tests which are to some extent equivalent to novelty and non-obviousness tests, whereas these latter tests should be applied *after* the Section 101 test has been passed.

To return to *AMP v. USPTO*, Myriad appealed the District Court decision and the case went before the patent-specialist Court of Appeals for the Federal Circuit (the 'Federal Circuit'). Then a remarkable event occurred: the United States, appearing as an *amicus*,[9] argued that the claims to 'isolated DNA', other than the cDNA claims, were *not* patent-eligible. The thing that was isolated was simply

9 A 'friend of the court', a party not directly involved in a case but wishing to draw attention to materials and arguments that may assist the court in making its decision.

the natural product retaining its primary functionality, i.e., the information in the nucleic acid sequence.[10]

The Federal Circuit decided that the 'isolated DNA' claims were to patent-eligible subject matter (*AMP v. USPTO* 2011). AMP unsuccessfully asked the Federal Circuit to reconsider its decision *en banc*,[11] before appealing to the Supreme Court.

In April 2012, shortly after issuing its decision in *Mayo v. Prometheus* concerning the patent-eligibility of certain *method* claims, the US Supreme Court instructed the Federal Circuit to rehear the *AMP v. USPTO* case and to take the *Mayo v. Prometheus* decision into account. The second hearing before the Federal Circuit was held in July 2012 and the court issued its new decision in August 2012. (*AMP v. USPTO* 2012) The language had changed a little from the first decision, and the judges' positions were a little clearer, but the outcome was the same – 'isolated DNA' was patentable. The case duly went back to the Supreme Court and a final decision can be expected to issue in 2013.[12]

Federal Circuit Judge Lourie's position was the most pro-patentee, essentially that the DNA molecule that is isolated is not the DNA molecule that exists in nature, i.e., it passed a 'novelty-type' test. According to him, *any* difference is enough, or alternatively put, the isolated DNA molecule is something new made by man.

Judge Moore was clearly more torn, deciding on each type of DNA on a different basis. First, with regard to cDNA, she argued that the claimed sequences did not exist in nature and that the cDNA molecule had a 'distinctive character and use', with 'markedly different chemical characteristics' from the natural molecules.[13] Second, for the claims to 'isolated DNA' where the sequences *did* exist in nature, she found '[t]o the extent that the majority rests its conclusion on the chemical differences between genomic and isolated DNA (breaking the covalent bonds), I cannot agree that this is sufficient to hold that the claims are directed to patentable subject matter.' Third, for the oligonucleotide DNA molecules,[14] Judge Moore found that these had 'markedly different properties which are directly responsible for their new and significant utility … It is not the chemical change alone, but that change combined with the different and beneficial utility that leads me to conclude that small isolated DNA fragments are patentable subject matter.' Lastly, for the larger 'isolated DNA', her position was that to hold these to be not patent-

10 The case for the US was put by the Department of Justice – the USPTO did *not* confirm that it agreed.

11 That is with *all* the Judges of the Federal Circuit taking part in the decision rather than just three as is usual.

12 See footnote 4.

13 All the quotes from the Federal Circuit decision are from *AMP v. USPTO* (2012). In this instance, Judge Moore is referring back to the *Diamond v. Chakrabarty* decision of the Supreme Court.

14 Short DNA sequences, in this case consisting of 15 or more nucleic acids, unlike the many thousands in the full length BRCA genes.

eligible after so many years was such a major change that it should come from the legislator. In the oral hearing, the counsel for the US had stressed that courts sometimes can and do and should change longstanding practice, but this was a step too far for Judge Moore.

The third of the three judges, Judge Bryson, was of sterner stuff. 'We are therefore required to decide whether the process of isolating genetic material from a human DNA molecule makes the isolated genetic material a patentable invention. The court concludes that it does; I conclude that it does not.' Judge Bryson drew on the bases for the sufficiency of the differences over the natural found in various US Supreme Court decisions, including *Diamond v. Chakrabarty*. Mentioning the Supreme Court's references to inventive concepts in *Mayo v. Prometheus*, Judge Bryson appeared to endorse what we would call an 'invention over the natural' test: '[i]n cases such as this one, in which the applicant claims a composition of matter that is nearly identical to a product of nature, it is appropriate to ask whether the applicant has done "enough" to distinguish his alleged invention from the similar product of nature. Has the applicant made an "inventive" contribution to the product of nature? … Here, the answer to those questions is no.'

Unlike Judge Moore, Judge Bryson considered that the established financial interests of the biotech industry did not justify dodging the question as to whether isolated DNA should be patentable. Quoting from the Supreme Court's *Diamond v. Chakrabarty* decision, 'Congress has performed its constitutional role in defining patentable subject matter in [Section] 101; we perform ours in construing the language that Congress has employed', Judge Bryson concluded that Federal Circuit judges 'have the same responsibility and should not shy away from deciding the issues of the law that the parties have brought to us'. (*AMP v. USPTO* 2012)

Indeed, the issue was clear – in what *sense* does something have to be 'markedly different' from a product of nature to meet the hurdle of patent-eligibility of Section 101 as interpreted by the Supreme Court? As Conley and Makowski rightly note:

> [D]espite its nominal chemical distinctiveness, [isolated DNA/RNA] is functionally indistinguishable from natural DNA and RNA. It contains exactly the same genetic information as its natural counterpart. It can do precisely the same work as a naturally occurring gene—protein synthesis—and it employs precisely the same processes to do it, whether in the body or in the laboratory. … In other words, what is claimed is whatever it is that does the work of the naturally occurring … gene. … [T]he non-coding regions do not participate in doing the work. … In evaluating the materiality of the differences between claimed DNA sequences and their natural counterparts, we are left with chemical distinctions versus informational and functional identity. Critically, it is these informational and functional properties that are the whole reason for seeking DNA patents. Researchers isolate, purify and synthesize DNA both as an intermediate step in the process of gene identification and as a tool for building proteins. Thus, [the] DNA [for which patents are granted] is identical to its natural counterpart with respect to the qualities that researchers deem most significant, and distinct in ways that can be fairly characterized as incidental. (Conley and Makowski 2004: 35)

It is interesting to reconsider, in this light, the statement from the US Supreme Court in *Funk v. Kalo*, approved in *Diamond v. Chakrabarty*, that some 'discoveries are manifestations of nature, free to all men and reserved exclusively to none', a statement which was made in the following context:

> [P]atents cannot issue for the discovery of the phenomena of nature … The qualities of these bacteria, like the heat of the sun, electricity, or the qualities of metals, are part of the storehouse of knowledge of all men. They are manifestations of laws of nature, free to all men and reserved exclusively to none. He who discovers a hitherto unknown phenomenon of nature has no claim to a monopoly of it which the law recognizes. If there is to be invention from such a discovery, it must come from the application of the law of nature to a new and useful end. (*Funk v. Kalo* 1948: 130, emphasis added)

In *Mayo v. Prometheus*, as Judge Bryson rightly understood, the Supreme Court had realised that 'markedly different' does not mean simply chemically or visibly different; it means inventive over the natural product or law.

Conclusion

In terms of access, consent and patent-eligibility, patent law has clearly taken little notice of ethics. To some extent that may be changing, but three steps are clearly needed in relation to inventions based on human genes and their uses:

First, in order to prevent the 'storehouse of knowledge of all men' (to quote the US Supreme Court) from being emptied, not only natural products and laws *as they appear in nature*, but *also* anything which is obvious in the light of those products and laws when they are discovered, should be excluded from patent-eligibility.

Second, in order to respect the autonomy of persons whose isolated body material forms the basis of patent applications, where inventions are based on genetic or other bodily information from patients or research participants, that information, or the material from which it was derived, must come *only* from persons who were clearly informed that patents might be sought on results of the research, who clearly *understood* that such patents might be enforced against others, and who nonetheless gave clear consent.

Finally, governments should be free to, and should accept their obligation to, override patents to protect the health and well-being of their citizens, e.g., by issuing compulsory licences to make genetics-derived healthcare products and services available in an affordable manner. Unlike the two previous suggestions, this one is not concerned with the *grant* of patents for human genes and their uses, but instead with their *enforceability* once granted.

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PART III
Participating in the Social Laboratory

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Chapter 9
Understanding Participation: The 'Citizen Science' of Genetics

Barbara Prainsack

Introduction: The Role and Value of Public Participation in Science and Technology Studies (STS) Scholarship

Questions about the adequate role, the benefits, and the disadvantages of public participation in tasks that have traditionally been carried out by state institutions or other elites have been discussed controversially in the academic literature. In disciplines such as development studies or political science, for example, some scholars have shown great resistance to the inclusion of non-professional expertise, because they fear that standards will be compromised if the authority of the professional experts receives competition from 'lay people' (e.g., Cooke and Kothari 2001; Lippmann 2011 [1927]). The field of Science and Technology Studies (STS) is different in this regard:[1] Concerns pertaining to public participation in science – including its governance, regulation, and 'translation' into practical applications – have marked the history of STS. In the mid-1990s, Alan Irwin, a key thinker in STS, used the term 'citizen science' (CS) to refer to the need for scientists and members of the public to work together to tackle complex challenges such as sustainable development. For such a collaboration to be fruitful, Irwin argued, the domain of science needed to take seriously the knowledge and understandings in the public domain, not only vice versa (Irwin 1995). Seven years later, in 2002, Harry Collins and Robert Evans published a (now heavily cited) paper distinguishing three different 'waves' of science studies. Each wave, so the authors argued, corresponds with a particular configuration of concerns about public participation in science within the STS scholarship (Collins and Evans 2002). Within the first wave, which peaked before the 1970s, science was seen as a truth-generating force that could provide solutions to many of the problems that societies were facing. During the second wave, the capacity of science to 'produce truth' was increasingly questioned, assuming that the value and accuracy of scientific advancements could only be assessed in retrospect. Policy makers were in the difficult position of having to decide what kind of scientific evidence to use for policymaking without being able to wait until the scientific dust had settled. Because of this difficulty, science studies scholars within the second

1 For relevant discussions on this aspect in the field of anthropology, see Harding 2008; Palsson 2008.

wave argued that science and technology policy must become more participatory, a quest that has become known as addressing 'the problem of legitimacy'. But how participatory was participatory enough? And what tasks should and could be devolved to non-experts? The third wave – one that Collins and Evans called for, more than they diagnosed it – was meant to provide a counterweight in areas where the second wave had gone too far: Scholarship associated with the third wave, Collins and Evans argued, has stopped focusing on whether scientific truth is possible, but instead moved on to questioning what roles (different kinds of) expertise should play in policy making, and in public discourse more generally. Not surprisingly, Collins' and Evans' call for such a third wave of science studies has attracted a lot of criticism, such as Brian Wynne's (2003) contention that Collins and Evans had missed the main problems pertaining to the legitimacy of science, including that institutions have largely neglected issues of public *meaning*. Controversies about Collins and Evans' suggestions regarding the roles of different kinds of expertise in enhancing the legitimacy of science are ongoing (see also Collins and Evans 2007; Lynch 2008); the saliency of these debates only underlines that questions about public participation in science continue to concern STS at its core.

Recently, however, some of the practices on the ground have changed. It is now easier than ever for non-professionally trained people to participate in the governance, regulation, and translation of science, as well as in some of the core activities of science itself. Science has always been a collective endeavour, of course, and also one where non-professionally trained people have made important contributions: Prior to the era of highly institutionalised science in the 19th and 20th century, the generation of scientific knowledge relied heavily also on autodidacts who lacked standardised credentials (so-called 'amateur'[2] science). From the 19th century onwards, however, the agendas and hierarchies of science were increasingly (and ultimately exclusively) dominated by professionally trained experts.[3]

The developments of the last few years could be seen as an indication that the pendulum is now swinging back again, and that non-professionally trained people start to assume a more central role within core activities of science again. Digital media and open-source databases, for example, have broadened the scope and number of people who now contribute to science, and accelerated the pace at which they can do this (see also Prainsack et al., in this volume). Also due to Internet and the increasing uptake of portable electronic communication devices that are easy to use and carry around, the threshold for contributing to the creation of scientific knowledge is arguably lower than ever. People without professional scientific training contribute in many ways, for example, by photographing and describing

2 For a discussion of some of the connotations and contestations of the label 'amateur' in the context of citizen science and public participation, see Rogers 2011.

3 For a more detailed discussion of the difference between amateur science and citizen science, see Tocchetti (2013).

plants or animals in particular habitats and uploading images to databases; by classifying the shape of photographic images or sounds; or by transcribing hand written historical records into digital databases.[4]

How can we understand and classify the contributions to science that these 'citizen scientists' make? In this chapter, I will start to develop a schema for understanding and classifying CS projects. This schema is destined to aid our exploration of whether – and if so, how – CS makes science more socially robust (i.e., corresponding with dominant social, political, ecological, and commonly shared values; see Nowotny et al. 2001), or whether it makes it 'better' in any other way, and to whose benefit. I will then discuss the US-based online personal genomics testing service 23andMe against the background of this schema, and conclude by addressing the question of why 23andMe, despite containing the strongest participatory features of all personal genome testing services on the market, has also attracted most fervent criticism.

Virtual Experts – CS in the 21st Century[5]

2011 was a 'golden year' for citizen science: Numerous papers in top-tier academic journals acknowledged the contribution of non-professional scientists to the creation of scientific knowledge,[6] a book was published about the crowdsourcing[7] of scientific discoveries (Nielsen 2011), and public media featured computer game players solving a problem that scientists had been pondering for a long time (Khatib et al. 2011). Since then, interest in CS has expanded further, and is on the way of becoming a household name (Larson 2013).

Many particularly illustrative examples of the participation of others than professional scientists in the creation of scientific knowledge have come out of the health domain. This is rather unsurprising: Who would be more motivated than a patient or a family member seeking new treatments? A question that has only begun to be addressed is whether, and if so, under what circumstances, CS[8] produces

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4 For an overview of active 'citizen science' projects, see, for example: http://www.citizencyberscience.net/projects [accessed: 20 March 2013].

5 This and the subsequent section draw strongly on Prainsack (2013a), and on Prainsack (2012a).

6 These papers reported on the results of patient-organised observational studies, for example (see Wicks et al. 2011. See Epstein 1996 for an early precursor of this phenomenon).

7 The term crowdsourcing is a composite of 'crowd' and '(out)sourcing'.

8 Shirk et al. (2012) discuss the different nuances in the way in which the term 'citizen science' has been employed in North America and Europe respectively. The authors themselves prefer the term 'Public Participation in Scientific Research' (PPSR), which they define as 'intentional collaborations in which members of the public engage in the process of research to generate new science-based knowledge' (Shirk et al. 2012: 2). I prefer the term 'citizen science' because it ascribes central agency to non-professional scientists, rather than labelling them mere participants in science (which is supposedly led by others,

'better' results than conventional science carried out exclusively by professionals; either because the results are produced faster, or because they lead to more socially robust applications,[9] or because they solve previously unresolved questions. There are different approaches to answering this question, many of which draw upon the literature on social networks. Journalist James Surowiecki, for example, in his book *The Wisdom of Crowds* (2005), suggested four criteria that need to be met for a 'crowd' to make intelligent decisions: First, independence of individual opinions from peer or other influences; second, decentralisation of expertise in the crowd; third, diversity of opinion; and fourth, aggregation (i.e., some mechanisms of turning individual opinions into a collective decision). While Surowiecki did not speak about the creation of scientific knowledge specifically, physicist-turned-writer Michael Nielsen, in his book *Reinventing Discovery* (2011) did; moreover, Nielsen started to unpack the question of how aggregation should take place in order to maximise the supposed intelligence of the crowd (see also Shirky 2008). Having looked at a number of successful CS projects in a wide range of domains, Nielsen distilled a set of criteria that have helped CS initiatives to produce what he considered good results: All successful initiatives discussed by Nielsen provided instant feedback to participants; moreover, tasks were divided into modules that could be tackled by different groups of people simultaneously (so that tasks could be separated and recombined); and there was always some level of coordination. The question of how exactly coordination takes place (e.g., emerging from bottom up, or top down), and how much coordination is too little or too much, still remains unanswered; hopefully empirical research into CS projects will shed light on this question in the coming years.

Understanding Participation – Towards a Typology of CS Initiatives

What all CS initiatives share in common is that they involve the participation of non-professional scientists at the stage of funding, data collection/generation, analysis, interpretation, application, dissemination, or evaluation. There are great

namely professional scientists). My use of the term 'citizen science' in this chapter should not be understood, however, as implying that all 'citizen science' is indeed led by non-professional scientists.

9 Also here, by 'more socially robust' I mean that culturally shared paradigms or representations, social values, dominant preferences of people, etc. are 'designed into' particular applications. For example, the development of a drug or treatment could contain considerations of the delivery of the drug or treatment that meets the needs of those who will use them (e.g., using tablets instead of liquid medicine when many of the patients do not have refrigerators). Another example would be the development of innovative ways of drug testing that could reduce the pain and suffering inflicted on animals. While social robustness is typically most relevant in the context of the application of science (Stelling 2013), it could also become relevant when deciding what scientific questions to address in the first place (e.g., in the domain of medical research, the prioritising of creating findings that would make a difference in the lives of many people, or the particularly vulnerable).

differences, however, in the activities and formats typically subsumed under the label of CS. While some are led by non-professional scientists at every stage of the project, in others, 'citizen scientists' have no decision making power with regard to core strategies but they contribute merely as data collectors, or even only as funders. The schema presented in Table 9.1 helps us to take a deeper look into the kind of participation that different CS projects involve. It helps to systematically explore how coordination is organised, and agency distributed, in particular CS projects; how 'open' they are; and what kind of entrepreneurial and innovative potential they utilise and foster. The answers to these questions establishes a basis upon which we can explore the question of whether – and if so, how – CS produces results that are better in any way than knowledge created by professional scientists.

The first cluster of questions focuses on *coordination*: Who is involved in agenda setting, in determining the execution of the main idea, and the procedural aspects? Who decides – and how? – what counts as results, what 'good' results are, and what should be done with them? Who decides on intellectual property-related questions?

The second set of questions focuses on the practices and modes of *participation*: Who are the participants of the project, and what characteristics do they have in common? Why and how do they participate? What are the (technical, geographical, language skills, etc.) requirements for participation? How much, and what kind of, training, expertise, experience, skill, and what talents and capabilities, are required to participate in the project? Are there cultural, institutional, or other differences in the perceptions and framings of core issues and stakes among actors at various levels? Very relevant in this context is also Shirk and colleagues' typology of interactions between participants and professional scientists in CS projects, distinguishing between contractual, contributory, collaborative, co-created, and collegial CS projects, according to the 'degree of participation' (Shirk et al. 2012: 4, Table 9.1; see also Ely 2008). My sole, yet arguably important, criticism of these categories is that they treat CS projects as something static. This limits their analytic value with regard to projects whose modes and practices of participation have been changing rapidly. We need to treat the dimensions of participation in CS as something fluid and flexible, as are the understandings of community underpinning it.

Notions of *community* are the subject of the third set of questions in our table. Questions we should ask include: What forms of community pre-existed this project, if any? Which new communities does the project facilitate or give rise to? What are the constitutive factors for the feeling of belonging on the side of the participants?

The fourth dimension is dedicated to evaluation: How, and by whom, is it decided what 'good' outcomes are? Note that 'outcomes' may be different from the aforementioned 'results'; outcomes may include the wider societal, educational, and economic impact. What happens to the results of these evaluations?

Table 9.1 Criteria for the classification of 'citizen science' projects

Coordination: Who has influence in:
1. Agenda setting
2. Determining the terms of the execution of the idea/procedural aspects
3. Deciding what results are (and what 'good' results are)
4. Deciding what will be done with results
5. Deciding on intellectual property questions
Participation:
6. Who participates (demographic and social parameters of those who participate)? Why, and how do they participate?
7. How much, and what kind of, training, skill, or expertise is required to participate in this project?
8. Are there cultural, institutional, or other differences in perception and framing of core issues and stakes?
Community:
9. What forms of community pre-exist this project, if any? Which new communities does the project facilitate or give rise to? What is the constitutive factor for the feeling of belonging on the side of the participants?
Evaluation:
10. How and by whom is it decided what good outcomes are?
11. What happens to the results of these evaluations?
Openness:
12. Do participants in the project have access to the core datasets?
13. Can participants in the project edit the core datasets?
14. Is the contribution of participants adequately acknowledged in published materials?
15. Are datasets made publicly accessible (open source/open access)?
16. Are main findings made publicly accessible (open source/open access)?
Entrepreneurship:
17. How is the project funded?
18. What is the role of for-profit entities in this project? Are these small, medium-sized, or large entities, and where are they located?
19. How are for-profit and other interests aligned in this project (and/or do they conflict, and where?)

Source: Author, adapted from Prainsack 2013a.

Openness is the notion that questions in the fifth cluster revolve around. A certain level of openness is a necessary condition for CS, but never a sufficient one. In short, the more publicly accessible every stage of the creation of scientific knowledge is, the more 'open' is the project. An imaginary project carried out by one Nobel laureate working entirely alone at her lab or at her desk, yet who makes all her data, her lab journals or research notes, and her findings, publicly available, would qualify as an open science project, but not as a CS project, because it does not include substantive contributions from non-professionals.

When thinking about how open a particular CS project is, questions we need to ask include: Do participants have access to core datasets? Can they edit the core datasets? Is the contribution of participants adequately acknowledged in published materials? Are datasets publicly accessible without price or other barriers? Are the main findings freely accessible? In practice, strongly participatory projects will always entail a certain level of openness, because otherwise, active participation by a wide range of non-professional contributors would be impossible.

The sixth dimension of our schema is entrepreneurship. How is the project funded? What is the role of for-profit entities in this project? Are these small, medium-sized, or large entities, and where are they located? Finally, how are for-profit, communal, and other interests and stakes aligned in this project (and do they conflict, and where)?

Answers to these questions may be different for various stages of the project. Some CS projects start out as grass root activities that are later bought by a commercial company and turned into something different. Other projects may change their mission after a few weeks, months, or years, for a range of possible reasons having to do with the internal organisation of the project, or external factors (e.g., new scientific advances, new technical opportunities, etc.). When analysing a particular CS project with the help of this schema, questions in every category should be considered separately for every stage of the project, as answers may vary.

Projects where the role of citizens is limited exclusively to data collection have been criticised as using citizens as 'brain soldiers', rendering them a 'cognitariat' (Toffler 1983; see also Cornwell and Campbell 2012); they often volunteer their time to carry out tasks that average human brains do better than computers, namely the filtering out of 'noise'. Sometimes they also donate their biological material, a situation that has been discussed in the critical social science literature as a form of 'biocapital' (Sunder Rajan 2006), 'biovalue' and 'clinical labour' (Cooper and Waldby 2013), or in the context of 'biosocial relations of production' (Palsson 2009).

The extent of 'grassrootedness' – i.e., how much influence non-scientist participants have over the aim, design, and utilisation of results in a project – tells us something about the emancipatory potential of a CS project. For example, does the project draw primarily on the creativity of people from outside professional science and academia? Does it empower people who would not normally engage with this field of science, and who would normally have no, or very limited,

access to datasets? Etc. The emancipatory potential of a project does not prejudice, however, how successful it will be in terms of the standards and metrics of traditional science. The overall assessment of the success of a CS project will always depend on what the main unit of analysis is: the degree of 'democratisation'[10] of science (whatever is meant by this term in specific instances), the education of citizens (see e.g., Bonney et al. 2009), or the solution of a pressing scientific issue in a way that advances the status of knowledge according to the standards and references of the traditional scientific system itself.

Scholars and commentators have been both enthusiastic and concerned about the emergence of CS in the health domain. Some authors (e.g., Angrist 2010; Nielsen 2011) welcome CS as a process of empowerment of patients and citizens. Other authors, however, are concerned that the replacing of professionally trained experts, such as clinicians and medical researchers, by 'amateurs' participating in the production of authoritative knowledge, may compromise the quality of both the science and the clinical applications emerging from them, because amateurs are not trained in scientific methodologies and may compromise the data quality and/or analysis. Hauke Riesch et al. (2013), in a study drawing upon interviews with professional scientists in a specific CS project, found such concerns articulated by their interviewees. This stance, of course, draws on the assumption that professional scientists all have solid methods training, and that they are less likely to 'taint' data than non-professionally trained people; this is a plausible yet unproven assumption, especially against the backdrop that amateurs contribute to the creation of scientific knowledge with the help of online tools where safeguards for data quality can be 'designed into' the application.

Also the political dimension of CS deserves closer attention. For example, it could be argued that models of participation in CS projects – especially those which are 'run', or coordinated, by companies, state authorities, or other actors

10 A paradox here is that the one ingredient that virtually all authors discussing participatory models in science and medicine see as crucial for a project to succeed, namely a certain amount of coordination, very often comes at the cost of participation. A very similar problem lies at the heart of any socio-political entity; it has been addressed by social contract theorists in their justification of state authority. Social contract theorists did not seek to provide an accurate historical account of how state authority came into being, of course, but they sought to justify it morally and politically. The core tenet of social contract theorists is that state authority is needed for any large and complex social structure to survive; if no centralised coordination equipped with the power to enforce its rule did exist, then people would be each other's 'wolves', as Thomas Hobbes famously put it. I argue that we should not be tempted to assume that this reasoning can be easily transposed to participation in science and medicine. Social contract theorists spoke of rule-making at the core of organising the basic structure of our social and political life; they did not talk about *any* rule-making in *any* organisation or institution. It is for this reason that I think we should use calls for, or claims of, the 'democratisation' of science with caution: Science, especially when it is publicly funded, should be publicly accountable; but this is different from saying it should be 'democratic' (Prainsack 2012b).

which are not primarily acting in their capacity as non-professionals and citizens – bear strong resemblances with many Web 2.0 enterprises. Google, for example, famously combined the prioritisation of user experience with reliance on user-generated information (Google's algorithms draw on how many times users access particular websites), and now dominates the market (Auletta 2009). According to this more pessimistic view of CS initiatives, people contributing to science in projects where they will not share the profits engage in value co-creation for the initiators of the project (Arvidsson 2008; Bonsu and Darmody 2008), whether these are for-profit companies, or traditional academic and scientific institutions who receive the main credit for the discoveries made by citizens.

We should not assume, however that all those who participate in projects where participants have only limited influence in project design are being exploited. For many, being part of something useful, being acknowledged publicly in publications, or learning about the scientific area in question is enough of an incentive to participate, and a satisfactory reward. While there certainly are initiatives aiming at profiting from the unpaid labour of people, not every instance of citizens participating in such projects should be read as an instance of false consciousness. As research with users of genetic testing services offered online has shown, motivations and benefits for users are diverse and complex, including the quest for entertainment, playful engagement with information, unspecified curiosity, and the desire to contribute to something meaningful (McGowan et al. 2010; Vayena et al. 2012).

The following section will take a closer look at 23andMe, one of the companies offering personal genome testing beyond the clinic (BTC; this concept captures practices of dealing with medically relevant information or data in ways that transcend the clinical domain; see Prainsack and Vayena 2013). I will examine how this company fits into the bigger picture of CS. 23andMe mobilises the rhetoric of CS and collaborative problem-solving very prominently; but does it entail any tangible devolution of agency from professionals to users (citizens)?

CS and Personal Genome Testing: The Case of 23andMe[11]

23andMe is probably the most widely known BTC genomics company (see also Groves and Tutton 2013: 7). It was one of the first services that did not test only particular genes but that considered markers across the entire genome. It launched in 2007 with considerable media attention, and was named 'Innovation of the Year' by *Time* magazine in 2008 (Hamilton 2008). 23andMe started out as a mere genome testing service: customers could order 'spit kits' online, received their saliva collection kit by mail, returned it to the company, and were given access to

11 The section draws upon observations of the development of 23andMe since its launch in 2007, as well as conversations and e-mail exchanges with users. An analysis of blogs, websites, and academic literature on 23andMe and other 'direct-to-consumer' genomics companies complemented these data (see also Prainsack 2011).

their results online a few weeks later. The company used a custom-made chip to test (initially) roughly 500,000 single-base variants – so-called single nucleotide polymorphisms (SNP) – across the genome to infer personalised genetic predisposition profiles for a range of diseases, drug metabolism and carrier status, phenotypic traits, as well as information on probable genetic ancestry (Prainsack et al. 2008).

Some commentators had suggested from the start that what 23andMe was doing, besides providing a personal genetic testing service, was building up a database that could serve as a resource for research (Lee and Crawley 2009; see also Harris et al. 2013). It was apparent that such a database could only be useful if, in addition to the genomic information of its customers, it included phenotypic, lifestyle-related, and other relevant (medical and other) information as well. In 2008, 23andMe introduced a research feature called '23andWe', which encouraged customers to 'vote' for diseases that the company promised they would then prioritise in their research brokering efforts. Customers were also asked to contribute their own lifestyle and other relevant information for research purposes. Around the same time, 23andMe started to offer free spit kits to people who had been diagnosed with diseases that the company decided to focus on. As a result, 23andMe increased the number of their users to roughly 150,000 by 2012 (Anon. 2012); moreover, they started publishing academic papers reporting on the results of their research, mostly replicating associations between genetic markers and phenotypic traits (Eriksson et al. 2010) as well as diseases (Tung et al. 2011) on the basis of phenotype characterisations that stemmed directly from their users.

In 2012 the company became the subject of severe criticism when it became known that they had filed a number of patent applications (see Sterckx and Cockbain, in this volume).[12] 23andMe sought to control the damage by insisting that they had informed customers in their Terms of Service, and in their consent document, about their intent to pursue intellectual property rights (see Vorhaus 2012a). This reference to a few instances of technical jargon in the small print on the website did little to appease the concerns of some outraged commentators (summarised in Vorhaus 2012a; see also Rimmer 2012). Although very few participants in this discussion accused 23andMe of doing something illegal by filing patent applications, a widely shared sentiment seemed to be that the company had acted immorally, and that it had been dishonest by not having communicated their intention to secure patents openly to their customers (and indeed to the wider public). Indeed, it is plausible to argue that it was ill-suited for an organisation that utilised the CS rhetoric as heavily as 23andMe does, to capitalise on the achievements that were made possible with the help of their customers, 'behind their backs'. In other words, the free labour (see Palsson in this volume) that the customer base had put in, in the name of communal benefit, was 'rewarded' by the

12 The controversy was fueled anew when the company was awarded a patent to a method for gamete donor selection that could enable clients of fertility clinics to have a say in what traits their future offspring was likely to have; see Sterckx et al. 2013.

realisation that the company not only did not intend to share any financial profits with those who had provided the data, but also that it could be erecting barriers to research by charging licensing fees for patents that were granted (Rimmer 2012). What made the situation worse was that some patients had joined 23andMe upon the recommendation of their patient support group, hoping that the contribution of their genetic and other personal data would help accelerate the development of treatments; the suspicion that 23andMe may in fact be planning to limit research by enforcing intellectual property rights on tests and applications developed by them may have felt to some of them like a slap in their face.

When thinking about 23andMe along the lines of the dimensions outlined in Table 9.1, a mixed picture emerges. In terms of deciding what exactly will be done with the results, and devising strategies regarding the sharing and exploitation of research findings, 23andMe operates like the for-profit company that it is: Participants have little say in these matters, despite participatory rhetoric (see also Prainsack 2013b). With regard to the openness and transparency of research carried out with the data of their customers, and the actual policies regarding access to data and findings, however, 23andMe has strong participatory elements: The company is in an active dialogue with their participants on these matters, and often implements their suggestions (see also Prainsack 2011; Reardon 2011). Customers can also download their raw data from the website and export it to any other repository, private or public initiative, or personal computer they want to. Also in terms of agenda setting – i.e., deciding what diseases and traits 23andMe-facilitated research should focus on, how personal genetic risk calculations are presented on the website, and what research questions should be included in the '23andWe' research feature, participants have significant influence. In sum, 23andMe is a for-profit company that includes CS elements. This mix of features, in addition to the fact that their business model seems to be continually evolving, is not untypical for web-based enterprises in the Web 2.0 era and beyond (Auletta 2009; Topol 2012). All the elements that we find in 23andMe – the alignment of a for-profit orientation with a seemingly genuine commitment to facilitate disease research and create collective benefits, the simultaneity of top-down and bottom-up practices and elements; centralised and de-centralised decision making structures, and frequent modifications in the company's goals and missions – characterise a wide range of social media, online games,[13] and web-based enterprises. If CS initiatives become more widespread, and increasingly adopted into mainstream science and research institutions, these characteristics could become common features of most web-based CS initiatives as well.

13 The field of 'serious games', which include scientific discovery games is growing very fast, and has been hailed as a highly potent tool for education, and for the dissemination of research findings. The biggest funder of medical research in the UK, the Wellcome Trust, for example, started a 'Gamify your PhD' initiative in 2012 (Wellcome Trust 2012). See also McGonigal 2011; Nielsen 2011.

The Self-Tracking Leviathan: What We Still Need to Figure Out

When we compare 23andMe to other BTC genomics services, such as the Icelandic deCODEme[14] (decodeme.com) or the German bio.logis (bio.logis.de), against the criteria formulated in Table 9.1, we are faced with an interesting question: 23andMe, which has been the subject of intense criticism in public and expert circles, seems to have more strongly pronounced participatory features than any other BTC genomics company: deCODEme and bio.logis, for example, have never had research features where users can suggest questions, or fill in surveys to contribute non-genetic data for analysis. Users cannot download raw data to run their own analyses, and bio.logis also selects very carefully the conditions and traits it tests for: It commits itself to testing only genetic variants for carrier status of inherited diseases or conditions for which further preventive and therapeutic measures can be taken, and refrains from testing for predispositions to conditions that would necessitate invasive interventions (such as predispositions for breast cancer). In short, users have more agency in shaping (some) elements of the 23andMe service than they have in relation to other BTC genomics services. Nevertheless, 23andMe seems to have attracted much more criticism than any of the other services. Is it only because 23andMe is the most widely known such service? Perhaps this is the main reason indeed; there are no established parameters according to which such an hypothesis could be tested. Another plausible explanation would be that the high level of criticism is related to the companies' financial and legal set-up. It is widely known that 23andMe's founder, Anne Wojcicki, was married to Google-boss Sergey Brin (the couple separated in autumn 2013, and both partners publicly insist that this separation does not affect Google's investments in 23andMe). This fact puts the company in an unfavourable light with those who treat the marriage of health and surveillance, and of big data and big money, with suspicion. Thus, 23andMe is seen by some people as a kind of Big Brother disguised with white coat and a stethoscope.

An additional explanation of the fervent criticism that 23andMe has attracted, however, could lie in their strong emphasis on participatory features, which challenge the gate-keeping position of clinicians to health-related genetic expertise. So far, clinicians have been the gate keepers to genetic information in the health domain, and any challenge to this prerogative has generated resistance. Moreover, the concern on the side of many clinicians about the risks of online genomics testing when no clinicians are involved – primarily concerns regarding needless anxieties or groundless relief on the side of test-takers – seems to reflect not only resistance against losing power and influence, but also a genuine concern about the wellbeing of patients. Within this rationale,

14 deCODEme stopped selling tests direct-to-consumers early in 2012, as a result of their takeover by Amgen (see Vorhaus 2012b).

it is perhaps unsurprising that 23andMe has attracted so much criticism: Other services, such as the Personal Genomics Service offered by the Frankfurt-based bio.logis, for example, do give users direct access to their test results (i.e., test-takers do not need to go through a clinician), but they leave relatively most of the other core categories that underlie the organisation of medical systems and discourses, intact: the distinction between 'science' and 'the public', between 'knowledge producers' and 'knowledge recipients', and between the medical and the non-medical. 23andMe, in contrast, contributes significantly to the blurring of all these categories. I argue that 23andMe's services are *not merely* the result of a particular business strategy, but they *also* embody several normative stances: (1) that access to data is an end in itself, and that it is not the role of the service provider, but of the end user, to decide on the utility of the data;[15] (2) that data do not flow only in one direction – from the service provider to the user – but also vice versa (the notion of 'prosuming' captures this well; see Toffler 1990); (3) that in a system that relies on data contributions – and partly also contributions to data interpretation and analysis – from volunteers, the definition of expertise is changing. Following this rationale, it would be plausible to argue that 23andMe has faced so much criticism also because it unsettles existing orders and makes controversial territorial claims, much more than other BTC companies do.

Another plausible explanation for the intense criticism that 23andMe has faced *in the more recent past*, of course, is their filing for patent applications without being proactively open about this.

But 23andMe's questionable intellectual property strategies aside, if the explanations offered in this section are valid, then one of the main strategic 'mistakes' of 23andMe was to become a hybrid: It does not fit any traditional categories – such as commercial enterprise, patient group, clinical testing facility – but it combines elements from all of these. There seems to be a tendency in public discourse to applaud participatory practices in bottom-up, grassroot, 'amateur'-driven projects, but not in commercial enterprises, nor in 'proper' academic institutions. Such a tendency could be explained by the hegemony of traditional understandings of expertise and authority: In organisations dominated by professional experts, their expertise should not be diluted by giving 'amateurs' too much say. In organisations and projects initiated by 'amateurs', in contrast, increasing the number of decision makers is seen as compensating for the supposed lack of *quality* of decision making (because decisions are made by other people than professional experts). This observation applies to wider instances of public participation in areas that are traditionally seen as the prerogative of those who hold esoteric knowledge, of course, and not only to CS.

15 This, of course, is also one of the central paradigms of Google (Auletta 2009).

Conclusion

We still need to do a lot of work to obtain a better understanding what CS is, and how it will affect the creation of scientific knowledge as we know it.[16] There is also an evident need for systematic empirical and conceptual explorations of the circumstances under which CS projects generate good outcomes in the sense that outcomes are academically or scientifically more accurate and better, and more socially robust, than the results of traditional ways of scientific knowledge production in health. Moreover, researchers will hopefully also explore according to what parameters the results of CS should be evaluated and assessed. An important question is how CS represents a significant change in how we assess and enact relevant expertise and authority when we create scientific knowledge, and how it does or should affect the ways in which we discuss and support participation in science. To complicate the situation further, contributions to generating scientific knowledge by non-professionals is typically neither a discrete nor an isolated activity but it is interwoven with other kinds of engagements, such as learning, gaming, passing time, and sometimes also profit-making. Also those of us who are not primarily interested in CS, but instead in how and why people engage with online genetics and genomics, can benefit from looking at these engagements against the backdrop of old and new modes of 'amateur' participation in science.

Acknowledgements

I am grateful to Jennifer Fishman, Michelle McGowan, Gisli Palsson, Silke Schicktanz, Tamar Sharon, Sharmila Sousa, Sara Tocchetti, Gabriele Werner-Felmayer, Hendrik Wagenaar and the participants of the 'Genetics goes Online' workshop in Maastricht, NL, on 12–14 September 2012 (organised by Anna Harris, Susan Kelly and Sally Wyatt) for helpful comments on the manuscript, in particular, Ine van Hoyweghen.

16 Hauke Riesch et al., in a study on 'citizen science' informed by interviews with professional scientists, draw attention to professional scientists worrying that labour- (and thus cost-) intensive work will increasingly be outsourced to volunteers under the label of 'citizen science'. As one interviewee said, the unpaid volunteers 'are taking my job away from me' (cf. Riesch et al. 2013; see also Cohn 2008. Note that both studies dealt with cases outside of the health domain).

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Chapter 10
LabouringMe, LabouringUs

Gisli Palsson

Focusing on personal genomics services (in particular 23andMe and deCODEme[1]) which offer personal services to people who wish to assess genetic risks for common diseases and to explore geographies of ancestry, this chapter extends the notions of labour and relations of production beyond 'natural' resources in the classical sense to the extraction, reproduction, and exchange of bodily material and information, to *biosocial* relations of production (Palsson 2009a). People who subscribe to personal genomics services tend to be seen as engaging in either recreation or consumption, not in labour activities. While a growing body of literature has drawn attention to the labour carried out by women in the context of artificial reproduction (see, for instance, Dickenson 2009), a similar perspective has not been developed with respect to personal genomics. I suggest that the labour carried out when subscribing to personal genomics services largely goes unrecognised in both the industry and the pertinent literature and that this needs to be rectified. If one takes this perspective seriously, one is bound to ask how such labour contributions can be properly acknowledged and rewarded and what kinds of regimes of governance and property this would entail. This is particularly acute if one considers the growing evidence of relational, entangled bodies recently accumulated by epigenetics, macrobiotics, and related fields.

While it may be argued, in a late-Foucauldian fashion, that the users of genomics perform labour on themselves, seeking to know themselves and to care for their bodies, their labour also needs to be situated in the biomedical mode of production involved, an hybrid complex of living material, digital information, and social relations. Addressing the realities of life itself in late-modern times in terms of labour processes and relations of production, I argue, helps to characterise the different arrangements involved in the production and circulation of biosocial value.

So-called 'life itself', the brute matter of living organisms, has become one of the active zones of economic production. Immortal cell lines are a case in point, reproduced on a global scale for a variety of purposes independent of the original host. At the same time, the capacities of the fragmented body have been turned into instruments for production, redefining labour and agency. Not only do all kinds of agents and instruments labour *on* life, through the political economy of life itself, life itself *does* all kinds of labour. One expanding labour front, I suggest, is genomic labour, involving the production of biosocial value which is material and informatic at the same time, both natural and social.

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1 deCODEme is no longer operating (see also Prainsack, in this volume).

Classic theories of political economy developed by Adam Smith, David Ricardo, Karl Marx and many others, tended to assume a labourer developing and applying his or her skills *in situ*, typically in an agricultural field or an industrial factory. For them, labour activities typically took place in the home or a nearby field or factory, in a local community embedded in a larger political economy. There have been exceptions, of course, throughout human history, with partial or temporary separations of the site of labour activities and the site of home. In fisheries and hunting and gathering, for instance, much production takes place far away from what are normally taken to be the geographical boundaries of the local community. Also, there is the case of pastoralism, where the two sites, so to speak, travel together; the herders and their camps regularly being on the move along the same tracks, following the seasonal fluctuations of the land, the bodies of their animals, and the political regimes of their neighbours. Nevertheless, in agrarian and industrial discourse the sites of labour and home have tended to be more or less permanently collapsed, a single life world or habitat. This is true as well for many (other) 'folk' theories of the forces of life and the generation and circulation of value, including the South American *casa* or house economies described by Gudeman and Rivera (1995) and the medieval Scandinavian *óðal* or estate described by Gurevich (1992).

In recent years, however, the spatial relations of labour activities have been radically changed as a result of complex and interrelated developments, including the growth of the World Wide Web, the network society (Deleuze 1995; Hardt and Negri 2000), and virtual migration (Aneesh 2006); 'paradoxically', Aneesh notes, 'the new space of transnational labour has reversed its relationship with the worker's body. Rather than move the body across enormous distances, new mechanisms allow it to stay put while moving vast quantities of data at the speed of light' (2006: 2). Call centres of the kind studied by Aneesh underline the ability to perform work at a place other than the site of the acting body. What travels, here, is not the worker, but her voice and presence, in its digitalised form of bytes and megabytes.

Not only have the sites of labour and production increasingly been separated, the capacities of the body have been fragmented and turned into instruments for production, redefining both human labour and human bodies. Thus, the famous HeLa cell line taken from Henrietta Lacks who died from cancer in 1951 are reproduced on a global scale for a variety of purposes independent of the original host (see Landecker 2007). 'Pluripotent' stem cells, endowed with the capacity to generate a variety of body tissue, represent another example. The 'same' body, then, in a sense, performs labour at two or more sites simultaneously. Not surprisingly, these complex tournaments of biosocial value have become active zones of politics, posing intriguing questions about biopolitics, place, and agency.

In order to draw attention to the significant economic role of women in the reproductive sector of biomedicine, some scholars have adopted an expanded notion of labour. Waldby and Cooper recast the gift economy for reproductive material as a form of unacknowledged productive work: 'We want to argue

that women who donate or transact their biological material to the regenerative medicine industries are engaged in a form of labour, even though the terminology of labour is not used in these contexts' (2010: 8). The labour involved, they suggest, goes unrecognised or is denied partly because 'it takes place at the level of women's biological embodiment, and hence it is readily naturalised, in much the same way that women's emotional labour in the service industries is taken for granted as a given feminine attribute' (2010: 9). Rethinking the rhetoric of altruism often associated with assisted reproduction, they both draw upon and go beyond feminist analyses that have applied the logic of alienation to the context of the home and the family. For them, a major characteristic of contemporary relations of reproduction in biomedicine is '*a denationalization of the reproductive sphere and its exposure to global precarious labour markets*' (Waldby and Cooper 2010: 12; emphasis in the original).

While the 'clinical' focus is highly productive and illuminating, it should not blind one to the importance of immaterial goods. A broadened notion of *biosocial* value is essential, I suggest, for understanding the coproduction of the hybrid complex of genomic material and information that Prainsack and I have referred to as 'genomic stuff' (Palsson and Prainsack 2011). Thus, the users of personal genomics services often contribute both a biological sample (a swap or a spit) and a variety of information on background and life style. What do personal genomics services consist of and how do people become implicated in new labour processes as they subscribe to them?

In his 'Theses on Feuerbach', the sixth thesis, Marx observed that 'the essence of man is no abstraction inherent in each single individual. In its reality, it is *the ensemble of the social relations*' (1998: 573; italics added). How might our understanding of personal genomics services and related blurring of the material and the immaterial in the production and reproduction of life itself benefit from the expansion of Marx's notion of the ensemble, by speaking of ensembles of biosocial relations (Palsson 2013)? The empirical part of the discussion is partly based on my own experience of requesting a genome scan and exploring the results.

Decode Me!

Before going on, it is pertinent to briefly describe the case of personal genomics, my key empirical site, represented by deCODEme, 23andMe, Pathway Genomics, Navigenics, and similar services. These services tend to claim that they 'democratise' genomics both in the sense that they offer test kits for a low price (ranging from $250 to $2,500), within the reach of the public, at least not just the research elite and the wealthy, and in the sense that analyses and interpretations of genome scans are now a matter of intense public discussion through all kinds of media, including web browsers and blog sites.

In October 2008 I signed up for the 'complete' scan offered by deCODE genetics, curious to explore the analyses it offers, to see what this might reveal about myself and my roots, and to find out how anthropological expertise was

implicated in the project (Palsson 2009b, 2012). Two weeks after I sent my cheek swabs and the relevant forms, I received an email from the company. The results were now available and I would be able to access them through the password provided. Since then, I have regularly received messages from the company alerting me to both updated and new conditions, to further analyses of traits and health risks. Once I logged on to see the results, I was urged to 'have fun browsing [my] … genome', 'dig into [my] … DNA', explore my ancestry and my 'genetic risks', play with maps and other visuals, search for specific genetic variants (SNPs or 'snips'), and download my genotypes for 1,2 million SNPs, a 33Mb datafile (deCODEme 2008).

The search for ancestry has six key features: The first, the 'Atlas', provides a comparison of one's genetic code with that of people from all over the world, based on several hundred thousand genetic variants and more than 1000 reference individuals from 50 different populations worldwide (see Figure 10.1). The Atlas compares my genome to reference populations throughout the world, ranking regional clusters (1 to 6) in terms of their relevance for me, in the order of genetic similarity: Europe (1), South West Asia (2), East Asia (3), America (4), Oceania (5), and Africa (6). In each case, I can zoom in on the population involved. My genome, not surprisingly, turned out to have most in common with European reference groups (a genetic similarity of 83,99 per cent), in particular those of Iceland, the Orkneys, France, and Russia. More astonishingly, the second feature, 'ancestral origins', indicates that judging from chromosomes 1 to 22 my ancestry is no less than 7 per cent East Asian, 16 per cent according to the X chromosome, considerably higher than for most Icelanders. I found this an interesting and puzzling revelation. To speak of 'genealogical dis-ease' (Rapp, Heath, and Taussig 2001) – to use a term developed by anthropologists studying what people make of genetic information about their roots and ancestry – would, however, be an overstatement.

The analysis of deCODEme of mitochondrial DNA establishes one's place in a matrilineal family tree spanning 170 thousand years. It turns out I belong to 'mitogroup R*', a category shared by 4.8 per cent of deCODEme users all of whom can trace their mitochondrial DNA to a woman thought to have lived about 60 thousand years ago, probably somewhere in the Near East. Analysis of my paternal DNA, on the other hand, shows that I belong to 'Y-group R1a', a category shared with 10.3 per cent of deCODEme users tracing their Y chromosome back to one man who is thought to have lived about 10 to 15 thousand years ago, probably in Western Asia. A further feature allows users to explore their 'map of kinship', a visual representation of genetic space on the basis of principal component analysis. Given this evidence, I occupy a somewhat marginal position, neither firmly within the European reference group nor any of the others, probably reflecting the puzzling observation mentioned above about my East Asian ancestry.

The main service, however, offered by deCODEme is that of analyzing the genome with respect to specific traits and health risks. For some weeks I resisted the lure of the health results. Both of my parents had struggled with cancer and I

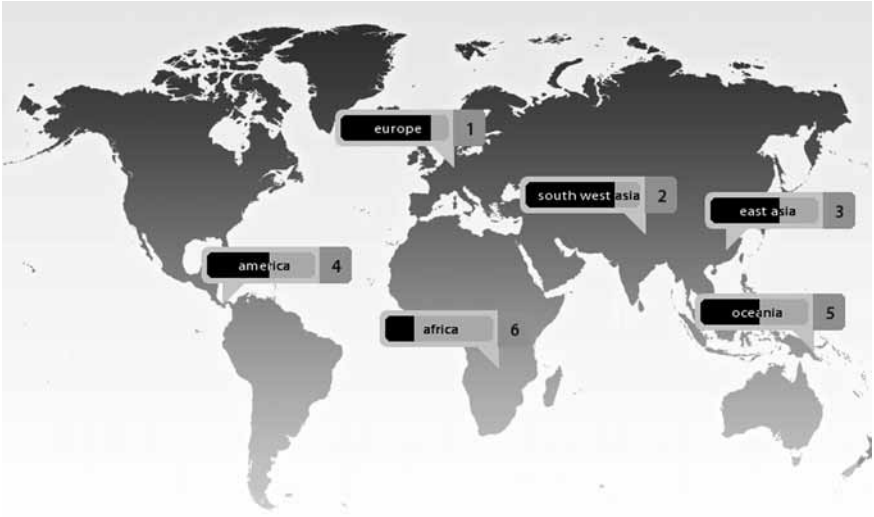

Fig. 10.1 The Genetic Atlas (according to deCODEme)

wasn't that interested in the kind of fortune telling offered by personal genomics. My results for the 50 diseases and traits currently covered are based on calculations comparing my genome to sequences of participants in studies published in the scholarly literature. To access results for some diseases I was invited to read about the genetic and medical details and to sign a statement about informed consent, by clicking on 'Accept'. I need not bore the reader with the personal details. Suffice it to say that some of the information provided sounds trivial (no alcohol flush reaction), some of it resonates with what I thought I already knew (I am less likely than the general population 'to become nicotine dependent (15 per cent or less)'), some results are encouraging (I have low lifetime risk for some diseases, much less than for males of European ancestry in general), and some details may promote the hypochondriac in me to request further medical information. When presented with these results, I was offered details on the mathematics of risk analysis. Also, I was invited to 'zoom in' on my genomic landscape, focusing on a part of a chromosome and the location of specific mutations reportedly responsible for potential traits or diseases. Again, there are some surprises and some food for thought.

The Genome Browser of deCODEme allows users to compare their complete data with friends and family. While my reference group of friends and family includes both hypochondriacs and anthropologists, so far they have seen few good reasons to participate and, as a result, there isn't much to compare. The website, however, allowed me to examine my genetic sharing with three 'famous' men, including Kári Stefánsson (the President, CEO, and co-founder of deCODE genetics). Here, the level of 'sharing' is visually indicated by colouring the relevant bits of the chromosomes.

No doubt personal genomics is becoming a family affair as well as a global concern, a form of 'recreational' genomics. At any rate, a thriving imagined community (Anderson 1983) of the users of personal genomics projects has been developing on the Internet. A number of websites testify to a lively discourse on the issues involved, including thinkgene.com, dna-forums.org, Eye on DNA, Urban Semiotics, and Dienekes' Anthropology Blog. The last one is 'dedicated to human population genetics, physical anthropology, archaeology, and history'. Judging from these websites, there is more interest in exploring ancestry than health risks. Perhaps users are reluctant to reveal their health risks in public, although they may be keen to download the relevant information for their own purposes. Some of the websites referred to are focused on specific personal genomics projects while others are more general. Users engage with the goals of personal genomics, analyses of their own genome, and comments expressed through the expanding virtual community of the Internet.

Technologies of the Self

In some of his last writings, Foucault (1988) shifted his attention from systems of domination to the agency and experience of the individual, drawing attention to the particular kind of subjectivity characteristic for the modern age and what he called 'technologies of the self'. It seems reasonable to argue that personal genomics represent one example of technologies of the self. Indeed, the genomics of ancestry is often assumed to provide an important avenue into identity and personhood. As Pinker observes (2009): 'Affordable genotyping may offer new kinds of answers to the question 'Who am I?' – our ruminations about our ancestry, our vulnerabilities, our character and our choices in life'. Significantly, Sykes book on the tracing of ancestry (2001) which opens with the question 'Where do I come from?' closes with a chapter on 'A sense of self'. This point is also underlined by the co-founder of 23andMe, Anne Wojcicki: the 600,000 genetic markers interpreted by 23andMe, she argues, are 'the digital manifestation of you' (see Hamilton 2008). Knowing where we come from, we apparently also know who we are (see also Quitterer, in this volume).

The companies involved in personal genomics tend to emphasise consumers' access to medical knowledge and their relative independence of the medical establishment. Thus, the claim by deCODEme: 'we wanted not only to empower the public, but also to give students, academics, physicians and other professions with an interest in genetics a chance to get a more in-depth view of their code and genome' (deCODEme 2008). Indeed, users draw their own conclusions and engage in dialogues with genomic experts, sometimes becoming experts themselves in the process (see, for instance, Soo-Jin Lee and Crawley 2009). In a sense, then, this is science from below (Harding 2008). One example is SNPedia. Drawing upon summaries of peer reviewed articles presumed to be relevant for given genomic data, it allows users of different testing services to

pool personal data, to learn more about their own genotypes, and to explore the effects of variations in DNA. The consumption of personal genome data, as Prainsack points out, is also sold as an experience (Prainsack 2011). Indeed, for me submitting samples, browsing the websites, downloading the results, and discussing them with family and colleagues was both an experience and an opportunity to look at potentially relevant medical information that my family doctor was unlikely to have access to. Interestingly, when I presented my results to my family doctor she appeared not to know of the deCODEme service. This is not, however, the end of the story. As we will see, one should not be too easily seduced by the rhetoric of democracy and the care for the self. It is important to draw attention to the labouring consumer.

Labouring Lives

In his writings on labour and economic production, Marx sometimes referred to nature as the 'inorganic body' of humans: 'The universality of man is in practice manifested precisely in the universality which makes all nature his inorganic body – both inasmuch as nature is (1) his direct means of life; and (2) the material, the object and the instrument of his life-activity' (1959: 75–76). For Marx and most of his contemporaries, labour activities were, by definition, directed at the extra-somatic, external world. Engels, however, suggested one might think of the body itself as the product of labour. Drawing upon an evolutionary perspective, he argued that 'in a sense, we have to say that labour created man himself' (2007: 25). '[T]he hand', he went on, 'is not only the organ of labor, it is also the product of labor' (p. 26). The modern world of biomedicine has made Engels' man-makes-himself perspective more pertinent than he could have imagined. Thanks to their labour activities, humans are now able to reproduce their own bodies, as part of the 'inorganic body' of 'nature'. Not only do modern bioindustries produce a variety of 'biologicals', agents extracted from or generated by biological material, these biologicals perform their own labour. Stem cells, for instance, are increasingly cultivated outside the human body, producing organs as 'spare parts' for humans (at least this is the vision of regenerative medicine).

Broadening the feminist perspective of Dickenson (2009), Waldby and Cooper (2010), and some others, I suggest that the labour carried out by *both* men and women when subscribing to personal genomics services largely goes unrecognised. Also, this labour is both material and 'immaterial', contributing personal information regarding life style, diet etc. as well as DNA material. Moreover, in contrast to the neo-liberal 'body shopping' represented by nannies and cleaners who physically move to the site where they are needed (Boris and Parreñasd 2010; Constable 2009; Freeman 2011), the providers of genomic material and information are virtual migrants, in Aneesh's sense (2006), at someone's service, contributing to transnational biobanks and databases that can be operated from anywhere anytime through the aid of the internet and computing machinery.

In the process of requesting and using personal genomics services, then, consumers facilitate, either directly or indirectly, the construction of gigantic DNA assemblies, coproducing knowledge of genomic differences. The spokespersons for 23andMe, unlike most of the other projects, have been quite open about the issue of alternative – e.g., research – uses of their data. Wojcicki suggests signing up for 23andMe is 'a great way for individuals to be involved in the research world … You will have a profile, and something almost like a ribbon marking participation in these different research papers. It will be like, 'How many *Nature* articles have you been part of?'' (Venturebeat 2007). This is highlighted in a comment on one of the web sites: '23andMe will be sitting in one of the largest genetic databases on Earth. And there's no opting out …' (Venturebeat 2008). Arguably, the people contributing cheek swabs to personal genomics services are engaging in co-working, a collective labour process that ultimately results in large-scale biobanking. Spitting saliva and providing cheek swabs, after all, is biosocial work, potentially contributing to the global networks and hierarchies involved in the manufacture of biosocial value.

deCODEme is part and parcel of its mother company deCODE genetics, whose purpose is to advance biomedical research and pharmaceutical development. Although the company seems to have no plans to directly draw upon its personal-genomics data in its biomedical research, a closer integration might take place later on. There are also strong financial and technical links between 23andMe and the giant Google which may be indicative of new, hybrid forms of biobanking and bioinformatics. Whatever their current ambitions, personal genomics projects are likely to connect with larger biomedical projects in the future. Given the possibility of hacking genomic data (Aldhouse and Reilly 2009), the clients of personal-genomics companies may eventually be contributing to projects beyond the awareness and control of the services they have contracted.

The possibility of linking a variety of scattered biomedical databases is not that remote. Thanks to the development of bioinformatics and the internet, it is no longer necessary, or even feasible, to assume a central 'hub' with monopoly of access. Already, there is much talk of 'federated' databases; such databases 'are a more complicated solution in terms of the required technologies, but they bring certain advantages that cannot be endowed by a centralized database' (Thorisson, Muilu, and Brookes 2009: 13). Record details from remote sources may now be directly searchable by other computers taking part in federation.

Perhaps the social network of Facebook helps to illuminate the issue of labour and co-working in the context of personal genomics. Here, users' expressions of 'likes' with respect to particular retailers or services are routinely translated into advertisements, as a result of which Facebook collects profits from retailers' payments. How could users' work be acknowledged? A lawsuit filed in California in 2011 (ANGEL FRALEY et al. Plaintiffs, v. FACEBOOK, INC., Defendant) provides some clues. The plaintiffs argued that Facebook users were not sufficiently informed of how their 'likes' translated into profits. In response, Facebook proposed a settlement which would involve informing users about sponsored

stories and a payment of $10 million to research and advocacy groups that work on digital privacy rights. A federal judge in California, however, rejected Facebook's settlement offer, requesting clarification on what would count as adequate and fair in such calculations suggesting that lawyers might have 'bargained away something of value' (The United States District Court for the Northern District of California San Francisco 2012). Presumably the judge was referring to the co-working of users.

Biosocial Relations

Hardt and Negri emphasise the vital dimensions of biopolitics, focusing on the production and reproduction of life itself; for them, it would be misleading 'to treat the new labouring practices in biopolitical society *only* in their intellectual and incorporeal aspects. The productivity of bodies and the value of affect … are absolutely central in this context' (2000: 30). Perhaps it is necessary to react to the informatic, textual trend associated with the mapping of genomes and the 'code of life'. Indeed, life itself – in the form of stem cells, tissue, and organs – is a central component in the production of biovalue, a point highlighted by the notion of 'clinical labour' (Waldby and Cooper 2010). Often, however, it is difficult to maintain a rigid distinction between the corporeal and the incorporeal.

This is particularly relevant for current gene discourse. As Keller (2000) famously argued, the concept of gene is highly unstable, and varies from one discipline to another. For Rheinberger, similarly, the gene belongs to a class of fuzzy 'objects' that cannot be assigned a precise meaning; in his view, the usefulness of boundary objects does not rest with a clear definition from the outset: 'indeed it can be rather counterproductive, to try to sharpen the conceptual boundaries of vaguely bounded research objects while in operation' (Rheinberger 2000: 221). Keeping in mind the fact that the genome is sometimes regarded as informatic assembly and sometimes as a material thing, it seems to make sense, in order to avoid unnecessary ambiguity, to simply speak of 'genomic stuff' (Palsson and Prainsack 2011); in other words, leaving aside the issue of materiality versus meaning. The notion of 'biovalue', then, seems too restrictive when dealing with genomic stuff.

To capture the complex implications of personal genomics services it seems pertinent to draw upon the notions of biosocial value and ensembles of biosocial relations, notions that seem to resonate with many indigenous accounts of personhood and relatedness (Palsson 2008). Users of these services are often contributing both tissue and information on phenotypic characteristics, health, drug use, and life style – information that in the future will probably be updated interactively to increase the efficiency of the machinery of schemes such as deCODEme and 23andMe.

Prainsack (2011) suggests that if we are witnessing a 'participatory turn' in genomics we need to ask what it involves. She makes a distinction between 'early adopters' and 'regular consumers' of personal genomics service, to

highlight their different motivation and work. While early adopters may play a significant role in shaping the project, regular consumers are likely to be rather mute or passive. Contributors to genomic services and biobanks are not only doing work in the sense that they provide genomic stuff of critical importance, also they are engaged in the coproduction of biosocial networks. This is an issue highlighted in several recent works (see, for example, Deleuze 1995; Levina 2010; Thacker 2005). Drawing upon her work on 23andMe, Levina suggests that 'life in the network society requires of its denizens a constant contribution to the growth of the network. …. Members are encouraged to think of themselves as dividuals, or nodes, in the network' (2010: 2). If network subjectivity, she continues, 'is conceived in terms of dividual bodies and identities, then each body – reduced to its information – can be abstracted from its social and cultural context. It becomes, in a sense, a free-floating signifier' (Levina 2010: 7). Such a perspective seems to resonate nicely with the notion of humans as ensembles of biosocial relations.

Entangled Bodies Entangle Services

With the new genetics and personal genomics, the biological gaze has been turned inward to the management and mining of the human body. While, the gaze has shifted from the outside to the inside and the 'inside' is often externalised for gazing at, such a distinction should not be rigidly maintained. Growing evidence suggests that the human genome is fundamentally mixed with the microbiomes of other organisms. The human body carries with it a number of microbes constituting about 90 per cent of the cells in the body, containing some 99 per cent of its genes. Interestingly, the so-called Human Microbiome Project that seek to map the microbiomes of the 'superorganism' anticipates the establishment of microbial observatories worldwide for the purpose of monitoring the 'microbial ecology of humans' (Turnbaugh et al. 2007: 809), for linking microbiomes to the planetary environment, and for facilitating sustainability. Life itself, its molecular structures and biosocial ensembles have entered the grand and seamless 'organism' of the globe, the world of the Gaia (Lovelock 2000). Such developments pose profound challenges for research, medicine, ethics, and all kinds of biopolitics. A recent editorial in *The Economist* concludes that genetics has been preoccupied with the 'wrong' set of genes and while nobody knows where the 'microbiome revolution' will end up it is 'clear that turning thinking inside-out in this way is yielding new insights into seemingly intractable medical problems, and there is good chance cures will follow' (*The Economist* 2012).

However, there is too much uncertainty on the horizon for anybody to be able to meaningfully predict where things are going. One major source of uncertainty relates to what might be referred to as the 'ecological' shift represented by escalating interest in microbiomes and epigenetics. Significantly, a recent US

National Academy of Sciences report, 'Towards Precision Medicine' (The National Research Council 2011), presents the microbiome and the epigenome as fundamental expansions in the mapping of health, as equivalents to the nested layers of the GIS maps of Google. If recent excursions into the 'human' genome have been fixated on the 'wrong' genes and humans are best seen as superorganisms, as biosocial assemblies of biological and social inheritance (to the extent that such a distinction has any value), will personal genomics be relegated to history as a trivial, narrow-minded exercise? What could possibly be 'personal' in mapping of the genomes of trillions of bugs in our guts and exploring their implications for our health – and who would bother to engage in such co-working?

Alternatively, one might envisage, assuming growing public awareness of the role of microorganisms, environments, and nutrition for health and development, that personal genomics will 'simply' become more complicated, casting its nets much wider than before, vastly expanding its range. The latter scenario, it seems, would call for enhanced governance concerns and greater respect for the "prosumers" of personal genomics, the blurred collectivities of producer and consumer (Toffler 1980), emphasizing contexts of community, political economy, and environment.

Conclusion

There is a rapidly growing interest in personal genomics, for the purpose of reconstructing our past and celebrating our emerging biosociality and for managing our lives and our future. Day by day, the companies involved offer additional services on their web sites, further details on diseases and traits, higher resolutions of data, and more powerful machines, diagnostic chips, visual presentations, and interactive features. While the genomic hype has faded a bit and some of the key players, in particular deCODE genetics and 23andMe, have experienced financial difficulties, there may be good grounds, however, for arguing that personal genomics will continue to thrive. It seems unlikely that the narcissistic pleasures and hypochondriac anxieties involved in the exploration of ancestry and the genetics of health risks are withering away, given the central place of the human body in late modernity. Also, there are immense financial stakes and concerns on the global level, for biotechnical and pharmaceutical companies. Moreover, the quality, magnitude, and comprehensiveness of knowledge can only increase with time. The power of computing machinery continues to expand and cheap complete sequencing is within reach. As a result, one may expect personal genomics projects to expand, realigning experts and consumers, institutions and disciplines, including genomic anthropology.

No doubt, personal genomics of the kind discussed here involve an element of empowerment. Some qualifications, however, are needed. Prainsack et al. (2008) argue that while relaxing the genetic protectionism rampant in recent

decades may be a good thing, giving people an opportunity to become active governors of their genomes, the arguments about individual freedom, informed choices, and the unregulated genomic marketplace should be taken with a grain of salt. For one thing, they disguise the fact 'that personal genomics is pushing the individualization of responsibilities one step further' (Prainsack et al. 2008: 34). Anthropology and related fields can play an important role on this front by exploring what such individualisation means and what people expect from genomics, providing 'thick' descriptions (see, for instance, Hinterberger 2012; Nelson 2008; Santos et al. 2009).

Another qualification relating to the agency of the users of genomics services is also essential; this concerns the labour they perform *for* personal-genomics services rather than their opportunity to comment, interpret, and engage in a dialogue on methods and results. It is pertinent, in my view, to broaden the notion of the biosocial in order to highlight biosocial relations of production, the labour processes and hierarchies associated with emergent biocapital (Palsson 2009b). Historically, the discourse on labour, property rights, and 'resource' governance has described the characteristics of the regimes in question in terms of rather simple binary dimensions: stationary vs. mobile, aquatic vs. terrestrial, biological vs. physical, material vs. intellectual. Along with some other body issues, including surrogate motherhood, organ transfer, and biobanking (Dickenson 2009; Gottweis and Peterson 2007), genomic stuff seems to invite new dimensions and considerations. For one thing, with the new genetics, the development of biomedicine, and the expanding production of biocapital (Lock and Nguyen 2010; Palsson 2007), the very notion of the 'biological world' has been destabilised as nature is increasingly subject to artificial, human refashioning (Landecker 2007; Rabinow 1996).

Personal genomics, as we have seen, represents a series of developments in genomics, biomedicine, informatics, and neoliberal economies. Rooted in the personalised medicine characteristic for population biobanks founded during the last decade or so, aiming to produce medicine geared to individual genomes, it is now firmly embedded in network society. In some respect, the focus on labour in this context may be narrow and restrictive, economizing complex and diverse developments. Consumers, after all, seem to get some rewards, in terms of belonging and sociality, indulging in play and recreation. Also, one may argue, there is a crucial difference between reproductive and genomic labourers in that the former, unlike the latter, take serious health risks with considerable emotional and physical involvement.

Consumers of such services, however, I have argued, are not simply passive recipients of goods, they are actively co-working, contributing labour from different virtual sites at a variety of scales and hierarchies with different forms of alienation and exploitation. These biosocial relations need to be mapped in detail in comparative contexts, partly for their own sake to better understand the scene and partly with respect to policy and governance. Following Toffler (1980)

who launched the notion of prosumption, Ritzer and Jurgenson suggest (2010: 17) that social theorists of production and consumption (e.g., Marx and Baudrillard) have too strongly distinguished between these two spheres, suffering from either a productivist or a consumptionist bias. While 'prosumer society', they argue, is nothing new, 'a series of recent social changes, especially those associated with the internet and Web 2.0 (briefly, the user-generated web, e.g., Facebook, YouTube, Twitter), have given it … greater centrality' (Ritzer and Jurgenson suggest (2010: 14)).

There is a vast terrain to explore in personal genomics and related fields, it seems, through the analytical lens of labour. The contribution of the 'consumer' is negated twice, rhetorically and financially, in the fact that the consumer *pays for* contributing and *has no share* in the profits derived from patents claims and other potentially collective products of personal genomics. Recognising the roles and complexities of co-working and the immaterial labour involved, in the communal spirit of solidarity that increasingly characterises bioethical discussions (Prainsack and Buyx 2011), it seems likely that new forms of management and public engagement will be explored for personal genomics. Perhaps in the near future governmental and non-governmental agencies along with private companies and users' organisations will experiment with some forms of DNA cooperatives, moving not only from me medicine to we medicine, to paraphrase Dickenson (2013), but also, to draw upon the fashion of naming in the industry, from LabouringMe to LabouringUs.

Acknowledgements

A draft of this chapter was originally presented to a session entitled 'At your service: Affective and 'immaterial' labour in the global economy' at the Annual Meeting of the American Anthropological Association in New Orleans (17–21 November 2010). I thank the organisers, Akhil Gupta and Purnima Mankekar, the discussant, Anne Allison, and other participants for stimulating comments and discussions. Other drafts were presented to the workshops 'Genetics as culture in a consumerist age' at the University of Innsbruck (27–29 October 2011) and 'Genetics goes online' at Maastricht University (12–14 September 2012), organised by Anna Harris, Sally Wyatt, and Susan Kelly. Barbara Prainsack, Silke Schicktanz, and Gabriele Werner-Felmayer, and Erik Aarden offered useful comments and suggestions. The chapter partly draws upon arguments developed in my article 'Decode me!: Anthropology and personal genomics' published in a supplement edition of *Current Anthropology* (2012, S5), 'The biological anthropology of living populations' edited by Susan Lindee and Ricardo Ventura Santos.

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Chapter 11
Making Responsible Life Plans: Cultural Differences in Lay Attitudes toward Predictive Genetic Testing for Late-Onset Diseases

Aviad E. Raz, Nitzan Rimon-Zarfaty, Julia Inthorn and Silke Schicktanz

Introduction

'Que sera, sera – Whatever will be, will be – The future's not ours to see,' so we are told by the famous oldie sung by Doris Day in, ironically enough, the movie titled 'The Man Who Knew Too Much' (1956). What will life be like when we get old? Predictive genetic testing for late-onset diseases is an attempt to provide some partial answers to that unanswerable question. In cases of colorectal cancer 'running in the family', where an effective treatment exists, predictive genetic testing is recommended by medical experts even for minors (Duncan et al. 2008). However, with some late-onset diseases –such as Huntington's–being untreatable, the availability of predictive testing has re-opened the discussion concerning 'the right not to know' (Erez et al. 2010).

In this chapter we focus on predictive genetic testing of adults for late-onset diseases. This domain of genetic testing is seen by providers as increasing the range of choices open to the healthcare consumer. A typical example is predictive genetic testing for breast and ovarian cancer of healthy, pre-symptomatic individuals (Levy-Lahad and Friedman 2007). Our research highlights the cultural diversity of lay (as opposed to expert) moralities concerning predictive testing, and how these public configurations interact with the top-down biomedical worldview of needs and benefits as well as with the Foucauldian approach to genetic testing as a uniform and unilateral regime of biogovernmentality (Lemke 2002, 2005; Novas and Rose 2000). This research also examines how public, interpersonal and subjective configurations of predictive testing reflect and interact with particular cultural repertoires (Raz and Schicktanz 2009a, 2009b), as well as express forms of resistance, avoidance, and criticism of genetic testing. Lay deliberations of predictive genetic testing, involving discussions of responsibility for the self, self-care, and responsibility for family members, are used to highlight relevant moral grammars (in the Wittgensteinian sense) and their embedding of social norms and individual behaviours. We examine these diverse expressions by looking at arguments developed by lay people in Germany and Israel, examining

the complex ways in which different cultural backgrounds, as well as experiences of being affected, influence the ways in which people make sense of predictive genetic testing in the context of making life plans.

Genetic Testing in Israel and Germany

Within Europe, Germany and Israel generally represent contrasting legal regulations and professional outlooks, particularly in relation to reproductive, prenatal genetic testing and pre-implantation genetic diagnosis (PGD). German genetic counsellors are more cautious regarding the use of prenatal diagnosis for selective abortion, while Israeli geneticists are often in favour of it (Hashiloni-Dolev 2007). While German patient advocacy groups like *Aktion Mensch* are critical of prenatal testing (Hashiloni-Dolev and Raz 2010), many disability activists in Israel support it (Raz 2004). Although the German legal situation has changed in 2011 by allowing, for the first time, pre-implantation genetic diagnosis (PGD) for serious early-onset diseases, its regulation is still very restrictive in relation to Israel, where PGD is allowed for HLA typing (the creation of 'sibling donors') and family balancing under specific conditions (Grazi et al. 2008).

Two Paradigmatic Cases of Predictive Genetic Testing for Late-Onset Diseases: Huntington's Disease (HD) and Colon Cancer (CC)

Huntington's disease (HD) is a dominantly inherited disease for which predictive testing can inform whether, but not precisely when, the disorder will manifest itself in adulthood. Testing for HD is not generally recommended, in part because no preventive or curative treatments are available, and because HD has a relatively low prevalence of five cases in 100,000 inhabitants (Hawkins, Ho and Hayden 2011). In contrast, colon cancer (CC) is a common disease in Western countries, with over 25 cases per 100,000 inhabitants. It is the second common cancer in women and third common cancer in men in industrialised countries. The rate of CC with a genetic basis is estimated between 5–25 per cent, with overweight and malnutrition as contributing external risk factors (Duncan et al. 2008). While HD testing implies high predictability without treatment, CC testing is characterised by increased susceptibility with often successful treatment, including radio- and chemotherapy and surgery or preventive care by healthy nutrition, exercise and early detection by regular checks. In HD, knowing one's genetic status and predicted age of onset can eliminate doubt and assist in making life plans, but the prospect of developing a fatal disease can be far more stressful than the uncertainty (Erez et al. 2010).

This may explain why only a relatively low percentage of those with a family history of HD have opted to be tested – with a conflict whether or not to know and to tell kin (Konrad 2003, 2005; Taylor 2004). Conversely in colon cancer, those with a family history of the condition are often referred to genetic counselling

(Duncan et al. 2008). Testing relatives at risk for CC is recommended in Israel, particularly for preventive considerations (Rosner et al. 2009). Similarly in Germany, there is high approval of testing for CC with at-risk persons holding a more favourable view of the testing (Berth et al. 2002).

Methodology

Our methodology draws on the concept that the empirical social study of attitudes provides descriptive 'facts' that can be understood as normative statements (Haimes 2002; Rehmann-Suter, Porz and Leach-Scully 2012). The convergence of social science and ethics intended by this approach is both of an epistemological and methodological nature (Haimes 2002; Haimes and Williams 2007). This allows integrating those perspectives which are often neglected or marginalised in the dominant expert discourse (Schicktanz 2012).

First, we add to the expert discourse of abstract ethical principles and formal policies the moral arguments of lay people, which are often ambivalent, informal and 'unprincipled' – a morality without theoretical foundation which is nevertheless the morality we 'live by'. Second, we add a methodological focus on social context – in our case lay, affected, religious, and national groups. In addition to providing empirical data for ethical analysis, the sociological analysis of these focus groups enables new questions to be asked, such as 'why are these issues defined as ethical concerns by these people in these times and these places?' (Haimes 2002). Such questions can then be further discussed as indicators of broader concerns and comparative trends within Germany and Israel. The juxtaposition of the two countries is expected to highlight the context of national variation and pluralism, as well as to offer a more fine-tuned examination of group diversity and similarities within the contexts of being affected and of religiosity (Raz and Schicktanz 2009a, 2009b). This methodological design is used to examine how cultural (national and religious) contrasts exist alongside shared positions which might reflect a common sense of being affected by disease-based experiences.

Our focus groups (FGs) were composed in a manner that reflected our interest in understanding personal experience with a genetic disease and with the medical system. In both countries we recruited respondents who were either patients or close relatives of a patient (i.e., 'affected') or had no experience of a particular disease, test, or treatment (i.e., 'non-affected', or 'lay'; see Schicktanz et al. 2008). We also had a FG composed of modern-religious respondents (Christians in Germany and Jews in Israel) in each country, which provided a source of comparison concerning the bioethical arguments of the other (secular) groups as well as to the expert bioethical discourse, where religious arguments often play a major role. We conducted eight FGs (N=60, 17 males (28 per cent), 43 females (72 per cent) with volunteers recruited in Germany and Israel during 2010–2012. In each country, two FGs comprised people affected by genetic diseases (participants had clinical symptoms, or were diagnosed as carriers, of a genetic disease, or

had a first degree relative who is sick with a genetic disease), and two FGs with non-affected lay people – one group of secular lay people and another group of modern-religious respondents. Similar scenarios and questions were used in all FGs. FG meetings in Israel and Germany usually included seven to nine people and lasted about two hours. In terms of education, groups were mixed with a slight tendency to higher level education, probably reflecting self-selection preferences. The participants' age range was 22–80 with a heterogeneous composition in each FG. Respondents affected by genetic diseases were recruited from self-help and support organisations of and for people with genetic diseases. German and Israeli respondents not affected by genetic diseases were recruited by disseminating flyers and ads in urban public places, and in modern-religious associations for recruiting the modern-religious participants.

The FGs included scenarios of testing for HD and CC, with questions about whether to test, when, and whether and whom to tell about test results. The discussions were audio recorded, transcribed, and translated into English. The transcripts from each country were analysed thematically using ATLAS.ti®, and compared cross-nationally in order to uncover discursive themes and categories of themes recurring within and across groups (Bloor et al. 2001). In the CC scenario, participants in Germany and Israel were asked to imagine that one of their parents was diagnosed with CC and that they are in their early 20s and consider taking a genetic test that can be performed at any stage of life, even before the onset of symptoms. Participants were told that the test will tell them whether they have a mutation that correlates with disease onset and which could, under specific environmental conditions (whose characteristics are still unknown) increase their probability to develop CC later in life. It was also explained that at an early stage, CC is often curable. Participants were then asked about their arguments for and against taking this genetic test, and about sharing test results with others.

Participants in Germany and Israel were also presented with a scenario asking them to imagine that one of their parents was diagnosed with HD, a severe neurological disease with degenerative symptoms starting around middle age. They were told that although the disorder itself is not fatal, complications such as pneumonia, heart disease, and physical injury from falls can reduce life expectancy to around twenty years after the onset of symptoms. It was explained that there is no cure for HD, and full-time care is required in the later stages of the disease. Participants were then told that a genetic test can be performed at any stage of life, even before the onset of symptoms, and that the chance for them is 50 per cent to have the gene. If they have it, it will, in virtually all cases, cause this disease to manifest itself in later life. Participants were asked about their arguments for and against taking this genetic test and sharing test-results with others. Quotes presented in the following section illustrate the range of responses with regard to two major themes that emerged from the analysis: self-knowledge and self-responsibility ('to know or not to know') and responsibility for kin ('to tell or not to tell').

To Know Or Not To Know?

A major argument among all the respondents in favour of knowing certain genetic risk factors was the availability of preventive or curative strategies:

> My ambition, I suppose like that of all people, is to live to 120 and live a healthy life as much as possible. Because you can perform a surgery to cure the disease when detected early, I would rather perform the test as early as possible in order to undergo the operation in time, and not discover the disease when it's too late. (Israeli female, lay group)

In Israel, the view in favour of testing in case of potential medical treatment was apparent in the case of CC across all groups. It was expressed by a majority of 66–85 per cent (see Table 11.1). While the majority of the modern-religious respondents were in favour of taking the test when medical treatment is available, they did not relate this attitude explicitly to their religious stance. The largest support of this view – that tests should be taken only when medical treatment is available or prevention is possible – was expressed in the two affected groups in Israel. In these groups, 64 per cent (9/14) of the respondents also said that they will change their lifestyle and increase their medical surveillance as a result of a positive test:

> If I am found to have the gene I will perform the medical tests for early detection of colon cancer, to increase the chances of prevention or cure. I do not think there are any arguments against such a genetic test since if colon cancer is treated following early detection this may improve the effectiveness of prevention. (Israeli male, affected group)

Only one or two Israeli respondents in each group were against taking the test in the context of CC. They also mentioned the right not to know:

> I can still monitor my health without the genetic tests, can't I? This will still allow me to treat the disease in time if needed. (Israeli female, affected group)

This minority view *against* taking the test even in a situation where treatment is available also contained references to the ambivalence resulting from the uncertainty of the expression of CC even for carriers. A few Israeli respondents supported taking the test because 'it contributes to the family,' arguing that responsible parenthood meant that one had to take care of his/her health:

> I think that for your children you have a responsibility, your life is no longer just yours but also your commitment to your children and that's why I think every person, especially if he has children, needs to take the test if he belongs to a risk group. I have no objections to the test and I think that whoever raises these arguments is irresponsible. (Israeli female, affected group)

Table 11.1 Number of respondents per focus group, in Germany and Israel, who supported testing for CC and HD and sharing test results with family members

FGs / Attitudes	German non-affected (n=11)	Israeli non-affected (n=8)	German modern-religious (n=9)	Israeli modern-religious (n=6)	German affected (n=12)	Israeli affected (n=14)	Total German (n=32)	Total Israeli (n=28)
Support testing for CC	4	6	1	4	4	12	9	22
Support testing for HD	2	5	2	1	1	3	5	9
Support telling family members about test results of CC	2	4	1	6	6	11	9	21
Support telling family members about test results of HD	2	3	3	5	3	11	8	19

In Germany, in contrast, only a minority was in support of taking the test, expressed in the context of CC by only 11–33 per cent of the participants. Arguments mentioned by many German respondents as part of the view against taking the test included the problem of probabilistic knowledge that is not helpful, anxiety, and the danger of discrimination: 'I know from experience that if you know something like that, it will be a burden' (German female, lay group).

Another argument against taking the test stressed the existence of risk as a normal part of life that should not be medicalised:

> Well, I would also not take the test. In principle. Because I find that we must not live in a society where everyone can know what he or she gets, or will get. We should live in a society where we allow the existence of risks. Just like crossing the street. (German female, lay group)

The largest support (4/12) of taking the test for CC was expressed in the affected German group:

> Well, I am very much in favour of early diagnosis because I learned much about cancer during my rehabilitation time. For three times, I was surrounded by 300 cancer patients. Then you'll know what early or late detection means. There is a great difference. I was diagnosed early and therefore it was curable. But someone who does not undergo cancer screening for five or even three years, even if he/she is a high risk patient… I find that, shall I say, somewhat negligent. (German female, affected group)

In both Israel and Germany, the HD scenario was approached with much more ambivalence, highlighting the type of information that is regarded as helpful or not in the context of predictive genetic testing:

> In this case [HD], I'll skip the test. If there is no cure, I would not want to know – I'd rather live my life peacefully and without fear. The disease anyway bursts, and it's better not to know about it beforehand. I cannot find arguments for the test. (Israeli female, lay group)

In Israel, the view in support of taking the test for HD was expressed by a very heterogeneous range of 16–62 per cent of the participants, depending on the group. Support was highest in the non-affected Israeli group and lowest in the modern-religious Israeli group (see Table 11.1). Many amongst those who were supporting this view in the non-affected lay groups argued in favour of knowing the result in order to be able to be better prepared in terms of life planning that is being conducted together with other family members:

> This question is very difficult. Yes, I would check. I want to prepare for the future, for my home, for family and financially. I think it's my responsibility as a mother to know in advance and prepare for it, responsibility towards my family. (Israeli female, lay group)

It was common amongst Israeli respondents, but not German respondents, to speak about their motivation for taking the HD test in the context of their children, as one Israeli respondent summarised it:'[Taking the test] is my responsibility as a mother.' Israeli respondents brought up prenatal testing in this context even though the moderator focused only on adult predictive testing. They argued that it is crucial to know about one's 'faulty' genes so as to know which diseases to test for prenatally or avoid having children. The option of adoption was not mentioned by any of the Israeli respondents, and outspoken attitudes supporting selective abortion were not met with criticism, as the following exchange illustrates:

> Of course, if there is likelihood that I pass to my children any illness I would like to know about it, so I can monitor the foetus during pregnancy and have an abortion if necessary … As part of my responsibility as a parent, I am responsible to my child's health and this responsibility extends to the pre-fertilisation stage. (Israeli male, affected group)

Another group member responded to this by saying:

> I'm all in favour of taking the genetic tests, if the disease could pass to the foetus, I think it is right to do it, I would not want to have a child with an illness or disability, who would suffer throughout his life and be mistreated by society. Of course if it is some mild issue that it is possible to live with, there is no real problem and I would not rush to test, especially if testing could endanger me or cause any bodily harm to me. (Israeli female, affected group)

Interestingly, the majority in the affected group was ambivalent or even against taking the test for HD, referring in this context to the right not to know and to anxiety, since there is no treatment. Compared with the CC case, this argument highlighted a view of self-responsibility to one's health/own body:

> Because there is no cure for the disease, in this case [HD] I see no reason to do the test. I will have to live with the knowledge that my parents have the disease and hope that it will not attack me… The body will wait for something which may never take place. If there was a remedy, I would attach great importance to conducting the test, but since there is none – it does not contribute anything to me, and I think that such news could just make me more anxious. (Israeli male, affected group)

A large majority (5/6) of the respondents in the modern-religious group similarly spoke against taking the HD test, mentioning the right not to know:

> I see no reason to check if I'm a carrier, if there is no medicine or vaccination that could reduce the confrontation, the struggle with the disease, or to prevent an outbreak. I have an instinct to say no, this is not something I would turn to religion about – and I also have a feeling that if you know, you would only become more stressed … so it's more relaxing not to think about it. (Israeli female, modern-religious group)

Another argument raised by the modern-religious respondents was stigma, especially in the context of dating:

> Being HIV positive is much more stigmatising. But being a carrier can also stigmatise you, especially if you are in the dating stage and have to talk about this. (Israeli female, modern-religious group)

Another group member responded to this by saying:

> Yes that's true. So we should recommend to young people *not* to test for Huntington's – they can do this much later in life. (Israeli male, modern-religious group)

In Germany, the view in support of taking the test for HD was expressed by only 0–22 per cent of the participants. Arguments given in favour of taking the test included the certainty of the genetic prediction as a potential source of comfort, and an important source of information for planning one's life:

> Well, Huntington's is an extreme case. Since there are no …treatments, it merely concerns knowledge. You have it, or you don't. I am not sure, but I believe that I would undergo a test in this case. Because, first, I know that my parents, or my mother has Huntington's. That means, that in any case the Sword of Damocles is hanging over my head, and I have a 50 per cent chance that it is this or that way. That means, the test is done, and there is a 50 per cent chance that I am well. I can continue living joyfully. And a 50 per cent chance that I have this disease. This then is indeed terrible. But at least I am aware of the situation. And I personally think that it is better for me if I know that I can still live ten or 15 years without a disability. And then, I believe, there are five more years of nerve disorder where one loses motor functions and the like. I think I can plan my life differently for this time if I know that I have actually still 50 years ahead. (German male, lay group)

An additional argument in support of taking the test was raised by a few respondents mentioning that for them the question of whether or not to test would arise only in connection with the wish to have children, as we discuss below in more detail.

A slightly higher proportion of the German respondents were against taking the test for HD. Arguments mentioned against taking the test included unnecessary anxiety:

> The question is whether I need this exact time specification to live a good life. Already if I know that this could probably be, I could try to live the life in my current as good as possible and as happy as possible. I don't have to know: In 15 years it will be my turn. I find that this time specification will drive one crazy. It is enough to know that this disease lies ahead of you. That is difficult enough to cope with. I don't have to assign a date to it. (German female, lay group)

To Tell or Not To Tell?

The formal right to know or not to know, as advocated by some lawyers and ethicists, tends to neglect the social implications of such knowledge. Individual disclosure of genetic status hence often leads to a new dilemma: whether one should convey this knowledge to relevant others. Therefore, it seemed rather natural for the participants to explore the social consequences of such knowledge. Interestingly, they spoke about this as a personal question of morality, or more explicitly of responsibility. However, the concept of responsibility varied according to different underlying norms such as love and care for others, duty and social obligation for those who are dependent, or self-identity as a family member. Even the decision not to tell could be framed as conveying responsibility to protect others from psychological harm.

Among our Israeli participants, considerable support for telling family members about the test results was apparent across all groups and was expressed in the case of CC by a majority of 50–100 per cent of the participants. The largest support of this view was expressed in the modern-religious group, where 100 per cent of the respondents said they would share the test results with close family members, corresponding with the strong family-oriented characteristics of this population (Shalev, Baum and Itzhaky 2012). In the case of HD, Israeli respondents' support was relatively high but also very heterogeneous, expressed by 37–78 per cent of the participants. Support was especially high in the modern-religious group (five out of six participants said they would share test results with close family members). Although almost all of our respondents were against taking the test themselves, they still said that if they were to take it, they would share the results with others:

> In this situation there is no doubt that my children need to know. If you already have children you should tell them so they will have the opportunity to be tested themselves. I think it's a huge burden for a child and I would not tell them before I felt compelled to. When? I do not know, probably around age 17. On the one hand I know I want to save my child from this knowledge, on the other hand I think it's his right to know. (Israeli female, affected group)

Arguments raised in the context of telling others about test results included responsibility for one's spouse and kin (sharing relevant information for life planning):

> I would tell my immediate family. I will definitely share it with my partner who needs to be aware of the risk and the possible impact of this on the continuation of our life together. (Israeli male, lay group)

Telling kin was perceived as important because they need to be medically informed and take the test themselves:

> I would inform my immediate family, if I am found to be a carrier that is; they may be at higher risk now and so they need to decide for themselves if and how to take care of their bodies. They should be made aware that they are at higher risk than the general population. (Israeli male, lay group)

Interestingly, the feeling of responsibility for their children was expressed also by taking the opposite stance: Some felt that they should share the test results only with their spouse and not with their children so as to not worry them too much:

> I would not tell my children so that not to make them worry. I think they have enough worries, I would not want to add. (Israeli female, lay group)

In Germany, only a minority – in all groups – supported the idea of sharing test results of CC with family members. Between 11 and 50 per cent of all respondents held the view that this should be done. A similar proportion of respondents in the lay and modern-religious groups were against telling family members, and the majority either expressed ambivalence or did not voice an opinion regarding this issue. In the case of HD, support of sharing results with family members dropped to between 18 and 33 per cent of the participants:

> Would then not the obligation exist to tell it to the others so they, too, can take this test? For example, to my own children? To suggest to them that they too could suffer from this genetic defect, or something similar. At least, to open up the possibility for them to take, or not to take, the test for themselves. (German male, lay group)

Another respondent countered this view by expressing worries about telling others:

> Oh God! That is definitely 1984 for me. George Orwell. That is totally frightening and I am horrified. I will probably dream of that tonight… But if this would push through, you would take away the person's responsibility to keep this to himself [*sic*] because others might possibly be harmed by it. That is for me a sign that it is very difficult for us to accept uncertainty in life. There are things that simply happen. [In the past], I rode without a helmet. Now everybody rides wearing a helmet, and sometimes people look at me with disapproval if I don't wear one. I also ski without helmet. But there are countries where it is already forbidden to ski without a helmet. This means that I don't have this freedom anymore. And I am afraid that this will keep going on. On and on. (German male, lay group)

German respondents who spoke against telling their family members emphasised the anxiety that such information may cause, and the unwillingness to share what was strongly perceived as personal information. Few German respondents discussed in this context the decision not to have children, or – in case one carried a gene for HD – the decision to adopt children:

> I also decided not to have children. But that has nothing to do with genetic testing. But let alone this: One has to deal with it. Not to have children is an inner effort. […] If I had a fifty-fifty, or let's say an eighty-twenty probability, then I would say: I refrain from having children: Then I'll adopt. I have five stepchildren. Then, I would rather adopt a child. To give the child a chance, instead of – sometimes religions see this somewhat differently – having my own child that would be severely disabled or something like that. I would probably blame myself throughout my entire life. (German male, lay group)

Some German respondents also referred to the potential risk of being discriminated against by employers, friends and society. Once again, relatively the largest support of telling family members was expressed in the affected group:

> Well, I think if it's like that, it's good to be able to share that fear [of the disease] with somebody else. (German female, affected group)

Moreover, some affected German respondents also spoke (in a parallel manner to the Israeli respondents) about the right to know of family members, which made them consider telling as their duty and part of their responsibility towards kin:

> Well, I'd consider it to be my duty. Yes. Well, I've always kept my relatives informed how my disease made its way through the genes of my family. (German male, affected group)

The majority of Israeli respondents did not express worries regarding potential genetic discrimination, and many agreed that the State and HMOs should finance these tests as part of public health. For some, however, support for telling spouses and/or parents was accompanied by reservations concerning not telling children and the rest of the family:

> I would tell only my husband or my mother. I would not tell friends or even close relatives; I suppose they would still accept me as I am … But inside, they surely would treat me differently, feeling sorry for me and taking care of me more … I'm wondering if it's right in this case to tell the rest of the family that this is a terrible disease that accompanies fear for life. (Israeli female, affected group)

Some German respondents who were in favour of telling family members also spoke about responsible sharing of information:

> We are somewhat supported by doctors or whomsoever, but our relatives will then be alone with me and this announcement. (German female, affected group)

Other German respondents mentioned in this context that in addition to telling their family members about their test results, they would also inform them about relevant information centres where counselling is available. Some expressed mixed feelings, agreeing in principle that family members should be told but also sharing how, when they were told by their family members, this was for them a burden at the time because they were very worried about the results of the test they were going to take.

Conclusion

What can these findings tell us about the complex ways in which different cultural backgrounds, as well as the common experience of being affected, influence the ways in which people make sense of predictive genetic testing for their life plans? A major argument in favour of taking the test was the availability of preventive treatments. In Israel, this view was apparent across all groups and was expressed by the majority. In Germany, support of taking the predictive test even when a pre-emptive treatment exists was, in stark contrast, a minority view. This finding appears to reflect a significant cultural difference between lay people in the two countries. The Israeli respondents stressed the benefit of the test as leading to pre-emptive treatment; while the German respondents emphasised that the treatment is available irrespective of the person's knowledge about his/her genetic status, and so the test does not make a difference. A parallel finding reflecting cultural differences was apparent in relation to telling others. In Israel, support for telling family members about test results was expressed across all groups by the majority. In Germany, support of telling family members about the test results was a minority view in all of the groups. In the case of testing for HD, the same picture emerged (as in the context of CC) when comparing Germany and Israel, but with overall lower percentages of support for testing and for telling others, as a result of the lack of medical treatment (see Table 11.1).

The findings demonstrate how lay concepts of responsibility are used as complex vehicles for meaning and values deemed important for the participants in the context of predictive genetic testing. This finding is theoretically remarkable, substantiating theories about how (bio)medicalisation implies a somatic responsibilisation that is understood as a 'regime of the self' (Rose 2007: 134). At the same time, this finding goes beyond this theoretisation in illustrating the need for further differentiation. Importantly, respondents seemed to draw upon different (implicit) understandings of responsibility involving multi-faceted configurations of various types of relationships (responsibility towards self, or others, or society at large), various types of temporal orientation (future-oriented or past-oriented), and various normative frames (rights and duties, for example). The theme of responsibility for oneself (self-responsibility) was mentioned by many Israeli respondents in the context of supporting testing but usually also with reference to one's family, namely that responsible parenthood meant taking care of your health also for the sake of your children. In contrast, German respondents mentioned self-responsibility as strongly connected to the perceived personal beneficence of the predictive test, in terms of the difference that knowing the genetic information can make for the individual. They also pointed out in this context concerns about the medicalisation of risk that should be seen as a natural part of the human condition.

These differences are arguably connected to broader cultural scripts. Self-responsibility, in the Israeli groups, was understood as being primarily the responsibility to stay healthy (for people's own needs as well as in the context

of their commitments to family members). In Germany, self-responsibility also meant to accept risk as part of life, and therefore to organise one's life around these risks – including the social acceptance of diversity and illness. This also had to do with how much certainty and agency participants wanted, and how they integrated predictive testing into this. Israeli respondents prioritised health care, and were anxious about possible illnesses, while German respondents argued that probabilities are not helpful and that society should not be built on certain ideas of health control.

In case they were found to carry a severe genetic disease, the desire to have healthy children led many Israeli respondents to support testing the foetus; whereas German respondents spoke about deciding against having children or otherwise preferring adoption. For participants in both countries, their hypothetical life plans in such a situation was connected with was they perceived as responsibility, but interestingly, with totally different implications. Such dialectical focus on the cultural grammars behind individual and inter-personal concepts of responsibility provides a helpful, although little explored interface, for bridging some of the gaps between experts' formal ethics of principles and our lay moralities, and between theoretical and empirical bioethical analysis (Schicktanz and Raz 2012).

The conceptualisation of responsibility in lay moralities that emerges from this study is not all personal and idiosyncratic but rather framed by broader socio-cultural and ethical narratives. In English and Latin etymology, responsibility denotes an individual emphasis on self-determination: a responsible person is someone who is "answerable," that is, who *responds* to accusations raised in front of a court or in parliament; whereas in Hebrew etymology, responsibility reflects relational support, as in standing behind someone (Schicktanz and Raz 2012). This more nuanced, culture-based understanding of responsibilities can be used to flesh out a concept that so far has remained very abstract, for example in the work of many communitarians, and develop this concept further on an empirical level. For philosophers of ethics like Baylis et al. (2008), concrete expressions of relational solidarity in the context of public health ethics are to be found in our accepting of responsibility for ourselves and our actions; in our willingness to be held accountable for others (especially the weakest and most disadvantaged in society); and in our awareness of mutual vulnerability and interdependence. However, because responsibility is always about our relationships to others, it is neither purely communitarian nor strictly liberal, but rather occupies a third, hybrid space of morality in-between these two opposites.

Our findings highlight and contextualise these different and hybrid meanings of responsibility as embedded in cultural grammars of individualism/collectivism as well as acceptance/rejection of disability and illness as part of biodiversity and social life. In the case of Israeli respondents we see a relational responsibility

blurring the boundaries between the individual and his/her family, reflecting a more traditional, family-oriented, close-knit society that highlights the importance of genetic kinship ties (Birenbaum-Carmeli 2010; Hashiloni-Dolev and Shkedi 2007). In the case of German respondents, we see a self-responsibility based on self-determination, reaffirming the boundaries between the individual and his/her family, reflecting a more liberal-oriented, loose-knit society that highlights the importance of social parenting (Hashiloni-Dolev and Shkedi 2007; Wiesemann 2010).

Being affected, in contrast to the abovementioned cultural grammars, was found to be connected with a line of arguments that in some cases provided more uniformity. In the case of testing for CC the largest support for knowing and for telling (the common view amongst affected as well as non-affected respondents in Israel) was expressed in the German *affected* group, highlighting the role of 'being affected' in producing a phenomenological source of uniformity that transcends national diversity. In Israel, as other studies have shown, 'genetic anxiety' (or 'responsibility,' depending on one's perspective) is constructed by professionals, experts and the public to provide a collective frame of risk in which the 'elective' uptake of genetic testing is exceptionally high and seen by many as moral duty (Remennick 2006). The moral argument regarding the duty to know reflects, in the case of Israel, a confluence of public and professional (medico-legal) worldviews promoting genetic testing as a collective agenda (Zlotogora et al. 2009). The moral argument regarding the right not to know due to anxiety and stress, supported by the German non-affected lay groups (but not by many in the affected groups) may also reflect a romantic tradition of scientific scepticism as well as a dystopian view of medicine and criticism of genetic testing (Hashiloni-Dolev 2007). Overall, this study demonstrates that a multifaceted awareness of the variety of worldviews, including the attitudes of those affected by clinical symptoms or by being a carrier, and of those not affected, warrants attention by sociologists, bioethicists and all those who are interested in a more nuanced understanding of genetics as social practice.

Acknowledgements

The authors would like to thank all the respondents who participated in this study. The study was made possible thanks to the support of the German-Israeli Foundation, GIF Grant No. 1023–317.4/2008, 'Cross-Cultural Ethics of Health and Responsibility: Expert and lay perspectives regarding bioethical dilemmas in Germany and Israel'. We thank Barbara Prainsack and Gabriele Werner-Felmayer for helpful comments on this manuscript.

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Chapter 12
Genetic Responsibility Revisited: Moral and Cultural Implications of Genetic Prediction of Alzheimer's Disease

Silke Schicktanz and Friederike Kogel

At first glance, genetic testing for a late-onset disease such as Alzheimer's Disease (AD) might not pose any new ethical questions as compared to existing genetic testing for diseases such as Huntington disease or breast cancer where their use has been widely discussed in ethical debates.

However, what makes the case of AD so specific is, at least, due to three mutually-reinforcing conditions: First, AD is seen as one of the most common and socially threatening diseases (see Box 12.1); second, AD in particular, and dementia in general, are culturally very differently framed: norms and values vary across cultures, religions, nations, etc., and influence how the social impact of AD is assessed and addressed; and third, the ethical und public debate concerning genetic risk assessment for this fatal disease is still in its infancy.

In this chapter, we focus on genetic testing for AD (see Box 12.2), as it is highly debated in the fields of gerontology and dementia care. Recently, direct-to-consumer genetic services such as 23andMe have started to offer tests for the genetic predisposition for AD (Scott 2012; see also Box 12.2):

> 23andMe first began allowing customers to learn about their genetic risk for late-onset Alzheimer's disease a little more than a year ago. We report on variants of the APOE gene (e2, e3, and e4) and provide a risk estimate for the disease for customers with European ancestry. The 23andMe report also includes detailed background information on Alzheimer's including other known risk factors for the disease, such as obesity and high cholesterol.
>
> Although some are critical of giving people access to this information, many people say that despite their fear of Alzheimer's they would rather know their risk than remain in the dark about the danger. For some knowing their APOE status and Alzheimer's risk may encourage them to engage in activities to stave off the disease, or prompt them to participate in clinical research that could lead to more information about the causes and possible cures for Alzheimer's.

Particularly the latter statement can be read as an imperative to prevention and risk planning underlying the rhetoric of 23andMe: 'Take a more active role in managing your health. Knowing how your genes may impact your health can

help you to plan for the future and personalize your healthcare with your doctor' (23andMe 2013).

As this slogan must be seen in conjunction with an overall trend in Western, industrialised public healthcare of shifting the responsibility to stay healthy or prevent illness to individual citizens, the case of genetic testing for AD is an illustrative case to reveal and understand the ambiguities of such tendencies. On the one hand, there is a social risk of blaming patients for unhealthy lifestyles or of generating sufficient hype for hope and making plans where planning is not possible. On the other hand, the idea of planning one's life in a responsible manner, and of envisioning one's own future, is an important moral resource that ensures good social relationships and can be seen as part of our moral agency.

Box 12.1 Dementia and Alzheimer's Disease

Dementia is an acquired syndrome characterised by a progressive loss of intellectual abilities. Symptoms include inter alia loss of memory, orientation, and language skills and changes in behaviour, personality and judgement. According to the current state of neurological knowledge, Alzheimer's disease (AD) is the most common cause of dementia (Alzheimer's Association 2012), but other also important causes are Parkinson Disease, Frontal Lobe dementia, Lewis Bodies Disease. The most prevalent so called late-onset form of AD mainly affects people at older age (approximate cut-off point at age 65). A small percentage of AD patients suffer from the familiar early-onset form. It affects individuals before the age of 65, many of them in their 40s. What the psychiatrist Alois Alzheimer described for the first time in 1906 (Alzheimer 1907) was retrospectively early-onset AD. The patient died aged 51 after several years of progressive mental decline.

According to the demographic change in the age pyramid we observe a significant increase in prevalence (proportion of people with the disease) of AD worldwide. Alzheimer's Disease International (the worldwide federation of AD associations) estimated that 35.6 million people worldwide lived with dementia in 2010 (Alzheimer's Disease International 2009). This number is expected to double every 20 years (World Health Organisation and Alzheimer's Disease International 2012).

AD related damage to the brain is caused by significant amounts of so called amyloid plaques and the neurofibrillary tangles of the tau protein. Early symptoms include memory decline and depression, while in later stages orientation, language and personality are affected. Patients in the final stage lose the ability to communicate and become bed-bound. At the present there exists no treatment known to stop or to significantly slow down the disease (Alzheimer's Association 2012).

In the following section we provide a theoretical-ethical framework for this latter consideration. We will start with a brief summary of how AD and prediction are framed by a controversy in science and public health of how to deal with ageing societies in the Western, industrialised world. Secondly, we provide a comparative analysis of existing expert reports vs. social-psychological studies on lay attitudes toward genetic tests for AD. Thus, we provide both an ethical interpretation of paternalism vs. self-determination with regard to the underlying assumptions of risks and chances of planning and prevention in the context of AD. Thirdly, we argue for a revised notion of 'responsibility' as a relational-ethical concept. We argue that such a revised notion is a fruitful tool to analyse both ethical as well as social issues. In this way, we comment on the increasing use of terms such as 'responsibilisation' (Rose 2007). In the social sciences, responsibilisation was introduced as a critical paradigm to excoriate the notion that citizens should take up the idea of later-life planning and prevention by changing their lifestyle and habits. The critical undertone of such an analysis is often driven by the concern that such an ascription of preventive responsibility to individuals can lead to people being blamed for bad health, while the societal factors contributing to the situation are being neglected. Another concern related to this critique identifies such an individual responsibilisation as part of a neo-liberal bioethical ideology which tends to frame all ethical questions as issues of individual choices. We share some of these concerns. We suggest, however, a refined conception of 'responsibility' with regard to the philosophical and analytical dimensions to enrich the overall understanding and use of this term.

AD: A National Threat or Just a Myth?

The consensus on the multi-causality, as well as the difficulties inherent in diagnosing and classifying AD (Ballenger 2006), have led some gerontologists and sociologists to question whether AD can be considered a discrete disease entity at all (Whitehouse and George 2008), and whether it has a physiological basis that can be treated. It is controversial whether AD and dementia more generally, are part of the process of physiological ageing, or whether they represent a pathological state. Many people and entities, including several national and international patient organisations do not agree with the 'radical' interpretation that dementia is nothing but senility and normal ageing, and that it has been unduly medicalised. For them, AD is a pathological assembly of serious symptoms even if causes and treatment options remain unclear (Alzheimer's Association 2013).

Box 12.2 Genetics of Alzheimer's Disease

Over the last 20 years, research has revealed a rather complex interaction between the individual genetic make-up and environmental factors. Within genetic research, it is suggested that AD is genetically heterogeneous as it is influenced by a variety of (additional) genetic risk factors (Ertekin-Taner 2010).

Late-onset AD

With regard to late-onset AD, the major genetic candidate associated is the gene for apolipoprotein A (APOE), a cholesterol carrier in blood, which is located on chromosome 19. The APOE ε4 allele enhances the deposition of amyloid β-peptide (Aβ) – the primary component of senile plaques – in brains of AD patients. Variants of the APOE gene still count as the important genetic risk factor, even if recent genome-wide association studies revealed more potential genes involved, all linked to cholesterol metabolism in brain (Ertekin-Taner 2010). Sortilin-related receptor (SORL1) is one of these genes and evidence that SORL1 gene variants are associated with an increased risk for AD was reported (Ertekin-Taner 2010). Still most of these candidate genes require further validation.

In the early 1990s, the ε4 allele for APOE, one of three variations (ε2, ε3, ε4), was found to be associated with increased risk for the disorder (Saunders et al. 1993). This finding has been replicated in various independent studies and across different ethnic backgrounds.

The APOE ε4 allele is linked to a greater susceptibility for developing late-onset AD but it is neither necessary nor sufficient to cause the disease. It correlates with the age of onset: The odds that carriers of one ε4 allele (ε4/ε3) have AD is two to four times greater than that of APOE ε3/ε3 carriers. In carriers of two ε4 allele (ε4/ε4), the risk of developing AD is even estimated to be six- to 30-fold increased (Ertekin-Taner 2010). Another gene variant, the ε2 allele, is associated with decreased risk. However, we only have knowledge of statistical correlations between the APOE genotype and the likelihood of developing AD.

Early-onset AD

For rare (less than one per cent of all AD cases), early-onset AD, three genes, APP, PSEN1 and PSEN2, were identified (Ertekin-Tanner 2010) as involved in the expression of the disease. In contrast to late-onset AD, the familiar early-onset form usually follows a typical autosomal-dominant inheritance pattern. Mutation in three genes, APP, PSEN1 and PSEN2, nearly always cause the disease (Ertekin-Tanner 2010).

Genetic risk assessment plays an important role in AD clinical trials to identify at-risk populations or to test any efficacy of potential treatments for APOE ε4 allele carriers vs. non-carriers (e.g., the MIRAGE study by Yip et al. 2005).

The contested field of AD[1] impressively illustrates how a missing causality-treatment-paradigm for a disease can lead to a paradigm shift in health research. Most recently, major efforts and expansive funding focus on tests for diagnostic and predictive purposes. However, this shift could imply that allocations of costs for care for the elderly are cut back. Recently, much attention has been paid to neuroscientific biomarkers – mainly PET scans of amyloid load[2] (Jagust et al. 2009) – but research on genetic predictors has not yet been completed. Both biomarker-based approaches, whether they are genetically or neuroscientifically oriented, aim to identify asymptomatic individuals with an 'increased risk' for AD. Recent hypotheses suggest that degeneration in the brain may begin years before first clinical symptoms occur. The hope expressed by scientists is to develop methods for diagnosis and treatment to be used before irreversible neurodegeneration occurs.

Recent guidelines published by the largest world-wide patient advocacy organisations, the US-based *Alzheimer's Association* and the *National Institute on Aging* (NIA), define a new stage: *preclinical Alzheimer's disease* describes a stage in which biomarkers are measurable but memory and behaviour are not yet affected (Sperling et al. 2011). As receiving a diagnosis is a turning point or 'status passage' establishing an illness identity (Glaser and Strauss 1971), the introduction of the new classification of AD as a 'preclinical disease' needs in-depth social and ethical analysis. This is particularly important in the case of a disease that remains 'fuzzy' and that is related to significant social stigma and disenfranchisement, and that is associated with a loss of social meaning and social roles (Beard and Fox 2008, for an overview of ageing in general: Cole 1992).

Professionals' Paternalism vs. Lay People's 'Wish to Know': The Risk of Knowing

As recent campaigns of the US *Alzheimer's Association*, 'A world without Alzheimer's' and 'End Alzheimer's' illustrate,[3] the major aim of attempts of genetic risk profiling is to establish future public health interventions and to promote risk-reducing behaviours. However, no validated ways of prevention of AD or modification of the risk to develop this condition currently exist. General preventive guidelines refer to activating brain and physical activity, keeping to a Mediterranean diet, as well as avoiding brain damage (e.g., by avoiding sports

1 In this sense, AD might not be seen as a contested illness (see Barker 2010), as it is accepted and even defended by the biomedical community, but it is contested by others, such as critical gerontologists.

2 PET (position emission tomography) scanning of the brain is a neuroimaging technique that produces three-dimensional pictures of functional brain structures. Measuring the amount of the AD related protein 'amyloid' (see Box 12.1) in particular brain areas may indicate in pre-symptomatic stages whether a patient will likely develop AD in later life. However, also healthy persons can show a load of amyloid.

3 See: www.alz.org.

such as boxing or rugby). These measures help to protect not only against AD, but also against many other conditions, including cardiovascular disease or type-2 diabetes. Any kind of preclinical pre-symptomatic AD test research operates in a field where one can at least fear that the gap between diagnosis and therapy is growing rather than decreasing.[4]

In the following we summarise the position of the professionals saying that the current lack of clinical utility of genetic predisposition testing for AD justifies the discouragement of genetic risk assessment. The argument is based on two assumptions: First, that in the absence of clinical utility, tests can have no other utility (see below). Second, that genetic testing against this backdrop potentially causes harm, and bears the risk of stigmatisation and discrimination. These risks outweigh the interest of individuals to know their genetic predisposition status. However, whether genetic risk assessment will very likely harm individuals must be critically examined and then this potential harm has to be weighed against the wish and right to know.

Professionals' Attitudes toward APOE Testing

When the link between the APOE ε4 allele and AD was established in the early 1990s, the scientific community, particularly in the US, deliberated on the value of these new findings and their implications. Different scientific Consensus Conferences[5] were held between 1995–1999 to determine whether these findings supported the use of genetic APOE testing for diagnosis or prediction of AD (Alzheimer's Association and National Institute on Aging 1998; American College of Medical Genetics and American Society of Human Genetics 1995; Brodaty et al. 1995; McConnell et al. 1999; Post et al. 1997; Relkin et al. 1996a, 1996b). All recommendations contain the conclusion not to test for APOE. Genetic testing for AD was neither to be performed in routine clinical diagnosis nor as a predictive test. Accordingly, other western experts, including the German expert community, relied on guidelines established in the US (Müller et al. 2004). Major arguments against genetic testing for AD in clinical diagnosis concern the limited sensitivity and specificity[6] of APOE testing, since the APOE status alone cannot provide

4 This is generally a problem of biomarker research. For further details see Chapter 1 of this book.

5 Here, consensus conferences refer to conferences of scientific experts held to discuss key questions in certain disease areas in order to reach a consensus over scientific controversies. It aims to provide clinical recommendations, but also to establish new epistemologies (see Solomon 2007).

6 Sensitivity and Specificity are binary classification measures to assess test results. Sensitivity or recall rate is the proportion of true positives. Specificity is the probability of correctly determining the absence of a condition.

certainty about the presence or absence of AD.[7] Moreover, genetic risk assessment in asymptomatic individuals or predictive testing is not encouraged due to the lack of treatment and prevention. Scientists and clinicians, however, admit that APOE testing used in association with other diagnostic tests can add confidence to the clinical diagnosis and, hence, could be used to confirm the diagnosis of AD in symptomatic patients (Mayeux et al. 1998).

The position in the existing guidelines against genetic testing for AD does not only rely on statistical or medical limitations but prefers ethical statements. The disclosure of a genetic risk for AD might have adverse effects on the psychosocial status of the person, as well as on his/her relatives (e.g., the 'possible disruption of a family relationship' due to genetic information, McConnell et al. 1999). Other concerns regard potential negative effects on insurability (Relkin et al. 1996a). Several consensus statements emphasise that the possible gain in confidence of diagnosis has to be weighed against ethical concerns (e.g., McConnell et al. 1999; Van Gool 1996). Overall, this position can be classified as an expression of a paternalistic position to protect patients from social and psychological harm. As a member of a consensus conference later pointed out, this willingness to protect patients from harm might have been the underlying reason which led experts to position themselves against testing (Quaid 1998: 126):

> This tendency to interpret genetic risk factors as deterministic underlies the concern of counselors that individuals will make life decisions based on information that the counsellors believe is not truly relevant. … Is this paternalism? I suppose it is, but I feel that it is justified paternalism. Widespread genetic screening for susceptibility to disease is a new enterprise, and the issue of how to respect individuals' autonomy while fulfilling one's professional role in helping clients identify what is in their overall best interest still needs to be worked out.

It is notable that in previously presented recommendations, socio-empirical evidence is missing for such ethical concerns. A decade later, several studies tried to explore attitudes and experiences of patients with the APOE genetic test and critically revise earlier assumptions. The study by Green et al. (2009) suggests that people receiving risk information for AD generally do not experience significant distress (see below). Based upon these new insights, a paradigm shift in experts' positions occurs. Most recent guidelines now relate to the affected person's right of choice. While genetic susceptibility testing in general is still not supported by most health care professionals, discursive space is opened and the patients' right to make their own choice and to be tested if they wish to – even if healthcare professionals recommend the contrary – is addressed (Goldman et al. 2011; Howe 2010). Recent guidelines of the American College of Medical Genetics argue:

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7 Notably this is an interesting distinction to professionals' attitude against Huntington disease genetic testing, where high test validity is rarely used as argument for testing.

> Despite its limited utility, patients express concern over their risk and, in some instances, request testing. Furthermore, research has demonstrated that testing individuals for apolipoprotein E can be valuable and safe in certain contexts. … If a patient wishes to pursue testing despite genetic counseling and recommendations to the contrary, testing may be considered at the clinician's discretion. (Goldman et al. 2011: 597)

Socio-empirical Studies of Affected and Non-affected Persons' Attitudes

The market-oriented practice of direct-to-consumer genetics originally lets us assume that there might even be demand by lay persons.[8] So far, recent surveys and qualitative studies can help to assess empirical assumptions underlying ethical concerns expressed by professionals, but also might refer to previously undetected areas demanding ethical consideration.

Most existing data are based on quantitative survey approaches conducted in the North American context. Early studies relied on hypothetical test scenarios that assessed interest in genetic risk assessment (Cutler and Hodgson 2003; Frost et al. 2001) and the willingness to pay for such testing (Neumann et al. 2001). To date, only few studies have been conducted in European countries (Chors and Meins 2000; Illes et al. 2006; Welkenhuysen et al. 1997). Most recent studies rely on the context of a 'real–life' clinical trial where APOE testing was offered to family members of AD patients. The US REVEAL (Risk Evaluation and Education for Alzheimer's disease) study can be considered the main source of empirical data addressing social and ethical issues. This study is an ongoing multicentre, randomised, controlled trial examining the social impact of genetic risk assessment for AD (see e.g., Green et al. 2009). Over 40 publications are related to the REVEAL study – most of them using quantitative findings from 'real-life' scenarios. There also is qualitative data which enriches the quantitative REVEAL findings concerning participants' background, experiences and motivation (Hurely et al. 2005; Lock et al. 2007). In the following section we summarise the major findings based on three major questions: Is there any public interest for knowing one's own risk for AD, and if so, what are the motivations? What are the observed social-psychological and behavioural changes in those who underwent testing for the APOE ε4 carrier status?

A major research aim of the REVEAL study was to explore the interest in testing of potentially affected groups in North America (Roberts et al. 2003). Adult children of AD patients were approached, and – if they agreed to participate in the study – were referred to a genetic counselling session. During this session, they were informed about the uncertainty of the genetic information and the lack of prevention options. Despite these caveats, over 80 per cent of the participants retained interest in genetic risk assessment. Levels of interest, the knowledge about testing, and attitudes, however, differed according to the ethnic groups involved

8 In late capitalism, it is important to mention and reflect upon the issue of market-driven demand instead of assuming that markets are always driven by existing demands.

(Akinleye et al. 2011; Hipps et al. 2003). For example, African-Americans were less interested than Caucasian-Americans and anticipated fewer negative consequences from a positive test result (Hipps et al. 2003). These findings were confirmed by Akinleye et al. (2011) but were explained on the basis of knowledge about and fear of the disease: African-Americans were less knowledgeable regarding genetics and less concerned about developing AD. Whether these findings are really specific for AD can be questioned as there were also similar reports for other genetic susceptibility tests: Compared to Caucasian-American women, African-American women with a family member with breast cancer were less likely to pursue genetic testing for BRCA1/2 mutations indicating a higher risk of breast and ovarian cancer (Armstrong et al. 2005).

Not only knowledge of genetics may explain these interesting differences in attitudes, but also the broader context of healthcare should betaken into account as a factor. According to continuous evidence, ethnic and class disparities exist in most areas of healthcare in sectors of American life (Institute of Medicine 2002). Hence, other ethnic groups seem to use medical services at lower rates than Caucasian-Americans (Ashton 2003). Possible relevant factors for that are not only the limited access to public education, including health education, but also little access and contact to any kind of healthcare and by this, a less medicalised attitude to health and ageing.

Other studies suggest that there are considerable cultural or even national differences in the interest of pursuing genetic risk assessment for AD. In this context, the Harvard School of Public Health and Alzheimer Europe (the worldwide federation of *AD* associations) conducted an important study in 2011 in five countries. Surveys were conducted in the US, Germany, France, Spain and Poland to assess the willingness to seek early diagnosis for AD among nationally representative samples with participants aged 18 and older (Alzheimer Europe 2011). Overall, the study found significant public interest in a pre-symptomatic testing for AD. When confronted with the question whether they would want a test which could tell them if they will get AD in a phase where symptoms of the disease have not yet appeared, half or more of the people in every country answered that they were 'likely' (very likely or somewhat likely) to undertake such a test. To be more precise, in France 65 per cent, in Germany 51 per cent, in Poland 78 per cent, in Spain 71 per cent, and in the US 65 per cent gave an affirmative answer. This general interest in testing was, however, likely triggered by the fact that the majority of interviewees in all five countries believed treatment was currently available, or would be in five years. Genetic counselling and critical information can reduce such interest in testing, but supposedly cannot eliminate it entirely: in a representative German sample, 38 per cent were still interested in susceptibility testing after genetic counselling, while prior to this approximately half of them (47 per cent) were interested in genetic risk assessment for AD (Illes et al. 2006).

Overall, these first results indicate that it is important to critically reflect on the underlying popularisation of genetic explanations of diseases as such, and a particular disease in the respective public, if we want to gain a better understanding of the attitudes of potentially affected persons and lay people.

Akinleye et al. (2011) speculated that African-Americans may be less fearful of suffering from AD because they have 'greater access to extensive support networks' (e.g., family, church). One important reason for seeking genetic risk assessment for AD might be the consideration of the need for future care and financial planning (see below). Hence, existing social networks as well as a lack of financial planning can both be seen as plausible reasons. They point toward relevant, but different cultural and socio-economic frameworks that have to be considered from an ethical standpoint.

Participants in the REVEAL study were also asked about their motivations and reasons for pursuing genetic risk assessment. The survey indicated that the need to prepare the family for the illness was the most important predictor of actual pursuit of testing (Roberts et al. 2003). Qualitative data confirmed that the wish to plan for the future was one important reason for people to participate in the study and to learn about their APOE status. The wish to plan and prepare the family was important to the adult children of parents with AD who participated in this study (Hurley et al. 2006: 379), as the following examples illustrate:

> Roberta: When they wanted to know if I wanted to do the program, I go, sure, I want to see where I'm at. Because I can make some decisions in my life that I could take care of everything before and not to have everybody else stress about it. I figured I needed to know because what if I get it? Who's going to take care of me?

> Olaf: I think probably for the future of my family and my kids because I know I've had to play a big part in my mother's care, so I'd like to be prepared and really have all the ducks in a row and know what is going to happen. That's why I was interested.

Particularly women seemed to be interested in the idea of arranging long-term care insurance and of arranging personal affairs (Roberts et al. 2003). This is backed by the *Shriver Report*, based on a poll and 502 phone interviews conducted in 2010 by the Alzheimer's Association with members of the organisation who are often caregivers: 39 per cent mentioned that they had no choice but to become a caregiver and 36 per cent expect that their spouse and children would take care of them if they were to develop AD (Shriver and Alzheimer's Association 2010).

According to the REVEAL study people receiving genetic risk information for AD will not experience adverse effects in terms of anxiety, depression or test-related distress even if they test positive for the APOE ε4 allele. This study tested the impact after 6 weeks, 6 months, and 1 year after disclosure (Green et al. 2009). Qualitative data from interviews with participants in the REVEAL study explained this by readjusting the importance of genetic information. Genetic factors were regarded as just one of several possible causes of AD and family history was regarded as more important for risk assessment than genetic testing as such. As the participants have already had their family history 'observed', it might not be surprising that the individuals showed few if any subjective changes in their social interactions or emotional status (Lock et al. 2007).

Interestingly, genetic risk information altered people's behaviour in some relevant respects. US participants who learned that they are APOE ε4 allele-carriers were more likely to purchase long-term care insurance (Taylor et al. 2010; Zick et al. 2005). This is particularly relevant in healthcare systems, such as in the US, where public healthcare and Medicare only cover parts of healthcare; it often remains expensive and hardly affordable for many lower income patients.

Despite the explicit information on lack of prevention options, people who learned about carrying an APOE ε4 allele changed their diet or used dietary supplements (Veranelli et al. 2010).

Implicit Lay People's Moral Responsibility toward Ageing

In sum, whether the liberal legal right to know always merges with the wish to know is questionable in many cases, but we observe a considerable public interest in genetic predictive assessment, which reflects the reasonable wish to plan one's later life. It corresponds with an increasing awareness of ageing and the willingness to take responsibility for adjustments in older age (Schweda and Schicktanz 2012). Such responsibility sometimes might be connected with a naive lay interpretation of genetics, but sometimes rather motivated by serious concern about oneself and for those we love. This concern is reasonable and pragmatic when considering plans for additional healthcare insurance or advance care directives. This wish for planning later life is – in accordance with our hypothesis motivated by moral underpinnings of responsibility – directed less toward the state, but more toward one's family and oneself. It is based upon the idea that one can be the co-author of one's life story by taking responsibility ahead of time for future events. However, it is important to note that not only genetic information but also biographical experience (e.g., being a caretaker for parents with dementia) and genealogy (such as family stories) are important. Both influence the imagination of future scenarios, such as the social consequences of suffering from dementia. Hence, the genetics of AD must be embedded in a broader framework of culturally negotiated images of family relationships, images of ageing, and 'memory' loss as a dramatic process of self-loss.

We do not reject the possibility that scientific knowledge and health policies frame these images. We are mainly interested, however, in how lay people cope with this and develop their own moral framework to deal with the ambivalence of late-modern society, influenced by such powerful scientific images. Hence, we ask what kind of 'responsibilisation' is ethically reasonable in such a situation. This question arises from the assumption that people have at least generally a right to plan their life. It should be taken into account, however, that it is difficult to stick to these plans, so this may be an illusionary or ineffective endeavour. Moreover, we have to reflect upon the appropriate means and necessary structures to make such prospective responsibility meaningful and feasible for those who are willing to take responsibility**.**

Theorising Emerging Responsibilities in the Context of AD Prediction and Ageing

In the last decade, the legal-ethical expert discourse brought forward the 'right not to know' as well as the potentiality of psychosocial harm when discussing genetic testing of late-onset diseases (Andorno 2004). The 'right not to know', as well as its counterpart 'the right to know', are both expressions of the liberal protection of self-determination. They originate from a practical orientation in bioethics to serve and advise legal and political discourses. However, while both are important, we should ask whether the narrow focus only on rights (and hence, on corresponding duties, e.g., by the state or medical professionals) is ethically satisfactory. In our opinion, this limitation could result in bioethics becoming restricted to a narrow-gauged quasi-jurisprudence. To prevent this development, more complex bioethical approaches are needed to ensure the integration of social-relational issues with individual understandings of a 'good life'.[9] Especially in the field of ageing and ageing medicine, bioethical approaches regard individual perspectives as shaped by social images and vice versa. The idea of ageing as 'gained years' (Imhof 1981) has led to complex and trend-setting demographic scenarios. Many of us hope to lead an active life up to the age of 100, or worry about spending long years in nursing homes or in social isolation. Particularly the latter is a prominent image of the lives of people with AD and dementia. These images are not only fed by political rhetoric but are intensively reiterated in movies, literature, and popular culture.

Different Notions of Responsibility in Bioethics

A theoretically enriched concept such as the one of responsibility provides us with the opportunity to address issues related to genetic testing for AD or dementia that cannot sufficiently be captured with the right to know, or the right not to know. The concept of responsibility has undergone its own bioethical evolution (see Schicktanz and Schweda 2012: 137). Its more recent history started in the 1960s–70s with the notion of collective forward-oriented responsibility, often directed toward future generations, as discussed, for example, by Hans Jonas. In the late 1970s, however, an intensified discussion of professional responsibility arose and was accompanied by the idea to strengthen patients' autonomy. A third stage, starting in the 1990s, focused on the interrelation between social and individual responsibility, often by emphasizing retrospective accusations and the role of causality. The various stages also illustrate different foci on different agents: the human species, the individual

9 This is particularly important because bioethical issues are a moral-epistemic-anthropological hybrid (Schicktanz 2009) in itself. This phrase signifies that the way we describe a 'bioethical' problem does not only contain moral conflicts, but is also already embedded in various anthropological assumptions of how to separate private vs. public, social vs. individual, human vs. non-human, etc. and depends on epistemic considerations of what kind of knowledge is certain, accessible, or objectively true. In other words, a bioethical problem needs to be critically reflected in its own right.

doctor, or the individual patient. A crucial shortcoming of each period can be seen in the problematic reductionist focus on only one or two agents. Instead, we need to consider a web of responsibilities.

In the context of genetics specifically, 'responsibilisation' should not be equated with 'genetic responsibility'. The latter was introduced by Lipkin and Rowley (1974), who argued for positive reproductive eugenics as a form of collective responsibility. What is discussed today as the phenomenon of ageing societies and its threat to future generations was then feared as the threat of overpopulation and ecological disasters. While previously used as a proactive term to mobilise the public, the meaning of responsibility has been transformed into a general term of critical social science. This critical notion implicitly condemns healthcare policies when they lead to the internalisation of individual feelings of guilt or self-restriction with regard to one's own reproductive and preventive health decisions (Lemke 2006). The corresponding biopolitical strategy holds individuals accountable and responsible for their health. The broader picture of this development includes cuts in social welfare and public health provisions since the 1980s. For the political philosopher Iris Marion Young (2011) the emergence of individual responsibility within such a particular framework and this, in this context has an absolving function; it pins responsibility on one agent and absolves others; here mainly the state and government.

Reducing the paradigm of responsibility resting on a concept that assigns guilt to individuals is problematic in two major respects. First, it is indeed important to assume that individual citizens are actually 'responsible' for ageing-related diseases, as it reveals a highly reductionist and misleading understanding of responsibility *and* the complex process of ageing. This is due to the fact that such a reductionist account of responsibility relies on assumptions of causality (X is responsible for Y because X determines the result of Y) which cannot be applied to a multi-causal process which is not even well understood. In a more action-oriented understanding where responsibility is used as a moral-legal term to determine guilt, there must be evidence of the assumption that person X acted deliberately and thus caused consequence Y. In the absence of such evidence, any social practice which leads to blaming individuals for their illness must be critically rejected. The problematic one-sided use of 'responsibility', however, does not justify its total elimination from our moral set of norms. Instead, we can offset this problem by developing alternative models of responsibility. In order to do this, we develop a broader concept of 'genetic responsibility'. Such an approach favours a web of different forms of responsibilities in which different moral subjects and different moral standards are linked to each other.

A Meta-ethical Relation Concept of Responsibility

As we have previously argued (for a broader overview see Schicktanz and Schweda 2012), responsibility is not only a valuable but an indispensable bioethical concept. Essentially, one should recognise its complexity and diversity as a basic (meta-

ethical) concept with various philosophical, legal and social meanings. A general formula underlying all notions of responsibility that can de- and prescribe human actions embedded within social relations and social structures could be: *Someone (a subject) is in a particular framework and retrospectively or prospectively (temporal direction) responsible for something/someone (object) against someone (norm-proofing instance) on the basis of certain standards (norms) with certain consequences (sanctions or rewards).*

The seven-relata formula of this general concept covers the most important normative aspects of different (more applied) models of responsibility in the bioethical context (in particular contexts more relata can be useful).

Understanding the Implicit Notions of Responsibility in the Field of AD Genetics

Applying this heuristic concept to one particular interpretation of genetic responsibility for AD illuminates its reductionist and problematic implications: *The individual is responsible for developing AD (or respectively his or her health in old age) on the basis of the norm of voluntary decisions with regard to his or her lifestyle (self-determination) towards society – and if this fails, he or she is blamed by society, or social healthcare is denied.*

As effective treatment is not available and a correlation between concrete lifestyle changes and the prevention of the disease has yet to be proven, this ascription of responsibility is indeed unjustified, and even illogical, given that the causal factors of AD are largely unknown (Yoder 2002). However, rather than dismissing the notion of individual responsibility in general, we argue that the fact that some people may feel 'responsible' and therefore want to know their risk for AD should be understood as a practical dimension of care, and as an expression of compassion for family members. It signifies a future-oriented mode of responsibility.

A simple general critique of 'responsibilisation' disregards these motives. Within a family context, for example, it seems reasonable and morally acceptable if a daughter wants to assume the responsibility of caring for her father with AD based on her social relationship with her father in terms of the norm of care and love. If she does this, the reward is likely to be (or should at least be) social recognition and gratitude from her father or other family members.

This notion of responsibility elaborates the link between moral motivations, social-family context, and expected consequences. As the philosopher Iris Marion Young (2011) pointed out, this *prospective* meaning of responsibility motivates moral behaviour and social improvement beyond a language of punishment, duty, and blame. However, it is important to stress that the argumentation outlined does not impose a duty to provide care under all conditions. Not all family relations are harmonious, symmetric and intact. Hence, if these conditions and motivations are not fulfilled, no such responsibility can be presumed. As mentioned previously, women in particular tend to be responsible care workers for elderly family members. This social practice (often involuntary) should not be equated with a moral notion based on the voluntary desire to provide care.

Overall, these theoretical considerations of the ethical value of a concept of responsibility can be supported by the empirical findings described above. Affected as well as not yet affected persons see an important value of knowing one's risks, particularly regarding the possibility of 'planning' later life. This planning focuses mainly on long-term healthcare planning, including health insurance and the composition of advance directives for the case that patients are not able to express their own will. This motivation and behaviour can be interpreted as an act of prospective responsibility. It is likely that this anticipation of future scenarios – how will it be to live with a late-onset disease, what kind of challenges will it involve for us and for those we love – is influenced by in part personal and in part social experiences of ageing. When we think of what ageing means to us, one of the first images that comes to mind is that of our grandparents or parents ageing. Hence, in contrast to many other bioethical dilemmas which are only abstract and socially distant, as they are ethical dilemmas only to others, bioethical dilemmas related to ageing easily become part of our own social experience. They may possibly become apparent to us in different roles (as a child, relative, or partner) and from different perspectives (as a patient or caregiver). The cultural practice of interpreting genetics should be contextualised within much deeper rooted practices of interpreting genealogies (Weigel 2002). Genealogies not only encompass knowledge of family relations, but also assumptions of what is 'typical' for a particular family. Especially when concrete genetic factors are unknown, or genetic knowledge is vague, genealogies can serve as heuristics for the imagination of trajectories of the later life course. Thus, assumed genetic factors in ageing-related diseases can be linked to our biographical and genealogical experiences. The case of AD might even be more illustrative as a form of severe forgetfulness which easily activates such images, as many families share various experiences of senility and social problems emerging from everyday situations of forgetting. Such situations can result in emotional conflicts and can lead to accusations of carelessness or even deliberate ignoring. Hence, the anticipation of dementia is full of social images and practical experiences for each of us; this gives us the feeling of being lay experts.

The wish to know one's fate in ageing does not only relate to family responsibilities but also strongly depends on socio-political frameworks such as healthcare provision, reliable insurance practices and legal regulation of proxy appointment and advance directives. Obviously, any reasonable actions to plan also entail socio-political responsibilities. Only if the state and legal framework provide a reliable, stable and fair system, individual planning can take place reasonably.

Conclusion

AD is a serious, multi-faceted very late-onset disease for which genetic susceptibility tests are seen as controversial in their efficacy. However, recent social-empirical findings indicate that some affected persons still wish to know. These developments necessitate an accompanying critical and broad debate

to ensure that neither individuals nor families overestimate the conditions and opportunities of dealing with this knowledge. However, the wish to plan later life, especially by composing advance directives, appointing a family member as a proxy or taking out additional health insurances might be particularly relevant for a disease that later threatens the capacity of complex decision-making and communication – at least in its advanced phases of development. Whether genetic susceptibility tests can be seen as adequate and appropriate tools, should be thoughtfully considered. Other developments in neuroimaging, even if not yet sufficiently validated, could prove to be more promising for the purpose of pre-symptomatic testing. Independent of whether neuroimaging is technically more promising, the social and ethical debate will remain the same: Do I want to know, and what happens if I do not want to know? So far, genetics as a social practice can serve as a paradigm for the reflection of other techniques of prediction.

However, critical attempts are necessary so that the wish to know does not become a duty to know. Blaming individuals for their responsibility in a case where neither treatment nor prevention is available should be rejected as ethically inappropriate. Social and political responsibility have to be spelled out in more detail to provide sufficient healthcare and regulative governance and to support affected persons and family members who are willing to take prospective responsibility for their care.

Acknowledgements

Silke Schicktanz's research on the ethics of prediction of AD was partly funded by the Alexander von Humboldt Foundation. She would like to thank Robert Knight, Berkeley, for his support. Friederike Kogel has received a short-term dissertation grant from the University Medical Center Goettingen. We like to thank Julia Perry for her help with language editing, and Barbara Prainsack and Gabriele Werner-Felmayer for helpful comments on earlier versions of this manuscript.

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Index

Note: Page numbers in **bold type** indicate illustrations.

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