

**UK Users' and Genetics Clinicians' Experiences
of Direct-to-Consumer Genetic Testing**

Teresa M. D. Finlay


School of Social Sciences
Cardiff University

Doctor of Philosophy

June 2015


Declaration

This work has not been submitted in substance for any other degree or award at this or any other university or place of learning, nor is being submitted concurrently in candidature for any degree or other award.

Signed  (candidate) Date 19th January 2016


STATEMENT 1

This thesis is being submitted in partial fulfilment of the requirements for the degree of PhD.

Signed  (candidate) Date 19th January 2016


STATEMENT 2

This thesis is the result of my own independent work/investigation, except where otherwise stated. Other sources are acknowledged by explicit references. The views expressed are my own.

Signed  (candidate) Date 19th January 2016

STATEMENT 3

I hereby give consent for my thesis, if accepted, to be available for photocopying and for inter-library loan, and for the title and summary to be made available to outside organisations.

Signed  (candidate) Date 19th January 2016

Acknowledgements

This project would not have come to fruition without the help and generosity of many people to whom I extend my heartfelt thanks. My decision to embrace doctoral study at a relatively late stage in my varied career has provoked surprise from some but interest and enthusiasm from many more. I would specifically like to mention the following people because they have directly influenced my production of this study. First and most important are the study's participants who volunteered their time and shared their interests in genetics so freely. Their insights were fascinating and it was a privilege to spend time talking with them. Second, the ESRC made the project feasible financially and for that I will be eternally grateful. It has resulted in significant knowledge generation and an invaluable and revealing learning experience for me.

Dr. Karen Melham, Professor Jane Kaye, Dr. Ann Ewens, Dr. Helen Walthall and Dr. Harriet Teare collectively helped me get started on the study and kept me going through it.

My supervisors Professor Adam Hedgecoe and Dr Michael Arribas-Ayllon have been a constant source of balanced support and challenge throughout. My special thanks to them for the intellectual challenges and personal help they provided and for their wisdom in both the subject area and accomplishing a PhD. Their genuine encouragement of my endeavours in a new discipline and their generosity in sharing their knowledge and expertise so enthusiastically are deeply appreciated.

Thanks to Professor Angus Clarke for his support of the project, for helping with clinician recruitment and NHS R&D approval along with Professor Dhavendra Kumar. Thanks also to Dr. Anna Harris and Professor Christoph Rehmann-Sutter for inviting my involvement in academic networks relevant to my work. I learnt so much from colleagues at workshops in Maastricht and Lübeck and am immensely grateful to have had those enriching experiences.

My fellow occupants of Room 101, Dr. Rebecca Dimond and Heather Strange have become firm friends as we have shared our work and experiences of PhD study. Thanks to them for their encouragement as I made the transition from health care to sociology. CESAGen(E) researchers, particularly Mel Evans, have always been welcoming despite my very intermittent appearances. Liz Renton, Corinda Perkins and especially Vicky Parkin provided consummate professional guidance through the administrative requirements of applying for and completing the Sosci doctoral programme. Anna Jones generously provided authentic Welsh hospitality, local knowledge and indispensable cwtches.

Thanks to family and friends for their patience and emotional support but particularly to my brother Robert whose persistent exhortations to "crack on" have kept me going. I could not have done any of this without my husband Bob whose belief in me has been life changing and fundamental to my recent achievements.

Finally, my thanks to you, the reader. I sincerely hope that you find this work as interesting and thought provoking as I have. Any mistakes or obscurity in the work are my responsibility and I welcome feedback.

Abstract

In the last decade personal genomics has been available to the public by direct-to-consumer marketing and sales. Different tests are available including single nucleotide polymorphism (SNP) genotyping. SNP genotyping measures variation in nucleotides at specific points in deoxyribose nucleic acid (DNA) and can be analysed for information about ancestry, physical traits, risk of susceptibility to common complex diseases, genetic disorder carrier status and drug metabolism. SNPs have been analysed in human populations to associate variation with particular traits and common complex diseases, though the association data for disease risk is known to be unreliable. Some claim that direct-to-consumer genetic testing embodies a positive shift from medical hegemony to a market-oriented system while others are apprehensive about the lack of involvement of medical professionals and purchasers' lack of understanding of probabilistic genetic information. These different views represent the dispute over SNP genotyping sold directly to the public that this study focuses on.

My thesis explores this emerging technology using the Social Construction of Technology to investigate the experiences of a group of early adopters of the technology in the UK. I contrast their experiences with those of a group of UK genetic clinicians and examine participants' respective understandings of SNP genotyping and its possible implications for the NHS.

Whilst the data largely mirror the extant literature, they give an insight into the importance of social factors in influencing decision-making in relation to adopting or rejecting new technology. I discuss the data's common themes of knowledge of genomics, the importance of social networks in understanding and engaging with new technologies, and personal versus collective medicine that characterise both groups' experiences. To conclude I explore these themes in relation to the concept of biosociality.

Contents

Declaration	ii
Acknowledgements	iii
Abstract	iv
Contents	v
List of Figures	viii
List of Tables	viii
Abbreviations.....	ix
Chapter 1 : Introduction	1
The German geneticists	1
The problem of DTCGT	4
The project	8
Research question, aims and objectives	10
The thesis structure	11
Chapter 2 : The history of direct-to-consumer genetic testing	14
Conditions of possibility for DTCGT	14
Technology	15
Ideology	22
Moral order	40
Regulation	42
Conclusion.....	49
Chapter 3 : Theoretical and methodological approaches to the	
sociological study of genetics.....	51
Researching genetic testing	51
The Social Construction of Technology.....	53
Symmetry.....	58
Interpretative flexibility	59
Expectations	60
Reflexivity	61
Ethnography and interviews	63
Interviews as accounts and conversations	67
Producing the study: notes on methods	68
Approvals	69
Recruiting participants	69

Users	71
Clinicians	79
Collecting data.....	85
Interviews.....	88
Analysing the data.....	89
Conclusion.....	92
Chapter 4 : Socialising DNA	93
Networks, expectations and responsibility	94
Users' networks and new genetic identities.....	97
Socialising DNA in the wings of the clinic.....	110
Conclusion.....	126
Chapter 5 : Personalising DNA.....	128
Expectations and reflections on DNA.....	129
Users' knowledge, interest and curiosity	130
The tipping point	144
Embodying DNA.....	148
Identity and embodiment	149
Genetic risk as embodiment	161
Conclusion.....	168
Chapter 6 : Testing the NHS	170
Responsibility	170
Empowerment or expertise	179
Regulation	186
Common ground	193
Conclusion.....	197
Chapter 7 : An unresolved dispute and future possibilities	199
The findings.....	200
Interpretative flexibility	Error! Bookmark not defined.
Networks and expectations.....	203
Individuals and identity work.....	205
Individualised or collective medicine.....	206
Biosociality and expertise.....	209
Expectations and future directions	212
Bibliography	215
Appendices.....	239

Appendix 1: School of Social Sciences REC Application	239
Appendix 2: School of Social Sciences REC Approval	244
Appendix 3: Cardiff and Vale University Health Board, R & D Committee Governance Review Approval	245
Appendix 4: Email to Recruit Clinicians	246
Appendix 5: Information Sheet for Users	247
Appendix 6: Information Sheet for Clinicians	250
Appendix 7: Consent Form for Users	253
Appendix 8: Consent form for Clinicians	254
Appendix 9: Interview Schedule for Users	255
Appendix 10: Interview Schedule for Clinicians	258

List of Figures

Figure 2:1 Genetic Predisposition DNA Testing	19
Figure 2:2 Myriad Genetics Advertisement.....	26
Figure 2:3 Sciona Company webpage with news of Body Shop partnership	28
Figure 2:4 “23andWe”: 23andMe’s research community webpage to November 2013	33
Figure 2:5 23andMe point-of-sale in UK health and beauty shop, May 2015	36
Figure 2:6 GeneticHealth UK website home page.....	38

List of Tables

Table 3:1 User Participants’ Information	74
Table 3:2 Summary of User Participants’ Information.....	78
Table 3:3 Clinician Participants’ Information	81
Table 3:4 Summary of Clinician Participants’ Information.....	85
Table 3:5 Symbols used in data transcription	92

Abbreviations

BBC – British Broadcasting Corporation

BSGM – British Society of Genetic Medicine

BRCA 1 and *2* – Breast cancer genes

CE – Conformité Européenne

CLIA – Clinical Laboratory Improvement Amendments (USA clinical laboratory regulation certificate)

CNV – copy number variant (abnormal number of copies of a section of DNA)

DNA – Deoxyribose Nucleic Acid

DTC – Direct-to-Consumer

DTCGT – Direct-to-Consumer Genetic Testing

FDA – Food and Drug Administration (US Government)

ESRC - Economic and Social Research Council

EU – European Union

FH – Familial hypercholesterolaemia

GAO – Government Accountability Office (USA)

GP – General Practitioner

GU – Genomes Unzipped

GWAS – Genome-wide Association Studies

HD – Huntington’s disease

HGC – Human Genetics Commission (UK)

HGP – Human Genome Project

HRA – Health Research Authority

IHGSC – International Human Genome Sequencing Consortium

ISOGG – International Society of Genetic Genealogists

IVDMD – In vitro diagnostic medical devices

MHRA – Medicines and Healthcare products Regulatory Agency (UK)

NHS – National Health Service (UK)

NICE – National Institute for Health and Care Excellence

PD – Parkinson’s disease

PHG Foundation – Public Health Genomics Foundation

REC – Research Ethics Committee (equivalent to an Institutional Review Board (IRB), Ethics Review Board (ERB) or Independent Ethics Committee (IEC))

SACGHS – Secretary’s Advisory Committee on Genetics Health and Society (USA)

SCOT – Social Construction of Technology

SNP – Single Nucleotide Polymorphism

SSK – Sociology of Scientific Knowledge

STS – Science and Technology Studies

UK – United Kingdom

USA – United States of America

Chapter 1 : Introduction

This thesis examines Direct-to-Consumer Genetic Testing (DTCGT) in the United Kingdom (UK) context from the perspective of genetics clinicians and users of this technology. Its purpose is to examine the way these two groups of actors are shaping personal genomics in this country through their discourse and the impact of their engagement with it on the UK's collective national healthcare provision. I commence this written presentation of the project with a story about an encounter I had during my time as a doctoral student. I have decided to share this story as it represents a watershed moment in my thinking about the sociology of science and a salutary illustration of the basis for the disputes that are at the core of this study.

The German geneticists

I began to really appreciate first-hand the importance of 'the social' in science during a state-funded workshop on Genetic Transparency which I attended in 2013 during the course of the PhD study. This week-long workshop was held at Lübeck University for Europe-wide doctoral and post-doctoral researchers and focused on the ethical and social implications of human genomics and genetic medicine. Different guest experts attended each day and presented their work alongside that of the participants, with discussion following each presentation and at the end of each day. The week culminated in the development of a book proposal for a volume on Genetic Transparency to be written in the year following the workshop with contributions from all the participants and experts (Dreyer et al, forthcoming).

My presentation was scheduled for the first day of the workshop at which the guest 'experts' were two German geneticists who presented their work on genomic sequencing at the beginning of the day. In addition a clinical geneticist specialising in cardiac genetics was in attendance as one of the facilitators of the workshop. It is notable that my presentation was only the second of the workshop participants', the first being given by a German

physician and doctoral student who is researching DTCGT from a clinical medical perspective, making mine the first non-positivist presentation of the workshop. I presented my early findings from the users' data in this project and discussed these in the context of expertise, arguing that users appear to have at least interactional expertise in personal genomics, based on Collins's and Evans's work on expertise (Collins and Evans 2007).

If the convenors of the workshop hoped for lively debate, my presentation gave it a flying start. In the group discussion following my presentation there was heated debate between the geneticists and workshop participants about the nature of genetic science and the validity of my work. After the session, the three geneticists physically prevented me from leaving the room for the break and continued to offer their criticisms of my work despite my peers' robust defence of it in the earlier discussion. Rather than commenting on what I was proposing, they protested that my work was scientifically flawed. Why had I interviewed only 16 people? Had I not done a power calculation? How could I judge expertise when I was not an expert (i.e. a geneticist)? It was a good thing that they were not examining my PhD thesis, as it would fail because I clearly had no idea what I was doing or how to be scientific. The geneticists seemed to imply that I was no expert, either by credentials or experience, and thus was acting as an impostor.

I am used to my research topic prompting discussion; usually it is about people's initial reactions to the idea of DTCGT, as they have rarely encountered the concept before. On this occasion a knowledgeable audience were reacting even more strongly and this was striking. The concept of expertise seemed to be at the core of the group's individual reactions to my suggestion that early adopters of DTCGT in the UK have similar levels of expertise in personal genomics as genetics clinicians, though it is likely to be of a different nature. The well-rehearsed debates about DTCGT seemed to be represented in the room. The scientists represented sceptical and paternalistic views about DTCGT, suggesting that it was nonsense and should not be accessible by the public while most workshop participants voiced support for a more objective view of the actors involved and challenged the geneticists' categorical, authoritative

rhetoric, both in relation to DTCGT and their criticisms of my work in particular.

The German story is interesting because it illustrates professionals' vigorous anger in response to this technology. I wondered why they seemed so livid, what was the cause of their visceral response? Reflecting on the incident subsequently I thought that their response could indicate tribal behaviour, particularly when their professional norms and status were subject to challenge, both in terms of disciplinary and ethnic differences. It showed that being culturally bounded within their positivist worldview, the scientists involved had little understanding of qualitative research, let alone the sociology of science and technology.

I was in a similar place at the beginning of this study: aligned to healthcare professionals, disapproving of public engagement with genetic technologies, which I rationalised with bioethical concerns. That is, I had an asymmetrical or partial perspective. This incident (which endowed me with a certain notoriety among my colleagues at the workshop) served to show me that my views had altered in that I could stand apart from these scientists and still appreciate their views having held similar ones myself until recently (albeit not in relation to qualitative research). Most importantly, I could appreciate the cultural and social basis for their views rather than interpret them as a personal attack. However, I was caught off-guard and felt physically intimidated at the time. I was disappointed that I had not been able to persuade them that there was an alternative perspective of genomics (particularly given the premise of the workshop). In hindsight I realise my sociological naivety contributed to this situation and I would certainly take a different approach to such a situation now.

This experience proved a watershed in my learning during the course of this project. It demonstrated to me the powerful influences that social factors have on identities, boundaries, knowledge and beliefs. In adhering to their positivist view that science constitutes privileged, objective knowledge, the German geneticists unintentionally demonstrated the reverse that is that their views are partial and subjective. It emphasised the importance of acknowledging the wider social contexts of different actors' constructions of their experiences or values and their influence on

developments in science and technology, including my own, as I hope this thesis will show.

The problem of DTCGT

DTCGT has evolved over the last decade, both as a result of the completion of the sequencing of the human genome and the genome-wide association studies (GWAS) that ensued and as an extension to the direct-to-consumer marketing of drugs in the United States of America (USA) which started in the 1980s (Gollust et al 2002). Initially offering metabolic and nutritional advice based on the analysis of a few specific nucleotides in an individual's deoxyribose nucleic acid (DNA), dramatic improvements in technology and reduction in costs mean that DTCGT now has a much wider capability and is more widely available, being mainly traded via the Internet. The DTCGT market offers numerous types of test including the more familiar monogenic tests to predict risk of genetic diseases such as familial breast cancer or Huntington's disease (HD), tests offering health improvement information (often linked to marketing of health improvement products), tests for physiological responses to drugs (pharmacogenomics) and tests for susceptibility to common complex diseases such as type 2 diabetes mellitus, osteoarthritis or hypertension (Borry et al 2010). The focus for this research project is the testing that claims to establish susceptibility to common complex diseases, which is usually sold in the context of a genome-wide scan of hundreds of thousands of single nucleotide polymorphisms (SNPs), also known as SNP genotyping¹. These tests simultaneously analyse the variation in single nucleotides at thousands of points across a whole genome (genetic material of an organism) to provide information about various aspects of genomic data including ancestry, physical traits, genetic disease, carrier status, pharmacogenomics and common complex disease risk (Leachman et al 2011).

As the phrase suggests, DTCGT refers to genetic testing sold directly to the public without the involvement of healthcare professionals. Tropes of

¹ Unless explicitly stated to the contrary, I use the term DTCGT in this thesis to refer to tests that involve SNP genotyping rather than alternative kinds of genetic testing.

personalisation are fundamental in order to appeal to the market and companies selling DTCGT emphasise the advantages of accessing information for making personalised health and lifestyle decisions in an autonomous, confidential manner. They do so by linking personalisation to genetics using deterministic language (Arribas-Ayllon et al 2011a). By appealing to the 'personal' in personal genomics in an overt way, DTCGT companies emphasise the appeal of individual autonomy, promising to empower the individual with information about their genome. Moreover this model of accessing health information is promoted as providing convenience and privacy, being accessed directly by the purchaser from any convenient Internet connection (Borry et al 2010).

The vision of autonomy and empowerment created by DTCGT companies has deliberately raised expectations, as is often the case with emerging biotechnologies (Hedgecoe and Martin 2003). These expectations have been reinforced by the lobby for the democratising benefits of personal genomics in terms of its availability to the public and the empowerment associated with accessing one's own genetic information rather than having access restricted by paternalistic healthcare professionals (HGC 2003, McGowan and Fishman 2008, Juengst et al 2012). However, this aspect of genetic testing is controversial because there are forceful counterclaims related both to the science that SNP genotyping is based on and to the problems associated with genetic information being available without the conventional associated genetic counselling.

The SNP genotyping employed in DTCGT is controversial because the data from GWAS that provide the basis for calculation of the meaning of variance have low validity and reliability, having been studied only in discrete populations. Whilst the raw data yielded is thought to be highly accurate, the choice of SNPs for analysis varies between companies, and the interpretation of the results is much more uncertain being based on GWAS studies whose populations may vary physiologically from the individual whose genome is being compared (Kraft and Hunter 2009, Leachman et al 2011). Additionally, DTCGT is widely reported as having poor clinical utility because the SNP variance is only one minor factor that contributes to the aetiologies of common complex diseases; epigenetics

and environmental factors provide a much greater influence on the risk of disease (Janssens et al 2008, Ng et al 2009, Leachman et al 2011).

In addition to concerns about the validity of SNP genotyping, there are concerns about the public's ability to appreciate the nature of the information a SNP genotype provides or to interpret the information provided by DTCGT without the help of genetic counsellors. Results for health-related aspects of the test are expressed as relative risks, which it is argued are difficult for the public to understand because they tend to individualise risk (Lloyd 2001, Paling 2003, Collins et al 2011). Given the companies' promissory rhetoric and the lack of conventional counselling, fears have been expressed that people may overwhelm healthcare facilities to obtain professional help with interpreting their genome scan results. Without it they may on the one hand suffer needless anxiety or family disruption and possibly seek drastic intervention to avoid disease. On the other hand they may adopt a complacent approach to their health in the light of test results based on information of poor clinical utility (Wolfberg 2006, ACMG 2008).

Concerns about the public's understanding of genetic information accessed via companies whose rhetorical style is seen as genetically deterministic aligns with suggestions that genetic determinism increases in association with new medical technologies in molecular biology. The companies' focus on the importance of genetic information corresponds to the potential for geneticisation in that it supports the genetic basis for identity and individual problems (Rapp 2000). In addition their appeals to individual responsibility for health and disease prevention using genetic information supports Kelly's assertion that geneticisation accompanies the upsurge of individual surveillance and responsibility for health (Kelly 2007). Apprehensions about the potential rise of genetic determinism are linked to the historical legacy of the eugenics movement and have no doubt directly influenced the conservative nature of conventional clinical genetics with its emphasis on confidentiality, informed consent and non-directive counselling (Fox Keller 1992, Hogarth et al 2008). This powerful moral interpretation, whilst risking being judged as paternalistic, informs the negative expectations of DTCGT that are circulated and are in contrast to the positive ones centred on empowerment and autonomy. It underpins healthcare professionals' and

particularly genetics clinicians' views about access to health-related genetic information, despite DTCGT's companies' assertions that their tests are not 'health' tests, a claim made ostensibly to avoid regulatory constraints (Vorhaus 2010a). However, many have argued that the public will perceive them as such; some studies' findings support this view (McGuire et al 2009, Mavroidopoulou et al 2015).

This dispute has the potential to undermine the public's confidence in the healthcare establishment because DTCGT users' expectations of personalised genomics may be in conflict with current healthcare provision. This is particularly true in the UK where the principal provision of by the National Health Service (NHS) is free at the point of access and there is less financial onus on individuals to take responsibility for their health care. While the public may be increasingly autonomous in seeking information from DTCGT, they may also continue to expect conventional healthcare support for test interpretation and subsequent intervention. Difficulties achieving this either due to ignorance of genomics on healthcare professionals' part or a lack of capacity in healthcare service provision, could adversely affect both the public and healthcare providers in terms of expectations and patient-clinician relationships (McGuire and Burke 2008). The importance of keeping the public's trust and engaging users, DTCGT companies and healthcare professionals in developing new relationships within a more democratic approach to health care are suggested as a more realistic way forward as the genomic era progresses (McGowan and Fishman 2008, Patch et al 2009, HGC 2010).

Personalised medicine

Having outlined the problem of DTCGT that has resulted in this study of DTCGT in the UK context, it is important to clarify how I use the phrase "personalised medicine" through the thesis. The phrase is important in relation to DTCGT because since the completion of the Human Genome Project (HGP) the rhetoric of personalisation has been used to link expectations about genomics to advances in medicine, scientific research and commercialisation of healthcare for the public (Arribas-Ayllon et al 2011a). Richard Tutton has demonstrated how the concept of the personal in medicine significantly pre-dates the HGP, appearing as a common theme

in patient-clinician relationships for decades. However the rhetorical emphasis on personalisation was adopted by journalists and then more widely used in response to developments in genomics (Tutton and Jamie 2013). There are two predominant aspects to personalised medicine both of which refer to customisation; the first aspect relates to the development of drug treatments tailored to particular genotypes, whether the individual's or the genome of their cancer cells. The second relates to the capacity for individuals to be empowered by their genomic knowledge and to make healthier decisions about their life-styles (Tutton 2014). DTCGT companies that include pharmacogenomics testing use both these aspects to market their products to the public. But they capitalise on the latter, appealing to individual responsibility for health and the importance of empowering genomic information in decision-making. The narratives about personalised medicine are significantly more complex than this brief outline suggests and I shall discuss this further in relation to the study's findings and conclusions later in the thesis. However, for now I use the phrase personalised medicine to refer to both the aspects of customisation outlined above, as this is the rhetorical position of the DTCGT users and 23andMe, the company they purchased their health-related SNP genotyping from.

The project

The debates surrounding DTCGT demonstrate the current lack of stability of this technology. This instability and the lack of empirical work on DTCGT in the UK context provided the impetus for me to undertake this research project. Initially, my interest in personal genomics was informed by my experiences of nursing people with colorectal and breast cancer. I wondered if the public would be tempted to use this technology to try to ascertain their (and their family's) risk of developing cancer. Patients I encountered were concerned for family members, as is commonly experienced in clinical genetics counselling (Chadwick 1999). However, these people also appeared to misinterpret the aim of national screening programmes with their emphasis on risk prediction rather than treatment or prevention, as David Armstrong has explored (Armstrong 1995, Armstrong and Eborall 2012). In my experience people appeared to assume either that screening offered protection or, alternatively, that it would not provide any useful approach to their family's personal risk management, being a

population-based initiative rather than a personal one. Thus, I wondered, would SNP genotyping for disease risk provide them with an alternative route to trying to manage their risk of cancer?

On the basis of my early interest in DTCGT, I developed a research proposal to investigate the uptake and experiences of users of DTCGT internationally, based on the bioethical principles of autonomy and trust. I was unsuccessful in gaining funding for the project on that basis. Prior links with Cardiff University then proved fortuitous in presenting the opportunity to investigate DTCGT from a sociological perspective; a somewhat different approach to researching the topic resulted and is presented in this thesis. As my account of my experiences in Lübeck demonstrates this opportunity has profoundly influenced my thinking about science and genomics. Delanty's suggestion that emerging genetic technologies make it implausible to separate science from society is resonant as it reinforces the argument for pursuing a sociological approach to studying this contentious topic and the actors who influence the debates about it (Delanty 2002). Accordingly it seems appropriate to use Pinch and Bijker's social construction of technology (SCOT) as the framework for the study. My decision to do so assumes MacKenzie and Wajcman's understanding of the meaning of technology. They suggest that technology encompasses **knowledge** or understanding about technology as well as the artefact itself, and that that knowledge is socially influenced by individuals, groups and contexts, and by extension, their discourse about the technology (MacKenzie and Wajcman 1999). DTCGT provides information that is open to interpretative flexibility and which is thus shaped by actors in relation to its quality, utility and validity. These actors' understandings provide an additional dimension to technology along with the artefact and the information it provides. This aligns with the assertion that genetic testing is not simply a laboratory procedure but also a "social practice" (Arribas-Ayllon et al 2011b:3) or socio-technology.

Throughout this thesis I refer to actors shaping DTCGT technology on the basis that discourse is part of how technology is used and that technology encompasses more than the artefact itself. In doing so I am able to conceive this research as a study that investigates how DTCGT is socially constructed and shaped by groups closely associated with it in the UK

context. This has enabled a wider, more objective and arguably more interesting, investigation of personal genomics and the actors involved with it. Rather than examining DTCGT from the perspective of its impact on the public and their relationships with healthcare professionals, I am instead able to provide a broader examination of the various ways in which the two principal groups of actors engaged with DTCGT in the UK talk about and, thus, shape the discourse, knowledge and thus the technology of personal genomics. These two principal groups are the members of the public who buy DTCGT² and the genetics clinicians³ who may be called upon to counsel them.

Research question, aims and objectives

So, the research question that this study addresses is

“What are UK users’ and genetics clinicians’ experiences of DTCGT?”

The study aims to establish users’ and genetics clinicians’ contributions as relevant social groups to shaping personal genomic technology in the wider social context of the NHS in the UK. More specifically it examines users’ motivations for engaging with DTCGT and both users’ and genetics clinicians’ expectations of DTCGT and how these are influenced. Users’ and clinicians’ views about direct access to personal genomic information are explored and how users make sense of complex risk information is ascertained. The nature and scope of genetics clinicians’ involvement with users of DTCGT and the implications for the NHS are examined. Finally, the implications of the study’s findings for personal genomics technology in the UK context and their influence on the possibilities for stabilisation of this technology are explored.

To complete this introduction I will go on to outline the thesis chapters.

² The people who bought DTCGT and participated in the study can be described as early adopters of the technology, being similar to those described by McGowan et al (2010). Detail about recruitment of these participants is given in Chapter Three. However it is important to note here that the approach taken to recruitment has inadvertently resulted in participants who are activists with outspoken views about DTCGT and that this will have affected the data collected.

³ Whilst General Practitioners are the public’s first point of contact for healthcare expertise, genetics clinicians are the most likely HCPs in the NHS to be knowledgeable about and consulted to interpret DTCGT. They were thus likely to make a more direct contribution to the debates about personal genomics at the time this study started.

The thesis structure

In order to set the scene for the project in more depth, Chapter Two outlines the procedure by which SNP genotyping is achieved and how consumers can purchase a test. This description is situated in a history of DTCGT through which I examine the conditions of possibility that have enabled its reification. Consideration is given to the three principal areas of influence, namely the technological, ideological and moral influences that contributed to the development of personal genomics services being sold directly to the public. The chapter is completed with an examination of regulatory influences on personal genomics and of how, until recently, the vacuum provided by the lack of regulatory oversight facilitated the development of DTCGT.

With this important context in place, Chapter Three proposes the study design. Using a review of the sociological research into genetic testing, I situate this study in the canon of Science and Technology Studies (STS) generally and genetic testing research more specifically. This exploration of other researchers' work extends the context for my research from Chapter Two's background, by providing sociological insights into genetic testing. It simultaneously provides a platform from which to justify my theoretical approach, using the SCOT framework and the tools that have guided my thinking and decision making in relation to the data collection and analysis. Methodologically I demonstrate how the use of interviews is an established approach in sociological research into genetic testing and is particularly apposite for studying genetic testing communicated in the online environment. Having outlined the theoretical and methodological approaches to the study, the latter part of Chapter Three presents detail about the study design, including approvals gained, information about the participants, how interviews were conducted and the approaches to the data analysis.

Chapters Four, Five and Six explore each of the three overarching themes from the data. In Chapter Four the concept of socialising DNA is the focus. Participants' beliefs and ideals about genomic information and the influence of networks and expectations are examined. The uncertain new technical

world of SNP genotyping is informed and facilitated by the expectations of the technology that its enactors have presented in an attempt to stabilise it. The effects of these expectations and the censorious ones circulated in professional genetics networks are examined for their effects on the participants developing understandings of the technology and their discourse about it. In addition, I explore the concept of responsibility for health or patients' welfare, which provides a moral standpoint which the participants support in their networks and construct their views of DTCGT.

The theme in Chapter Five is personalising DNA, one which aligns closely to Novas's and Rose's concept of the "somatic individual", whose imagining and embodiment of DNA results in an altered individual (Novas and Rose 2000:487). Here, the participants' perceptions of DNA in the context of SNP genotyping are explored in relation both to its influence on their individual identities and embodiment of its information (in both groups) and to the clinicians' work to align their expectations with their counselling practice. In contrast to Chapter Four, this chapter explores the individual, internal and personal aspects of participants' expressed ideas about DNA in the context of SNP genotyping.

Chapter Six considers the tensions between users and clinicians in their respective support for personal or collective medicine. Responsibility is a feature on which this tension turns and I explore how users and clinicians are diametrically opposed in their interpretation of the relevance of genomics to the NHS. Common ground is found in their consideration of pharmacogenomics, but this represents only little glimmer of hope for resolution of the dispute that personal genomics presents to these two groups of actors.

The final chapter brings the findings of the study together and draws conclusions from the three main themes in the data in relation to biosociality. This discussion leads to suggestions for possible future work in this area, either in genome sequencing or in theory development in the areas of citizenship in the context of people's engagement with genomics.

Having presented an introduction to the thesis, I will go on to explore the historical background to DTCGT in Chapter Two. This is important for

understanding the context in which early adopters of the technology and genetics clinicians have come to occupy different positions in the debate about personal genomics.

Chapter 2 : The history of direct-to-consumer genetic testing

Genetic testing has long been the subject of sociological research generally and science and technology studies specifically. This century's developments in genomics have enabled commodification of genetic information in the shape of DTCGT, which provides additional scope for study of the implications and effects of this development on individuals, groups and organizations with any interest in its capability. In order to provide the context for this study, this chapter will examine the history and development of DTCGT to see how the stage has been set for users' and clinicians' involvement. In addition, the on-going influence of historical events on technologies' evolution supports my decision to adopt this approach (MacKenzie and Wajcman 1999). I have decided to do this by examining the conditions that prepared the ground for DTCGTs emergence, examining the scientific, social, economic and political factors involved. This wider social context is important for my subsequent examination of how users and clinicians are shaping the technology, as Klein and Kleinman (2002) suggest. First I will examine the conditions of possibility that were in place in order that DTCGT could evolve, then I will go on to explore the regulatory landscape and the main events that feature in it.

Conditions of possibility for DTCGT

Three factors contributed to the emergence and existence of DTCGT as it is currently provided. These can be broadly grouped into the technological, the ideological and the ethical (or moral order).

- 1) Technologically, the developments in genetics early this century in the HGP and subsequent GWAS and the increasingly available access to information technology and the Internet have been key to making DTCGT possible to provide and to access.
- 2) The ideological aspect concerns the shift to neoliberalism, specifically health consumerism, which is changing public expectations in

relation to information and healthcare provision. The role of companies and consumers in shaping DTCGT is crucial to setting the scene for how UK users and genetic clinicians are shaping DTCGT.

3) The problematisation of acquiring genetic information continues to provoke debate about associated ethical issues and society's response to these, its moral order. However, DTCGT's unclear ontological status has added fuel to these discussions that is unmatched by consistent regulatory decision-making or the ability to apply such regulation effectively. Thus, a regulatory vacuum has resulted, facilitating the developments of DTCGT companies' activity. The actors involved in the debates about regulating DTCGT will contribute to the role of society and culture more broadly in influencing DTCGT as this section will show (Bijker 2010).

I will now go on to examine each of these areas in more detail, starting with technology in order to set the context for DTCGT and how it is undertaken.

Technology

The three principal technological factors that influenced the early emergence of DTCGT were all becoming part of wider social consciousness during the last decade of the twentieth century. These were the Internet, the HGP and GWAS. In order to provide background for this study of DTCGT, I will first outline what types of information DTCGT provides and the process of purchasing, testing and receiving results. Then I will discuss the developments and influence of the Internet, the HGP and GWAS on the emergence of DTCGT.

DTCGT – the procedure

DTCGT companies sell genetic tests for various purposes. Types of testing include whole genome or exome sequencing, where the sequence of nucleotides along the length of the individual's genome (entire DNA) or exome (DNA that codes for functional proteins) is elicited. Tests can also identify paternity, single-gene mutations, or carriers of single gene disorders. Genotyping compares markers in an individual's DNA with another individual's markers or a reference sequence and contrasts them in

order to calculate probabilities of the person having particular traits or developing common complex diseases (Sander 2000). SNP genotyping includes testing for

- ancestry
- inherited characteristics such as tongue rolling or ear-lobe shape
- metabolic responses to certain drugs and nutrients
- exercise physiology for training and nutrition planning
- susceptibility estimates for common diseases with complex causes including cardiovascular disease, various cancers and Type 2 Diabetes Mellitus
- pre-symptomatic, predictive carrier or diagnostic tests for certain single gene mutations causing diseases such as cystic fibrosis or breast cancer due to *BRCA* gene mutations

(Chapman 2010)

DTCGT is simple to purchase. Having accessed a DTCGT company website and paid a fee online, the customer sends their sample of saliva in a tube, a buccal swab or a blood spot from a capillary sample on porous paper to the company's laboratory. The procedure for analysing samples involves extracting DNA from the sample and copying (amplifying) it several times. It is then spliced into sections by enzymes and each section is tagged with a fluorescent marker. The tagged sections are introduced to a glass slide chip on which are millions of small sections of DNA known as probes. Each of these is complementary to one of the SNPs being tested for. When introduced to the chip, the customer's spliced DNA sections will pair with the probes on the chip where the nucleotide sequences are complementary. Pairing results in activation of the marker indicating which SNPs the customer's DNA contains (etc Group 2008). In due course the customer receives notification of results, mostly via a secure personal webpage on the company's website. Depending on the company and the service purchased, a qualified doctor may or may not be involved and there may be an option to receive counselling. Some companies notify users regularly about updates to the results as more SNPs are identified by

GWAS and the customer's DNA is compared against these (McBride et al 2010)⁴.

The phrase "new genetics" is frequently used in relation to genomics in general and DTCGT in particular. This is without its meaning having been clearly defined, but its deterministic value is related to the shift in genetics from analysis of chromosomes and isolation of single genes to the molecular level of genetics from which the HGP and GWAS have developed (Conrad and Gabe 1999). These will be discussed after I have considered the information technology developments that have supported the emergence of DTCGT.

The Internet

From 1991, the worldwide web made access to centrally stored information available to anyone with access to the Internet. The Internet provides interlinked computer networks that enable access to electronic communications media. Using the Internet and browser software, documents and 'spaces' can be navigated via the worldwide web. Web 2.0 was developed following incremental technological developments and this facilitates interaction and user-generated content to be displayed, contributing to the development of online social networking and commercial activity. As hardware costs fell from the end of the 20th century, people have been increasingly able to use the Internet for personal, commercial and professional communication, information storage and access and trade (Ward 2006, O'Reilly 2009). More recently, access to the worldwide web and social networks has been significantly aided by the development of a range of wireless hand-held devices. These developments have enabled almost ubiquitous access to the Internet and social networks and, in the information age, play a key role in democratization and personalisation, particularly in relation to Web 2.0. Its personalised mode of access and interaction has made the Internet the perfect vehicle for DTCGT (Foster and Sharp 2008, Arribas-Ayllon et al 2011b).

⁴ SNP genotyping differs from DNA sequencing in that it analyses single nucleotides at various points across the individual's entire genome, rather than analysing all the consecutive nucleotides in a gene, or the whole genome.

The Human Genome Project and GWAS:

Most influential on the capability of DTCGT was the project to sequence the human genome. Begun in 1990, the project was undertaken by the International Human Genome Consortium at university laboratories in six countries with parallel work being done by Celera Genomics, a biochemical technology company founded by Craig Venter (Wright et al 2011a). Completion of a functional map of the human genome was completed earlier than anticipated in 2003 (IHGSC 2004).

Genetic sequencing was facilitated by biotechnological developments; the first was the invention of the polymerase chain reaction (PCR) in 1983. This process uses temperature changes and the enzyme *taq polymerase* to first break up DNA into a single strand and then create copies of that DNA sequence from single nucleotides labelled with molecules to enable their identification and sequencing of the strands of DNA. The development of DNA microarrays followed, which utilize specific sequences of DNA as probes. When mixed with a solution containing spliced sections of DNA, these probes bind to complementary sections of DNA, capturing them for subsequent sequencing. These two developments laid a foundation for further developments, including next-generation sequencing, which enabled genetic sequencing on an increasingly massive and cheaper scale (Wright et al 2011a).

GWAS were made possible by mapping the human genome and the biotechnology advances in sequencing apparatus; their aim is to draw correlations between human genotype variations and diseases by genotyping DNA from large numbers of donors to human biobanks. Genotypes of people with or without the diseases of interest are analysed and compared for nucleotide variations that can be linked to the diseases (Kaye et al 2009). The genotypes of SNPs for an increasingly large number of conditions and traits are being established through GWAS. These data provide the template information against which DTCGT companies compare customers' genotypes and calculate disease risk for complex multi-factorial diseases (Edleman and Eng 2009, McBride et al 2010).

DTCGT companies have deliberately used deterministic language in their marketing material, as shown in Figure 2.1. However, related to the difficulties of ‘personalising’ relative risk estimates from a study population, there are a number of issues associated with SNP analyses for common complex disease risk evaluation relating to the tests’ utility and validity. A genetic test is said to have analytic validity when it accurately detects the genetic anomaly being tested for; clinical validity relates to how well the genetic anomaly indicates presence of disease; clinical utility indicates the ability of the test to provide information about diagnosis and treatment that is of use to the affected individual (Holtzman 1999). DTCGT is widely reported as having low clinical validity and utility in comparison with predictive genetic testing for single gene defects (Kuehn 2008, Van Ommen and Cornel 2008, Caulfield et al 2009, Edelman and Eng 2009, Kraft and Hunter 2009, Patch et al 2009, Annes et al 2011, Evans et al 2011).

HOME DNA TESTS & PRICES ANCESTRY BY DNA ABOUT DNA TESTS WHY ARE WE DIFFERENT HELP & SUPPORT

Genetic Predisposition DNA Testing

Genetic Predisposition DNA Test
Price: £239.00
Testing: Applicant only
Timeframe: 19-21 working days

ORDER HERE

International Biosciences provide genetic testing to determine your genetic predisposition for many important health conditions including cancers, cardiovascular conditions, diabetes and obesity. Knowing which genetic disorders and diseases you are at risk from will allow you to plan to reduce it by making lifestyle changes such as diet and exercise.

[Click here to view a sample Genetic Predisposition Report.](#)

[Click here for step-by-step instructions detailing how to do a Genetic Predisposition test.](#)

Verified and listed by
dna
testingchoice.com™

Figure 2:1 Genetic Predisposition DNA Testing
(International Biosciences 2015)

The analytical validity of some companies' SNP analyses has been questioned. Janssens et al (2008) reported on the lack of sufficient evidence for SNPs being useful for disease risk information. Ng et al and a Government Accountability Office (GAO) report both showed that comparison of test results on the same DNA yielded different results for disease risk estimates from some of the larger DTCGT companies (Ng et al, Kutz 2010). Another study by population geneticists demonstrated that the algorithms used by the personal genomics companies 23andMe, deCODEme and Navigenics resulted in substantially different risk predictions due to the use of different SNPs to analyse for the same conditions, different risk calculation formulae and different reference population risks to factor into the calculations by each of the companies (Kalf et al 2013). This bears out Martin Richards' experience of getting different results in his auto-ethnographic study (Richards 2010). Underpinning these institutional variations in reference data used to ascertain customers' SNP variants is the problem of the quality of the human reference genome. The original sequence was neither accurate nor contiguously assembled and has resulted in a less than optimal baseline sequence (Mardis 2010)⁵. Moreover, SNP analysis ignores other genetic factors that may affect an individual's propensity to develop disease such as epigenetic effects and copy number variants (CNVs) both of which alter the DNA and its expression (phenotype). SNPs are thought to contribute no more than about 10% to the overall risk of disease; environmental factors are a much more powerful influence on an individual's disease risk (Ng et al 2009).

These difficulties highlight one of the principal debates related to the technological aspect of DTCGT. Using terminology associated with genetics, DTCGT companies imply the deterministic nature of their products. Petersen and Bunton while analysing the uncertain meaning of the phrase "new genetics" suggest that the implications of new molecular genetic knowledge align with a Foucauldian concept of "bio-power". This new knowledge increases the level of control over the human biological life cycle, influencing life from conception to death (Petersen and Bunton 2002).

⁵ The human reference genome is being improved with ongoing sequencing work by the Genome Resource Consortium (Wellcome Trust Sanger Institute 2014)

But the obvious alignment of bio-power with DTCGT companies' discourse in marketing genotype analysis is undermined by the stochastic and contingent nature of the results of SNP genotyping.

I would argue that the problematic nature of DTCGT is well known among geneticists, bioinformaticians and related scientists, the "core set" as described by Collins and Evans in their polemical paper on expertise in science studies (Collins and Evans 2002:242). But other stakeholders in the wider DTCGT community view the capability of DTCGT with less uncertainty whether negatively or positively. On the one hand the majority of healthcare professionals are used to operating within a cause-effect-treatment model and understand Mendelian genetic patterns of inheritance. They are less used to the muddier waters of genomic SNP analysis as contributing partial information to risk assessments for complex diseases and thus tend to simply dismiss it as either useless or misleading (Heschka et al 2008, Boddington 2009, Ormond 2009). On the other hand, DTCGT companies appear to have traded on the likelihood that their customers will have limited understanding of what their genotyping test is really capable of telling them (Patch et al 2008, Kaye 2008). Users are thus thought to be convinced of the certainty and determinism of the information DTCGT offers in relation to disease risk. It is likely, however, that early adopters may well approach DTCGT with less certainty of its capability than was originally thought, as early research found (McGowan et al 2010) and as explored by Harvard psychologist Steven Pinker in his *New York Times* article about his own experiences of genomic testing (Pinker 2009).

Pinker neatly undermines the deterministic nature of personal genomics in several ways. He outlines the subjective way DTCGT company scientists choose GWAS data for associations between SNPs and phenotypes, the absurdity of testing for some of the traits included when obvious phenotypes either indicate the same information or contradict it – in his case his genotype for male-pattern-baldness that is not phenotypically evident, and the difficulties of translating relative risk data. The advice to consume healthily and exercise more applies as the intervention for almost

all his results but, as he notes, he did not need to pay for SNP genotyping to know that (Pinker 2009).

However, Pinker also writes about individual curiosity, the entertaining aspects of personal genomics and the democratic argument for freedom of access to personal information rather than paternalistic regulation, which he and others support (Pinker 2009, Vorhaus and MacArthur 2010). This emerging democracy is key to the ideological conditions for DTCGT, which I will discuss next.

Ideology

Neoliberalism and healthcare consumerism

Since the latter half of the 20th century Western governments have led the development of neoliberal approaches to national leadership and globalisation. Often attributed to Margaret Thatcher and Ronald Reagan, recent approaches to neoliberalism are characterised by the central role of the state in ensuring an emphasis on privatisation, deregulation, free trade and competition both within and across state borders (Davies 2014). The resultant emphasis on capitalism has influenced healthcare provision (to varying extents in different countries) and provided opportunities for choice and individualised consumerism of healthcare services, notably in North America where DTCGT first developed and is largely based. It also results in the inequity characteristically associated with neoliberalism, in this case in access to health care and opportunities for responsible behaviour. However, devolvement of responsibility for health care to individuals places responsibility for health, health promotion and disease risk management on citizens some of whom have become autonomous consumers of healthcare services (Arribas-Ayllon et al 2011b).

US citizens in particular, have become increasingly autonomous healthcare consumers with growing expectations of their entitlements to healthcare provision (Robinson 2005). These expectations and their effects on consumption patterns have contributed to the development of new markets for new health technologies and services (Green 1991). Consumer

expectations are in turn influenced by numerous companies marketing health-related products directly to consumers, using all available media. For example, since the early 1980s pharmaceutical companies have advertised prescription medicines to the public in the USA using consumer choice as a tool to influence prescribing decisions (Gollust et al 2002). This has had the added effect of engaging the public with their health and with prescribed pharmaceuticals for treatments of common illnesses. Whilst prescribing remains the preserve of healthcare professionals, advertising to the public has not only positively influenced pharmaceutical companies' income but also contributed to engaging the public in their health and prescribed pharmaceutical treatments for illness (Huang 2000).

The situation is slightly different in the UK with NHS healthcare provided by compulsory subscription. Whilst medicines sold directly over-the-counter are advertised direct-to-consumer (DTC), DTC advertising does not influence drug prescribing *per se* because it is illegal to advertise Prescription-only-Medicines to the public in the UK and the EU (Ventola 2011). Prescribing by NHS employees is restricted to generic (non-patented) versions wherever possible and the National Institute for Health and Care Excellence (NICE) is directly involved in influencing the availability of many prescription drugs for cost-effectiveness (NICE 2012). However, the public are increasingly encouraged to manage individual risks to their health by participating in nationally advertised health promotion schemes related to diet and exercise and disease screening programmes such as those for hypercholesterolaemia, hypertension and breast or colorectal cancer (NHS 2011).

The increasing democratisation of health care has influenced individuals' relationships with healthcare providers and the traditionally paternalistic and authoritative nature of healthcare professionals. In the UK, this was influenced significantly by the last conservative government of the 20th century; in an attempt to reduce costs and improve management in the NHS they published the "Patient's Charter" (DoH 1991a) and their white paper "The Health of the Nation" (DoH 1991b). These strategic documents, whose principles have been built on by successive governments, served to

facilitate a shift in thinking about health care from illness to health and a shift of power from healthcare professionals towards individual citizens by giving them rights, choice and simultaneously, responsibility for maintaining their health (Klein 2010). Consequently, both citizens and healthcare professionals have autonomy for their practice in relation to managing health; citizens for promoting their own health and managing risks to it, clinicians for providing resources for health promotion, screening and intervention. Whilst the aims of both groups superficially appear to be congruent in this respect, this dynamic shift challenges traditional medical hegemony and the public's increasing autonomy may be in conflict with the current, resource-strapped NHS. This challenge could affect both the public and healthcare professionals in terms of their differing expectations impacting on the patient-clinician relationship, which has previously been based on trust. I will examine this issue further in the later section on Moral Order.

More autonomy in relation to consumption of health care requires individual responsibility and knowledge of health and disease in order that citizens effectively manage their health and the potential risks to it. The media largely fulfil this need providing a conduit for information between scientists or healthcare providers and the public. There is a plethora of information in the media about health and illness and about scientific and technical advances in relation to them, specifically in genetics and genomics, with almost daily publications. However, the media's reductionist, over-simplified interpretations often significantly undermine scientific objectivity. This is exemplified in a short piece on the British Broadcasting Corporation's (BBC) Internet site entitled "Scientists crack the human code"; in just 750 words it refers to the HGP, the basic biology of DNA, key people involved in the project including Craig Venter and John Sulston, and the announcement jointly by US and UK leaders at that time, Bill Clinton and Tony Blair (BBC 2000). Assumptions are made that it is on the basis of this style of information dissemination that the public's self-education in current science and health care is based, in particular their knowledge of genomics and its relationship to health screening.

At the beginning of the 21st century the stage was set for DTCGT by the interplay of various factors. The public was primed to manage their individual health and risks to it and had increasing expectations of developments in genetics as promised by the media and companies marketing their products directly to the public. The emergence and commercial activity of these companies were able to exert considerable (albeit varying) influence on DTCGT technology owing to their organisation and investment as Williams and Edge note (1996). An examination of the companies involved in marketing DTCGT follows.

Myriad Genetics

One of the first companies to engage with the DTCGT market was Myriad Genetics. Myriad Genetics is a diagnostic biotechnology company, founded in 1991 by scientists researching genetics of breast and ovarian cancer. As part of an international research collaboration, Myriad developed predictive genetic tests for the *BRCA1* and 2 genes, in addition to other malignant disease genetic markers. The company took the unprecedented step of applying for patents for *BRCA1* and 2 mutation testing and exerted its rights to a monopoly on testing and for mutations and drug susceptibility in these genes in the USA; the patents were reduced in Europe (Conley et al 2011). Myriad began marketing these tests directly to consumers and their doctors at the turn of the century and launched advertising campaigns in some states in the USA in 2002 and 2007 (Caplan 2007).

Fig 2.2 shows an example of a Myriad advertisement from the 2007 campaign, which uses powerful language to persuade younger women of many backgrounds to arm themselves with knowledge about their genetics to avoid breast and ovarian cancer. Their abbreviation of the slogan 'Be Ready Against Cancer - BRAC' is almost identical to the abbreviation *BRCA*, the name for the breast cancer genes. Myriad's campaign simplifies the information on risks of developing breast or ovarian cancer omitting to say that most breast cancers are not hereditary (and thus that testing is not relevant for most women). All breast cancer causes are conflated with being hereditary and linked to the *BRCA* genes, implying that most women

will be affected but that testing will empower and possibly even protect them - “Cancer doesn’t have to be inevitable”.



Figure 2:2 Myriad Genetics Advertisement
(Caplan 2007)

The launch of their *BRCA* test marketing strategy in 2007 was described by the company as a public health education exercise. However, their simplistic, commercial approach was characterized by a lack of detailed information about testing, its outcomes or disadvantages. The clinical utility and validity of tests was not referred to and thus the implication of false positive or negative testing was not addressed. Neither did they refer to the possibilities for subsequent expensive and potentially inappropriate testing and mutilating treatment for women and their families who might be affected. In addition the problems associated with false reassurance and complacency following a negative result are ignored (Matloff and Caplan 2008); the company’s current website covers none of these aspects.

Hull and Prasad cleverly compare this marketing approach with the central theme of the play ‘Wit’ in which the central character learns to interpret healthcare professionals’ rhetorical devices as providing false hope rather

than truth. Hull and Prasad criticize Myriad's early emotionally orientated marketing strategy for their distortion of what tests can offer and potentially harming future customers by implying certainty from testing. They illustrate the advertisement's lack of information in relation to the specificity and sensitivity of the tests, or suggestions to seek medical advice prior to testing (Hull and Prasad 2001).

After a long-running legal wrangle with the US Supreme Court, Myriad Genetics lost its rights to hold patents of DNA but its almost exclusive provision of *BRCA* testing in the USA appears to have continued regardless (Wagner 2013). This and the widespread criticism of the company's marketing strategy do not seem to have undermined the company's trading, although their business is largely US focused, 86% of their revenue being from *BRCA* testing in the USA (Conley et al 2011). Arguably, their high media profile could be seen to equate to free advertising.

Sciona

At the same time, a UK company made a relatively brief and ultimately unsuccessful foray into DTCGT. Sciona, then based in the UK, sold tests for nutrigenomics. DNA was analysed for SNPs in up to nine genes thought to be linked to metabolism and customers were offered nutritional and lifestyle advice on the basis of the results. The company's activities raised concerns about the questionable use of hype and hope in their marketing strategy, particularly because of the lack of evidence for the utility of the tests. In addition, the advice being given to purchasers was similar to any health improvement advice, namely to eat more fresh fruit and vegetables, consume less saturated fats and alcohol and take more exercise (Editorial NG 2002, Meek 2002, Vineis and Christiani 2004).

Further factors came to light about Sciona that were of specific relevance in the UK. The tests were being sold on the high street in the Body Shop chain of stores (see Figure 2.3). This was embarrassing on two counts. First, the Body Shop uses its ethical approach to business as a marketing strategy and the ethical debate provoked by DTCGT being sold on the high street did not sit comfortably with that company policy. Second, the UK

government had invested in the company on the basis of its innovation, possibly informed by Tony Blair's earlier publicity with Bill Clinton on the completion of the first draft of the human genome sequence in 2000 (Newton 2001). This was provocative to scientists, including Professor Wolf of Dundee University, Dr Bingham from Cambridge University and Paolo Vineis from Turin University, none of whom felt the validity or utility of the tests were meaningful. Finding that public investment had been awarded to a company who were using other scientists' early genomic data raised concerns about the different standards required for research in public versus private companies (Meek 2002, Vineis and Christiani 2004).

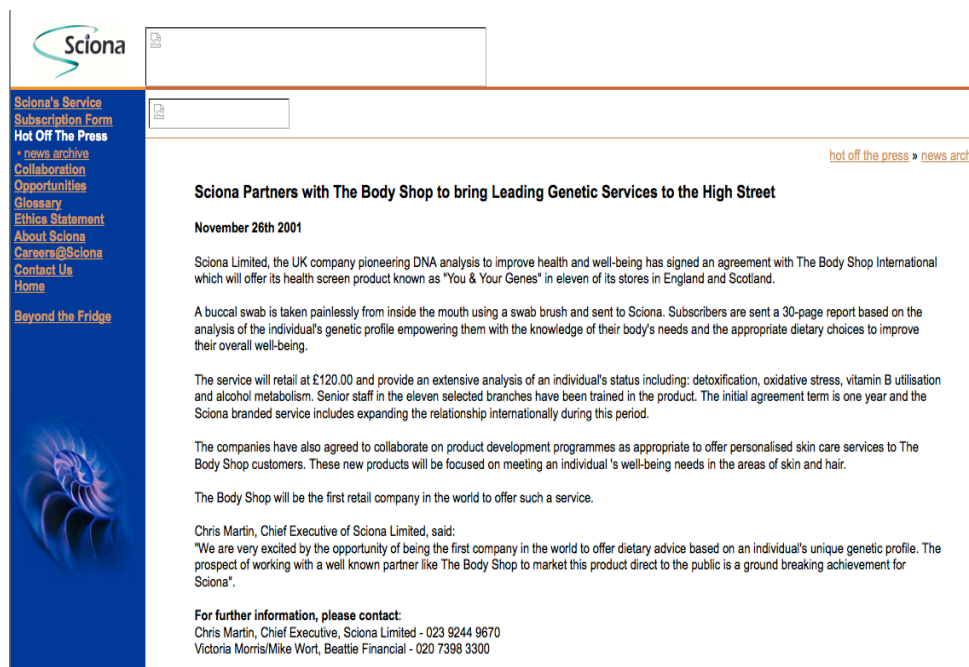


Figure 2:3 Sciona Company webpage with news of Body Shop partnership (Wayback Machine 2013)

GeneWatchUK, a not-for-profit group monitoring genetic technologies for the public interest, raised objections to the test and its availability to the unsuspecting public in Body Shop stores. GeneWatchUK's concerns related to misleading marketing, meaningless test results, poor evidence base and lack of regulation similar to that which would apply to genetic tests used in clinical genetics in the NHS (GeneWatchUK 2002, GeneWatchUK 2004). Sciona's marketing in high street stores was short lived. Despite having been approached by Sciona, several other well-

known companies including Boots the Chemist, John Lewis and Marks and Spencer decided not to sell the kits and the Body Shop chain removed them from its shelves within months of first stocking them. Sciona subsequently re-located to Colorado in the US, though they continued to market to the UK on the Internet, but the company ceased trading in 2009 (GeneWatchUK n.d, Chapman 2010).

Leading DTCGT companies 2007-2013

From the plethora of publications and weblog entries, 2006-2013 saw an increase of activity in relation to DTCGT, as the market initially blossomed and then began to shrink again as the global economic crisis unfolded. In 2007 the principal companies selling health-related tests started their DTC business, namely deCODE genetics, 23andMe and Navigenics, as well as Knome, which offers whole genome sequencing (Lenzer and Brownlee 2008). In their rhetorical move of appealing to the “personal” in personal genomics in an overt way, both with some company names such as 23andMe, deCODEme and Knome (my emphasis) and with marketing strategies that highlight empowerment, convenience, privacy and autonomy, the DTCGT market asserts the desirability of the neoliberal, autonomous self who Petersen had earlier proposed as a self-assessing, caring and improving individual (Petersen 1996). Personalisation is linked to consumerism in the promises companies make to empower the individual with information about their genome and their health to prevent disease and improve quality of life (Kaye 2008, McGowan and Fishman 2008, Nordgren and Juengst 2009, Arribas-Ayllon et al 2011b). The Erosion, Technology and Concentration Group (etc Group, a civil society organization addressing socioeconomic and ecological issues surrounding new technologies) also highlight the *personal* emphasis that companies make, but neatly contrast this with the *impersonal* nature of the GWAS data that DTCGT analyses are generated from (etc Group 2008).

These DTCGT companies’ products played to the democratising autonomy of neoliberal citizenship by being available to anyone (with sufficient disposable income and an Internet connection), without the gate-keeping and appointment systems of conventional healthcare providers or without

informing health insurers (Foster and Sharp 2008, Gurwitz and Bregman-Eschet 2009). Associated advantages include learning about genomics, genetics and disease with a view to gaining knowledge for risk management and improving one's health and lifestyle (Matloff and Caplan 2008, McGowan and Fishman 2008). This section will consider the main companies' contributions to the personal genomics evolution between 2007 and 2013.

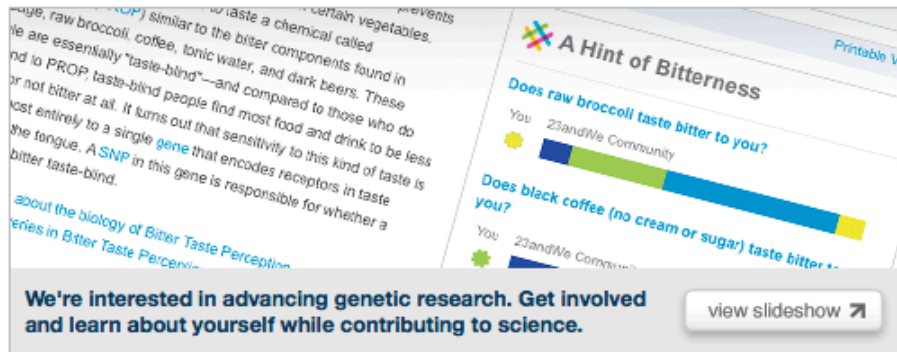
deCODE genetics was established in Iceland in 1996 by Harvard neuroscientist and geneticist Kari Steffanson with the intention of researching genomics and pharmacology to benefit patients. deCODE initially secured funding from the pharmaceutical industry company Roche to use Iceland's unique genetic database for pharmacogenomics research and development. deCODE floated on the stock market in 2000 and developed commercial DTCGT later that decade with deCODEme, using its databases to research genes and pharmacogenomics for various conditions, including Alzheimer's disease. Failing to keep pace commercially with its research success, the company was brought down by the Icelandic financial crisis. Following a bankruptcy declaration in 2009, the company re-launched but still failed to convert its research success into revenue, despite its uncompetitive DTCGT⁶ (Vorhaus 2010a). In October 2012 deCODE was taken over by US biotechnology company, Amgen, who saw deCODE's databases as crucial to their pharmacogenomics research and development programme. Amgen/deCODE genetics withdrew its DTC genotyping test deCODEme to new customers in January 2013. The shifting fortunes of deCODE and this takeover serve to exemplify concerns voiced about data protection in relation to genetic samples and information. Whilst people's DNA samples remain in Iceland for now, at the time of the takeover there was disquiet in Reykjavik about a US company having control of Icelandic (and other customers') genetic samples and their data protection is only as good as the current legislation (Herper 2012, Vorhaus 2012).

⁶ deCODE sold the most expensive and least comprehensive SNP genotyping test on the DTC market.

Navigenics, launched in 2007, offered personalised genomics in its Health Compass product, but the company took the more cautious approach to its services by requiring a physician to order the test for a patient and by offering a genetic counselling service to patients. This tempered approach to the DTCGT market was presumably adopted to increase sales and pre-empt possible future regulatory constraints, but may also have been associated with one of its founders, physician Professor Agus. With its more moderate approach to DTCGT, Navigenics notably partnered with the Scripps Translational Research Institute and the Mayo Clinic for studies into the effects of genetic disease risk estimates on personal genomics users. Findings from both studies indicated that effects on users' anxiety were minimal and likely to abate over time (Bloss et al 2011, James et al 2011). Attracted by Navigenics's highly accurate sequencing technology in its Clinical Laboratory Improvement Amendments (CLIA) certified laboratories, Life Technologies Corporation, a global biotechnology company, acquired the business in August 2012, following which Navigenics's personal genomics product was no longer sold (Vorhaus 2012). Here too the issue of data protection and confidentiality is of concern, potentially to both Navigenics's customers and clinicians. Former customers accessing the Navigenics website are reassured that their data are accessible for three years but that their sample will be destroyed, as will their data, after August 2015. Physicians are similarly informed that patients' data will be available for three years but will then be destroyed and that, in line with the consent gained from patients prior to testing (including a consent form in addition to direction to Terms and Conditions), their data will not be sold on to third parties and will only be used for the purposes they consented to (Navigenics 2013).

23andMe was launched in 2007 in California by biotechnologists Linda Avey, Paul Cusenza and Anne Wojcicki, with financial backing from Wojcicki's husband Sergey Brin's company, Google. Significant changes to their product and marketing strategies followed a Cease and Desist order from the Food and Drugs Administration (FDA) in 2013, which I shall address later in this section. However, from their launch in 2007, the company's unique selling point in DTCGT was their focus on research and the social aspects of genomics for personalised ancestry and health testing.

Using the tropes of personalisation and empowerment common to DTCGT marketing, 23andMe engages customers in the 23andMe community with blogs, discussion boards and forums on their website. Here users can share and discuss information, experiences and questions with others, capitalising on the potential for social networking that these Internet sites enable. Prior to 2013, 23andMe's commitment to research was foregrounded with invitations to customers to be part of research that will "benefit us all" in the future (see Figure 2.4), simultaneously appealing to the personal as well as an altruistic sense of contribution. Users were invited to engage in "citizen science" by voting to prioritise the company's research projects and by submitting their genomic data and online surveys for phenotypic information to be used in genomic and pharmacogenomics research. Awarded *Time* magazine's Invention of the Year in 2008 for making its product accessible and affordable, 23andMe have led the market in DTCGT with arguably the most competitively priced, cost-effective product available.



A Hint of Bitterness

Does raw broccoli taste bitter to you?

You 23andWe Community

Does black coffee (no cream or sugar) taste bitter to you?

You 23andWe Community

We're interested in advancing genetic research. Get involved and learn about yourself while contributing to science.

[view slideshow ↗](#)



23andMe isn't just about you. Our research arm, 23andWe, gives customers the opportunity to leverage their data by contributing it to studies of genetics. With enough data, we believe 23andWe can produce revolutionary findings that will benefit us all.

Get involved in a new way of doing research.

- Direct research by participating in studies of conditions and traits you care about.
- Join an effort to translate basic research into improved health care for everyone.
- Support 23andMe's efforts to discover new genetic associations that could shed more light on your data.

Participate in research while exploring your own genetics.

- Take surveys that collect important data for scientific research.
- 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.

Join our research communities.

- **Parkinson's Disease:** Recent discoveries suggest that genetics plays a greater role in Parkinson's disease than was previously thought. Help us understand how both genes and environment affect risk for PD by joining over 5,000 people that are already part of this movement.

Figure 2:4 “23andWe”: 23andMe's research community webpage to November 2013

(23andMe 2013)

23andMe promotes its self-professed “democratic” approach to personal genomics and research by using social networking and has engaged user participation (users’ votes) to influence the research it engages in. However,

its relationship with its user community was significantly dented in 2012 with the company's announcement of their award for a patent for sequencing SNPs associated with Parkinson's disease (PD). The commercialisation of customers' free SNP data to develop a patent with the intention of improving the company's income by enforcing its application seemed contradictory to the company's repeatedly voiced philosophy of genomic democracy (although users had agreed this as per the Terms and Conditions small print). In addition, it is not clear if the 23andMe community voted PD to the top of the company's research agenda or if the fact that Sergey Brin's family have mutations linked to PD was a more significant influence. The Michael J Fox Foundation, a research organization set up by the Hollywood actor who developed young-onset PD at 30, is now partnered with 23andMe, using its customers' genomic and survey data to contribute to PD research (23andMe 2012a).

Having lowered the price of their test kit, from \$999 at their launch in 2007 to just \$99 in 2012 (23andMe 2012b), 23andMe launched a USA television advertising campaign in the late summer of 2013. The aim was to increase their customer numbers from 400,000 to a million but unfortunately this goal was undermined when the FDA issued the company with a Cease and Desist letter in November 2013 (Conley 2013a). This reiterated the Administration's earlier decision to class 23andMe's personal genome testing as a medical device requiring regulatory approval that includes evidence of the test's clinical validity and utility for the risk information it provides. Issuing the Cease and Desist letter in 2013 appears to have been provoked by 23andMe's failure to cooperate with the regulatory process (Brice 2013, Prainsack 2014a). The filing of a Class Action Lawsuit against 23andMe, also in November 2013, suggested that the decision to file the suit was deliberately timed in order to capitalise on the FDA's letter, which it relied on as part of the evidence supporting the suit (Conley 2013b). Subsequently, 23andMe has continued to market testing to USA residents that only includes ancestry testing and raw SNP data (if desired), although they do invite participation in health research on purchase of a test kit.

However, whilst cooperating with the FDA and having achieved approval for their test for carrier screening for Bloom's syndrome (Janssens 2015), the company launched bases in Canada, the UK and the European Union (EU) in 2014. They are now selling modified versions of their former "full" range of health, carrier, trait and ancestry test to residents of Canada, the UK, Denmark, Finland, Ireland, the Netherlands and Sweden, as the Public Health Genomics Foundation had forecast earlier in 2014 (Brice 2014). In a surprising commercial move, the company also started selling test kits in a UK health and beauty retailer's stores early in 2015 (see Figure 2.5). This is surprising on two counts. First, it is unclear why the company has chosen to market tests in a retail setting in the UK, although their press release claims this will "improve accessibility for UK customers" (23andMe 2015). Second, the only other attempt at shop-based retailing of personal genetic tests since Sciona's brief partnership with the Body Shop was Pathway Genomics' test kits in Walgreens' shops in the US, which provoked significant reaction at the time and never got off the ground, as I will describe in the next section. Responses to 23andMe selling tests from Superdrug in the UK have included what are becoming the expected statements of concern from organisations such as GeneWatch (GeneWatch UK 2015) and in this instance, the Royal College of GPs (Meikle 2015), but essentially this move has resulted in comparatively little reaction. It is yet to be seen how successful sales will be from this outlet.



Figure 2:5 23andMe point-of-sale in UK health and beauty shop, May 2015
(My photograph)

Given the varied fortunes of the personal genomics market, basing a business model solely on selling personal genetic testing direct-to-consumer has been shown to be an unlikely proposition for commercial success. Indeed Williams and Edge’s concept of “veto power” appears to have been of some influence here in that consumers have not adopted SNP genotyping in droves (Williams and Edge 1996: 878). The announcement early in 2015 that 23andMe has signed deals with large pharmaceutical and biotechnology companies (including Genentech and Pfizer) to provide access to their databases of customers’ genetic and health information finally made the company’s business plan public. Collaborating and selling access to customers’ data for research and pharmacological developments has been key to 23andMe’s business model since its launch, but has only recently been publicised (Herper 2015).

Other DTCGT companies

Other contributors to the DTCGT market include Knome, who offer whole genome sequencing and Gene By Gene (which includes their formerly separate groups DNA DTC and Family Tree DNA), who offer whole genome or exome sequencing (without interpretation of the sequence), mtDNA sequencing, single-gene tests or carrier screening through a physician and ancestry testing DTC. Harley Street-based GeneticHealth offer online testing and were the company featured in the controversial ITV reality television programme “The Killer in Me” in 2007. With a similar trope relating to empowerment through knowledge, GeneticHealth’s website offers SNP testing and uses endorsement from its partnership with ITV to promote its services (see Figure 2.6). However, it is notable that dates on the company website are several years old (© 2005), calling into question its current authenticity (GeneticHealth 2005).

Premium Male **Premium Female** **Obesity, Diabetes and Weight Loss** **Nutrition Gene** **Pharmaco Gene**

Personal Health Management

"The best service to British Clients is GeneticHealth's because of the personal consultation. It does not present results online but in documents."

Financial Times - [How to spend It Magazine 2009](#)
Read the full article →

Order a Kit **Register Kit** **Doctors Area**

Welcome

Medical research is increasingly clarifying the link between an individual's genetic makeup and their risk of many age related diseases. The interaction between your genes and the way you lead your life, "your environment", can play a crucial role in determining your future health.

At GeneticHealth, we provide state of the art genetic testing that specifically looks at a group of selected genes that have been shown by medical research to be implicated with your susceptibility to several age related diseases.

We look at this genetic data in context with your family history and lifestyle and advise you on your level of risk for each of the major disease areas.

GeneticHealth Videos

BBC News This Morning

"The Killer In Me" - ITV

Last year GeneticHealth proudly partnered with ITV for a one off documentary on genetic screening
Read more →
Watch video →

Latest News

GeneticHealth has proudly partnered with ITV for a one off genetic testing documentary "Killer in Me"

More news →

What the media is saying about us

The Interpretation of Genes (FT How to Spend It)
DNA flaw means two thirds of us can't stop eating (Daily Mail Dec 11th 2008)
A Life Decoded (BBC Focus Magazine Summer 2008)
Have you got the fat gene? (The Daily Mail, 20th Nov 2007)

All media →

Request Call Back
Click here →

Download our Brochure
Find out more about our genetic testing services...
Download Our Brochure →

Login

Username
Password
login →

What our customers are saying...

Send information to a friend →

Figure 2:6 GeneticHealth UK website home page
(GeneticHealth 2005)

Pathway Genomics, based in California, offer genotyping (SNP analysis) in a similar model to Navigenics', tests being ordered by physicians and genetic counselling being offered pre- and post-testing. In 2010 this company had an interesting, if short and unproductive foray into high street store marketing of its genetic tests. It partnered with the large US pharmacy chain store Walgreens, who agreed to sell DNA test kits directly to customers in their stores. No kits were ever sold because publicity about the Walgreens/Pathway Genomics deal resulted in the FDA sending a letter to the company noting they had no approval for their test, which appeared to be a medical device and resulted in the kits being withdrawn. Subsequently, similar letters were sent to online DTCGT companies (FDA

2010), to which they responded in private discussions and agreements with the FDA.

Whilst not substantively different from other DTCGT companies' SNP genotyping tests, The Walgreens/Pathway Genomics CLIA certified test seems to have crossed a divide from those available online, to being sold in shops. It is unclear exactly what the nature of this divide is as the issue was resolved by terminating the business agreement between Walgreens and Pathway Genomics but, in tandem with Sciona, it appears that having genotyping test kits physically, publicly available on the high street, (at least in the USA) constituted a greater threat than that posed by individual citizens procuring them online with the privacy of their own computer terminal. This is possibly because a legal loophole in the USA meant that DTCGT was at that time not necessarily seen as a *clinical* test because it was sold online to the public for various uses (Vorhaus 2010a) but when sold by a pharmacy chain store for disease risk testing, the implication that it is a clinical test became more difficult to avoid.

Celebrity testing

Although DTCGT companies appear to be falling like ninepins in this post-economic downturn decade, having appeared in number in the late 2000s, the media often referred to high profile "spit parties" and celebrities' experiences of being tested in their coverage of DTCGT as an emerging technology. With potential customers being attracted to DTCGT by promises of personalised, genomic information that would provide previously unknown information about their health (and/or ancestry), the use of celebrity as commodity (Ferris 2007) was also being marshalled to add to the hype and marketing of this deterministic new technology. Amongst many others, celebrities including Oprah Winfrey and Steven Pinker had stories about their genome analyses published in the media. Interestingly, both stories were characterised by the lack of validity of their tests: Winfrey was led her to understand that she was of South African origin and she publicly announced her membership of the "Zulu nation" whilst on a trip to South Africa in 2005. This was later undermined by geneticists and historians who pointed out that there was a lack of detail in

genotyping at the time Winfrey tested and that the Zulus were not involved in slavery migration, which was from West Africa rather than South Africa (BBC News 2005). Steven Pinker's article on testing describes few surprises and several discrepancies between his genotype and phenotype, presenting a more pragmatic interpretation of the value of genotyping to the user (Pinker 2009).

In a prophetic publication in the Hastings Centre Report, Silverman described the expansion of genetic testing from medical laboratories into private enterprise and forecast the proliferation of genetic testing marketed directly to the consumer. Whilst Silverman did not envisage direct-to-consumer testing *per se*, his article sets the tone for subsequent authors' publications on DTCGT. He does so by raising concerns about the impact of genetic testing on individuals' behaviour and lifestyle choices in the light of results seen through the deterministic lens DTCGT companies portray genotyping information with. He ends his article suggesting that regulation is the only "moral influence that can be brought to bear" (Silverman 1995: S17). This will be examined in the next section on Moral Order, the third condition of possibility for DTCGT, after which I shall go on to explore the history of regulation of personal genomics.

Moral order

In liberal society moral order is central to a system of obligation and accountability for productive relationships between individuals and groups in society. Relationships between the public and healthcare professionals have been based on trust, whether on a more or less paternalistic basis, but the neoliberal movement towards health consumerism threatens this status quo. DTCGT is particularly problematic in this regard because of the tension it sets up between the public (users) and clinicians. Individuals in both groups are accountable to society, healthcare professionals being additionally accountable to patients, employers and their professional bodies. As part of this accountability the public are being encouraged to take responsibility for their health and manage their risk of disease. DTCGT is sold to facilitate this risk management and is seen as having personal utility for many purchasers (Khoury et al 2009, Bunnik et al 2011, Tutton

and Prainsack 2011). However, the autonomy lay individuals are exercising in relation to DTCGT challenges clinicians' accountability to them, because many clinicians view the public's choice to buy genotyping as lacking utility, being misleading and potentially harmful (Wade and Wilfond 2006, Wolfberg 2006, Van Ommen and Cornell 2008, Kraft and Hunter 2009). Alternatively, there is potential for doctors' ignorance and scepticism to undermine the public's confidence in the healthcare establishment, unless they take up Farkas and Holland's call for doctors to assist the marketplace and consumers by guiding them through DTCGT with information (Farkas and Holland 2009). Keeping the public's trust and engaging users, DTCGT companies and healthcare professionals in developing new relationships within a more democratic approach to health care are suggested as a more realistic way forward as the genomic era progresses (McGowan and Fishman 2008, Patch et al 2009).

However, the ethical problematisation of DTCGT relates to the lack of clarity of its ontological status. Whilst it has emerged from research into human genomics, it is difficult to reduce DTCGT simply to the procedure of SNP analysis. The nature of genetic knowledge is wider than its biological basis because it affects people's lives, their decision-making, their families and their health risk management. In this way, DTCGT has the capacity to cause harm in relation to anxiety, drastic interventions for the purposes of managing the risk of disease or complacency and lack of disease risk management, all on the basis of a test that is of doubtful clinical utility (Wade and Wilfond 2006, Wolfberg 2006, HGC 2007, Katsanis et al 2008, Wallace 2008, Patch et al 2009). Bunnik et al (2011) highlight how novel data are being produced by GWAS research, which both add to and alter previous DTCGT results, so customers may have to change their views about their health in light of shifting results and may be repeatedly exposed to an emotional rollercoaster each time they are notified about new GWAS associations with their SNPs. Concerns for the relatives who may be affected by genetic testing results, the vulnerable or those subject to surreptitious testing (testing people without their knowledge) and, in particular, children are all potentially difficult and can be viewed in juxtaposition to companies advertising fun for the family or purchasing

testing for others (whatever the motive) (Borry et al 2009, Kutz 2010, Udesky 2010, Vorhaus 2011).

Privacy is suggested as a positive attribute and is related to autonomy in relation to buying a genotype test online. However, breach of confidentiality in relation to individuals' genotyping data is problematic because potentially sensitive information used by third parties for discriminatory reasons may have significantly harmful effects. Despite the enactment of the Genetic Information Non-Discrimination act in the USA in 2009 (Conrad 2009), there are concerns about individuals' insurance and employment should DTCGT data get into the wrong hands. As I alluded to earlier in relation to the take-overs of deCODE genetics and Navigenics, privacy and confidentiality could be threatened when companies go bankrupt and the fate of customers' samples (if kept) and data is uncertain, regardless of the purchasers' country of domicile (Foster and Sharp 2008, etc Group 2008, Udesky 2010).

Anthropologist Mary Douglas suggested that shared classifications are central to moral order in human society and, without shared or commonly accepted understanding, disapproval is expressed and rituals are enacted to restore order (Davis 2008). The equivalents in respect of DTCGT are the debates for and against DTCGT published in the popular and professional media and the numerous calls for oversight of some kind to protect the public from harm. The history of overtures to policy and regulation of DTCGT are discussed next.

Regulation

The debate and activities related to policy and regulation of DTCGT illustrate the contingent and evolving nature of this technology. Their examination is particularly important in setting the scene for this study because of the interdependence of research into the social construction of technology and its influence on public technology policies (Williams and Edge 1996). Regulation of DTCGT needs to be understood in the context of a spectrum spanning those who view DTCGT as information which

neoliberal citizens have a right to access directly should they so choose, to those who view it through the same lens as clinical genetic testing and thus expect similar regulatory frameworks to apply for the protection of patients. Whilst some are anxious to avoid being seen as paternalistic, those at the regulation end of the spectrum call for government regulation of DTCGT companies, including control of the threshold of clinical utility of tests, laboratory quality standards, informed consent, test ordering and interpretation using healthcare professionals and protection of consumers from discrimination (Wallace 2008, Evans and Green 2009, Patch et al 2009, Wright et al 2011b).

A number of authors' calls for regulation refer to a lack or failure of oversight and refer to public requests for involvement of healthcare practitioners and government oversight of testing. However these are usually *vox pop* surveys rather than based on users' direct experience (Lenzer and Brownlee 2008, Boddington 2009). The more moderate view in relation to regulating DTCGT supports voluntary standards and ethical practice, the adoption of which would support companies' marketing power and potentially prevent less scrupulous companies from moving premises to another country where regulatory controls are less restrictive (Gurwitz and Bregman-Eschet 2009, Hauskeller 2011). Some question whether regulation is required at all (Russo 2006, Wright and Gregory-Jones 2010); others propose patience while the innovative nature of DTCGT companies' investments in personalised genetic medicine reap rewards in relation to the development of useful tools for users as is arguably emerging with the take-overs of deCODE Genetics and Navigenics in 2012.

The overall lack of regulation of DTCGT on both sides of the Atlantic illustrates the influence of agencies on the shaping of DTCGT and relates to the specific challenges DTCGT presents, which are summarised as follows. Globalisation of information and trade enabled by the Internet has vastly widened the scope of access to DTCGT and undermined the power of local legislation to enforce regulatory oversight across borders (Jordens et al 2009). The fast moving pace of change with biotechnologies associated with genomics and, thus, products on sale and the DTCGT

commercial landscape make the moving target of DTCGT difficult to define and legislate for. The lack of consensus about what DTCGT constitutes makes oversight difficult. Different groups view it as personal information, an educational or recreational resource, or a clinical diagnostic test and each would require a different approach in relation to any oversight, which is no doubt why the patchwork approach to any regulatory oversight persists. I will now go on to examine the principal events of regulatory activity that bring us to the current state of assorted approaches to regulatory oversight.

In the USA at federal level a number of factors contributed to a perceived lack of legal or political governance of DTCGT. The Food, Drug and Cosmetic Act (FDCA) prevents relevant companies from making false claims about and requires demonstration of the safety of their products. The FDA has responsibility for enforcement and this includes diagnostic medical devices; however, tests made by individual laboratories and marketed directly to consumers are not classified as medical devices but are regarded as “home brews” (Editorial NG 2002:553). Additionally, the CLIA requires specific certified analytical validity standards to be achieved by all testing on human tissue undertaken for diagnosis or treatment since 1988 (Fraker and Mazza 2010, McBride et al 2010), but it has no statutory or regulatory requirements for proof of clinical utility or clinical validity (HGC 2003). However, many US DTCGT companies apparently managed to avoid these requirements early in their evolution. This was largely due to the FDA loophole and because the nature of the tests offered was unclear as they were being sold directly to consumers rather than being administered by healthcare providers, and partly because the oversight requirements were rarely used (Vorhaus 2010b).

In 2006 the Genomics and Personalised Medicine Act commenced its development in Washington, later to become the Genetics Information Non-discrimination Act (GINA) proposed by then Senator Barrack Obama and signed into law by GW Bush in 2008 (Vorhaus 2010b). Early in GW Bush’s presidency, the Secretary’s Advisory Committee on Genetic Testing was reconstituted as the Secretary’s Advisory Committee on Genetics Health

and Society (SACGHS), which published its report into regulation and genetic testing in 2008. It highlighted the inconsistencies in oversight of existing FDA and CLIA requirements, the lack of understanding of the DTC tests available, the inconsistencies between tests offered and the dubious clinical utility of DTCGT. As a result, public health offices in New York and California sent Cease and Desist letters to DTCGT companies in their jurisdiction. Publication of the report resulted in more federal and state-level discussion about regulation and some increased surveillance and action at state level, which led to the closure of some companies and procurement of licenses to operate by others (notably the larger companies, 23andMe, deCODE genetics and Navigenics). Some companies (notably Navigenics) stopped selling directly to consumers but continued marketing DTC while selling via healthcare professionals or clinics (Vorhaus 2010a, Evans et al 2011).

In the UK, the Advisory Committee on Genetic Testing that was established in 1996 was subsumed into the Labour government's Human Genetics Commission (HGC) in 1999. In 2002 the HGC began a consultation into regulation of genetic testing. The report from the consultation considers the issues related to all genetic testing and makes recommendations for controls of genetic testing. These suggest prohibition of DTCGT because of concerns about lack of counselling and uncertain or poor clinical validity and utility of tests available (HGC 2003). In the same year the European *in vitro* diagnostic medical devices (IVDMD) directive regulating diagnostic tests came into effect in the UK, to be enforced by the Medicines and Healthcare products Regulatory Agency. However, the IVDMD directive is enforceable only within the UK and not on kits bought by the public from abroad, thus highlighting the difficulties of regulating tests sold on the Internet from other countries, as noted by both the HGC and the FDA (HGC 2003). Similar concerns were expressed in other Western countries but none were in a position to regulate tests bought by the public online from other countries.

In 2010 the GAO published a report into its second investigation into DTCGT companies (Kutz 2010). The report was the result of a two-part

investigation into DTCGT and revealed genuinely worrying detail about some personal genomics companies' practices. Notably, the initial investigation highlighted companies' use of misleading marketing information, the lack of clinical validity and reliability of tests (by submitting samples from fictitious people and obtaining different results for the same samples) and results that differed from the individuals' phenotypes (as with Steven Pinker). In the second part of the investigation undercover GAO staff contacted companies assuming the role of customers and the resulting report exemplified conversations in which advice about the deterministic nature of tests for disease risk was supported by a company representative, in one instance, asserting that the customer's test result put them at high risk for developing breast cancer. In another instance, a caller was encouraged to submit her boyfriend's sample for testing surreptitiously, in order to give him a surprise gift. However, the effect of this sensationalist report was to backfire on the GAO's presumed intention to force legislation to regulate the industry, because its inconsistent approach and unrepresentative content (no companies were identified with specific examples of poor practice) rendered it confusing and ineffectual for informing regulation (Vorhaus 2010b, Ray 2011).

Publication of the report prompted a flurry of activity in political and legal circles, in commercial genomics companies and in blogs posted by those with interests in DTCGT. An important milestone was the congressional meeting with representatives of the main stakeholders (companies, the FDA, the GAO and congress), in which the deficiencies in the GAO report were revealed, as it resulted in confusion and inappropriate assignment of all DTCGT companies' practices to the lowest common denominator of the reports unidentified examples. However, the FDA then issued letters to individual DTCGT companies inviting private discussions to clarify the status of the companies' products in relation to requirements for FDA approval. As a result, companies took various approaches to either changing the products to reduce the disease trait testing or only providing tests at a doctor's request. Notably 23andMe continued to trade as before, promoting its democratic and altruistic philosophy to attract customers. However, despite this, no change in the federal regulation of DTCGT ensued at that time (Jostins and MacArthur 2011). Subsequently, the FDA

issued 23andMe with a further Cease and Desist letter in 2013. In their view this was provoked by 23andMe's failure to engage with the Administration's process for regulation. As a result, the company stopped providing health reports to new customers in the USA and instead set up off-shore bases from which to sell testing to residents of other countries not affected by the FDA's jurisdiction, as described earlier in this chapter.

In the UK, the House of Lords Science and Technology Committee reported on DTCGT in 2009 and suggested that the HGC draw up principles for practice, which, due to the perceived low risk of DTCGT under the European IVMDD, could be voluntary (House of Lords 2009). Having published "More Genes Direct" in 2007 to update information on developments in the DTCGT field (HGC 2007), the Commission then published its "Common Framework of Principles for DTCGT Services", with the aim of providing a code of practice from which companies could trade and practice could be monitored when and if regulation was enforced. Covering all types of DTCGT, the Framework proposed practice standards in relation to transparency in marketing, informed consent and counselling, test and laboratory standards, test interpretation and data protection. However, the Framework seemed to ignore the potential for the Internet to provide DTCGT to the public across national (and jurisdictional) boundaries (HGC 2010). Like the SACGHS, the HGC was disbanded following a change of government and it is not apparent that their Framework has been adopted at national level in the UK or anywhere else. UK nationals are still at liberty to buy tests online without these principles being adhered to. In contrast, the Nuffield Council on Bioethics suggested that more research into the effects of DTCGT is needed before decisions are made, as supported by Prainsack et al 2008, Khoury et al 2009 and Leighton et al 2012 (Nuffield Council on Bioethics 2010).

In the EU in 2008 the Council of Europe produced an additional protocol to the Convention on Human Rights, which essentially advised that genetic tests must be administered under medical care and with counselling (Council of Europe 2008). This could have significant implications for DTCGT in Europe, except that the Protocol is essentially not binding in Europe unless member states sign up to it, which a number have not,

including the UK (Borry 2008, Kaye 2008). In 2009 Germany passed national legislation limiting the return of genetic tests results to physicians, preferably with counselling, although it appears that this is only applicable to tests bought from German companies within Germany's geographical jurisdiction (Vayena and Prainsack 2013). Similar legislation exists in France and Switzerland but it is not clear how this is enforced with the online market (Borry et al 2010, Skirton et al 2012).

Guidance published by various professional medical associations on DTCGT calls for some level of oversight and the involvement of healthcare professionals at some level, from discussion about testing to ordering and interpreting tests (Skirton et al 2012). The basis for these proposals, which are supported by others in ELSI academic circles, appears to be the association of DTCGT with clinical genetic testing and potentially diagnostic properties and the need to protect the public from the attendant possible harms, which they are thought to be unaware of. This much is evident from the statement released by the Royal College of General Practitioners when 23andMe started to sell tests in a UK health and beauty retailer's shops (Meikle 2015). However, the need for protection is not borne out by research findings. A small number of US-based studies have established that knowledge about genetic risk for disease appears not to alter behaviour, beliefs or perceptions of control, or cause distress to individuals in the medium to long term as will be discussed further in Chapters 4-6 (Heschka et al 2008, Marteau et al 2010, Bloss et al 2011, Collins et al 2011).

DTCGT can be sold in the UK at the time of writing, owing to the testing being undertaken in a non-EU country and the kit not being classed as a medical device under the current EU directive for IVDMD; in addition, the collection device itself is marked with the Conformité Européenne (CE) approval symbol (Burton 2015). However, the IVDMD directive is being revised to a regulation that is unlikely to permit loose interpretations that have followed from scientific and technological developments since the directive was first produced. This may restrict the sale of genetic tests direct-to-consumer, but the 2012 draft regulation is still under negotiation at the European Council (GeneWatchUK 2015).

In contrast to the professional view, the little that is established about the public view indicates that DTCGT is generally viewed in relation to its *personal* utility, as individual information that is interesting, potentially useful and the consumer's right to be able to acquire without imposed restrictions (Vorhaus and MacArthur 2010, Gordon et al 2012). These two juxtaposed views partially illustrate the foundations for the on-going uncertainty in relation to regulation of DTCGT.

Conclusion

Whilst DTCGT has emerged from research into human genomics and has a basis in biological science similar to more conventional genetic testing, it is difficult to reduce it simply to the procedure of SNP analysis. This is in common with clinical genetic testing, because the nature of any genetic knowledge is wider than just its biological basis due to the effect that knowledge potentially has on people in terms of their health risk management, their decision-making and the implications for their families (Featherstone and Atkinson 2012). However, companies market testing as providing vital knowledge to consumers and this sets up tensions between the public who use it and HCPs, who are sceptical of its value and concerned about the possible harms that knowledge might cause. Furthermore, there is the possibility of a potential drain on resources by an increase in referrals to clinical genetics services for counselling for DTCGT users. Given that these referrals result from private individual testing transactions and are additional to the current workload plans of NHS clinical genetics services, they have the potential to disrupt services and the trust-based relationship between the public and their HCPs as the two groups' expectations diverge.

Research conducted into DTCGT to date fails to support the concerns about harm to users or significant additional impact on healthcare services that have been emphasized in professional and academic literature since personalised genomics became available. Weblog posts support McGowan's and colleagues' findings that most users are educated, informed individuals who are curious and unlikely to be surprised by test

results or to regard them as deterministic (McGowan et al 2010, Bloss et al 2011, Collins et al 2011). There is little empirical evidence of the impact on genetics clinicians, though much of the literature implies a lack of adequate knowledge about genetics or genomics in most general or family practitioners (Jordens et al 2009, Ormond 2009).

Almost all the empirical work published to date reflects either provisional studies (research conducted on groups who have not actually engaged with DTCGT but are presented with possible scenarios) or is focused on North America. In the face of this notable lack of research I feel it is important to examine the implications of personalised medicine from the perspectives of people engaged in obtaining genomic screening for disease risk in the UK and genetics clinicians who may be called upon to help them with the results. The next chapter will discuss the methodology and methods I have chosen for this study in order to elicit how the discourse about DTCGT is being socially constructed and shaped in the UK context.

Chapter 3 : Theoretical and methodological approaches to the sociological study of genetics

Genetic testing has long been researched by sociologists. In this chapter I shall review the sociological research literature on genetic testing to show how sociologists have investigated genetic testing previously, in order to situate and justify my study design. I shall relate the overarching theoretical and methodological concepts of my study to that literature and the work of STS scholars that has informed my use of SCOT as a conceptual framework. I shall complete the chapter with details of how the study was performed.

Researching genetic testing

Informative research into people's responses to predictive testing for genetic disease includes a number of quantitative studies that attempt to measure individuals' psychological responses to testing. Psychologists Theresa Marteau and Susan Michie and colleagues have conducted numerous survey-based studies to ascertain what psychological effects result from suspecting a familial genetic illness and being tested (or not) to establish genetic status (Michie et al 1997a, Michie et al 2001, Michie et al 2002). These studies employ structured interviews and some include Quality of Life measurement questionnaires to assess psychological status, such as the Spielberger State-Trait Anxiety Inventory (Marteau and Bekker 1992). Non-directiveness of genetic counsellors has also been examined from a quantitative perspective by recording consultations and questioning counsellors and consultands before and after consultations, using structured interview schedules or questionnaires (Michie et al 1997b). These studies are of relevance here because, whilst they are based on a realist perspective, their empirical work contributes to understanding people's experiences of genetic testing and counselling.

One of the earliest research projects into the impact of genetic susceptibility testing on perceptions of disease risk was part of the multi-centre Risk Evaluation and Education for Alzheimer's Disease (REVEAL) study. One element of the REVEAL project was a randomised controlled trial that sought to establish participants' (adult children of people with Alzheimer's disease) perceptions of their risk of developing Alzheimer's disease after having had their risk estimated (Roberts et al 2005). The intervention arm of the study had disease risk estimated with inclusion of Apolipoprotein E genotype, unlike the control arm whose status was calculated on age, sex and family history only. The *APOE4* variant of the gene is thought to contribute approximately 50% to the genetic risk of developing Alzheimer's disease (Farrer et al 1997). Findings concluded that perceptions of risk are influenced by genotyping in those who tested *APOE4* negative, but not those who tested positive (Marteau et al 2005).

Continuing the quantitative theme and directly related to DTCGT (although not strictly 'sociological' research), Howard and Borry sent questionnaires to genetics clinicians in the EU to investigate clinical geneticists awareness of, and attitudes towards commercial genomics. Perhaps unsurprisingly, their survey found that this group of clinicians feel that it is inappropriate to conduct this kind of susceptibility testing without counselling (Howard and Borry 2013). Their work shares this approach with a number of studies of genetics clinicians and potential early adopters in the UK and North America conducted by bioethicists and healthcare researchers aiming to access disparate and relatively unfamiliar groups (Cherkas et al 2010, Gray et al 2009, Giovanni et al 2010, Kauffman et al 2012, Leighton et al 2012, McBride et al 2009, McGuire et al 2009).

However, survey research could be judged as being sterile, in the sense that it de-contextualises its data and the subjects who provide it from the social aspects of life that inform them, as Wynne suggested (Wynne 1992). Howard's and Borry's work provides a good example of this. While their findings could be said grossly to mirror those from my study, their pragmatic approach to obtaining European-wide data with a survey is unable to obtain the in-depth nuanced views about the social aspects of clinicians' views that this study has done by interviewing UK-practising

clinicians. In a similar vein, RCTs in health-related research could be said to provide data from a context in which it is so manipulated (in order to achieve the requisite control of variables) as to render it meaningless in the context of the populations for whom it is likely to be relevant (Rothwell 2005). Thus, much of the sociological research into genetic testing adopts a qualitative, constructivist model in order to elicit the contextual, social aspects of people's experience.

I adopt a constructivist approach in common with many well-known sociologists who, whilst acknowledging the existence of physical reality in the shape of biological or medical knowledge, emphasise the importance of studying people's multiple and socially organised responses to reality (Barnes 1982, Nicholson and McLaughlin 1987, Atkinson 1995). Anne Kerr notes that constructivism enables consideration of how people's social worlds influence their engagement with technology in general and genetic testing technologies in particular (Kerr 2004). Constructing understanding of how their knowledge and experiences shape their responses to new technologies and thus the technologies themselves, will be covered next.

The Social Construction of Technology

The broad design of this study is to take a constructivist approach to understand how the new technology of commercially obtained SNP genotyping is being shaped by the discourses and actions of principal groups of actors engaged with it in the UK. Discourses in this context are understood as practices of sharing and developing beliefs and understanding through discussion, evaluation, interpretation and sense-making with the purpose of producing knowledge about this technology (Lessa 2006). Rather than taking a realist position and researching the physical reality of SNP genotyping and the potential implications for disease or trait phenotype, which undeniably exist, I am instead seeking to construct knowledge about the social processes that influence individuals in their uptake or dismissal of this emerging technology in the UK. In other words, I am viewing SNP genotyping as a whole system, or assemblage, that is socially constructed and shaped by the beliefs, interpretations, talk and practices of the actors involved with it (MacKenzie and Wajcman 1999),

rather than simply as a laboratory test. Consequently it seems appropriate to use the SCOT framework to analyse how this emerging technology is being shaped in the UK context. I shall now examine the developments in STS that informed the development of technology studies generally and then examine the development of the SCOT framework in particular, considering relevant examples in order to justify its use in this study.

The study of scientific knowledge (SSK) with its characteristics of impartiality and symmetry towards both truth and falsity in scientific beliefs emerged in sociology in the second half of the 20th century. The application of the principles of symmetry and impartiality were the basis for both Bloor's and Collins' respective programmes of work in the Strong programme and the Empirical Programme of Relativism (Collins 1981, Bloor 1991). In a rejection of both technological determinism and linear models of technological development that was in keeping with the developing work in science studies, it was proposed that SSK principles are just as applicable to studies of technology, given that both scientific and technological knowledge claims are socially embedded (Pinch and Bijker 1984, MacKenzie and Wajcman 1999). Several approaches to the study of the relationship between technology development and society have been proposed including the Social Shaping of Technology (MacKenzie and Wajcman 1991, Williams and Edge 1996), Actor Network Theory (Latour 2005) and the Social Construction of Technology (Pinch and Bijker 1984).

The Social Shaping of Technology approach assumes mutual shaping of society and technology (MacKenzie and Wajcman 1999); pivotal decisions at certain points during development of a technology influence which paths are subsequently followed as a result. This approach to technology studies was used by MacKenzie in his study of nuclear missile guidance technology in which he demonstrated how the technology's accuracy was influenced by organisational and political factors rather than simply scientific or technological ones. MacKenzie suggested that the relationship of certainty about a technology's capabilities is aligned to people's understanding and proximity to it. Those who are directly involved in its development are less certain of its capabilities knowing its potential failings, those who are institutionally aligned to it or may use it are more certain of it whilst those whose loyalties are elsewhere are likely to be uncertain about

it (MacKenzie 1990). Whilst MacKenzie's work on nuclear missiles guidance technology does not follow the SCOT framework *per se* it is relevant here because the relationship of proximity to technology and certainty in its capabilities can be applied to this study's participants as I shall go on to illustrate in the section on expectations.

Pinch and Bijker outlined their Social Construction of Technology framework advocating that both scientific and technological artefacts are social constructs that are influenced by social groups whose members influence the development and stabilisation of technology. As a direct result of the influence of SSK, they advocate that symmetry is the core principle in analysing technological developments, that both successes and failures should be examined in the same way so as not to privilege either or to draw conclusions about success or failure based on whether or not a technology works. This principle of symmetry is applied across the four main concepts that make up the SCOT framework

- interpretative flexibility,
- relevant social groups,
- closure and stabilisation
- the wider social context. (Pinch and Bijker 1987)

Important sociological studies of technology have included Pinch and Bijker's well-known study of the bicycle and its evolution from early beginnings as the arguably dangerous Penny Farthing design favoured by speed-loving young men, to the more comfortable and stable version favoured by women, the basis of which design persists today. Their study of this evolution, whilst arguably not all-encompassing of the bicycle's long history starting as it does at the end of the 19th century, does examine the relevant social groups' influence on changes in design of the technology by dint of their interests in speed, safety, comfort and practicality (Pinch and Bijker 1994). More recently Paul Rosen extended Pinch and Bijker's work with an examination of mountain bikes, to illustrate his critique of SCOT. He cites Pinch and Bijker's lack of emphasis on the wider social context and its impact on technology development as problematic, as is their lack of detailed consideration of the membership and relative influences of the relevant social groups (Rosen 1993).

Similarly Klein and Kleinman's (2002) critique of SCOT focuses on the contrast between using an agency-focused approach rather than including examination of structural concepts in analysing technological development. Their concerns relate to assumptions that the characteristics of relevant social groups are homogenous and equivalent in their membership, rather than examining group membership for their capacity to shape technology owing to their political, professional or economic power. This concern is certainly relevant to this study where the relevant social groups being studied in relation to DTCGT have origins and particular characteristics that influence their discourse and thus influence the disputes about DTCGT and its uptake, as the data will demonstrate.

The SCOT approach facilitates productive investigation into the "black box" of genetics, using the phrase in the sense of "opacity" that Lynch et al (2010:17) refer to. SCOT can help develop understanding of how the relevant social groups of users and clinicians engage with genetic information in different and competing ways for social, economic, regulatory and professional reasons. Analysing these relevant social groups' use of interpretative flexibility and sense-making practices, allows these differences and their implications to be illuminated and objectively understood, as does the influence of the wider social context of the provision of health care in the UK's state-funded collective medical service. This understanding will illustrate the influence of human intervention on the current path that SNP genotyping is taking in the UK rather than assuming a technologically deterministic position that would absolve people from any responsibility in their engagement with it (Wyatt 2008). It will also illuminate the factors that are sustaining the disputes between the relevant social groups, or those that may offer opportunities for stabilisation of the technology in the resolution of the disputes and acceptance (or loss) of the technology. Given the potential impact of genetic information on people and their social networks, however that information is obtained, an exploration of the social construction of this technology seems the most pertinent approach to understanding DTCGT, and thus its impact in the UK.

Pinch and Bijker suggest that new technologies have different meanings for the different actors involved with them, citing users and producers as the key stakeholders involved (Pinch and Bijker 1987). Bijker adds to this by suggesting that the “technological frame” (Bijker 2010:69) is a group’s shared understanding of a technology is built by their shared experiences of knowledge or use of the technology. It can describe people’s actions and constructions of the technology in the sense of their knowledge about it, encouraging some actions or beliefs and discouraging others (Klein and Kleinman 2002). With this in mind it is an obvious decision to involve users of the technology in the UK as they will have direct experiential knowledge of DTCGT and thus some shared understanding that they associate with DTCGT (Pinch and Bijker 1984). In addition to including users, my decision to involve genetics clinicians was informed by evidence from the literature that this group would have a competing view of DTCGT compared with either users or producers of the product. In addition, being viewed as experts in human genetics, genetics clinicians may be called upon to interpret results and counsel patients in the event of any difficulty. This is because in the wider context of healthcare provision in the UK, genetics clinicians are the providers of professional advice and support about genetic conditions. Given that both users and clinicians constitute social groups for whom DTCGT has relevance, I felt it important to investigate each group’s understandings and contribution to discourses about the technology and the controversies that surround it, using SCOT as an interpretative framework. Representatives of these two groups recruited for this study, demonstrate power relations between groups and their competing discourses; they also illustrate the inter-group conflict, different technological frames and their potential influence on DTCGT. This more political understanding about how DTCGT is being shaped aligns with Klein and Kleinman’s critical review of SCOT and their suggestion that a more “structural” approach to analysing social groups and the wider context of technological development is required for SCOT to be comprehensive (Klein and Kleinman 2002:29). A more detailed description of the members of these groups is given in the Methods section later in this chapter, in accordance with Pinch and Bijker’s assertion that detailed descriptions of the relevant social groups are an important part of the analysis (Pinch and Bijker 1984).

Given that relevant social groups constitute the “unit of analysis” in SCOT (Oudshoorn and Pinch 2005), it is appropriate that this informs the theoretical approach of this study’s design, focusing as it does on the groups of users and clinicians. Having discussed the relevant social groups for the study, and described the importance of the wider social context of the NHS I now wish to explore the remaining concepts that have an important bearing on this study. These include taking a symmetrical approach to examining different actors’ contributions to shaping discourse about DTCGT, examining the interpretative flexibility applied to DTCGT, considering how expectations have contributed to shaping the technology, and adopting a reflexive approach. The relevance of each of these tools to this study and their importance in STS and sociological research in genetics warrants specific consideration, which I shall cover in the next section.

Symmetry

As I suggested earlier, the Strong Programme in the study of scientific knowledge is underpinned by the contention that all knowledge should be treated equally by sociologists (Bloor 1991). Whilst agreeing with Bloor’s principle of symmetry, Collins also notes that, where a controversy exists, the sociologist’s role is to observe the different aspects of the debate objectively and with symmetry in order to analyse it. No other approach is possible, as we (sociologists) have no way of knowing what the truth or point of settlement will prove to be, any more than those actors involved in the controversy have (Collins 1981). In applying this principle to the study of technology, this is certainly true in DTCGT, as the on-going debate about its advantages and risks attests. Both groups in this study have distinctive views about DTCGT and, by taking a symmetrical approach to the principal actors’ claims about DTCGT, I am able to observe and analyse the uptake of DTCGT in the UK context not in relation to its success or failure, but rather in considering how the relevant social groups view the technology and promote or undermine its success as a result.

This principle of symmetry has been a crucial tool in this study, by giving equal representation to groups with different, sometimes opposing, views and different types of influence in their respective spheres. Application of

symmetry has also confirmed my position as a researcher, which is not to know how the debate will be closed or what the *truth* about SNP genotyping is. I am not in a position to know if the science is flawed, whether clinicians are wrong or users are right, or vice versa. My role is to note their views and experiences in a symmetrical manner. Consequently this study has evolved into a very different one from the bioethical study that I first envisaged. This is as a result of the application of both symmetry and the related tool, interpretative flexibility, which I shall discuss next.

Interpretative flexibility

In conjunction with the use of symmetry to approach different actors interpretations of genotyping, observing how they use interpretative flexibility is critical to understanding how different views of science and technology influence debates about the technology. Potter and Mulkay refer to interpretative flexibility in relation to interviewing scientists about scientific theory; scientists interpret theory differently according to their audience, and what they are trying to achieve by articulating their thoughts at any given time. Variation in accounting can be used as an analytical resource for making sense of persistent disagreements when people fail to adopt a theory that is seen as “correct” (Potter and Mulkay 1985). Also, different interpretations of technology can influence the technology’s design and future use as Pinch and Bijker demonstrate with the example of the bicycle (Pinch and Bijker 1987).

Williams and Edge have criticised SCOT suggesting that it is difficult to explain how disputes about technologies achieve closure (or stabilisation) using interpretative flexibility owing to the infinite possibilities that could be put forward (Williams and Edge 1996). However, whilst I will go on to argue that DTCGT is not yet stabilised in the UK context, the concept of interpretative flexibility is crucial in this study’s data as it is clearly demonstrated in the inconsistent and arguably self-contradictory views expressed by many of the participants in both groups, as well as the overall differences in beliefs about DTCGT between groups. I will show how this points to the on-going nature of the disputes about DTCGT in the UK. Interpretative flexibility is also an important principle to apply in examining

the debate about genotyping and GWAS, in examining how these technologies are variably interpreted in the different arenas in which they are debated, and in relation to the varied interpretations of SNP genotyping different companies produce. Martin Richards notes this particular aspect in his autoethnography of DTCGT, in which he received different interpretations of his SNP genotyping from each of the companies he sent his samples to (Richards 2010).

While people's views of a new technology or theory may be contingent and variable, depending on their proximity to it as researchers, scientists or adopters, they are likely to use interpretative flexibility to articulate variable expectations of the new theory or technology, depending on whether their audience is fellow research scientists or outsiders (Borup et al 2006). Using interpretative flexibility to influence expectations and the uptake of genotyping has been fundamental in DTCGT and it is to the sociology of expectations that I turn next.

Expectations

Expectations have been shown to play a central role in creating and shaping biotechnologies (Bijker 1997, Brown et al 2000, Brown and Michael 2003, Hedgecoe and Martin 2003, Borup et al 2006, Groves and Tutton 2013). They lend a technological innovation structure and legitimacy, driving its design and re-design and clarifying the roles of those involved in its development. "Enactors", as Groves and Tutton refer to those creating the technology and expectations of it (Groves and Tutton 2013:182), or the "core-set" of scientists (Collins and Evans 2002:242) use interpretative flexibility to articulate expectations of their new technology depending on whom their audience is comprised of (Borup et al 2006). In personalised genomics this flexibility hides uncertainties about the technology.

Expectations evolve over time and alternate between hype and disappointment. This happens in the course of their being tested in arenas between their enactors and those who decide if and how technologies can be reified, referred to as "selectors" (Groves and Tutton 2013:182). Expectations of DTCGT are being tested by selectors in relation to the

ethical challenges it poses, its differences in approach compared with the clinical genetics model of testing and its technological contingencies. As a result, companies' products and the expectations that partner them have adapted and changed. The regulatory landscape has seen considerable activity as companies have sought to establish themselves or adapted their businesses to circumvent local regulatory restraints, as evidenced by 23andMe developing country-specific products for Canada and the UK in 2014 (Picard 2014, Gibbs 2014).

Examination of the sociology of expectations has helped me to understand early adopters' experiences of DTCGT as well as reactions from clinicians. Because consumers have little use for uncertainty, people need to be convinced by hope rather than dissuaded by truth, given that hope maximises possibilities while truth minimises them (Groves and Tutton 2013). Being 'further' from a technology decreases uncertainty about it, as I indicated earlier when referring to Mackenzie's certainty trough (Mackenzie 1990). MacKenzie suggests this may be an explanation for users' engagement with technology and this could be applied to the users of DTCGT in this study. Genetics clinicians, whilst knowledgeable about genetics are likely to be more uncertain about the technology's capabilities because they are not directly involved in its development or use and have competing interests in technologies used in clinical genetics around which they engage in boundary work. However, many early adopters of DTCGT are scientists knowledgeable about genomics. This could be because the wider scientific community only becomes privy to the detailed scientific knowledge about the technology late in the process and so only sees the less uncertain aspects of the technological innovation. Widespread publications about DTCGT have raised awareness about genotyping among those who are interested in genetics and represent the emergence of the kind of network that works to reduce uncertainty. For clinicians, scientists' interpretative flexibility may be at work in terms of presenting more moderate expectations.

Reflexivity

The problem of reflexivity in STS has been discussed by many but possibly most engagingly captured by Malcolm Ashmore. The thesis of his

thesis cleverly explores the regress induced by taking a reflexive stance in studying the sociology of science and technology, shedding light on the approaches the principal scholars in the field have taken (Ashmore 1989). Whilst also acknowledging its limitations, others stress the importance of reflexivity - that is researchers' transparency about their views and attitudes in relation to the research topic - when researching the implications of the new genetics for the public and genetics professionals (Kerr et al 1997, 1998).

I feel that it is important to consider it here in relation to my own journey into STS. Having come from a healthcare background, I am acutely aware that my views about science generally and genomics in particular have shifted radically over the period of this study. This is as a direct result of employing a symmetrical approach to studying the actors involved in DTCGT and applying this to my own views. As a healthcare professional my opinions about genetic testing were characteristically asymmetrical and partial but have altered over the last four years as I have become a sociologist. Adopting a relativist position towards science and technology has enabled me to be less judgemental which, in addition to the obvious advantages of adopting a more open perspective, has facilitated much wider learning and understanding than I could have previously imagined.

My awareness of this and the opportunity to apply the principles of symmetry and interpretative flexibility in action were brought into sharp focus by my experience with the geneticists at Lübeck that I recount at the beginning of the thesis. I feel it is important to acknowledge this change in order to provide some transparency in relation to my role and learning during the conduct of this study and the construction of its findings. My intention here is to align my attempt to be reflexive with Bourdieu. I am attempting to pay conscious attention to the effects of my own views in order not to attribute them to my participants or other actors involved in the study. I attempt to do this while simultaneously observing and incorporating my observations about these actors and their contributions to the study, either directly as participants or indirectly in influencing my thinking along the way (Bourdieu and Wacquant 1992).

Having considered the theoretical concepts that have informed my overall approach to this study I shall go on to consider the methodological aspects of the study design. By reviewing relevant sociological research in genetic testing I will justify the methods used for this study, showing how this approach has been successfully exploited in related research into genetic testing.

Ethnography and interviews

Prompted by the unprecedented developments in genetic research and diagnosis, many sociological and anthropological studies of genetic testing over the last half century have been undertaken, most of which adopt ethnographic approaches. This is apposite, given the importance of people *and* culture to these studies. In researching the work of genetic counsellors in an American paediatric hospital, Bosk gives a compelling justification for an ethnographic approach. He demonstrates how being present in the field facilitates the observation and description of everyday actions and understandings to expose the tensions and power relations that shape new genetic technology and practice (Bosk 1992). Feminist anthropologist Rayna Rapp conducted an impressively detailed and in-depth ethnography of women undergoing amniocentesis at three different centres in New York in the 1990s. Her ethnography explored what was then a novel genetic technology and involved consideration of the technological transformation of pregnancy, reproductive and disability rights and scientific literacy in North American culture at that time (Rapp 2000). Mike Lynch's and colleagues' ethnographic work on the history and developments of forensic genetics reveals society's investment in a deterministic interpretation of this aspect of genetics, despite its contingent foundations (Lynch et al 2010). All these researchers provide revealing experiences achieved through encountering and interviewing the different actors involved and making detailed observations of everyday practices and interactions in laboratories, clinics, mortuaries and offices. These rich and varied data provide powerful arguments for ethnographic research in clinical or forensic genetics, not least due to their challenging accounts and interpretations of practice in genetics made possible only by their immersion in the respective fields of enquiry.

Ethnography requires the immersion of the ethnographer in a social group for a period of time to observe behaviour and communication and interview group members (Hammersely and Atkinson 1995), also known as conducting fieldwork. Scott and Marshall describe fieldwork as any study that involves conversation with people, including asking them questions, whilst in a similar, if anthropological, vein Rabinow suggests that everything constitutes fieldwork (Scott and Marshall 2009, Rabinow 2007). However, despite these arguably flexible interpretations of ethnography, an ethnographic approach to studying actors involved in DTCGT would be very difficult to achieve. This is because “the field” in relation to DTCGT in the UK, in the sense that Goffman understands “the field”, is a difficult concept to define (Goffman 1989). In the context of DTCGT the field is an unknown territory; users engage with this technology using a computer to access the Internet at unknown and potentially multiple sites. Their communications with each other about testing are conducted in multiple sites, including the workplace, recreational group meetings and, more commonly, online communities, as user participants reported to me. For genetics clinicians, there are very few (if any) geographical locations where they meet to explore personal genomics as a group and consultations with users of DTCGT are few and far between. Clinicians explained that they usually heard about DTCGT through their own reading (also usually in an online environment) or at seminars or conferences when the experience was didactic and short-lived, as opposed to one of interaction in the field. So, in seeking to investigate actors’ understandings and contributions to shaping the discourse about DTCGT technology and the controversies that surround it, the most obvious approach was to simply ask them. Accordingly, I decided to conduct interviews with purposively sampled participants from groups of users and genetics clinicians. I shall go on to justify my choice of interviews as the method of data collection for this study by considering other researchers’ related work in this area of STS and medical sociology.

In her exploration of the sociology of genetic disease, Anne Kerr rationalises the importance of interviewing as a method for constructivist research into the sociology of science and technology. She observes that giving the public a voice equal to that of perceived experts in matters of science and genetics, illuminates their understanding more effectively

than conventional research methods eliciting public opinion (Kerr 2004). Conducted in collaboration with Sarah Cunningham-Burley and Amanda Amos, Kerr's earlier research into public and expert views of the new genetics adopts both interviews and focus groups. The latter are used to generate breadth and depth in discussing hypothetical scenarios relevant to new genetic technologies that might not evolve in a one-to-one interview discussion (Kerr et al 1998). However, interviews are specifically adopted in the parallel study with experts in order that participants' views may be individually expressed and related to what the individual feels is important (Kerr et al 1997).

Hedgecoe's research into pharmacogenomics draws on data from interviews with clinicians and researchers working on Alzheimer's disease, breast cancer and drug regulation and development related to these diseases. He situates his methodological approach within the STS research tradition, using interviews to privilege stories from groups such as clinicians, whose views are often ignored despite their ultimately fundamental role in the uptake of pharmacogenomics technologies. With this approach he is able to explicate the way pharmacogenetics is being shaped by clinicians and researchers, rather than users simply accepting the hype raised by those who seek to reify its expectations (Hedgecoe 2004).

Sociological research into people's understanding and experiences of genetic testing has relied on interviewing as a method for accessing the views of those potentially or actually affected by inherited diseases. Hallowell and colleagues have researched the effects of *BRCA1/2* mutations on women and men in several studies, all using interviews as the primary method of investigation. These interdisciplinary studies involving sociologists and healthcare professionals have been able to show how people and their relatives perceive risk and responsibility in relation to their potential risk of breast or prostate cancer (Hallowell 1999, Hallowell et al 2004), as well as facilitating participants' reconciliation with their genetic identity through narrative reconstruction (Hallowell et al 2006). A study into polycystic kidney disease, a less familiar but relatively common genetic condition, used interviews to establish the extent to

which this condition is “geneticized” by sufferers, their relatives and the clinicians who care for them (Cox and Starzomski 2004:137).

Research into genetic susceptibility testing also uses interviews to advantage in eliciting people’s ideas about the difference in implications of susceptibility testing from predictive testing, or their experiences of the former. Interviews conducted with participants in the REVEAL study, following their intervention for the RCT, demonstrated that genotyping did not substantially alter people’s embodied identity or lifestyle. Interestingly, it also indicated that these people felt that family history was a much more effective indicator of disease susceptibility than genotyping (Lock et al 2007, Chilibeck et al 2011). People interviewed after genotyping for susceptibility to deep vein thrombosis mostly thought that the test was useful for making alterations to drug regimens that might add to their risk of thrombosis. However, they did not change their lifestyle in any other way or view the tests as providing information that indicated counselling would be helpful (Saukko et al 2006).

In a smaller study, Harvey explored how clinicians in three professional groups responded to emerging genomics technologies implicated in type 2 diabetes mellitus. As in Hedgecoe’s work on pharmacogenomics, this interview study was able to show how practitioners contribute to the “mutual reshaping” of genomics technologies in the clinic (Hedgecoe 2008, Harvey 2011:309). In two similarly small studies, the first empirical work on DTCGT users and clinicians, experiences of early adopters of DTCGT in the US were investigated using interviews, as discussed in Chapter Two (McGowan et al 2010). The same researchers recently followed this with a similar study in which they interviewed clinicians about their use of genotyping in their clinical practice. These doctors have been re-positioned as the recipients of many US commercial genomics companies’ marketing and sales strategies following the concerns expressed about marketing directly to consumers. Despite these concerns, the study demonstrates that the clinicians involved rely largely on the commercial companies they contract with for understanding and interpreting genomic data. They describe having little additional, objective knowledge or judgement in relation to the genomic data provided (McGowan et al 2014).

As this review of sociological and related research into genetic testing demonstrates, interviewing is a method of long and commendable standing in STS and medical sociology. Potter and Mulkay stress this point in justifying its importance as an approach to illuminating scientists' interpretative practices (Potter and Mulkay 1985). In my study, interviews give both users and clinicians equal opportunity and space within which to construct their individual experiences of DTCGT. Consequently, it privileges the account of neither group in doing so, which is important for reasons of symmetry. Secondly, given the lack of a physical field in which to observe practices and communication, triangulation of interview data with participant observation data was neither feasible nor appropriate for this study.

Interviews as accounts and conversations

In seeking to construct knowledge about people's influence on discourse about personal genomic technology in the UK, I am interested in their ideas, their beliefs, the meanings they attach to these, their understandings about the sources that informed them and how they talk about these. My intention is not to establish what they do in relation to the technology. This has been documented elsewhere in various media, such as academic papers including auto-ethnographies, YouTube videos, company websites and the like (Richards 2010, Harris et al 2013, Harris et al 2014). Moreover, numerous sociologists have warned against conflating interview talk with representations of participants' actions, instead suggesting that interviews allow constructions and *interpretations* of their social action (Potter and Mulkay 1985, Atkinson and Coffey 2002, Jerolmack and Khan 2014). Thus, the talk in interviews can be seen to construct knowledge through the accounts and narratives offered. These narratives enable participants to employ an approach to discourse that accounts for their actions (Scott and Lyman 1968:46, Arribas-Ayllon et al 2011a). Garfinkel similarly suggests that people account for behaviour in certain social situations with descriptions or explanations of particular attributes displayed in those situations (Garfinkel 1967). Atkinson and Delamont stress the importance of rigour in relation to the use of accounts or narratives in qualitative research; that is, to avoid privileging personal accounts over other forms of data and to recognise storied aspects of

qualitative data as constructions that are culturally shaped (Atkinson and Delamont 2006).

Recognising participants' accounts as culturally shaped performances that retrospectively construct experiences and interpretations indicates the need to analyse the data thematically taking into consideration how participants talk as well as what they talk about (Holstein and Gubrium 2004). Data need to be carefully analysed for topics and in relation to the participants' rhetorical approaches, such as justifying or excusing their decisions or actions (Scott and Lyman 1968, Atkinson and Coffey 2002). In common with researchers in STS and medical sociology, whose work has been discussed earlier, my approach to analysis in this study is not only to examine the data for themes in terms of content but also to consider participants' discourse, including their adoption of rhetorical styles that denote the use of justifications or excuses. Establishing inconsistencies and variation in accounts is also important, as these contribute to participants' flexible interpretative practices in their construction of meaning about emerging technology (Potter and Mulkey 1985).

Having situated the broad design for my study within the context of an examination of related STS and sociological research literature, in the second half of this chapter I shall provide detail about the recruitment of participants, how the interviews were conducted and how the data were analysed.

Producing the study: notes on methods

In the second part of this chapter, I shall discuss the steps taken to perform the study, including gaining approval for the study, participant recruitment and data collection and analysis. I will address each of these aspects in the chronological order that they were undertaken. I describe this aspect of the study as a 'production' because of the social influences that mean scientific knowledge and technologies are produced rather than revealed.

Approvals

The aim of a Research Ethics Committee (REC) is “to protect the rights, safety, dignity and well-being” of all actual or potential research participants (HRA n.d: para 3). Research conducted by university staff and students requires approval from the academic school or faculty REC as well as, in the case of NHS patients or staff, approval for health and social care research from the relevant NHS Health Research Authority (HRA). Different requirements exist for approval depending on the status of study participants (that is the relative risk to them of the research depending on either the research intervention, their perceived vulnerability or both). Projects such as this one are arguably of negligible risk to participants and approval is required to interview the public only from the academic school REC. This was achieved without incident in January 2012 and I was able to begin recruiting DTCGT users to the study.

Involvement of NHS staff requires approval from an NHS Research and Development Committee (as opposed to a REC). The process for gaining approval to interview clinicians from the Research and Development Committee was considerably more tortuous than that for the school REC. But having successfully performed this rite of passage to gaining approval, I was able to recruit clinicians to the study from September 2012. Approvals documents can be found in the Appendices.⁷

Recruiting participants

This study involves purposive sampling, including snowball sampling, to access the relevant social groups involved in influencing SNP genotyping technology in the UK. The decision to involve DTCGT users and genetics clinicians as two groups with interests in this controversial technology is key to understanding how the technology is being shaped in the UK. Mel Bartley suggests that investigation of professional and patient interest groups in medical sociology is often a starting point for examining the social interests of actors in medical scientific knowledge and technology

⁷ The IRAS form submitted for approval by the NHS Research and Development Committee is not included as the PDF file is too large to import. Their approval letter is in Appendix 3: Cardiff and Vale University Health Board, R & D Committee Governance Review Approval.

and that this needs to be undertaken with symmetry and impartiality as the guiding principles of investigation, in keeping with the Strong programme of investigating scientific knowledge in health care (Bartley 1990). Thus, participants were recruited from two relevant social groups with interests in DTCGT technology but whose views are likely to represent opposing or conflicting interpretations and knowledge claims, that is, users of the technology and genetic clinicians who might be approached to counsel them. The juxtaposition of these two groups' views contributes to a symmetrical approach in the study. These two groups are involved to examine and contrast the different experiences and interpretations of DTCGT by the participants in both groups, giving equal attention to influential professionals and the public regardless of their perceived social positions in a hierarchy of power (Atkinson and Delamont 2006)⁸. In addition, the principle of impartiality is adhered to in my examination of the social nature of these actors' views and reported engagement with the technology and their knowledge about it. For this reason, participation was invited from members of the public who had bought DTCGT and genetics clinicians who had counselled people who had bought DTCGT and who were willing to talk about their views on the subject. The users represent a relevant social group directly associated with the technology but one that is relatively disparate and disorganised. In contrast the genetics clinicians are not directly involved with the technology but represent an organised relevant social group by virtue of their professional identity and work in clinical genetics. 17 users and 16 clinicians were purposively sampled and invited for a single interview; I shall go on to explain how this came about.

⁸That said, the role of power in each group's influence on DTCGT may not be as clear-cut as it first seems. The strength of professional power in shaping medical technology is challenged by the experiential knowledge of an informed and vocal group of early adopters of DTCGT technology. This group's origins lie in the production of DTCGT because SNP genotyping sold directly to the public requires users (consumers) to purchase and thus 'produce' it. This could be interpreted as investing them with a more cohesive and powerful impact on the technology. In contrast, the genetics clinicians' professional power has less impact because it is simply associated with DTCGT by virtue of their professional knowledge of human genetic testing.

Users

The Internet was the area used for recruitment, as it is the environment in which DTCGT is marketed and sold. It was therefore assumed that test users would be familiar with looking for information relating to personal genomics online and would thus find invitations to participate on the Internet. When I started the study, for this reason I felt confident that using the Internet was the best way to recruit users. Unlike genetics clinicians who can be found in geographical locations and whom I assumed would be easy to enrol, this group were an unknown entity and recruiting them was a venture into uncharted territory. I anticipated that recruitment of these virtual people, who Brown and Webster describe as without embodiment, would be challenging and would require several approaches, as Temple and Brown suggest (Brown and Webster 2004, Temple and Brown 2011). Accordingly, I approached researchers with experience in recruiting participants online and gained valuable tips, such as the use of 'Google Ads' linked to relevant search engine terms. My plan was thus to post about the study on several relevant weblogs, place a Google Ad linked to DTCGT companies and personal genomics interest sites and undertake snowball sampling via early volunteers, as outlined by Atkinson and Flint (2001).

The selection criteria for participation included being a UK resident and having purchased DTCGT that included health information. This was due to my interest in people's engagement with the health-related aspects of the technology and the relationship to HCPs and healthcare provision in the UK. It was initially unclear who would comprise this group of participants and thus it was surprising that contrary to expectation, recruitment of users was almost effortless. The majority of participants were recruited from one of two online groups, or were known to other participants and were approached by them and asked to participate. The groups are

- 1) the UK branch of the International Society of Genetic Genealogists (ISOGG) <http://isogg.org/>
- 2) the weblog genomesunzipped (GU) <http://www.genomesunzipped.org/>

A member of the UK branch of ISOGG found out about the study from my university research webpage within hours of it being published online and published a link to it on their website. Two members contacted me to volunteer within hours of this happening and were followed by several more over ensuing weeks. GU published my guest post to describe the study and appeal for participants on their weblog and several more participants were recruited from this source. As a result, the participants fell into two groups being primarily interested in either genetic genealogy (ISOGG) or personal genomics (GU), though a few were interested in both. Two participants were recruited outside of both these groups, one having heard about my study at a research conference at which I presented. Despite their different routes into DTCGT, all these people had bought a DTCGT that included health information obtained from 23andMe. Details about their individual backgrounds, tests done and primary motivations for testing are in Table 3.1. A summary of the user participants' information is in Table 3.2.

In a British Social Attitudes Survey report on public opinion about genomic science, Sturgis et al (2004) identified the socio-demographics of the section of the UK public more likely to follow debates about genomics. Their analysis of this group reported that the average age was 55-59 years, that women were more interested in genomics and that this group were interested in biology and educated to GCSE level or higher, were more likely to have a genetic problem in their family, were interested in politics, read a broadsheet newspaper, had access to the Internet and probably had higher knowledge of genetics (Sturgis et al 2004). There are some commonalities between these data and those recruited to the user group in this study. In the user participants' group the average age is 52 years. 12 of the 17 participants are female; this supports Richards's assertion that women act as genetic housekeepers in families (Richards 1996). Whilst one's education was unknown, the majority were educated to degree level (13). One of those who was not had an expressed interest in biology, as did the person whose education level is unknown, and 6 were educated to MSc or PhD level and were, or had been, working in genetics. Whilst I did not establish their political views or newspaper

preferences, I do know that they all had access to the Internet. 17 users were recruited and interviewed, the first 16 over a period of four months.

At the time I was recruiting to the study I was relieved to have so many expressions of interest in the study in such a short time and simply accepted the first seventeen volunteers as participants. In hindsight, relying on the Internet and snowballing to enrol users largely limited recruitment to two sub-sets of people who are particularly interested in personal genomics, that is people who work in genetics and genetic genealogists. This will have missed people who are more concerned with personalised medicine and health screening *per se* who would constitute a different relevant social group of actors with influence on DTCGT in the UK. As a result the structural characteristics of *this* relevant social group (individuals with knowledge of SNP genotyping) provide them with a more informed position from which to engage with DTCGT and to exert a specific kind of influence on the technology as a result of their informed position. It makes them more likely to be willing to share their experiences and enthuse others. Their enthusiasm for personal genomics and their informed starting point is likely to give a particular view of how DTCGT is being shaped that is less significant for the NHS. However at the time I recruited participants I anticipated that the experiences of any member of the UK public who bought DTCGT that included health information would constitute membership of this relevant social group of users of this technology. Whilst all the participants fulfil this criterion, in hindsight a different recruitment strategy might have resulted in a broader cross-section of users whose views of the technology may have been different and thus led to different conclusions about their influence on it, and the potential impact on NHS services.

However, it is worth noting that the users I recruited fitted the profile of early adopters that McGowan et al described in their study of early DTCGT users in the USA (McGowan et al 2010).

Table 3:1 User Participants' Information

Participant identifier and pseudonym	Age	Recruited	Ethnicity	Closest blood relatives	Education (highest award)	Occupation	Test(s)	Cost	Dates	Motivation	NHS Referral
UP1 Alan	66	ISOGG	White British (WB)	Daughters	BSc (Hons) Chemistry	Retired Pharmaceutical Industry	FT DNA 23andMe	\$99	2005 2010	Genealogy	N
UP2 Ann	53	ISOGG	WB	Sons	Degree	Editor, writer	FT DNA 23andMe	\$99	? 2010 approx	Genealogy	N
UP3 Barbara	61	ISOGG	Ashkenazi Jewish WB	Sons	MA	Local Councillor, retired social researcher	FT DNA 23andMe	\$299	2002 2009	Genealogy	General Practitioner (GP) Oncology
UP4 Carol	59	Friend of participant	WB	Son	PhD	Public Health Researcher	23andMe	\$99	2011	Curiosity genetics	N

UP5 Christine	45	Colleague of researcher	Italian Australian	Siblings	PhD	Public Health Lecturer	23andMe	\$99	2012	Health	N
UP6 David	65	ISOGG	WB	Son	FE Agricultural College	Farmer	Sorenson surname DNA, Y-DNA, FT DNA, Autosomal DNA 23andMe	unknown	2010 approx	Genealogy	N
UP7 Elizabeth	66	Wife of participant	WB	Son	Teaching Certificate	Retired Primary School Teacher	23andMe	Free (offer)	2011	Gift Genealogy	N
UP8 Fiona	33	GU	WB	Sisters	BSc (Hons) MSc	Scientist genetics	23andMe	\$99	Jan 2012	Curiosity genetics	N

UP9 Geoffrey	70	GU	WB	Son, daughter	BA	Retired publisher	Oxford Ancestry FT DNA 23andMe	\$99 + subs	2001 2002 2008	Genealogy	N
UP10 Helen	34	GU	WB	Parents	Incomplete BSc Physics	None, disability	23andMe	unknown	Oct 2011	Health	N
UP11 Jane	54	GU	WB	Sons	Degree	Finance Director	23andMe	unknown	Jan 2012	Gift Curiosity Genetics	N
UP12 Ian	53	GU	WB	Sons	Unknown	Retired	23andMe	unknown	Jan 2012	Gift Curiosity Genetics	N
UP13 Keith	60	ISOGG	Irish	Brothers	MSc	Database consultant	Sorenson Y DNA 23andMe	\$99 + subs	2010 Sept 2011	Genealogy	N
UP14 Kirsten	48	GU	WB	Daughter, son	PhD	Statistician genetics	23andMe	\$99 + 1 year subs fee	Summer 2011	Teaching resource	N

UP15 Laura	43	Email from participant	WB	Brother	PhD	Computer biologist genetics	23andMe	unknown	Summer 2011	Health	N
UP16 Maria	27	Email from participant	WB	Siblings	BSc (Hons)	PhD student genetics	23andMe	unknown	Summer 2011	Curiosity genetics	N
UP17 Nicola	45	GU	Irish	Daughter Sisters	PhD Molecular Biology	Commercial biologist	23andMe US private Exome sequence	\$99 \$900	2011 more recently	Looking for neurological diagnosis	N

Table 3:2 Summary of User Participants' Information

Age	Range: 27-70 years	Mean: 52 (rounded) Median: 53.5
Sex	Women: 12	Men: 5
Recruitment source	ISOGG: 5	Genomes Unzipped: 7
	Known to participants: 4	Known to researcher: 1
Ethnicity	13 White British 2 Irish 1 Ashkenazi Jew British 1 Italian Australian	
Education	1 unknown 2 further education level 14 Degree level 3 Masters level 6 Doctoral level	
Occupation	5 genetics 2 public health (research and teaching, experience in genetics) 10 various other occupations (non-genetics/biological sciences)	
Test	17 x 23andMe (all) 5 x FTDNA 2 x Sorenson Y DNA 1 x Sorenson Surname DNA 1 x Autosomal DNA 1 x Oxford Ancestry 1 x whole exome sequence	1 participant had 5 tests 1 participant had 3 tests 5 participants had 2 tests 10 participants only tested with 23andMe

Referrals	6 told their GP (5 in a consultation for something else) 1 referred to Oncology via GP	11 did not discuss test results with any healthcare professional
------------------	--	--

Clinicians

Clinicians were recruited to the study using a number of strategies. The initial approach was by email to British Society of Genetic Medicine (BSGM) members from this project's local NHS collaborator, a member of the BSGM Council. The BSGM is a professional society that represents healthcare professionals working in clinical genetics services in the NHS in the UK. Responses to the email were disappointing (n=2), so a combined approach to recruitment was used. This included leafleting delegates at a BSGM conference, snowball sampling by using study volunteers to suggest contacts in the field (Atkinson and Flint 2001) and contacting Regional Genetics Services in the UK to raise awareness of the study and ask for expressions of interest from the clinical genetics staff. Recruitment continued to be slow despite these additional approaches; my discussions with staff at Regional Clinical Genetics Services suggested that there was a paucity of experience of these consultations and that clinicians probably felt they were not eligible to volunteer. Consequently, the initial intention to recruit clinicians who had counselled users of DTCGT was widened to include any genetics clinician who had views about DTCGT and was willing to volunteer to discuss their views. These combined approaches resulted in 16 interviews being conducted over 11 months with genetics clinicians who had a variety of experience and professional backgrounds. See Table 3.3 for details about the clinician participants.

Genetics clinicians are healthcare professionals who take referrals and see patients and families for genetic counselling and testing. There are three professional routes to this role.

1. Medical doctors who wish to specialise in clinical genetics complete a postgraduate diploma for membership of one of the

medical Royal Colleges (either the Royal College of Physicians or the Royal College of Paediatrics and Child Health), after which they complete a 4-year specialist registrar training post in a regional genetics centre.

2. Registered nurses or midwives who have post-registration clinical experience, have counselling experience and have successfully completed a course in genetics are eligible to apply for trainee genetic counsellor posts.
3. Postgraduates with relevant degrees, such as genetics, biology, psychology or sociology, or allied health professional degrees including nursing and midwifery, may complete an MSc in Genetic Counselling and then apply for trainee genetic counsellor posts. (BSGM n.d.)

Participants were from a range of the Regional Clinical Genetics Centres in the UK. Northern Ireland was not represented but all other countries were, with the majority of participants practising in centres across England. Collectively they represented a range of catchment areas including the South Coast, the Southeast, the Northeast, the Northwest and London. There were no volunteers from the Midlands. A summary of the clinicians and consultations for DTCGT can be found in Table 3.4.

Table 3:3 Clinician Participants' Information

Participant identifier	Profession	Role	Time in genetics at interview	Education	Consultations for DTCGT	Tests consultands used
CP1	Scientist	Genetic Counsellor	12 years	PhD, MSc Genetic Counselling	GP queries x 2	Unknown
CP2	Doctor	Consultant and Professor Clinical Genetics	>13 years	Medicine, Clinical genetics training, PhD	GP queries Patient x 1	Ancestry 23andMe
CP3	Scientist	Genetic Counsellor	3 years	PhD, MSc Genetic Counselling	Patient x 1	23andMe
CP4	Doctor	Consultant and Professor Clinical Genetics	15 years	Medicine, Clinical Genetics training, PhD	Patient x 1	Sibling had personal genome test (not 23andMe, company unknown)
CP5	Nurse	Consultant Genetic	25 years	Nursing PhD	Journalists for TV	All 23andMe

		Counsellor, Reader in Clinical Genetics			x 3 (not NHS consultations)	
CP6	Doctor	Specialist Registrar in Clinical Genetics	5 years (PT)	Medicine, MSc and Clinical Genetics training in progress	Patient x 1	Sibling had Personal Genome Test (LifeDNA, Italy)
CP7	Scientist	Researcher, Genetic Counsellor	>5 years	PhD, MSc Genetic Counselling	None	
CP8	Scientist	Genetic Counsellor	2 years	PhD, MSc Genetic Counselling	None	
CP9	Nurse	Principal Genetic Counsellor	18 years	Nursing	Patient x 2	1) HD test 2) BRCA test (Single gene mutation tests from USA)
CP10	Doctor	Consultant	>17 years	Medicine,	Unknown	

		and Professor Clinical Genetics		Clinical Genetics training, DM		
CP11	Doctor	Consultant and Professor Oncogenetics	>17 years	Medicine, Oncology training, PhD	Patient x 2	1) Ancestry 2) Relative's tumour genome sequenced and analysed by another relative who warned of mutation and need for testing
CP12	Nurse	Genetics Counsellor	12 years	Nursing, MSc in progress	None	
CP13	Doctor	Consultant Clinical Genetics	>11 years	Medicine, Clinical Genetics training	None	

CP14	Doctor	Specialist Registrar in Clinical Genetics	1 year	Medicine, Clinical Genetics training in progress	None	
CP15	Doctor	Specialist Registrar in Clinical Genetics	3 years	Medicine, Clinical Genetics training in progress, PhD	None	
CP16	Nurse	Principal Genetic Counsellor	>20 years	Nursing, Midwifery	None	

Table 3:4 Summary of Clinician Participants' Information

Profession	Consultations for DTCGT	Types of DTCGT	
		SNP Genotyping	Other
16 Clinicians Interviewed	12 Consultations by 8 Clinicians		
Doctors = 8	Doctors = 6 (1x2, 4x1)	for common complex disease risk = 7	Single Gene Mutation = 2
Nurses = 4	Nurses = 5 (1x3, 1x2)	(23andMe = 5)	DIY tumour sequence = 1
Scientists = 4	Scientists = 1 (1x1)	(1 consultation with sibling of testee)	(consultation with relative of affected individual)
		(3 consultations with journalists who tested with 23andMe for media productions, not NHS)	
		for ancestry = 1	

Collecting data

In justifying the theoretical framework for the study I referred to accounts and conversations in the context of constructing knowledge about the relevant social groups involved with DTCGT, their interpretative flexibility and the wider social context of this technology. This was achieved by talking with participants: I undertook one-off, face-to-face interviews with the participants in a venue and at a time of their choosing, in all but three cases. Two of the users and one clinician were interviewed on the telephone, as this proved much more convenient for both parties due to their distant locations and busy schedules. I had hoped to use video and voice calling software for interviewing via the Internet in these situations but, after an abortive attempt in the first of these

interviews, I abandoned this idea. The requirement for both parties to have sufficient, consistent bandwidth to support an uninterrupted conversation could not be guaranteed. As a result, the conversation was repeatedly disrupted by loss of sound or vision or both and this undermined the potential for establishing any rapport or useful exchange of ideas. I felt that what was lost in terms of visual cues was a trade-off worth making to have a fruitful conversation.

This illustrates an example of what Silverman refers to as “trial and error”; it was entirely understandable to resort to telephone conversations with these three participants in the circumstances, but in hindsight the conversations we had were much less fruitful than the remainder that were conducted face to face (Silverman 2000:236). Without visual cues I found it difficult to build a rapport with the participant or to pick up on points to probe further and the conversations were thus more perfunctory. I found the lack of visual cues much more difficult than I anticipated and my concentration was noticeably affected by straining to hear people (I am slightly hard of hearing and I find the telephone the most difficult medium for conversation owing to the lack of opportunity for lip-reading). Following these experiences, which occurred early in the data collection period, I subsequently arranged to meet all the participants for interviews, despite their concerns about my having to travel. From these early experiences, I knew that the conversation would be much richer and more fruitful when conducted face to face.

Ethical conduct

I have already outlined how approvals for the study were gained, that is, the socially constructed regulatory process for sanctioning research in the UK and in the NHS in particular. However, the procedural activities that are required in order to fulfil the trust invested in researchers by RECs that Hedgecoe describes are distinct from conducting research in an ethical manner, particularly when interacting with participants (Hedgecoe 2012). Whilst this is closely related to reflexivity, the situations I encountered that were related to ethical conduct provided important moments in my relationship with the participants. Consequently, they

influenced the resulting data and my analysis of it, albeit indirectly, and are thus worthy of inclusion in this discussion.

Interviews started with completion of the participant's consent form. This relied on them having previously read the study information sheet, which was included in the original study information on weblogs and in email invitations to participate. I also sent the information sheet and the consent form to all the participants as attachments to the emails arranging our meetings, with a reminder to read them before we met. I was surprised to find that many participants had not read these documents prior to meeting me. However, they were all happy to sign the consent form, although one or two people did take the opportunity to read the information sheet that I brought to the meeting before doing so.

Signing the consent form provided an important opportunity for me and the participant to take part in an external activity together on a relatively neutral basis. It bridged the transition from meeting a stranger, with its attendant formality and social conventions, to the interview and its opportunities for disclosure of personal information. I came to appreciate this activity and the related conversation about it as opportunities to continue to develop a rapport and understanding between us before commencing the conversation about DTCGT. They also provided the opportunity to remind participants that the interview would be recorded for transcription but that they could ask to stop the interview at any time. More importantly, the transition hopefully helped participants to relax and thus feel more able to create their stories in their own manner (Charmaz 2000).

Confidentiality and anonymity are concepts that present challenges in qualitative research, as Walford and Saunders et al highlight, particularly when conducted within a specific interest group such as this study's, where certain actors are well known in the social groups concerned (Walford 2005, Saunders et al 2014). Signing the consent form provided an opportunity to explore the potential for identification of participants by others reading the results of the research and what steps I would take to avoid this. I have given the users pseudonyms to protect their identity as far as possible, whilst simultaneously trying to maintain some individual

character through the threads of data, as Seidman suggests. Clinicians are identified only by profession to protect their identity, while maintaining the distinction between the two groups of actors in the data analysis (Seidman 2013).

Interviews

As I noted earlier, the purpose of the interviews conducted in this study was to encourage participants to explore their experiences and thoughts about DTCGT. To facilitate this, I intended to take a relatively unstructured approach to the interviews in order to provide a conversational exploration of individuals' accounts and to probe certain points for further examination when indicated, as recommended by Kvale and Brinkman (2009). However, despite my wish to have as unstructured an approach to the interviews as possible, past experience of research interviews prompted me to develop an interview schedule as an aide-memoir of points to explore during the conversation, should these not have been covered spontaneously. This proved useful in more stilted interview exchanges (including those on the telephone), but the schedule figured less as my confidence interviewing increased. I opened and closed the interviews with broad questions to facilitate participants' engagement with narrating their stories from their perspective. This was particularly useful at the end of the interview when asking the question "Is there anything you thought I would ask you about that we haven't covered?" allowed participants to bring up points of importance to them which had not already been explored. In addition to seeking any final points that they deemed important in relation to the preceding conversation, it provided an opportunity to give participants the final word in their story, from the point of view of being able to choose both *what*, if anything, to add and *how* to close the interview. I also offered everyone the opportunity to follow up the interview with contributions on email if anything further occurred to them, but only one person did so, again someone I spoke to on the telephone. See Appendix 9: Interview Schedule for Users and Appendix 10: Interview Schedule for Clinicians.

Interviews lasted between 45 minutes and an hour in every case and were digitally recorded. I made written notes during the interviews to record

points that I thought warranted more exploration later in the conversation and immediately afterwards to record my thoughts about the location, the process and my impressions of the interview. I transcribed the interviews myself as soon as possible afterwards, usually within twenty-four hours. This enabled me not only to recall the interview and its context clearly and to begin to familiarise myself with the data, but also to have a written text to study and prompt my memory later. The transcriptions were limited to the recorded speech, omitting pauses, hesitations or other minutiae, as a discourse analysis is not the principal focus of this project.

In the following section I shall explore how I analysed the data, although, as Seidman suggests, separating data collection and data analysis into distinct activities is problematic, in that it is impossible not to think about the data as it is collected (Seidman 2013).

Analysing the data

Qualitative data analysis requires an organised approach to facilitate the sorting, managing and retrieval of relevant pieces of data as required. In line with many qualitative researchers' suggestions I took an iterative approach to analysis in order to elicit the themes within the texts of the interview conversations (Coffey and Atkinson 1996, Creswell 2013). In the initial stages of the analysis there was scope for trial and error in the approach I took to this, and, as a result, I experienced both lows and highs during my engagement with the data, which I will recount in my exploration of how the data were analysed.

During data collection, I undertook training in the use of computer assisted qualitative data analysis software (CAQDAS), specifically NVivo 10, as this software and the training to use it were available at the university at the time. Prior to conducting this project I have had some experience of analysing qualitative data on a few occasions, but the data sets were significantly smaller, so I had not used CAQDAS. Also I realised that there would have been significant developments in the software and I thought it would be beneficial to understand NVivo's functionality and be able to use it at least to manage the data, if not more extensively.

After the training sessions I decided to abandon my ideas to use CAQDAS. Following the experiential learning in the sessions (participants took their own data to sessions to use for training), it was clear to me that NVivo would only add more steps to the process and probably take longer. I also found it distracted me from the data *per se* and focused my attention on the software programme instead. To put it another way, I felt that NVivo would come between the data and me, thus bringing a metaphorical cuckoo into the nest of the project. Having made this decision, I find Seidel's references to the dark side of CAQDAS, what he refers to as "analytic madness", resonant with my impressions of it, despite his having written about it over two decades earlier (Seidel 1991:107). This confirmed my decision and consequently I analysed the data 'by hand'.

I transformed the interview conversations into texts by transcribing them, thus immediately giving me some familiarity with the content and food for thought. As transcripts were completed I read through them and considered the ideas that seemed to be emerging. With data from the two groups collected separately, I was able to learn from my initial approach to the first group's data and try a slightly different approach with the second.

From the interviews with users I generated paper copies of the transcripts. I used these to read, re-read, underline and make notes in the margins about emergent or *in vivo* codes, that is, those arising from the participants' discourse (Strauss 1987, Ryan and Bernard 2000, Creswell 2013). In addition, I made notes or memos about patterns and exceptions that appeared in the data and the ideas that occurred to me whilst reading the transcripts, and used the reflections I recorded about the interviews to inform my thinking with the data (Coffey and Atkinson 1996). Having completed this initial phase of organising and managing the data, I read it again alongside the literature for an inductive approach. With this iterative approach to reading, coding, memoing and referring to literature, the chaotic nature of qualitative data analysis was to the fore (Glaser 1999) and I identified with Adele Clarke's reference to "the sea of discourses with which we are awash" (Clarke 2003: 559). However, this period of thinking with the data led to the development of early categories and classifications that I recorded next to examples in the data. At this point I

moved from paper to screen, the former becoming too unwieldy. Thereafter I relied on electronic versions of both groups' sets of data, as keeping track of examples of the emerging classifications in the data and the codes, categories and themes was easier.

Once I had undertaken these early steps in the analytic process I commenced writing up my findings, still working with separate data sets from the users and the clinicians, connecting the emerging themes with the literature in a more direct way. This aspect of the analysis was a revelation to me, in that it was while writing that the true analysis began and themes began to emerge from the mire that Wolcott refers to (Charmaz 2000, Wolcott 2001). From the perspective of these themes I went back to the data and using writing to explore my thoughts as Latimer describes, I was able to interact with the data, the literature and my constructions from the data, and I found this process surprisingly enjoyable and liberating. In addition to writing about the data, I have presented it several times to different groups and audiences, which has similarly informed my thinking with and development of the data analysis. From this iterative process I have developed the analysis of the three overarching themes presented in the chapters that follow (Coffey and Atkinson 1996, Latimer 2007).

In the excerpts shown to represent my analysis I have used symbols, as indicated in Table 4.5, based on conventions described by Corden and Sainsbury (2006) and Roberts and Sarangi (2005). I have adopted transcription conventions where participants' short phrases are represented within my text in quotation marks, but longer excerpts of data are presented in indented plain text. Verbal hesitations not relevant to the data have been omitted by a "light tidying up" to aid the flow of the text in common with Corden and Sainsbury's findings in their research into the use of verbatim quotes (2006:18).

Table 3:5 Symbols used in data transcription

Symbol	Meaning
...	Ellipsis: represents analyst's edit of the data
[]	Square brackets: represent analyst's comment or modification of the excerpt
(())	Non-verbal communication

Conclusion

In outlining the conduct of this project I have described how I have undertaken it, the research traditions that informed the theoretical and methodological basis for the design and the realities of conducting this research as it has progressed. The few low points included the challenges of obtaining the required assurances from university administration in order to apply for NHS R&D approval, recruiting clinicians and feeling overwhelmed by data, all of which I share in common with many, if not most, researchers. These aspects were more than compensated for by experiences of recruiting users, developing a conversational interview approach and learning the value of writing in data analysis. Information technology challenges confirmed that talking face-to-face is the most rewarding mode of communication, but that handling data is better achieved electronically.

Having described my approach to the study and the issues I encountered during the process, I shall go on to provide a representation of the themes that emerged from the data in the three chapters that follow. In them I will show how users of DTCGT and genetic clinicians in the UK socialise and personalise DNA and test the NHS as a consequence.

Chapter 4 : Socialising DNA

As Novas and Rose have suggested, those who undergo genetic testing are not solitary individuals but are members of networks that pre-date their engagement with genetic testing (Novas and Rose 2000). This chapter explores the importance of social networks to members of both relevant social groups involved in this study, and their role in developing expectations and ideals in relation to genetic testing.

The chapter has two main sections: the first presents my analysis of how the users socialise DNA and how their initial awareness of DNA leads them to engage in networks of actors with similar interests, which develops and shapes their expectations of DTCGT. In the second section, my symmetrical analysis of the clinicians' socialisation of DNA shows how professional identity and networks in clinical genetics result in boundary work in respect of DTCGT. The moral work of the clinic and boundary work in relation to the scientific validity of SNP genotyping are used to frame clinicians' understandings of DTCGT from their clinical genetics perspective and, I suggest, contrasts with and parallels users' views where relevant.

Arguably social practices around any shared interest or belief are opportunities to share individual understandings and experiences with others. Thus a distinction between personal or individual considerations about an artefact and the act of sharing these with others in a relevant social group could be viewed as moot. However, I would argue that there is a valid distinction between Socialising DNA and Personalising DNA to be illustrated by the data in this study. This chapter will show the importance of social networks in building participants' technological frame of DTCGT and providing channels for them share their experiences. Actors' ideas and beliefs about DTCGT are developed and shaped and made visible to others by interactions in their networks. The nature of what is shared with others in socialising DNA is qualitatively different from individuals' personal reflections on DNA. Whilst elements of personal reflections coincide with what is shared socially, personalising DNA represents an internal re-imagining of identity incorporating newly found genetic information (whether that is the individual's or in the case of genetics clinicians

in respect of aligning new understandings of DTCGT with their identities as genetic counsellors). This personal imagining will not necessarily be shared, and if it is may not be represented in quite the same manner as Chapter Five will demonstrate. Approaching this study's data from the separate perspectives of socialising DNA and personalising DNA also provides one opportunity to illustrate participants' use of interpretative flexibility around DTCGT.

I start this chapter by analysing how participants' networks and expectations of DTCGT demonstrate socialising DNA. I will show how important the networks that facilitate this process are in shaping expectations and supporting the ongoing development of this group's identity practices after testing. The experiences of the two user participants who are deviant cases in this data will be incorporated in my discussion as a way of illustrating this theme (and that which follows), due to the sharp contrasts between them and the remaining user participants.

Networks, expectations and responsibility

For almost all the user participants a network was instrumental in introducing them to the possibility of DTCGT and engaging them in considering buying a test. Gibbon and Novas refer to sociality as identity practices and situate this in relation to Paul Rabinow's term "biosociality", which he coined in the 1990s (Rabinow 1996, Gibbon and Novas 2008). Rabinow used the subsequently popular term to suggest that the human genome project would provide a network of known genetic anomalies that would produce social groups with common (genetic) identities. Group members, with their common genetic identities and associated medical care and support, will also have commonly-held narratives and traditions as a result of their social processes of sharing, providing support and achieving a sense of empowerment and belonging within their group. The genetic aspect of their identity provides a vehicle for them to collectively "experience, share, intervene and understand" their genetic identity (Rabinow 1996:102). The possibilities for the development of communities such as those Rabinow envisages is greatly enhanced by developments in information technology. These developments facilitate communication and thus social processes among individuals who are geographically scattered.

Whilst the users in this study are not known to have genetic illnesses in the clinical sense, by engaging with DTCGT they reify their genetic identity. This gives them a common, biological focus around which to engage in social processes with others who share the same experience. They do this by socialising with others who are similarly interested and identified as such, both before and after testing, face to face in occupational or recreational groups and virtually through the Internet. The latter can be aligned to the “new collective formations” Rabinow and Rose envisaged (2006:204).

Critics of Rabinow’s concept of biosociality suggest that this is not a novel idea but rather a novel focus for more conventional (pre HGP) forms of sociality (Kerr 2004, Plows and Boddington 2006, Raman and Tutton 2010). There is some room for acknowledging this argument here in that the group of users recruited to this study appear to engage in sociality prior to buying DTCGT. This is through their membership of groups that introduce them to DTCGT and enable sharing of their common experiences and values in relation to it as I will go on to show in the data in this chapter. Their social practices could be described as simply social rather than biological. However the reification of their *genetic* identity and their engagement in social practices around this experience relates to a novel genetic technology, and although it precedes testing rather than resulting from it, it is arguably *biosociality* in the sense that Rabinow suggested.

By virtue of buying DTCGT that includes health information, users in this study are members of a relevant social group that will contribute to shaping DTCGT in the wider context of the NHS. As previously acknowledged in Chapter Three, by virtue of the approach to recruitment to this study the particular structural characteristics of this group is important as it explains their ability to engage in social practices around DTCGT using various means of communication (owing to their economic and educational status) and their knowledge of genomics. However, there is a clear heterogeneity within this group; sub-groups including the genetic genealogists and those who work in genetics are likely to have different views about DTCGT that they circulate in their social networks which will thus have different effects on the potential for stabilisation of the technology (Klein and Kleinman 2002).

The clinicians' networks are also fundamental in shaping their beliefs and expectations in relation to DNA and DTCGT in the context of the NHS. Though more implicit than explicit in the data in comparison to the users, practising genetic counselling provides these clinicians with membership of professional networks that influence their beliefs and ideals about DNA and how its characteristics should be revealed to patients. Genetic counselling is delivered in the UK by multidisciplinary teams consisting of doctors, suitably qualified or experienced nurses, midwives or allied health professionals and scientists (Skirton et al 2013). The clinicians in this study are doctors, nurses and scientists and are members of the NHS clinical genetic counselling network as well as of their individual professional networks.

For the clinicians in this study, socialising DNA is about promoting genetic counselling as the responsible way to help people decide whether or not to elicit genetic information and handle the consequences of that decision. This is shown in the data by participants' references to the importance of the moral work of genetic counselling. Boundary work is a key feature of this socialising theme in the data; this is expressed in relation to the work of the clinic by all three professions interviewed, but only by doctors and scientists in relation to the science of genomics. At first glance it might appear that because all the clinicians work as genetic counsellors, any consideration of their different professional backgrounds would not be pertinent to analysing the data generated. However, whilst the numbers of participants in each group is very small and the findings cannot be generalisable, there did appear to be professional differences in how clinicians focus on their beliefs about DNA and DTCGT. Thus, as with the users, it appears that different sub-groups within the relevant social group of genetics clinicians share slightly different understandings and may thus have slightly different effects on shaping the technology.

The groups involved in this study socialise DNA in the sense of being members of groups or networks interested in genetics and with common identities and cultures due to their interests and/or occupations. Bijker's introduction of the concept of a "technological frame" in relation to how social groups shape a technology refers to the communication about an artefact that is part of the socialisation around a new technology within its newly established relevant social groups (Bijker 2010:69). As users and clinicians

socialise DNA, each group constructs a technological frame in relation to DTCGT. Within the group of users and clinicians there are sub-groups with slightly different characteristics that influence their technological frames of DTCGT, but a common feature is their engagement or rejection of it through their sociality in groups. Starting with the users, I will explain how that socialising and the moral values held as a result are evident in the data.

Users' networks and new genetic identities

Brown and Michael suggest the association between uncertainty and people's proximity to technology shows a direct but inverse relationship; being 'further' from it decreases uncertainty about it (Brown and Michael 2003), also neatly described as "distance lending enchantment" (Collins and Evans 2002: 247). Potential users are less likely to have uncertainties about the future promise of new technology that might prevent them from engaging with it than the experts directly involved in its development. Brown and Michael indicate the importance of networks and activities in driving the expectations about new technology, which could also be equated with relevant social groups developing their technological frames around an artefact. This is key in considering how this study's group of users became involved with DTCGT.

For almost half the users the network that introduced them to DTCGT was ISOGG, a group to which many genetic genealogists belong and through which they share information about developments in their field, including the emergence of new technologies such as DNA testing for genealogical purposes. Ann is a key actor in the English group of ISOGG (a group administrator). A married woman in her 50s, she is very interested in and knowledgeable about genetic genealogy; she has published books on genetic genealogy and describes herself as a DNA enthusiast. Ann presented the initial entry point for herself and others she knows as being linked to their gradual movement from records-based genealogy to genetic genealogy and then on to package tests, which include both ancestry and health risk testing such as that offered by 23andMe.

A lot of genealogists have taken part in the health tests because we've been using the DNA tests for our genealogy research so it's a natural progression really, just out of

curiosity to see how it all works ... I got involved with ISOGG because they have a mailing list where group administrators, all volunteers, can get together and talk and share experiences ... I was just interested to see how the health aspects worked as well and having seen other people taking the test I knew how it all worked. (Ann UP2).

The sociality of the network is important for her and others interviewed, not just for learning about new technological artefacts and how they “work”, but also for sharing experiences that develop their technological frame in respect of DTCGT that includes health information, and shape expectations of it while getting support from others in their trusted community. Ann describes “a lot of genealogists” testing and links this to her own decision being a “natural progression”. This implies that she views herself as part of this group, from whom she has learnt a lot and with whom she identifies herself. This is a demonstration of her membership of a relevant social group that is shaping DTCGT.

The genetic genealogists in this study are all members of one or more genealogy organisations. With this shared interest, genealogists’ networks have spread the word about technological developments in genetic genealogy, engendering the expectation of increased personal information for genealogy *and* health purposes, widening their technological frame of DTCGT and influencing each others’ thinking and actions through this social network. Through this social network, individuals have encouraged and supported each other in venturing into DTCGT. Barbara, a woman in her 60s, has an 18-year long interest in genealogy owing to her family’s Jewish ethnicity and medical history. She describes how support from distant relatives found with genetic genealogy introduced her to 23andMe.

23andMe came into view and they were advertising themselves in terms of health testing and I thought well this might be interesting. And I was put in touch with them because I’d already developed close bonds on the family lines with two people in particular, both based in America who both tested with FTDNA [Family Tree DNA – ancestry

genomics company] ... and they got to hear about all these things. (Barbara UP3).

Barbara refers to developing “close bonds” with people who are already part of the network of adopters of SNP genotyping. These people had had experience of both genetic genealogy testing (with FTDNA) and had made Barbara aware of other possibilities, including 23andMe, which provided the additional information about disease risk. This was an important aspect of Barbara’s expectations of personal genomics owing to her family’s medical history. Through the sharing of interests and experiences, she gained information that shaped her understanding of testing and helped her decide to test with 23andMe.

Four of the users are men in their 60s, or older, whose primary interest is in genealogy; Alan, David, Geoffrey and Keith are all involved with online genealogy groups to varying degrees. They have all been aware of genetic genealogy for several years, having bought tests from ancestry testing companies in the very early stages of genetic genealogy. This demonstrates the importance of socialising in terms of their involvement in genealogy; others in their networks are involved and share the information that has led them to test with 23andMe. Geoffrey has had experience of genetic genealogy for over a decade.

I’ve been interested in them [tests] more from the genetic genealogy side of things for a long time ... when Oxford Ancestry were first doing it ... it was a mitochondrial DNA test, quite elementary ... and I suppose I was on email lists and things. FTDNA cropped up, about 10 years ago. They do mailing to the genetic genealogy-type people and so I got on from there. (Geoffrey UP9)

As with most other users, Geoffrey is “interested” in testing because of his genealogy hobby. He is technically knowledgeable about DNA testing and has gained information from genealogy groups that has developed and shaped his interest and actions in genetic testing. He expected to enhance his personal knowledge, primarily in relation to finding relatives as a result of testing with 23andMe; the health information was of secondary importance to him. For

genetic genealogists socialising DNA facilitates access to information about relatives and ancestral origins. Sharing information about genealogy is fundamental to its success, which is measured by increasing knowledge of relatives and their origins. Thus this subgroup of users circulate knowledge and experiences of DTCGT avidly in order to increase their ancestry data and thus influence uptake (and development) of DTCGT by others.

The majority of the remaining participants are members of occupational groups involved in genetics and genomics, are related to members of these groups who could be said to have recruited their relatives to the group, or follow genomics developments online, including on the genomesunzipped website. Five users are scientists who are regularly involved in discussions about SNP genotyping at work in relation to laboratory research, statistics, ethical issues and personal views about testing. Fiona, Kirsten, Laura and Maria are all involved in genomics research, working in teams of scientists and bioinformaticians and they all referred to testing in the context of their workplace or discussions with their colleagues. Most of them contrasted their respective decisions about whether to test or not with what colleagues were saying and, in Laura's case, referred to testing that was being offered to workers in her institution to compare with her DTCGT.

Actually we've had an offer to have some genetic testing done here on site as well. They just started offering ... a limited amount of SNPs that they're checking and so I've put in for that. (Laura UP15)

Helen's and Nicola's recruitment to networks is also evident, although their reasons for testing were different from others, both having chronic long-term conditions that they had failed to achieve diagnosis of by conventional methods in the NHS. Nicola is a commercial biologist who works in genomics and who described having contacts in the industry with whom she discussed genomics and who helped her think about testing and helped in her search for access to testing. She is thus a member of the sub-group of users interested in DTCGT by virtue of their work in genetics although she is also searching for personal health information. Helen's networks are less immediately obvious than other participants'. Helen has suffered from a chronic condition since she was in her teens. Now in her thirties, this illness disrupted her undergraduate

studies, leaving her unable to complete her degree or work since she was in her early twenties. She is socially isolated due to her incapacity and relies on the Internet for information about science and technology developments that might help her establish what is wrong with her and what interventions could be effective. She described having been interested in genetics for some time, attributing this to her mother's work in genetics (her mother having had a career working in genetics), and told me how she subscribed to genomesunzipped. She is knowledgeable about genomics and SNP testing and described how she spends time researching genetics and biochemistry on the Internet whenever she is able. Her expectation is to find answers to her problems through genomics as more knowledge is developed about the influence of genetics on chronic problems like hers. In the meantime she decided to buy a test from 23andMe.

I've been interested in genetics for ages anyway ... I found out about it [23andMe], I came across the genomesunzipped blog – I subscribe to that – and they kept saying about the 23andMe things and I thought I'd give it a go. (Helen UP10)

Again, Helen's interest is explicit and was frequently referred to in the interview. As with others, Helen's initial interest in genetics prompted her search for and engagement with genomics networks. She demonstrates this by describing her subscription to genomesunzipped, which exemplifies her alignment with a genomics network. She shows how this network introduced her to 23andMe; her phrase "they kept saying about the 23andMe things" demonstrates how she is drawn into the social world of SNP genotyping. The Internet gives Helen access to communities such as followers of genomesunzipped and 23andMe, which she has joined despite her physical isolation. She is thus able to participate in socialising DNA and shaping DTCGT by virtue of her contributions to her online communities as Dimond et al suggest (2015).

Whilst these two women's principal motivation for engaging with personal genomics is centred on their ill health, their investment of hope in personal genomics has been informed by occupational or self-directed learning and ongoing engagement with networks in the form of online genomics interest groups, both having been recruited to the study from genomesunzipped.

Two participants who were not recruited from genomesunzipped were academics in public health who were friends; Carol was encouraged to have a test by a biologist she knew well through work and she then persuaded her friend Christine to have one. Christine had wondered about taking a test for a while; hearing about Carol's interest and experience of testing helped her to research the products and decide to buy a test. Christine was reassured by the information and perceived support she got from Carol and 23andMe's website.

I did like 23andMe because they did have people commenting ... I read reviews from other people and I contacted them to know how easy it was and what was involved ... and I had [Carol] to talk to as well mind you, so I did actually have a little chain of people ... and even having them on websites was good to know. (Christine UP5)

Christine specifically pursued others who had tested for information on their experiences, thus demonstrating the influence of the social - her "little chain of people" - on her developing knowledge and ideas about testing. She contrasted her expectation of enhancing her understanding of her genetic identity with her initial uncertainty and need for reassurance, which she gained through interaction with Carol and other testers.

The majority of the users' engagement with personal genomics seems to be facilitated by their role as what Latour might describe as actors in networks that circulate expectations about personal genomic testing technology (Latour 2005). These expectations are uttered and become embedded in the networks' culture, albeit tentatively. Their roles in these networks point to the social nature of their learning and decision-making about testing and the importance of becoming part of a relevant social group in gaining access to the group's technological frame of the artefact. However, their membership of the networks and the existence of most of the networks in question precede their engagement with personal genomics for their own individual purposes. Rabinow's work on biosociality suggests that genetics will provide a network of identities that individuals will join, resulting in social groups based on genetic characteristics, the 'bio' becoming the 'social' (Rabinow 1996). However, I suggest that in this group of early adopters of DTCGT in the UK, it is their networks and the socialising therein that enable them to learn about personal

genomics and join other users in sharing meaning of DTCGT technology and influencing its development. It is this learning that facilitates their decision to buy a test and begins to establish part of their genetic identity – a kind of reverse biosociality wherein the social enables the foray into accessing genetic information.

Participants' expectations of SNP genotyping are primarily to increase their personal knowledge, satisfying their curiosity and interest through their personal genomics. For the genealogists, the allure of finding unknown relatives is an important aspect of their on-going identity practices, whilst for some of the scientists it appears to be elevating knowledge from the mundane to something more transcendent and relevant to them individually.

My assertion is reinforced by Jane's and Ian's experiences of testing with 23andMe, which were distinctly different from the other users' expectations. Jane and Ian are a couple with a family member who was a researcher in genomics at the time of this study's commencement. Jane and Ian both clearly state that the final decision to have the test was theirs, but the original suggestion and information about DTCGT came from their relative. It would appear that their relative has more in common with the social group of users involved in genetics who are early adopters of DTCGT whilst Jane and Ian are not part of this social group. Their alternative entry point to personal genomics resulted in this couple's distinctly different experiences from the other participants in this study, representing the different impact DTCGT appears to have on different types of users, and the possible contribution of these different experiences to the lack of stabilisation of the technology. Ian described his amateur interest in science and genetics, which may explain why some of his views are more related to the other users', but neither he nor Jane has had any direct involvement in genetics or related networks.

The test was a Christmas present from [name of relative] ... asked us whether we wanted to have it and we thought about it and discussed it and me and my husband said yes. (Jane UP11)

This was a Christmas present from [name of relative] although obviously they asked us beforehand. We [Ian and

the relative who bought him the test] both have an interest in science, we have a similar interest and we talked about it quite a bit, so we were sort of informed, myself more than my wife before we did it and I also have an interest in it just as a pop science thing as well, so I have some background. (Ian UP12)

Here Ian, like other users, employs the word “interest” several times in describing how he came to have a test. This repeated use of the word could be seen as a justification for his decision to agree to have the test so that his decision does not appear to be naïve and to show solidarity with the relative with whom he shares a common interest in science.

Jane and Ian having decided to have their tests, Jane’s family became intrigued by testing and her sister and mother subsequently bought 23andMe tests also. Whilst direct personal involvement in a relevant network is not a feature in this family’s case, their kinship and indirect involvement through Jane’s and Ian’s relative has informed their understanding of testing and drawn them into a wider network interested in personal genomics technology, so they have been influenced by their relationships and the family network. However, this couple decided to test on the basis of their relative’s suggestion and sharing knowledge of personal genomics with them, rather than on the basis of their own learning about personal genomics through genetic genealogy or their occupation, as with the majority of the other participants.

Jane’s and Ian’s lack of direct involvement in a genomics network explains their lack of exposure to the socialisation and networks that facilitated other people’s entry into thinking about DTCGT and others’ subsequent interest and curiosity as they learnt more about testing. So while Jane and Ian were introduced to DTCGT and were interested in it as a result of their relative’s sharing information about experiences of it with them, they had no wider networks to reinforce or ‘socialise’ them in personal genomics other than the immediate kinship of their family unit. As a result, their expectations of testing seemed different from the other participants’, being less specific and arguably vicarious, having agreed to be supportive of a relative’s enthusiasm for testing. They both refer to discussing SNP genotyping with their relative before testing;

the details of this discussion were not elucidated but it appears that they were not clear about how testing would work, what sort of information they would be given, or when they would receive it. Consequently, when confronted with results they were not expecting, both Jane and her husband Ian found this difficult to deal with. Jane explained her concerns, which related to genealogy, very early in the interview.

So, tell me about it then, what was it like, how did you feel about it at the time? (Teresa)

Well, umm, I suppose I was nervous about it but [name of relative] was very reassuring and didn't push us into it; it was our own decision and I thought that it was just going to be just one test and that would be it and one set of results. What I didn't expect was, and I don't even know if [name of relative] knew, was that it would be continuous. So I'm continually getting emails and they are continually looking at my DNA test ... and there'll be an email saying there are six new results. And so that was something I wasn't expecting, so to tell you the truth, umm, I don't look at them any more ... The surprise came in the ethnicity when I got an email from a gentleman saying that he had the same maternal genes as me so he thought that we were probably related. And then I went on and you can like map your ethnic origins and it came up with quite a strong probability that one of my grandparents was Jewish. And there's nothing known about that at all [in the family] and when my sister got hers done, hers said the same thing and my Mum's said not at all and so there was a bit of confusion there. That was very confusing for me and I talked to [name of relative] about it ... you know the whole thing really for a while affected my identity and made me very curious and I couldn't get to the bottom of it. My father's dead and he was brought up as a Methodist in ... in [x]-shire [England] and it's like an old family you know that you can trace back and I wondered whether it was on my Mum's side because her mother was brought up in London and in the East End but no, there was nothing on her side so there's

definitely something on my Dad's side. So you know that wasn't something that I expected at all and umm, would not, so the experience was more upsetting than I expected. (Jane UP11)

Jane's assertion that she was not expecting repeated emails with new information suggests that she may have anticipated more deterministic health information rather than the probabilistic, contingent data her results were updated with. She repeats the words "continuous" and "continually", implying intrusion by the test information into her life as Bunnik et al (2011) suggested people might. This is compounded by her distress in relation to the unexpected and unsought information about her ethnicity, as shown in her references to the confusion and impact on her personal identity. Unlike Jane, Ian was very positive about finding out the ancestry aspects of his test results. However, like her, he was not anticipating the updates to the test results and expressed his consternation about this repeated return to a state of uncertainty on receipt of 23andMe emails in dramatic terms.

So, probably about a month or two later I got an email from 23andMe and I foolishly thought that the test was the test and that's it. I didn't really, obviously you participate in this thing and your information is used but I didn't know they'd come back to me and say "Good news we've found more diseases you're going to die of" and it's like "Oh shit what's this?" And of course there's change and every time an email comes in every month or so there are like new diseases or changes to your propensity for the diseases, some have gone up, some have gone down and that is quite shocking actually and of the whole thing that is the worst part of it. It makes you realise that there is so much being found out all the time that you realise that your first test, the whole thing is in its infancy really, it was that and also they didn't do an Alzheimer's test but now they can do an Alzheimer's test and here's the results so here you go again. If you choose to read them you have got lots of information coming at you if you choose it, which is really good in many respects but it means you go through this ordeal quite often. (Ian UP12)

Why was it shocking? (Teresa)

I just foolishly didn't think they'd send me more information, it was just stupid really. In the modern day if you participate in something you get more information and that's good but it wasn't made clear, or maybe I just didn't read all the information, anyway. I don't think my wife knew either. (Ian UP12)

Unlike other users' experiences, Jane and Ian had not personally been members of genomics networks of any kind prior to testing, nor had they researched SNP genotyping. Consequently they were not party to enactors' expectations of the technology or socialised into networks that would have facilitated this. Brown and Michael refer to the relative certainty of end-users (or "selectors") of new technologies in their reference to MacKenzie's certainty trough (Brown and Michael 2003, MacKenzie 1990). It is possible that this couple were not in a position to be identified with those selectors as they had not been recruited as actors in the personal genomics networks early enough to develop their own expectations of DTCGT. Although they demonstrated understanding of the nature of the test, their reliance on their relative for that proxy engagement in personal genomics networks appears not to have provided the socialising and understanding that other users conveyed. They suggest that they found continuous risk information shocking rather than empowering, possibly because it generates more uncertainty about their health and risks of disease. This points to the different experiences different types of users appear to have of DTCGT, which in turn illustrates the potential for different technological frames and impacts on the technology.

Kinship was important for users who had a family history of chronic diseases. For these users there were expectations related to finding out what information their SNP genotype might reveal about their family health history or their own health concerns. Their expectations of testing in the context of the family were about gaining information that might shed light on a known family history of illness and were informed by the stories and experiences of the family's ill health that were shared among the family network, as Richards describes (Richards 1996). Here, the network and sociality are familial and focused on

shared DNA. In addition, all but one of the participants who expressed personal or family illness as a factor in their decision-making to buy a test were members of established networks in either genetic genealogy or their occupational groups, as previously discussed. The socialising aspect of these networks seemed to give them the confidence to pursue testing for common complex disease risk (as well as genealogy) to gain additional genetic information to add to their existing knowledge about themselves and their family's illness.

Christine, the lecturer in public health, had a family history of gastrointestinal tract malignancy, both her parents having recently been diagnosed; her hope for information about her family cancer markers was an expectation of DTCGT in the hope that she could avoid the fate of her parents.

We talked about 23andMe and I told her [Carol] about my unusual circumstances with my parents. So my mother died of [upper gastrointestinal tract] cancer ... which is one in a million. And unfortunately just recently my father's been diagnosed with [different upper gastrointestinal tract] cancer. So two in the family and even if that was the pathology it's rare. (Christine UP5)

Christine's discussions about testing with her friend Carol gave her an opportunity to explore her expectations of testing with a friend who was familiar with DTCGT. Here she is describing how her distressing experience of her parents' illness contributed to her discussion with Carol about DTCGT. She is sharing her family cancer history that she thinks may have a genetic component and discussing with Carol whether DTCGT could shed any light on this. In their discussion they are socialising DNA through talking about family illness anticipating that DTCGT may give Christine some information about her risks should she decide to test. Her expectation of finding out about any genetic component to her family history is presented in terms of her using the information to try and avoid developing cancer herself in future.

Laura, who works as a computer biologist in cancer genetics, had envisaged testing in relation to her own health issues and her expectations of obtaining

relevant information were brought into focus by her father's ill health and her brother's anxieties about the impact of their father's illness.

It's interesting from the point of view of looking at what I have tendencies towards and my family as well. My Dad's been pretty ill and my brother's been quite worried about his health as a consequence of his [own] genetic makeup ... we all realise it's just a tiny snapshot of your health and the things that contribute, but it's a start. (Laura UP15)

Laura refers here to her interest in what she has "tendencies towards", although she is aware that SNP genotyping is not deterministic of health outcomes. But she situates her genetic identity within her family, acknowledging her identity as part of that immediate network and its cohesive view of genotyping with her phrase "we all realise". She and her family have been socialising DNA by discussing their health problems and the possible genetic aspects of these as indicated by her brother's reported concern "as a consequence of his own genetic makeup". They have done this in the context of DTCGT and what it might reveal, albeit "a tiny snapshot", about their shared DNA.195

Novas and Rose argue that individuals are located in matrices of networks, notably in relation to genetics, in a "network of relations" (Novas and Rose 2000:490), a point that is supported here by these individuals talking about illness in the context of their family network. The family 'condition' and the role it plays in the family's story appear to be contributing, in part, to their thinking and expectations of their DTCGT.

It seems clear from this aspect of the data that the social aspect of knowing people who had tested, finding out about testing through others' experience and sharing one's own experience with a knowledgeable and sympathetic person are important factors in deciding to buy a DTCGT, whether the networks providing that support are occupational, recreational or family-based, or a combination of these. This is also a feature of the role of Bijker's technological frame in shaping these actors' actions and interactions. The networks seem to be established in people's lives prior to their deciding to buy a test, rather than being something they become part of having done so. So

rather than exemplifying Rabinow's biosociality, these users (apart from Jane and Ian) represent the social networks Brown and Michael suggest enable engagement with new technology by reducing members' uncertainty (Rabinow 1996, Brown and Michael 2003).

Having explored how users socialise DNA and are influenced by doing so, I shall go on to contrast how the clinicians' social world of clinical genetics and genetic counselling are instrumental in shaping their views of DTCGT.

Socialising DNA in the wings of the clinic

The focus on the clinic as the site for studying medicine and its relationship with patients has been crucial for demonstrating how medical dominance and power are exercised, although Paul Atkinson highlights the disproportionate emphasis of sociological analysis of health care on doctor-patient consultation (Atkinson 1995). However, it is important to note that much (if not most) medical work is conducted in arenas away from direct interaction with patients. Thus, the basis for the clinician participants' socialising DNA, as demonstrated when talking to me about DTCGT, are the ideals and beliefs developed and shaped in forums external to the clinic, although ready for and informed by practice in it. The idiom 'in the wings' also suggests that clinicians are waiting for consultations about DTCGT, which, for the most part, have yet to materialise for the majority of practitioners I spoke to. A low referral rate to clinical genetics services following DTCGT is in common with early, if non-generalisable, research findings (Giovanni et al 2010, Brett et al 2012).

For the clinicians, professional status as a healthcare professional (of whatever kind) and practice in genetic counselling provide the social milieu and networks within which their values and beliefs about DNA are shaped, shared, reinforced and confirmed. Whether their expressed values in relation to DNA and DTCGT are simply informed by the literature or are based on wider understanding and experience, professional identity and authority as genetics clinicians provides its own social context for these participants' beliefs and ideals about DNA. The status and professional power of this relevant social group could be influential in shaping DTCGT in very different way from the users, not least because of the influential role clinicians are perceived to play in the wider context of the NHS that this study is concerned with.

The clinicians referred to DTCGT from a genetic counselling perspective, comparing the two models of genetic testing in a moral context. The principles of genetic counselling to inform, facilitate autonomous decision-making by an individual and/or family members in a non-directive manner, gain consent and maintain confidentiality were aspects of testing referred to when considering their views on DTCGT. Information giving was the principle that featured most prominently in the interviews, as promulgated by the professional bodies that regulate clinical genetics in the UK (BSGM n.d.).

In talking about genetic testing, the clinicians promoted the morality of genetic counselling as embodied by their professions, demonstrating boundary work in relation not only to the morally problematic nature of DTCGT but also to its perceived technological deficits that contribute to the on-going difficulties with its stabilisation in the clinical community. Not only is DTCGT criticised (and neither stabilised nor accepted) within clinical circles, but also boundaries in relation to clinical genetics were expressed by all the clinicians regarding what is appropriate for patients to discover (about their genetics) or not – that is, the moral dimension to boundary work. In addition, the doctors and scientists also engaged in boundary work regarding what is scientific or not. Both these aspects of boundary work illustrate this relevant social group's capacity to shape DTCGT negatively and asymmetrically in relation to users more positive technological frame.

Joanna Latimer has written about how the medical profession increasingly aligns itself with science or identifies its work as scientific, particularly in respect of diagnosis as performed in the clinic and how medicine provides an obligatory passage point for the legitimisation of biosciences (Latimer 2013). This is important in respect of the clinicians' socialisation of DNA because it explains the position from which they feel able to make judgments about SNP genotyping and perform boundary work in respect of judging DTCGT to be scientifically flawed compared with clinical genetics. As Gieryn suggests, boundary work is a practice undertaken by professionals when trying to exclude rivals, protect professional autonomy and monopolise an aspect of practice (Gieryn 1983). It is also a means of constructing a social boundary around what members view as legitimate science (in this case clinical genetic testing) to distinguish it from non-science (SNP genotyping) with the authority

to do so facilitated by their professional influence. I also suggest it explains the clinicians' focus on identifying the problematic nature of DTCGT and the basis for their shared social struggle⁹ with the discourse of commercial genomics.

In this section I shall demonstrate how the clinicians demonstrate professional authority and engage in boundary work in order to socialise DNA within a moral framework and restrict genetic testing to their shared world of clinical genetics. I shall also explore how they use the arguments of flawed science and misleading marketing of DTCGT to present their social views of DNA as superior to that of the DTCGT model. Finally, I shall examine their concerns for the public, whose understanding of genetics many of the clinicians appeared to frame within a deficit model therein illustrating this relevant social group's assumption of being influential in respect of dismissing DTCGT in the wider context of the NHS¹⁰.

The moral imperative in boundary work for clinical genetics

The genetic counselling model's influence in socialising the clinicians' views about genetic testing is evident from all of the clinician participants' references to the non-directive counselling model when expressing their thinking about DTCGT. Most focused specifically on the concepts of diagnostic questions and informed consent and their centrality to genetic counselling, although a few also referred to helping people deal with difficult test results. The lack of opportunity to make people aware of the implications of genetic information and facilitate non-directive decision making through genetic counselling was referred to by many of the clinicians as an aspect of DTCGT that is of concern to them because no face-to-face pre-test counselling occurs. This participant, a scientist with extensive genetic counselling experience, represents a concern about the lack of counselling with DTCGT expressed by the majority of the clinicians.

I'm interested in what motivates people to go for the test. You know, why do people trundle up to genetics in the first place?

⁹ The struggle is 'shared' and 'social' in the sense of being commonly experienced across the different professions who provide genetic counselling.

¹⁰ A deficit model is one where it is assumed that the public have a knowledge deficit. In this case it is in relation to knowledge of genetics and genomics (Dickson 2005, Jordens et al 2009).

People have genetic tests for many different reasons and I do wonder then [if] that will probably be reflected in why people have DTCGT ... you need to have a conversation about what the test can and cannot do for you. You need to ask questions about the detection rate for particular conditions depending on the ethnicity and the type of defect, what is the rate in that population. The difficulty for the public is that it doesn't give you a yes/no answer. And also making them aware that certain genetic tests can affect life insurance. You need to make them aware that they need to be mindful of that. What answers were they wanting to the questions they were asking? So it's not the test that's important it's giving people the strategies to cope with uncertainty. The people who go for genealogy tests, what does that mean for them? What if someone who has a 50 percent Viking inheritance suddenly is a genotype that's recognised to be associated with Sudden Adult Death. Who owns that information? Does it get fed back? It's Pandora's box. Part of me thinks it's Pandora's box open and the worms are crawling out thank you very much and you can't put them back in. (Scientist CP1)

This excerpt illustrates the moral aspects of the boundary work she is engaging in, with her association of genealogical information to potential pathology and her reference to Pandora's box, an expression she was not alone in using. In referring to needing "to have a conversation", she expresses concern that people may test without being adequately informed and be harmed by receiving upsetting information, by subsequent discrimination, or by the uncertainty testing is likely to provoke.

The relevance of family in genetic counselling and information giving was presented as a specific aspect of the moral work of genetic counselling. Although one of the clinicians suggested that the lack of predictive value of the DTCGT negated the need for counselling about the potential impact on family members, this scientist represented more widely expressed concerns about individuals not realising the potential implications for relatives when thinking about genetic testing.

Our job is to raise the issues that people wouldn't necessarily think of or if they did think about them, might not really address. So yeah, I think what we do say to people is "If you get a result you don't want, then what will you think and how will you talk to your family about that and how will you think about your children who then become at 50 percent risk? Will it make a difference to your life and is that in a good way or not in a good way?" I think those are the things that you wouldn't necessarily [think about] if you were just paying 99 pounds on the Internet, you wouldn't necessarily go through that thought process. (Scientist CP7)

Participants highlighted the importance of counselling face-to-face to establish the consultant's understanding and facilitate decision-making, as the next excerpt illustrates. Some suggested that counselling was not always necessary and that telephone conversations could be a substitute in some circumstances. But the importance of interaction between professional and consultant was demonstrated by most clinicians, referring to people's need for information, time to think about the potential meaning of test results for themselves and their families and how counselling changes people's views of genetic testing. Here clinicians are engaging in moral boundary work by contrasting the questionable relevance of tests and lack of counselling or informed consent afforded by an online commercial genomics company with professional genetic counselling. This nurse describes in some detail the importance of face-to-face interaction in her counselling and its centrality to the moral work of genetic counselling consultations, points which other participants also made.

I think there is a role for telephone counselling but it doesn't anywhere equate to face-to-face. You can't read somebody's body language or the tone of their voice if you've never met them before to be able to pick up any concerns that they're not voicing but if you saw them face-to-face you would say, "You're looking confused there, would you like me to..." So that's really my main concern that they're going to be either getting a result that makes them feel that they're definitely

going to get a cancer and what do they do with that result? Or they're not going to get a result, which is far more likely and they're going to then not be any further reassured and still have this unrealistic perception of what their risk is. You know as part of our counselling even if we can't offer a genetic test we talk about health promotion issues such as lifestyle, diet, what screening opportunities are available etcetera but someone going DTC is not going to get that healthcare promotion side of things ... whereas, I would like to think that coming to see us they've always got a named person that they can contact that they have met that hopefully they feel comfortable with contacting ... and in actual fact our pre-test counselling should go over "If you get this result it means this, if you get that result it means that. If you get that result I will then refer you so that you can be followed up by this clinician; if not then it means that, talking about cancer again, it does not mean you will not get a cancer but it means that you're at population risk etcetera etcetera. (Nurse CP16)

As I will go on to demonstrate in Chapter Six in the section on Responsibility, clinicians were not necessarily opposed to the public buying SNP genotyping tests directly. However, the wider context of DTCGT in relation to the NHS is critical here. Clinicians' socialising DNA within the clinical genetics world was demonstrated through their moral boundary work to maintain their institution and authority by approaching personal genomics from their genetic counselling perspective, and preserve their roles in the collective medicine provision of the NHS. For many of the clinicians this view was reinforced by the questionable validity and utility of the data DTCGT provides in contrast to the marketing hype of the companies marketing testing. I will go on to examine these aspects of the data next.

Flawed science and misleading marketing

In the last two decades, advances in genomics have been foregrounded with hyped expectations that have so far not been realised, despite the vast amounts of data generated (Franke 2010, Groves and Tutton 2013). This hype,

whether about the scientific capabilities of genomics or about the potential and deterministic value of SNP genotyping as described by commercial marketing, was used by clinicians to contrast the problematic nature of genomic data compared with that generated in clinical genetics. A majority of the clinicians referred to the uncertain state of the science that SNP genotyping is based on, problematising it in relation to their knowledge of clinical genetic testing. Uncertainty about the meaning of genomic data was key to most expressed concerns about DTCGT specifically and genomics generally and was expressed by doctors and scientists, giving an insight into this groups' technological frame of DTCGT. In an example of socialising DNA from the clinicians' perspective, one doctor training to be a clinical geneticist contrasted what she referred to as the "wishy-washy" nature of DTCGT with the detailed research studies on sequencing specific cancer and cardiac disease genes, supporting the widely-expressed view that standard public health advice for healthier living is as useful as any information DTCGT may provide (Van Ommen and Cornell 2008, Henrikson et al 2009, Cherkas et al 2010, Dickenson 2013).

A consultant specifically related the uncertainties of GWAS data to diagnosis and clinical intervention, representing some clinicians' concerns about the science of SNP genotyping well.

On the whole we haven't seen this great flood of extra information yet that's had this impact from GWAS. And the SNP typing, the DTC, they are just the baby versions of them. They're just taking the output of them and sticking them on some kind of testing platform be it a chip or a nucleotide re-sequencing platform and generating data that I'm not sure then translates into any useful clinical intervention. You know if we're still struggling at the research discovery phase of these I think then taking this to tell you "Oh look we can tell you your future", is a bit naïve. It seems to me as well that many of these GWAS SNPs, the relative risks they come out with is somewhere between 0.9 and 1.1, or a relative risk of 1 point something and you know that's no relative risk at all in real terms. That's just teasing out round the edges ... and my criticism of them from everything I've read is so what? I don't

think much of them. So, it's a bit harsh but you can say 'snake oil'. (Doctor CP13)

Here too there is evidence of socialising DNA in the phrases "everything I've read" and this clinician's use of "we" referring to his department's experience of referrals for DTCGT interpretation.

Two counsellors, both with a molecular genetics background, contrasted the approaches to SNP genotyping that DTCGT companies use with their expectations of testing for similar diseases and questioned its validity, particularly in relation to the populations the GWAS studies data used by the companies were based on. This scientist's example of cardiac disease details the contrast and possible impact of a DTCGT test versus those her service would offer.

They say they would test for a 25 base-pair deletion in *NYBPC3* which is basically one of the four genes that we've found the majority of the mutations in. And actually that 25 base-pair deletion is not one that I encounter on a regular basis with my patients. So that's interesting. I could name two other mutations in *NYBPC3* that I would put at the top of the list if I was doing targeted mutation analysis but yeah, if you're only testing for one mutation in one gene, if you didn't know that we have thirteen genes that we currently test for that condition and the mutation can be anywhere in those genes then you might think "That's it, I haven't got that mutation and I'm never going to get that disease." But there's one in 500 people in the population who are affected by it so that's not true ... and they said it was more common in Indian populations or something like that so there's a founder effect there, or something like that, that will be more relevant to a different ethnic group. (Scientist CP8)

In pointing out the difference between sequencing genes for mutations that cause people's illness and looking for SNP variations, this scientist is expressing her concerns about the different methodologies of different types of

genetic test. She suggests that people who buy SNP analysis will gain inadequate information from their test results and could be falsely reassured.

The clinicians' criticisms of DTCGT for disease risk seem linked to their concerns about the deterministic approach that DTCGT companies take to marketing their health genotyping services and illustrates their alternative technological frame of DTCGT on scientific grounds. Some clinicians voiced concerns about companies' use of terminology that contradicts the uncertain nature of genomics knowledge, with the potential to mislead the public about the nature and value of the products being marketed. This consultant's interpretation of the misleading nature of DTCGT marketing directly highlights professional concerns about companies' deterministic rhetoric and their target market, a genetically deterministic public.

Yeah, I guess I have a view that I'm not against cutting out the health professional; that doesn't bother me so much as a feeling that a lot of people who use it at the moment are being misled. And they might only be being misled because their starting point about genetics is not quite realistic and that's such a common thing for people to think it's all about the genes. It is so common for people to think that a genetic test will tell you more than it often does but without being able to move someone back from that position a test can be quite misleading I think; particularly because it's commercial and they want you to carry on thinking that. They don't want you to think it doesn't do very much. (Doctor CP4)

The emphasis this doctor makes is on how the public are in danger of being misled by companies' marketing approaches, because they do not provide pre-test information in the detailed manner that a genetic counselling consultation would. She plays down the need for genetic counselling but simultaneously points out the necessarily deceptive nature of DTCGT marketing, implying that this could be harmful.

Uniquely, this nurse expressed her understanding of the business plan for 23andMe as being driven by biobanking, but contextualised this in relation to the companies' marketing practices, echoing other clinicians' concerns.

I didn't buy into the argument that direct-to-consumer tests were a terrible thing because there were all these terrible ethical problems and they [users] were going to be incredibly damaged by what they found out ... I suppose I personally feel that because nothing in there is very predictive, it wasn't actually a health thing at all and that it was not very useful ... I saw it as a consumer issue but with the challenges of getting enough information to people so that they understood what they were buying. And it seemed to me that actually that could never be met because if you gave the true information that what susceptibility testing offered was worthless then why would you buy it? If you gave the real information, you know "BRCA test has really significant information for you and your family, it's available on the NHS free, available under certain circumstances" why would you buy it? So for me it was, it was never on that basis that this was so dangerous it should be forbidden. I felt it should not be offered because it was useless and things were being misrepresented ... my impression is that most companies have gone out of business that were operating and that the major company 23andMe is actually doing something very different. It's not marketing health-related tests like the impression given, it's using that opportunity to get data and that potentially is what's going to drive its business from now on. Because it always struck me right back at the very beginning, where is the money in this? I couldn't see where the money was. (Nurse CP5)

This nurse initially dissociates herself from published criticisms of DTCGT and its potential harms. Instead she focuses on the morally problematic activity of companies selling useless products to gain data for future commercial uses under the cover of marketing useful information to the public (at least in the case of 23andMe).

Professional group differences

Clinicians who questioned the scientific basis of DTCGT were all doctors or scientists. The four scientists all discussed the problematic nature of DTCGT in relation to the inadequate nature of SNP genotyping as compared with the detailed genomics research they were either familiar with or engaged in. Most of them also expressed insight into the tensions between the paternalistic nature of genetic counselling and public engagement with technology, as I will demonstrate in the final section of this chapter. One nurse also referred to the lack of validity of SNP genotyping in the context of tests provided by personal genomics companies. However, she has a PhD in genetics, like many of the medical and scientist participants in the clinician participant group. The remaining three nurses, who have gained genetic counselling posts and registration by experience rather than undertaking the MSc in genetic counselling, did not refer to the molecular genetics or the scientific basis of testing at all in their interviews. Their concerns seemed more focused on the moral aspects of genetics, judging by their references to the lack of genetic counselling principles in DTCGT as compared with their genetic counselling responsibilities. This illustrates the influence of different professional perspectives on socialising DNA and shaping the DTCGT from the different sub-groups in this relevant social group. Whilst there is undoubtedly common ground between the different professions working in clinical genetics, each professional group has its own values and identity and thus its own version of the technological frame of DTCGT. Nurses' primary concern and their professional values focus primarily on caring, higher academic learning in sciences being a relatively recent development in nursing education. This is in contrast to doctors and scientists, whose identity is primarily founded on education in sciences. The foundations of these occupations are influential in the way the clinicians socialise DNA. One experienced doctor with leadership responsibilities noted differences between genetic clinicians on the basis of their professional background and education. This extract illustrates the differences between the professions well.

It's about understanding the data that the prediction's based on, not believing what the company actually report and looking up the evidence the results are based on. I think for some genetics counsellors that must be quite difficult if they don't come from that evidence evaluation framework like I do.

Some of them do, some of them very much do, but just not all of them. Genetic counsellors come to the profession from different places and they've been in it for different lengths of time and they have different skills. They've got very different backgrounds with different skills. Often those that come from nursing backgrounds for a long time are very good communicators and they have a really good understanding of disease and it's breadth and of pathology and how that pathology impacts on people and they understand well about the genetic stuff they know. Particularly when they are junior, the concept of things like relative risk and numbers needed to interpret studies, to come up with a conclusion, understanding the process, can be less in-depth than perhaps someone with an MSc in Genetic Counselling who has an epidemiology degree who will just come in in a different place. People who come in with medical training do to some extent get layered on the stuff about how you appraise evidence and how much evidence you need to appraise something and I think that is part of genetic counselling training but it's just not as embedded, not as deep. (Doctor CP2)

The doctor points out the different skills that different professions bring to genetic counselling. She implies that nurses do not have the scientific grounding to be able to interpret evidence and communicate risk clearly, although they have excellent communication skills and that this might be problematic for interpreting DTCGT. This observation points to the social differences between professional groups; it highlights the influence that professional cultures have on how genetics clinicians' technological frames of DTCGT differ. This is due to how they socialise DNA differently according to their profession and perceive those differences in their colleagues.

In terms of professional cultures and their influence on their members' views and practice, the other notable professional difference in the way clinicians socialise DNA is the manner in which the scientist counsellors and one nurse referred to the public's engagement with genomics, as I noted earlier. In the next and final section in this chapter I shall show how clinicians' and also some

users' beliefs about public understanding of genomics are part of their socialising DNA and how different groups had different ways of seeing this.

Public understanding

Durant et al (1996) suggest that the word 'public' is used in contexts where it can have one of three meanings. The meaning inferred in the phrase 'public understanding of science' is that the public are people who are outside the knowledgeable group in question. These people lack knowledge or understanding of that group's specialist, privileged knowledge and, in this instance, cannot socialise DNA in the same way the knowledgeable group do. In other words the public represent a social group with different understanding, knowledge and technological frame of DTCGT from the knowledgeable group, if indeed they have any. In the clinicians' opinions about DTCGT their meaning appeared to be congruent with this on the whole; that is, the public are people who are outside the healthcare professions and genetics and who lack understanding of either genetics or the privileged knowledge of the healthcare professions. In considering the issues related to DTCGT generally (rather than in relation to specific consultations), the clinicians referred to the public's knowledge and understanding of genetics mostly from a deficit perspective, as the preceding definition of the word 'public' suggests. Those who talked about this suggested that the public were unlikely to understand various issues relevant to genomics, and there were specific references to misunderstanding of relative risk or disease risk when expressed in percentages. This perspective was used to justify the need for the moral aspects of the boundary work exhibited, underpinning the need for genetic counselling so that the public would be able to exercise their autonomy from an informed perspective and not come to any (avoidable) harm. These clinicians were not alone in this view, as several of the users similarly suggested that the public, also referred to as "the man on the street", did not understand genetics. By aligning with adopters of genomic technology, they distance themselves from the public who, in their opinion, would not understand it. In their study exploring lay expertise about genomics prior to completion of the HGP, Kerr et al noted that the dominant deficit model of public understanding of genomics is key to the maintenance of boundaries by government, scientists and healthcare professionals (Kerr et al 1998). In addition, they found that lay participants in their study also subscribed to the deficit model in respect of the (rest of the)

public's understanding. Kerr subsequently noted that research participants who are recruited to studies as representative of 'citizens' often shift their identities towards a position of expertise, distanced from "the ignorant and amorphous public" (Kerr 2004: 123). This assumption does not appear to have shifted since then and is part of genetics clinicians' rhetoric in socialising DNA, as the excerpts below indicate.

Concerns about the public's understandings of genomics being based on a deficit model appeared, for the clinicians, to be influenced by practitioners' experiences of explaining genetic disease to patients and families in genetics clinics, but also perhaps by the more widely held view given that some users expressed it also. One scientist likened the public's understanding to knowledge gained from watching a fictional North American television drama about forensic medicine, whilst a doctor represented the more widely expressed understanding among the clinicians that the public think genetic information is deterministic.

People access tests for very different reasons and unfortunately I think the public do think its CSI. (Scientist CP1)

I think that people often start off in a position where genetics is really something very important and very deterministic. If they don't start from that position then I'm not so worried about it but I think it's really common in UK society to think that genetics determines much more than, much more in a clear way than I think it does. (Doctor CP4)

This expression of the public's deterministic understanding of genetics can be compared with Fiona's. Also a scientist, Fiona expressed opinions about the public's understanding of genetics that represents other users who are scientists also.

Even well educated people don't know very much about genetics and have a poor understanding about genetics. On one level that's a shame but at the same time it's not an easy thing to change. (Fiona UP8)

Concerns about people's knowledge and understanding were explicitly related to understanding risk by a number of clinicians who felt that the public were likely to misinterpret its meaning for them. One of the nurses expressed this clearly.

My fear is that quite often the information is put across to the patient and they become misinformed ... they get "You have a higher risk of cardiac problems" mixed up with "You're going to have a heart attack when you're older, or you're at risk of a heart attack." Well yes that's true but to a certain extent everybody is. There are lots of other parameters involved or appear to be in this mass-screening that people get and it's not often specific, it's in percentages. I suppose what I'm trying to say is people's perception of risk is very different and when you give risk in a percentage it very much depends on where that person's at as to how they'll take that on board and I think in some cases it can become misinformation for them. (Nurse CP9)

The nurse's views show how her experiences in nursing and genetic counselling have led her to conclude that patients' understanding of information is not always the same as that intended by the person giving it. She is concerned that the DTCGT companies' results will be misinterpreted by some and this implies that harm could potentially result without the provision of professional interpretation that genetic counselling would offer.

However, there were also some suggestions that the public *is* able to understand and deal with the information that DTCGT provides appropriately. A small group of clinicians, mostly scientists, referred to public understanding in more positive terms, demonstrating a perspective of the public as having resilience in relation to receiving genetic information. Also, some clinicians from other professions suggested that the public could access DTCGT without coming to any harm or causing them (the clinicians) concern. Neither an assumption that public knowledge is based on a deficit model nor understanding public resilience seemed to depend on experience of consulting with people who had used DTCGT. Indeed, there was also some ambivalence

with some clinicians expressing opinions that supported both points of view. This evidence of interpretative flexibility is resonant with Evans et al's exploration of the public engagement with genomics. They noted that individual actors involved in public engagement with genomics often had roles in more than one group, which suggested that ambivalence was more likely in actors' understandings rather than the habitually predicted pro-con dualism (Evans et al 2007). This could be interpreted as making the group's contribution to shaping the technology more complex as technological frames will vary somewhat between and within relevant social groups, as indicated in this study's participants. The following excerpt from one of the scientists is representative of an understanding of public resilience expressed by a number of other clinicians, framing it within a more objective view of the social world of genetic counselling.

That traditional route is obviously tried and tested but I can see how it might put some people off because... I don't know, it's time consuming, you have to speak to various different people; you can see that for some people it might be attractive to click a few buttons on the Internet, send off your 99 pounds... Because I mean let's say your sister had a child with cystic fibrosis. You might go on the Internet and say oh yeah that means I've got a 50:50 chance of being a carrier. So the traditional route would be go to the GP get referred to clinical genetics. Obviously you have to talk to your GP, you have to wait however long you have to wait and then come up to clinical genetics talk to someone, you have to wait again for a result and then you have to talk to someone again. That against "Well I'll just buy one of these, pay for one of these kits." I suppose it's just surprising to me that more people don't do that now I think about it because that seems a more private way of doing it. And you don't even have to tell anyone the result then ... it could be that we are making more out of this potential psychosocial risk, we are worrying too much about the impact. Maybe people can cope with these things better than we think they can or just that you know, going through a course of genetic counselling is not actually helpful. If they are upset ... then they'll talk to their

family or their friends and that helps them adjust to it rather than talking to us. (Scientist CP7)

This excerpt shows this participant's reflexivity in considering DTCGT. She highlights the negative aspects of genetic counselling as experienced by patients in her particular specialist area of practice. As she does so, she concludes that there are potential advantages to DTCGT for the public, thus indicating insight into the potential for the public's understanding and resilience and into alternative views of genetic testing and counselling.

Conclusion

This chapter has explored how DNA is socialised by participants in the study. It is clear that social networks are crucial in shaping the discourse about personal genomics. The technological frame of users may support engagement, as represented by the role networks play in facilitating users' adoption of it, whereas that of clinicians is more hostile towards it, as demonstrated by their performance of boundary work to distinguish and distance their professional genetic counselling service from the contingent and challenging aspects of DTCGT. In the wider context of the NHS, users' and clinicians' views about the relevance of DTCGT to health care can be summarised as support for either personalised medicine or collective medicine respectively and these aspects of the data analysis will be explored in detail in Chapter Six. But the networks of the different social groups that the users and clinicians in this study circulate within appear to be in tension. McKinlay and Marceau suggest that the golden era of medicine is over, or at least shifting to a somewhat less gilded state (McKinlay and Marceau 2002), in that its autonomy and power are being challenged from numerous fronts, including technological, administrative, economic and public challenges. The users' position in supporting the new technology of DTCGT could be interpreted as a challenge to existing medical hegemony, particularly when juxtaposed with the moral virtues of genetic counselling that the clinicians largely seem to promote.

This body of data would suggest that healthcare professionals working in clinical genetics continue to assume medicine's privileged status of power and authority, although some suggestions of public literacy in either genomics or healthcare decision-making are acknowledged. Alignment to a Parsonian

definition of professionalism, in which the ethics of care are based on service and altruism, is shown by many of these clinicians. However, these data would suggest that healthcare professionals in clinical genetics in the NHS perceive the divisions of knowledge and the relative power and authority attached to it along established boundaries of practice and expertise that other scholars have noticed remain intact despite expectations of change (Hedgecoe and Martin 2008). This supports the users' views of medical paternalism in relation to genetic and genomic knowledge. It also represents these groups' different technological frames of DTCGT as being based on the users side as being rights-based, democratising and challenging to medical paternalism whilst the clinicians' is based on moral boundary work, professional authority and their perceptions of flawed science.

Having considered the social aspects of participants' beliefs and values about DNA, the next chapter explores how participants think about DNA on an individual level and how their experiences of DTCGT has influenced their thinking about their identity and subsequent embodiment of DNA, either in their individual lives as users or in their practice as genetic counsellors.

Chapter 5 : Personalising DNA

Personalising DNA relates to the participants' individual ideas about DNA in the context of SNP genotyping and incorporation of its perceived information in their own lives or genetic counselling practice. Whether they were introduced to genetics through their education and work, or through genealogy, the participants almost all had an interest in DNA and genetics that provided the impetus for their further learning and engagement with testing. This learning in turn appears to have provided the scope for people to engage in identity practices, as described by Gibbon and Novas (2008), or to consider how SNP genotyping might affect consultands' identities. Their ideas and learning can then be incorporated into their lives, whether in the form of newly edited identities, life-style changes, or thinking differently about personal genomics or approaches to genetic counselling. As I noted in Chapter Four, socialising with others who have similar interests and knowledge will of course both influence and be influenced by individuals' expectations and embodiment of DNA. However, personalising DNA as represented by individual ideas and embodying of DNA, is different from external sharing in that it is about internal imaginings and identity practices rather than publicly shared understanding and beliefs which will be shaped by and during socialising. This chapter focuses on how the participants expressed these individual expectations and ideas about DNA and incorporated them into their lives. Sharing SNP genotyping results with family, though it could be construed as socialising, is discussed in this chapter in relation to embodying DNA, due to the inseparable nature of imagining one's own genetic identity in the context of kinship.

The chapter is made up of two main sections and will commence by considering participants' expectations and reflections on the DNA of SNP genotyping, what factors informed their thinking and led to users' pursuing further knowledge and engagement with the technology and clinicians' ideas about why people might decide to engage with DTCGT. The tensions between users' and clinicians' expectations will be illustrated by contrasting data from each group that relate to this theme. The second section will explore how participants embody DNA, that is, the impact results of DTCGT had on users'

lives and on-going identity practices will be discussed as will experiences of clinicians who counselled people who had questions about DTCGT results.

Expectations and reflections on DNA

In their exploration of DTCGT company rhetoric, Nordgren and Juengst point to the centrality of individual identity in contemporary post-modern society (Nordgren and Juengst 2009). Likewise, the users of SNP genotyping in my study seemed to be seeking genetic information with which to inform (and possibly enhance) their accounts of themselves. The future-orientated nature of thinking about and committing to buying DTCGT links to people's hope for information about themselves. They appear to hope that it will either be useful in relation to health or genealogy, or satiate their curiosity about themselves and their family and genomics more generally. The DNA test facilitates recreating their pasts as well as their futures in the light of their anticipated understanding of their extended (genomic) identity, as Brown and Webster forecast new medical technologies would (Brown and Webster 2004).

I earlier described the majority of the users in this study as early adopters of personalised genomics, people with what Jenny Reardon refers to as "learnedness". She describes early adopters as people at whom DTCGT companies initially specifically targeted their marketing strategies, in order to sell their tests and simultaneously increase their acquisition of data (Reardon 2011:97). The quality of learnedness I suggest is based on users' existing knowledge and inquisitiveness about genomics and their own bodies at the molecular level, that which Novas and Rose refer to as the "somatic individual", and which they can now begin to realise in the form of DTCGT (Novas and Rose 2000:485). I use the word inquisitiveness to mean an interest in how knowledge about their genome might influence their thinking about their identity, whether in relation to genealogy, physical traits, disease risk or carrier status.

Clinicians talk about DNA in the context of DTCGT by envisaging what the public's motivations and experiences of testing might be, wondering how these individuals fare on their solo genetic testing journey without the institutional support of genetic counselling and expert interpretation of results. For both groups, prior knowledge of genetics is fundamental; this is emphasised by the

contrasting experiences of the two users who did not conduct research into personal genomics prior to testing, as I will show when discussing users' reflections on and expectations of their DNA.

Users' knowledge, interest and curiosity

In this section, I will present data about how this particular social group of users developed knowledge of SNP genotyping by learning about it from the Internet and how this knowledge affected their thinking about testing and their individual genetic identities. The significance of participants' expressions of interest in personal genomics will also be explored and these two areas contrasted with Jane's and Ian's rather different expectations and ideas about their DNA.

Knowledge

Prior to obtaining results from testing, most users described reflecting at length on their expectation that SNP genotyping would reveal information about their genetic selves, whether in relation to genealogy, traits or disease risk and envisaged what this would be like from the reference point of their knowledge of genetics. It is clear that their knowledge and engagement with genetics was important in influencing their interest and expectations about their test results. Given the mixed background of the group of users, it appears that specialist prior knowledge was not a prerequisite for developing an interest in new genetic technology (assuming that only the genetic scientists fulfilled this criterion). Most users either felt they already had a sound working knowledge of genetics generally and SNP genotyping specifically, or spoke of developing that knowledge by undertaking extensive research into SNP genotyping before deciding to take a test. This aligns with Reardon's assertion that personal genomics companies deliberately sought to attract followers who were knowledgeable and able to consent to SNP genotyping (Reardon 2011). It also indicates that participants' learned, or extant knowledge of genetics equipped them with the vocabulary to extend their ideas about their DNA and entertain the possibility of reifying it by thinking about testing.

The following excerpt from David demonstrates his commitment to learning about genetics from a genealogical starting point, but he indicates that this then widened to include health testing as well.

I first of all took a free test with Sorenson which gave me a first insight into what it was all about and it was quite a long learning curve but worthwhile spending all that time learning what it was all about and I became more and more interested in it ... the primary reason for taking that test was for family history but I was also aware that there was a huge health side to the test and I was interested in both. I was interested in learning about any health implications good or bad. (David UP6)

David had no background in genetics prior to becoming involved in genealogy. From this starting point he became increasingly involved in his family ancestry, which led him into genetic genealogy, in common with many genetic genealogists (Nash 2004). He indicates that he taught himself about genetics over time and became increasingly engaged with it as he learnt more, becoming as fascinated by the health aspects as the genealogical, regardless of the implications of what testing might reveal about him.

Christine had more understanding of genetics from her science background and research in public health. Although she was initially introduced to DTCGT by a colleague and was apprehensive about confidentiality, she researched it thoroughly on her own before deciding to test.

I read the fine print and all the rest of it and I was happy to (go ahead). I read reviews from other people, I contacted them to know how easy it was and what fees were involved and the fine print. I was hesitant at the start to send samples off like that but then I thought it was what I wanted and it was quite easy. It was easy, it was clear. (Christine UP5)

Christine illustrates how she read about testing on the Internet and followed this up by contacting others who had tested. She describes practical concerns, which were partly informed by having worked with human samples in research laboratories earlier in her career. The simplicity of the testing process seems to have encouraged her to finally decide to buy a test to find out more about the

genetic aspect of her identity, which she had been contemplating for a while, as indicated by her saying “it was what I wanted”.

Christine’s example also demonstrates how learning about SNP genotyping enabled her, in common with other participants, to develop new knowledge and language associated with personal genomics. This individual learning and the connections made during its acquisition appear to provide the skills and opportunities to engage in socialising DNA in communal forums providing an example of where personalising and socialising DNA intersect.

Interest and curiosity

It appears that DTCGT had influenced participants’ expectations to varying degrees, as indicated by their descriptions of their thinking about testing before deciding to buy and then while waiting for results. Most participants referred to their interest in personal genomics. Some explicitly expressed curiosity and two appear to have developed an arguably obsessive interest, having taken genetic genealogy tests with every company they could easily access.

Use of the word “interesting” in conversation can have various meanings. It may mean indifference to what has been said but be voiced out of politeness, be used as a time-wasting tactic in polite conversation or actually convey the person’s interest in the topic in question, although on occasion this may be in a general, non-committal fashion (Urban Dictionary 2013). However, use of the word by these participants mainly appeared to convey people’s politeness and genuine interest in genetics, their imagining their own genome and learning more about it or their learnedness about personal genomics. All but two of the participants repeatedly referred to their interest in personal genomics generally and in their test specifically, in relation to both the process of testing and the results. Some users used the word repeatedly, notably Carol, David, Helen and Maria, demonstrating their genuine engagement, as shown in the following excerpt.

This is really interesting. I’d be just fascinated to know, just fascinated to know on a personal level what I’ve got and what I haven’t got. (Carol UP4)

Carol's repetition of and emphasis on her "fascination" suggests her interest and learnedness based on her genetic knowledge from her education and work in public health research. She suggests that she has envisaged that her SNP genotype will reveal certain characteristics about herself that have previously been hidden but which she may have encountered in her family and her work and have wondered about in relation to her own identity. This is akin to Armstrong et al's portrayal of the concealed aspect of an individual's identity being revealed by genetic testing. The genetic aspects of a person's identity exist but are unseen, buried in their cells; genetic testing reveals them, rendering them visible. Unlike situations in which people have to change their identity due to diagnosis of illness or disfiguration, genetic information or diagnosis reveals a previously inaccessible aspect of identity (Armstrong et al 1998).

Three users who work in genetics, Fiona, Kirsten and Maria, all talked about wanting to see their own DNA data, rather than only researching and working with others', to put their daily theory into personal practice. They have seemingly imagined what their own DNA might be like, in comparison to that which they encounter in their working lives on a daily basis, and are curious about what the reality of their own genomic information might reveal. Fiona is a genetics scientist in her 30s whose interest was related to her work.

I suppose I was intrigued, I think because of the nature of the field I work in I've known about these things for a fairly long time and it finally got to the point where it would just be interesting to apply the things I know because of my work to something that was actually about me as opposed to something else. (Fiona UP8)

Fiona demonstrates her interest and its relationship to her expectations of personal genomics in her use of the word "intrigued". Referring to genetic information that is hers rather than someone else's points to her hope to extend her own identity and individuality into the genetic so that she can have similar personal information to that she deals with daily. Maria, a young PhD researcher in genetics, has a similar wish to access her own genetic identity; she voices her imagining and motivation more strongly, using the word

“narcissistic” to focus her curiosity on her interest in her own genome, as opposed to those she works with in the laboratory on a daily basis.

So I'd been interested in it for a while and ... you know as a geneticist the idea of actually looking at my own SNPs was quite exciting ... I was interested to see how accurate it would be and if I would find anything out and it was getting cheaper and really I did it for a narcissistic curiosity. It really wasn't for disease risk information, though I did find that interesting as well, but you know as a geneticist the idea of actually looking at my own SNPs was quite exciting so that was the main reason really, curiosity. (Maria UP16)

Maria seems to be challenging the SNP genotyping test by saying that she wishes to see how accurate it would be, presumably by comparing the results to her existing self-knowledge, but she also seems to be imagining her genetic identity and individuality as separate from, but in the context of, the genetic material she deals with on a daily basis.

Juxtaposed with the word “interesting” is the use of the term “curious” which a few participants used in addition to Maria, in relation to their thinking about testing. Unlike the word “interesting”, which implies a justification and a more responsible and informed decision in relation to testing, “curiosity” could suggest something more trivial and unfocused than an (informed) interest. “Curiosity” was often used by participants as a kind of excuse for their decision to take a test (Scott and Lyman 1968). The way the word was used by most participants reduced the individual's responsibility in relation to deciding to test, due to their (often implicit) suggestion that giving in to curiosity was something they were not fully in control of, undermining informed decision-making. Kirsten, who works in bioinformatics, puts it well.

Genome testing's been around and affordable for a couple of years now; I did mine last summer so I was slightly late on the bandwagon. I did it because I was starting to do methodological work in this area, how to put together SNPs and environment models and I had a PhD student working in

it, so I'd done some of the research and read some of the papers about, you know, how we do it and what the problems are and giving seminars on the statistical aspects of it and you know, just curiosity in the end just got the better of me. (Kirsten UP14)

In this extract, Kirsten conveys the influence of her work on her decision to test, another example of putting the theory into practice by experiencing the reality of testing and of a prior knowledge base informing knowledge and the decision to engage with SNP genotyping. However, she does also acknowledge her thinking about her own genome in her throwaway remark "curiosity in the end just got the better of me" and suggests that her final decision to test was based on the desire to know exceeding her caution and self control.

Christine, the public health lecturer with an interest in genetics and a family history of cancer, referred to being curious about her results while waiting for them to be posted in her 23andMe account.

I was excited too ... well the whole possibility because I know how many tests there are and they had claimed there was a thousand SNPs and I was really, really quite curious by that stage. (Christine UP5)

The anticipation of having her genetic identity revealed is conveyed by Christine's excitement and her emphasis on her curiosity by repeating the word "really". She suggests that the quantity of information that will be available is important in reifying her DNA.

Interest and curiosity were taken a step further by a few participants, mostly genetic genealogists, who either described having several tests or specifically referred to an "addiction" to DTCGT. Genealogical DNA testing includes autosomal, mitochondrial and Y chromosome DNA analysis, comparing SNPs or haplotypes from the individual's DNA with others from similar lineages, cultures or historical groups. These tests are available from companies including Sorenson, Oxford Ancestry and Family Tree DNA and preceded testing for common complex disease risk. Those interested in genealogy are familiar with DTCGT, having previously tested with one of these genetic

genealogy companies or knowing others who have. Indeed, two of these participants described testing as addictive.

Having done four tests, David, a lifelong genealogist, was the most experienced user of DTCGT. It is important to point out that his wife, Elizabeth, volunteered the information in the interview that David has Asperger's syndrome and this may explain his engagement with genetic genealogy and DTCGT in a more intense way than other users'. However, this does not undermine his knowledge or interest in personal genomics and he expresses his curiosity vividly.

After my free test with Sorenson, I then took a paid-for test with FTDNA, a YDNA test on more markers and lots more flexibility and help from that company. They give you a better insight into what the results mean. And then the autosomal DNA test became available first of all from 23andMe ... so I hummed and hah-ed for a while but got so intrigued I couldn't resist ordering a test even though it was expensive. I just went ahead and placed an order and it's been really interesting ... I couldn't wait to see them [the results]. If you haven't taken such a test yourself you have no idea of the anticipation of waiting for the results, it's a great experience really. Waiting, waiting, waiting and then bang the results are there, you dive in and try and understand it. (David UP6)

David's striking explanation of his experience of testing illustrates his impatience to extend his understanding of his genetic identity. In a manner similar to Kirsten, he describes a loss of self-control in being unable to resist ordering a test. His description of waiting for the results of the test is emphasised by his repetition of the word "waiting", thus conveying his excitement about the future possibilities inherent in getting his test results in a very similar manner to Christine. The waiting culminates in his diving into the results to try and make sense of this new information about himself as part of his on-going identity practices.

Clinicians' imagining DNA

Some of the clinicians also referred to identity practices and reflecting on DNA from a personal genomics perspective. Most of them imagined DNA from the perspective of users of DTCGT, but a few others mentioned a personal interest in the concept of DTCGT, using similar tropes as the users. This was mostly prompted by the context of counselling a patient who had bought a test, but demonstrates the seductiveness of thinking about one's own DNA and genetic identity. Two scientists (CP3 and CP8) and a very experienced nurse (CP9) all referred to being interested in finding out more or having a DTCGT. However, they were all quick to be dismissive of disease-risk testing, due to its lack of utility and concerns about it provoking anxiety. This could be interpreted as an illustration of the distinction between personalising DNA and socialising DNA and the interpretative flexibility required for members of the clinicians group to move between these points. After considering DTCGT from the perspective of their own DNA they reassume their professional group's discourse and technological frame of DTCGT.

I would probably be quite interested in doing it [genetic genealogy] because it's so bloody vague it's not going to tell you anything in reality but I can appreciate that because of the field I work in and I have a better understanding of genetic tests than most people and I would be interested in doing that, but I also know it doesn't mean a great deal in all honesty. But I couldn't do it for anything else, personally ... we all have innate curiosity but there are so many uncertainties [in genetics] and I wouldn't want to get hooked on a piece of information that preyed on my mind that in reality was irrelevant. Because I'm just as vulnerable to doing that as anyone else, when you're worrying about your own health you do. (Nurse CP9)

This nurse's comments are interesting because of the contradictory nature in which she phrases them and her interpretative flexibility. She expresses her interest in uncovering genealogical aspects of her genetic identity, but then immediately acknowledges that it would be unlikely to tell her anything meaningful and then repeats both assertions in slightly different ways. She goes on to refer to her "curiosity". This indicates that she has thought about

her own DNA and genetic identity. However, she excuses it by assigning her interest to a commonly held human behaviour, describing it as “innate” rather than something she is specifically interested in, thus distancing herself as a genetics counsellor from the public who might engage with DTCGT. She seems to imply that she is curious to extend her identity into the genetic realm. However, she simultaneously expresses caution, possibly because she imagines herself in the place of her consultands, with the potential for being distressed by aspects of her disease-risk in a manner more linked to genetic essentialism. This view is similar to that proposed by Alexandra Plows, who suggested that geneticised accounts of one’s identity have the potential to eclipse the additional aspects that comprise that identity (Plows 2011). This nurse’s response is also indicative of the tension between the personal and professionally social, accessing her own DNA and engaging in the powerful moral boundary work of the genetic counselling clinic she did along with the other clinicians, as demonstrated in Chapter Four.

For some clinicians, personalising DNA was related to thinking about the perspective of people who want to engage with DTCGT. They talked about SNP genotyping in relation to the public, envisaging users’ motivations and experiences. Despite the intrinsic tensions between the moral boundary work of the clinic and imagining personal genomics I referred to above, three clinicians, two scientists and one junior doctor (CP8, CP9 and CP14) acknowledged people’s desire to find out about their genetic identity and their ability to think about genomics in a meaningful way.

I suppose they start from a position of wanting to know something and if they realise it’s commercially available you know, why not? I think from looking at the website and understanding it a bit more and also from anecdotal stuff that I’ve heard there are two groups of people. There are people who are going to approach it from a health point of view and there are people who want to approach it from the genealogy, ancestry point of view. (Scientist CP8)

This scientist acknowledges people’s interest and curiosity, their wish to discover aspects of their genetic identity, whether in relation to their health or

genealogy. Her personal research into commercial SNP genotyping had extended her knowledge of DTCGT and thus enabled her to imagine DNA from a different perspective. Another doctor anticipated, as did a number of the users, that people who would engage with DTCGT would be knowledgeable about genomics.

I would imagine that people doing the direct-to-consumer tests would have done a bit of reading and would have thought about it. I think your average person on the street who wouldn't have thought about it wouldn't go for it ... There will be people with a certain amount of awareness and the ability to look through these kind of Internet sites and know about family history and that kind of thing. So I think they'll need to have a certain level of knowledge or understanding before they go to the direct-to-consumer test. (Doctor CP6)

This doctor envisages that those who engage in genetic identity practices are likely to be knowledgeable about genetics or in a position to educate themselves about it. She uses the expression "person on the street" to refer to a public who are not knowledgeable or in a position to develop their knowledge outside the learned group in question (as I discussed in Chapter Four). This indicates her assumption that those who engage with DTCGT are less likely to be harmed by the experience on account of their prior knowledge, unlike some of the people she encounters in her practice who might be.

Worrying about DNA

Interest and curiosity about genetics appear to be dynamically associated with knowledge and learning, both prompting and being the result of people's discovery. The majority of participants framed their experiences of learning and thinking about personal genomics positively, describing how they imagined their DNA whilst anticipating the arrival of their results. However, as demonstrated in Chapter Four, Jane's and Ian's experience was different from other users'. The heterogeneity they contribute to this study's users' group could also be interpreted as belonging to a different relevant social group in relation to shaping DTCGT, one in which group members have less knowledge of genomics or understanding of the test's capacity to provide useful

information. They are the couple who had their DTCGT bought as a gift and I shall examine their alternative experience of anticipating their DNA, before going on to explore the expectations and reflections of those whose motivations for testing were related to health.

As outlined in Chapter Four, Ian described himself as having an interest in “pop-science” and genetics, which he shared with the relative who bought the tests for him and his wife. However, neither he nor his wife had any formal education in biology or genetics, nor did they undertake any research into genetics or personal genomics, beyond talking with their relative, prior to deciding to test. They were persuaded by their relative’s enthusiasm and agreed to testing on that basis but had not been socialised into the world of DNA testing which seemed to give them a different experience of DTCGT, at least initially. Jane described being nervous about testing beforehand and having no preparation for the reality of receiving the results. Ian describes having similar expectations of his DNA after Jane’s concern at her results, which were received two weeks before his. This gave him additional time to dwell on what his genetic identity might reveal.

Jane got her results first and she was really nervous and I hadn’t thought about that until she got hers. She looked at them at work and then at home and saw something in the email that made her think she’d been tested for something and she was really worried about it and it made me think this is quite a thing we’ve taken on, so by the time I got mine I similarly had my heart in my mouth and got really worried which is silly because we knew, I particularly knew, it would only raise your propensity by a matter of percentage points, it’s not a death certificate but ... (Ian UP12)

Jane’s reaction seems to prompt Ian to think about testing from a different perspective than previously, as indicated by the phrase “this is quite a thing we’ve taken on”. He then appears to consider the possibility that his genetic identity may contain something sinister and communicates his anxiety in relation to this in his use of rather powerful terms including the phrases “heart in my mouth” and “death certificate”. However, his subsequent use of the word “silly” suggests he later decided the anxiety was unwarranted.

Despite the difference in Jane's and Ian's experience of DTCGT, neither of them had initially been concerned about reifying their DNA through SNP genotyping and neither had specific health-related concerns that informed their imaginings or their decision to agree to accepting their relative's gift. A few of the users had specific health-related concerns that influenced their thinking and I shall discuss these next, before completing the section on Imagining DNA, by discussing the tipping point in the decision to buy a DTCGT.

Genetic identity and health

The concept of a genetic identity influenced some users' responsibility in relation to their health and this was used as a justification for testing. For Helen, Laura and Nicola, SNP genotyping was a way of getting health-related information independently of any healthcare professional, as this had proved unhelpful in their efforts to shed light on their chronic health-related problems. Barbara and Christine had pursued DTCGT partly out of concern about their family health histories, as had Keith, although his motivation was primarily for genealogical information. Rather than feeling anxious about potentially alarming information, most of these participants, along with the majority of users, referred to the disease-risk aspect of testing as knowledge to be used responsibly by themselves and possibly their relatives in the future. This could be explained as a normative responsibility to gain knowledge about their health through genotyping, which is rooted in understanding public health and a Kantian duty to the self (Jeske 1996). It also presents an additional biological facet to individuals' manifold identity practices, as Novas and Rose suggest in their discussion of genetic identity (Novas and Rose 2000) By the powerful associations with personhood, it also indicates a moral basis for people's thinking about their genetic identity and its implications for their biological kin. This is demonstrated in cancer genetics research including Hallowell et al's study of women with breast and ovarian cancer (Hallowell et al 2003). Responsibility in relation to one's health is also related to the third theme in this study, Testing the NHS and will be discussed in more detail in the section on Responsibility in Chapter Six.

Nicola pursued testing because she imagined her genetic identity contained discrete information about an inherited disorder that conventional medicine

could not and would not elicit. She was interested in her results for herself but also for her child, as she felt, in common with those who undergo genetic counselling and testing for inherited conditions, that this was a familial problem that could influence her child's future.

There's something going on in my family; I need to know what it is not just for me but also for my child so I can guide them in their career choice and things like that. (Nicola UP17)

The moral basis for her decision to test is framed in her responsibility for her own health, but she emphasises her responsibility in relation to the health and future well being of her child, a common justification for clinical genetic testing (Hallowell 1999). Results had implications for her child not only physically but also in relation to how Nicola parented the child to best deal with the potential impact of her genetic identity in future.

Barbara, the genetic genealogist, had a significant family history of malignancy on both parents' sides of her family. Her mother's death from cancer, in association with her Jewish heritage, was the original trigger for her interest in genealogy, as she had wanted to trace her family lineage and health history.

I'd already got hundreds and hundreds of birth, marriage and death certificates for my family so I knew the causes of death as well as the fact a lot of my family had died from different cancers which again is a common thing with Jewish genealogy, so on my mother's side out of four siblings three of them died from cancer and on my father's side, out of six surviving siblings four of them died from cancer, all different ones. So this concerned me. So I thought if this testing was available I'd like to know what genes I'd got or not. So I thought is it better to know or not know so I did think about this for a very long time, do I want to know or not because I had the option of, at that time on 23andMe you could opt either for just the family tree testing or the health testing or you could have both as a package. And I decided to go for both as a package. (Barbara UP3)

Barbara uses the phrase “better to know or not know” and emphasises her considered decision. This is indicative of her decision to exercise her autonomy and responsibility in relation to gaining information and potentially making choices about her health and lifestyle, which are overshadowed by her risk of cancer demonstrated in her family history and her Jewish heritage. This decision is in line with Chadwick’s assertions that genetic testing provides individuals with information that provides a basis for autonomy in relation to choice and responsibility, whether simply to oneself or to others (Chadwick 1999).

Helen and Laura both pursued SNP genotyping for information about life altering chronic health problems. Helen describes how she viewed any information as helpful and, as did Laura, imagined that gaining information about her genetic identity could hold potentially important information, if not now then in the future.

I got a test because I’ve been very ill for 17 years and unable to work or do anything since I was in my early twenties. I’m desperate to try and find out anything that might explain any of it or give some clue as to something I could do that would help because it’s just got worse and worse ... there’s family history that suggests it’s at least partly inherited. So I thought, hang on, other ones [SNPs] that are relevant to this are included in the 23andMe profile and I’m interested anyway and there’s the ancestry, which is interesting and there’s all the rest of it and something might be useful in future so I just did the whole lot. (Helen UP10)

Using the words “desperate” and “anything”, Helen shows how she has invested significant hope in accessing her genetic identity, imagining that it could provide some answers to her problems, if not now then in the future. This empirical discovery about herself aligns with Nordgren’s and Juengst’s observation that accessing one’s genetic identity, particularly when it is framed in the discourses of new technology and mainstream science, can be reassuring (Nordgren and Juengst 2009). This would be particularly understandable for Helen and Laura, who had failed to find diagnostic information, let alone treatments, elsewhere but who continued to invest hope

in the future utility of their test results. Their hopes in the future potential of SNP genotyping are different from Mokyr's assertion that early hope in new technology usually results in later disappointment in relation to the truth that is revealed (Mokyr 1991). Perhaps this is because the uncertainty of SNP analysis has already been revealed and thus the hopes invested in genotyping in its earliest stages have been modified for those coming to it a few years after companies like 23andMe started. The inherent lack of 'truth' or certainty in the test results leaves the way open for people's hope to shift from the DTCGT itself to future possibilities for the interpretation of their data or broader developments in genomic analysis for health (Arribas-Ayllon et al 2010, Tutton 2012).

Having discussed participants' learnedness and curiosity in relation to imagining DNA, I will now go on to explore the tipping point. This refers to the point at which users decided to buy a test and commit to establishing or extending part of their genetic identity, having previously contemplated personal genomics in imaginary and theoretical terms or in a more limited way.

The tipping point

Whatever the users' individual expectations of DTCGT and motivations for pursuing it, almost all decided to buy DTCGT at a certain point after a period of deliberation. I refer to this as the tipping point, although the metaphor is not being used in strictly the same sense that it is usually employed. Here I shall first outline the origins of the phrase and then discuss it in relation to the participants' decision to purchase a DTCGT.

In the 1950's sociologist Morton Grodzins wrote about the "tip point" or tipping point, in relation to changes in racial demographics in American cities (Grodzins 1957:34). He described both African Americans and real estate agents using the phrase to describe how the racial demographic of a population in a given area would "tip" (from a Caucasian to African American majority); once a threshold number of African Americans had moved into an area the majority of its remaining Caucasian population would move out. Gladwell used the term rather more recently in his book "The Tipping Point", since when it has become more widely used (Bhatanacharoen et al 2004). Gladwell refers to three rules that result in a sudden shift in group behaviour:

the Law of the Few, the Stickiness Factor and the Power of Context (Gladwell 2000). A dramatic change in behaviour requires

1. a few key people who drive or initiate the change by spreading the word about the issue by virtue of their knowledge, by knowing a lot of people, or by their charismatic nature and skills of persuasion,
2. the new thing to be memorable so that it sticks in people's minds,
3. the context in which it is happening to be apparent and relevant to others; that is, the environment needs to support the move.

I would not suggest a tipping point has been reached, in any literal sense of uptake of DTCGT in the UK. However, I would suggest that within the group of the UK's early adopters of DTCGT, there are key factors that relate to each of Gladwell's three rules. First, uptake of testing can be seen to relate to key people in genetic genealogy and genetics research who have acted as "key" selectors (my emphasis of Gladwell's rules) and been instrumental in setting up networks of support and information dissemination about testing (see Expectations in Chapter Three). Members of ISOGG and genomesunzipped spread the word about DTCGT within their respective communities, raising people's awareness of it. Second, 23andMe have a captivating approach to their marketing that is likely to "stick" in people's minds. This includes their tropes of democratisation of genomic data, their invitation to interact with their DNA and alter their life-styles in accordance with results and an apparently altruistic commitment to genomic research that their community embraces. Finally, and as will be seen from the data below perhaps most importantly, the "environmental context" has to be conducive, and I would argue that this context is the financial environment. The most common criterion mentioned by almost every participant as a deciding factor, after his or her period of deliberation about buying a test, was the cost. Whilst not everyone paid the same price for their tests, the majority paid a fee of \$99 because they purchased the test either when 23andMe were doing a promotional sale or during the period when 23andMe dropped the price of the test itself. However, the real incentive for most of the users appeared to be a price reduction or advertisement of a special offer, regardless of the actual cost of the test. This pattern of decision-making in relation to purchasing goods is supported by the economic theory of the Law of Demand in which the price of goods is inversely related to the demand for them; the cheaper a product becomes, the more people will buy it, assuming other factors remain constant (Hildenbrand 2014).

The decision to buy was made after a period of deliberation and the terms that describe the financial basis for the decision provide evidence of a justification for it. SNP genotyping became cheap enough to justify the purchase, rather than it being an expensive and impulsive decision. What this also indicates is that most people considered their decision to test for some time and followed the companies' websites, using their information and changes in prices to inform their decision.

The genetic genealogists' networks update them with current developments in genetic genealogy and genomics. ISOGG has a wiki where members post information and updates that facilitate the communication of current events and links to other similar online resources. The participants who are genetic genealogists either bought a test at the price it was advertised when they were particularly interested in it (Barbara and David) or, more commonly, described watching the updates on various websites and then spotting a reduction in price or a special offer (Alan, Ann, David, Geoffrey, Keith).

Ann, a local administrator for the England group of ISOGG, describes her decision to buy a test.

I eventually got involved when 23andMe had a sale and they were selling the test for 99 dollars just for a couple of days so I, coz I think it was 399 dollars at the time which I thought was, I just didn't think it was worth it, but at 99 dollars I jumped in to try it. (Ann UP2)

Ann's reference to getting involved "eventually" shows how she had debated buying a 23andMe test for some time (in addition to the genetic genealogy tests she had taken earlier). The special offer at a reduced, affordable price "for just a couple of days" gave her the impetus to buy the test.

David, Keith and Geoffrey describe similar situations, where advertisements of special offers persuaded them to buy after a period of thinking about testing with 23andMe (i.e. obtaining further genealogical information and additionally extending their genetic identity in relation to physical characteristics, disease-risk and carrier status). David judged his test to be expensive but managed to

get a free one for his son and another for his wife as a special offer, which he felt offset the price he paid for his own test.

Users who were scientists were similarly influenced by the price, as indicated by Fiona and Maria, who demonstrate the importance of the price in tipping them to their decisions to get a test. They had watched the prices reducing over time, whilst they were debating testing, and decided to buy the test when the price (for 23andMe) reached a point they considered reasonable.

And it [23andMe test] finally hit the price-point where I thought “Yeah, it’s not a lot of money”. (Fiona UP8)

Also influenced by price, although not paying for it personally, Kirsten selected her test on the basis of cost, having investigated various companies’ products.

So I did my homework and I looked at primarily 23andMe and deCODEme and deCODEme was well over 1000 dollars at the time ... I’ve got a slide in a seminar I give that shows the shopping basket with the figure down at the bottom and everyone is always appalled when I show it; and 23andMe was down at, I think I paid 200 dollars for my test, so I went for that one, just purely on price. (Kirsten UP14)

Although she says the price drove her final decision, Kirsten had also looked into what different companies offered and chose 23andMe as being cheaper whilst providing similar information. She justifies her choice on the basis of personal research, as well as the price, and seems to be trying to provide evidence of that justification by describing students’ reactions to her decision, which align with hers.

Carol and Christine both decided to buy a test after being introduced to testing by others and then researching SNP genotyping for some time. The basis for their decision was that it was affordable to them. Christine considered the price of DTCGT in the context of costs of sequencing, demonstrating how the amount of money involved is key to most of these users’ final decisions to engage with DTCGT.

I mean I certainly wouldn't pay 1000 pounds to have my genome sequenced properly, but paying 150 quid to find out SNPs and that, is accessible. (Carol UP4)

Unlike Carol, Helen and Laura were interested in WGS to shed light on their health problems. But their views on the costs of testing were still key in terms of tipping the balance in relation to their decision to test with 23andMe because for them it was an affordable option. Like Carol, they said that WGS, whilst important to them, was not affordable which perhaps equates to SNP genotyping being a justifiable expenditure.

Personalising DNA relates to people's imagination about their genetic identity facilitating embodiment and the experience of embodiment, influencing the imagination thereafter. Whilst acknowledging this dynamic, I feel it is necessary to separate imagining from embodying DNA in order to give due consideration to participants' thinking about DNA *and* their experiences of SNP genotyping as part of personalising DNA. Contrary to concerns about DTCGT's capacity to cause harm outlined in Chapter Two, the users in this study appear to view personal genomic information as expanding their identity practices and satisfying curiosity in a fashion that could be construed as relatively light-hearted, given their final decision to test seems linked to saving money. This is further supported by evidence of their general lack of behaviour change, which is demonstrated in the following part of this chapter. Next I will examine how users embody their DNA following SNP genotyping and the parallel experience of clinicians who were involved in counselling DTCGT customers.

Embodying DNA

In using the phrase 'embodying DNA' I refer to the process of using newly acquired knowledge of DNA (in this instance by SNP genotyping) to develop or change one's feelings, understanding of identity and corporeal behaviours or practices of the body. In proposing that genomes are "incorporated", O'Riordan suggests that genomes are embodied in individuals' lives in both thoughts *and* bodily practices, influencing them in multiple arenas rather than simply in altered behaviours (O'Riordan 2010).

In this section I shall initially consider users' experiences of testing and receiving results and the impact embodying DNA had on their identities. I will follow this with an exploration of the clinicians' experiences of embodying DNA, which focused on interpreting concepts for disease risk. This invites exploration of how referrals for consultations for SNP genotyping interpretation were made to their departments and how consultations were experienced, as well as consideration of how SNP genotyping is being mobilised in a different manner in one clinical setting.

Identity and embodiment

Although Rabinow's proposed biosociality argument implies that people adopt a genetic identity that goes on to influence their sociality, Novas and Rose suggest that genetic identity practices are only a part of people's identity within their multifaceted worlds and the plurality of both their identities and the networks they are engaged in (Novas and Rose 2000, Rabinow 1996). Novas's and Rose's suggestion is more congruent with my data, which show users' reactions to their DTCGT is an extension of their identity within the context of the multiple aspects of their identities, rather than simply a genetically deterministic one. Whilst participants described embodying DNA as being actively aware of this additional genomic aspect of their identity, few, if any, demonstrated any embodiment in the sense of anxiety or altered body practices that were sustained beyond a few weeks in common with previous research findings (Van Ommen and Cornell 2008, Bloss et al 2011, Kaphingst et al 2012).

To examine how the users engaged in identity practices through SNP genotyping, I will discuss how their embodiment of their DNA through learning and understanding, incorporating health information and discussing their results with family members contributed to their identity. Finally, I will explore participants' contradictory views about their DNA and how they subjectively contested the aspects they were unwilling to embody.

Many of the participants described genomic information as valuable to their identity. Alan, Ann, Laura and Maria all specifically referred to embodying their DNA in relation to valuing having this additional knowledge about themselves, suggesting that embodiment was empowering. For genealogists Alan and Ann

and chronically ill Helen there was an attraction in having their raw DNA data to ponder and manipulate.

I satisfied myself and others that you can interpret ... data applied to genealogy data and make connections. (Alan UP1)

Alan, a committed genetic genealogist and retired chemist, shows how he used his raw data productively. In respect of his genealogy he extended his knowledge of himself and his relatives and his ability to use genomic data effectively in doing so. His phrase “satisfied myself and others” demonstrates the relationship between personalising DNA and developing the language and skills to socialise it.

An extension of genetic identity was realised by the potential for finding relatives with genetic genealogy. Whilst most users found their genealogy results interesting or, in Jane’s case, distressing (as outlined in Chapter Four), of the genetic genealogists only Alan, Geoffrey and Keith reported finding useful data in the form of new relatives. The others either did not comment on it or felt it was disappointing, but this subset of participants was already familiar with genetic genealogy.

Others described their learning and understanding in relation to the process of SNP genotyping and how results are communicated, in addition to extending their own identity into the genomic. Ann, Fiona, Kirsten and Maria all valued having the opportunity to experience the process of testing from getting information and buying the test online to embodying their DNA through the practice of spitting and then mobilising their DNA using the Internet as the portal through which to access it. All these women referred to being interested in the process. Fiona specifically referred to the practicalities of producing a sample.

It was interesting just to go through the process and get the tube and generate quite that much saliva! (Fiona UP8)

Conveying the patience and effort required to produce the sample represents Fiona’s commitment (and by extension that of all the users) to move from imagining their DNA to embodying it. Kirsten expressed her experience of the

beguiling effect of the embodiment of her SNP data on the computer screen, despite her expertise in dealing with this type of data in her daily work.

When I drilled down into it I carry the [x] allele for [x condition] which of course confers a high risk and so most of it came from that single variant, so I started to get slightly more impressed at that point because I thought it is actually, you know I'm picking up something that I had some interest in or something I thought I should be at increased risk of. And then after that the numbers dropped off fairly substantially we were down to an increased risk of 1.23 for [site] cancer or something, which is neither here nor there really. But it's quite seductive when you look at that screen and it's your results there ... I was quite surprised how seeing those numbers - your numbers - in print on the screen gives them a level of value beyond what you might expect. (Kirsten UP14)

Her references to the seductive nature of seeing *her* results and *her* numbers on the screen shows the initial impact of embodiment of her genetic identity; her identity and risk materialise on the screen. In common with other users who were scientists, Kirsten was clear about the contingent nature of SNP genotyping data, but this description conveys how powerful the initial impression of her data was for her personally despite this knowledge. Carol and Christine, the public health researchers, described similar responses to their first impression of their results, being initially mesmerised by some of their disease-risk results. For all three this initial fascination appeared to abate within weeks of having the results, as I will explore in the section on Contradictions and contestation. Prior to that, the next section considers people's responses to the disease-risk aspect of their test results and how they embodied their DNA in relation to them.

Embodying DNA for health

Most users' reactions to the health aspects of their tests ranged across a spectrum from scepticism or disappointment, to being initially somewhat mesmerized by the disease risk information. In those for whom the main motivation for testing was health-related, a lack of relevant information resulted

in their returning to future hope and expectations. Only one user, Barbara, sought further advice from healthcare professionals as I shall demonstrate shortly. Participants who tested for genealogical or general curiosity viewed their health risk results as conveying interesting information but nothing they should take specific action for, other than the generally established knowledge that exercise and diet are the key factors for a healthy life. They largely viewed results as providing only part of the picture rather than being deterministic. They were also informed by and justified their results in relation to, their family health history, their lifestyle and environmental factors that are known to influence the onset of common complex diseases.

Some users relied on moral justifications for testing when referring to disease-risk results. Receiving results from the SNP genotyping appeared to have influenced some users' perceptions of their potential for developing certain common complex diseases, resulting in their expressions of the need to be vigilant and think about their lifestyle in future, or advise family members of how they might be affected but without provoking anxiety. This normative responsibility to gain knowledge about their health, which is rooted in public health and a Kantian duty to the self, may have been affirmed by 23andMe's reliance on the tropes of personalisation and responsibility in relation to health information; both genealogists and scientists suggested that the moral aspect of embodying DNA is relevant, irrespective of original motivations for testing.

David's references to finding this knowledge useful, both now and in the future, is representative of many of the users' views on this point.

It's nice to be aware of that and watch out for any signs in the future ... it's better to know than not know because you can do something about it. (David UP6)

He uses the phrase "better to know than not know". This is indicative of David's decision to exercise his autonomy and responsibility in relation to gaining information and potentially making choices about his health and lifestyle, although he had not in fact made any changes. This is entirely in line with Chadwick's assertions that genetic testing provides individuals with information that provides a basis for autonomy in relation to choice and responsibility, whether simply to oneself or to others (Chadwick 1999).

Having had extensive experience of consulting with healthcare professionals for her chronic health problem, Laura similarly identified that genomic data gave her the novel advantage of autonomy in relation to managing her health.

It's nice to know more about myself ... it doesn't worry me. I like to have as much information as possible. I know it's a tiny snapshot but I like having control ... it gives you a chance to plan. (Laura UP15)

Laura's quest for information shows her interest in extending her understanding of herself, here by embodying her DNA. She assumes the value of this information for health is limited but appears to value it in a qualitative sense, in that the impression of autonomy and control it provides are paramount for her.

Disease-risk information was of primary interest to a few of the users and I will consider these people's discussion about their health results next. Barbara, Christine, Helen, Laura and Nicola had all tested in order to try and reify their imagined genetic causes for their health concerns. These included a familial history of malignant disease or an experience of other chronic illness which conventional medicine had not been able to help them with.

Helen, Laura and Nicola were all interested in SNP genotyping as a means to extend their identity practices independently of conventional health care. Helen and Laura were interested in gaining *any* new information possible. When SNP genotyping did not reveal anything specifically related to their troubles, they both justified their decision to test with the interesting nature of the information gleaned, embodying their DNA by investing hope in the future potential of their genomic data. More specifically, Nicola described searching for a diagnosis, which arguably equates to establishing some certainty in the face of an indeterminate future. The following demonstrates the importance of extending her identity practices in this way.

I decided 23andMe offer genetic testing, I'm going to spit in a tube and send it off to them. And I did that and I got the results and I was fascinated by them. The reason that I found

it interesting was that when you have a condition for which there is no real clinical diagnosis you never really know whether you have it or not. I read a book on [disease]; the first chapter I went to was genetics. And in that chapter he said, "Why bother with genetics? We can't give you a cure even if we know what's wrong with you." And I threw the book in the fire in frustration because it's not just about getting a cure. It's not about directing drugs because of my genotype. It's about understanding what is going on in my family so that I know what I've got, you know and getting a diagnosis is almost just as good as getting a cure. So this was my thought process behind the whole thing. (Nicola UP17)

In common with many other participants, including Laura, Nicola is a biologist with extensive experience of genetics and so had knowledge and experience on which to imagine her DNA. However, it is clear from this excerpt that extending her identity into the genomic so that she could embody her DNA in the form of a diagnosis for her problem was her overriding concern at that time. She clearly indicates her frustration with a professional view that genetic identity is irrelevant due to the lack of effective treatment and pursued her identity practices in this arena, like Helen and Laura, to be able to have some autonomy in relation to her health.

For Helen, Laura and Nicola, their bodies have represented sites of contestation between numerous actors, including medical professionals, blood and tissue samples, technologies for investigating and embodying aspects of their anatomy and physiology, texts that circulate as a result of consultation and investigations and family members who support them. Their experiences have rendered them de-centred and transformed into objects for others to challenge as sites of social contest, as Foucault suggested in discussing medicalisation (Foucault 1994). These women have arguably embraced SNP genotyping as a means to extend and assert their identities through their DNA in a novel network, where they are in a position to exercise autonomy and authority as actors rather than as passive objects.

Barbara and Christine were both interested in SNP genotyping because of their family histories of cancer, although this was not their only motivation for testing. The elevated risk of a specific cancer in Barbara's test result linked directly to this family history of the same type of malignancy (not breast cancer) and this prompted her to seek medical advice.

And it seemed to show I had a particular gene which seemed to be the same one my father had because he died of [site] cancer. So then I thought, well what shall I do with this? I thought I'll go and ask my doctor (GP) and ask "What shall I do?" So I went to my doctor and they referred me to the [name of specialist cancer centre] Hospital being only down the road. So I went to see this lady who was the lead for breast cancer ... non-breast cancers were of more interest to me but the health community was concentrating on the breast cancer issue rather than the other ones for [Jewish] women. We did a family tree, I knew sufficient information to show her where all these cancers lay [who was affected in the family]... I took along the print that I'd got off 23andMe. So she explained to me, it's not got to be just one marker it's got to be a range of markers and they're going to interact with each other. And she explained that the biology would interact with the environment so you need to take into account what happens in your lifestyle what you eat and all this sort of thing. I was relieved, one marker wasn't it, there had to be more ... several people have told me since, rather more cancers were from an environmental background rather than a genetic one, but that the mixture of the two would set it off. (Barbara UP3)

Barbara describes her initial concern at finding a marker in her SNP genotyping results that seemed to correspond with a risk for the same malignancy that had caused her father's death. In trying to establish whether this marker would be a problem for her health in the future, Barbara was able to provide important family history detail at the consultation. Despite seeing an oncologist who specialised in breast cancer (rather than the site her family tree indicated), Barbara describes being reassured by the explanation that

environmental factors were as important in cancer development as the genetic ones and that genetic predisposition was polygenic. Her embodiment of her DNA shifted to incorporate the narrative from this consultation as well as her SNP genotype and family history, in line with Kelly's assertion that experiences including knowledge of risk and genetic counselling are significant in genetic embodiment (Kelly 2007).

When embodying their DNA, most users demonstrated integration of this new aspect of their identity into their existing, complex and compound identities, although this was often problematic, in that some aspects were more easily integrated than others. In the following section I will explore the contradictions in participants' justifications for incorporating or rejecting different aspects of their DNA.

Kinship and belonging

Sharing SNP genotyping experiences and results with others is an aspect of Socialising DNA as discussed earlier in Chapter Four. However, this activity is also part of embodying DNA, in that it facilitates people's sense of individually belonging to a group with whom they share knowledge, experience or embodied DNA (O'Neill 2007). Family bonds and memories contribute significantly to people's identities and are influenced by their embodiment of their DNA, which is why this aspect of the data is discussed here.

Whilst not everyone decided to share their results with family, the majority did so. Some users indicated that sharing their test results with family members both added a genetic dimension to the familial aspect of their identity and provided a vehicle (through the discussion about common physical traits) for more serious discussion about disease risk, or, in Jane's case, ethnicity. Christine, David, Elizabeth, Jane and Laura all told me about discussing results with family members who had been tested, looking for similarities and differences. Two genealogy group members, Ann and Keith, described sharing test information with others in their groups. Three people referred to the traits information specifically, which related to learning about themselves and family. For Christine, whose parents had both recently been diagnosed with terminal cancer, sharing her test results with her siblings (who do not have her science background) was important. She initially talked about discussing physical traits

and then went on to share her raised risk of particular cancer markers relevant to their parents' illness.

It was lovely to read about familial traits because on the side of the results they also talk about phenotypes, someone can curl their tongue in the family...so it was pitched at a level of seriousness but it was also quite fun [like] "Do you sneeze in the sun?" and that. We talked about [who had what in the] family ... there were [site] cancer markers and that was something I shared with my siblings as well, apart from the general practitioner mentioning to us we might have a familial problem ... having two parents suffer so much, thinking you don't really want to go down that way either. (Christine UP5)

Christine indicates using the trait SNP results to reconnect with her siblings through shared memories of family traits. She also said it was important that they were aware of her cancer marker results, as she interpreted this genetic aspect of her identity as supporting the warning from the family GP about a familial aspect to their parents' diseases. This suggested that Christine and her siblings would use this knowledge to exercise vigilance in relation to their health in future in order not to "go down that way either".

Barbara decided not to share her results (including her increased risk of cancer) with her family, including her two children who were in their twenties. She explained that they knew about the family health history of cancer and did not need to know about her DTCGT results at this time in their lives. Barbara appears to have confined her avid interest in genetic genealogy to her own identity practices, rather than sharing results and encouraging other immediate family members to test, unlike other users. This struck me as a contradictory decision compared with other users and Barbara's interest in her wider family's health history. However, the following section will demonstrate more specific examples of the thorny issue of users' selective approach to embodying DNA.

Contradictions and contestation

O'Riordan describes embodiment as problematic when it challenges the individual to have to assimilate unwanted characteristics into their identity

(O’Riordan 2010). Participants appeared to be selective about which aspects of their SNP genotyping to incorporate into their identities and which to reject or ignore, mostly relying on the argument that SNP genotyping is known to be of limited validity and utility to explain which to ignore, thus demonstrating interpretative flexibility (Bijker 1997). Having mulled over their results for a while, others suggested that they realised ignoring public health advice because of their test results was probably not sensible. Several of the users contradicted themselves when telling me about their interpretations of their test results. Their accounts bear a striking resemblance to O’Riordan’s descriptions of “preferred readings” of the genome, in that not all the users adopted the novel genomic aspect of their identity without disagreement or a lack of embodiment in terms of altered behaviour, despite expressed intentions to do so (O’Riordan 2010:4).

Nicola’s interpretation of her results demonstrates interpretative flexibility. She initially used her test results to try and explain the signs and symptoms of an undiagnosed complaint. The negative results in her SNP variant was later undermined when she was diagnosed with the condition she had suspected. This is why she refers to “how limited 23andMe are”, but she then immediately goes on to attribute a different physical abnormality to her test results as well as her tendency to overeat.

And I also had a lot of fun going through my 23andMe results because it’s, I do find it fun. I’m very much aware because of my background that it’s very, very limited in terms of scope across the genome. One of the questions in my own head, because I couldn’t get to see an NHS consultant, I was trying to self-diagnose, was “Is there any chance I could have [chronic disease]?” So I looked at the risks in the SNP call out [results] and it said I had no increased risk. So this really puts it all in context how limited 23andMe are but still it was a starting point for me. At least I could explain my [physical abnormality] and I also know I have a genetic tendency to over eat, it’s not just greed! So I do find it fun. (Nicola UP17)

This acknowledges the lack of utility of SNP analysis, but cannot totally discredit it in relation to traits that appeal, showing her embodiment of her DNA

in a discriminatory fashion and her essentialist interpretation of the results despite her knowledge of genomics.

Carol, Christine and Kirsten (all female scientists in their 40s) describe being initially captivated by some of their results, but this did not last, nor result in any changes in their understanding of the contingent nature of DTCGT. Carol, in particular, reacted deterministically to some of her results initially but then resumed her former lifestyle choices.

Having seen that I don't have my mother's diabetic genes, or risk factors, then I thought "It's OK to eat sweets" ((laughs)) because I haven't been eating them up until that point. It was like being given a free pass on the food front for a month and then I thought, "Just stop being silly and go back to what you were doing before." I've got over it now but it did make me think, "Yeah I can do different things behaviourally" because there are certain things; I mean I'm less likely to get melanoma than most people so I've been out in the sun ... it is impacting on my behaviour in potentially unhelpful ways, umm, but you know it's like, again next week I've got to go up and have breast screening which, umm, oh it's just awful. I don't have any *BRCA* genes so is there any point in my being screened? I mean there probably is because there are other causes of breast cancer but you know, it's making me reconsider those sorts of choices. (Carol UP4)

This example plainly shows how Carol has embodied her DNA in working to understand and assimilate its challenges to her identity. She seems initially to use the results to justify engaging in potentially risky behaviour (eating what she likes and sunbathing). This is despite her knowledge and understanding of public health and the role genetics plays in the aetiology of common complex diseases. She describes having to make decisions about her lifestyle, which encompass both her prior knowledge of public health, her knowledge of genomics and the novel genetic aspects of her identity. She seems to have achieved a balance, as evidenced by the phrase "I've got over it now but it did make me think", and later in the interview she justified her decisions by saying "it's SNPs so there could be mistakes, anomalies."

Many participants referred to making room for uncertainty in SNP testing, whether in terms of the inadequacy of current knowledge related to genomics and health risks or in relation to understanding the small part genotype plays in common complex disease development. Fiona, one of the scientists in her early 30s who has had a long-term condition since childhood, excused the anomaly between her results and her actual health as being due to bad luck.

That was the most surprising thing was that I was not even at normal risk for that I was at low risk for it. When you go and look at it there are several alleles some of which I'm on the negative side for and some of which I'm on the positive side for and they decided that the positive ones are more protective than the negative ones are bad and I suspect that actually means they are probably all independent things and I was probably just unlucky. (Fiona UP8)

Despite telling me that, as a genetic scientist, she had a clear understanding of the contingent nature of SNP genotyping information, this excerpt indicates that Fiona initially looked for genetic answers to her condition. She was evidently surprised that the results contradicted her phenotype, but she attributes this to luck, arguably identifying with a genetically essentialist interpretation at this point.

Brown and Webster suggest that because genetic testing is inherently linked to our sense of self and individuality, it is compelling in relation to both fascination and anxiety (Brown and Webster 2004). Whilst the majority of users clearly demonstrated fascination at the prospect of delving into their genetic identity, most conveyed a sense of pragmatism about the deterministic nature of SNP genotyping in relation to health. Whether this was influenced by their knowledge or research into the poor validity of SNP genotyping, or their unwillingness to embody their DNA by incorporating disturbing results and change into their identity (or both) is not clear.

Before moving on to discuss clinicians' embodiment of DNA, it is worth mentioning that Jane's and Ian's reported experiences of embodying DNA seemed to converge with other users' experiences, despite their obvious

differences in socialising and imagining DNA. Both shared their results with family members and expressed intentions to act on some results by employing health screening, demonstrating the normative responsibility towards their health that I discussed earlier in this chapter.

Genetic risk as embodiment

Clinicians' experiences of embodying DNA from SNP genotyping relates to helping consultands to understand and, in turn, embody their own DNA. Genetic counselling in this context helps people understand the concept of risk and the meaning of the numbers associated with their SNP variants. All the clinicians involved also described used their more conventional surveillance tools, including taking a detailed family history from the consultands, demonstrating an adherence to their group's professional procedures in approaching DTCGT that could be interpreted as strengthening their case in relation to their views of DTCGT.

In this final part of this chapter, I shall first outline how potential referrals for interpretation of DTCGT to genetics departments were handled in the departments the participants worked in, as this contributes to understanding the context of the limited number of consultations that occurred. I shall then go on to discuss how the clinicians embodied DNA in the consultations that were described to me, or in other ways and their learning and thinking as a result of those experiences.

Many of the clinical genetics professionals described how their departments had anticipated a deluge of referrals of patients for discussion of DTCGT that had not so far materialised. This was empirically supported by the difficulties recruiting clinician participants to my study, as outlined in Chapter Three. It is also unsurprising, given the evidence from the users' interviews that showed they did not seek referral. As demonstrated in Table 3.1 earlier in this chapter, of the 17 users interviewed only Barbara had been referred to a hospital consultant by her GP and that consultant was an oncologist specialising in breast cancer rather than a clinical geneticist. Literature published at the time personal genomics companies proliferated anticipated increased referrals to healthcare services for interpretation of personal genomics tests (etc Group 2008, Lenzer and Brownlee 2008, Jordens et al 2009). It is likely that the

clinicians in this study were aware of this and that it had influenced their expectations of having increased referrals of worried-well DTCGT users from GPs needlessly taking up scarce clinical resources. Along with the knowledge circulated about SNP genotyping this may have contributed to their understanding of DTCGT and its potential to cause people unnecessary anxiety. With the professional rhetoric associated with genetic counselling, this technological frame could be interpreted as employing their obligations to support their case against DTCGT in the wider context of the NHS by working to exclude DTCGT users from their clinical work.

Only one clinician reported that her department would see anyone who was referred. Half of the clinicians indicated that their departments would discuss the referral at triage, as with any other referral. They would only agree to see the person if their department's usual referral criteria applied; that is, the person being referred to discuss their DTCGT also had a family history of illness with a known genetic connection. The remainder of participants either did not know about their department's referral criteria or had no policy for DTCGT referrals because there had been none so far. Only one of the participants familiar with the referrals and triage for their department was aware of a referral being 'bounced' back to the GP (refused) and described the consultant's emphatic response to the referral.

We've had one patient that was discussed at the weekly meeting that someone said, "This came in from the GP who said the person wants to discuss their results." And I can't remember what test it was but it was a kit (DTCGT) and the lead consultant went "Nah, we're not touching it with a bargepole because we don't know what to advise, what to say. We don't want to get caught up in that." (Nurse CP12)

Though this excerpt reports someone else's response to the referral, the terms in which it was conveyed to me imply an authoritative, but simultaneously revealing, response. The phrase "we're not touching that with a bargepole" suggests a powerful message to the entire department that they do not want to be held responsible for any advice they may give these consultands, while the phrase "we don't know what to say" suggests that this novel technology is outside the realm of current clinical genetics practice in that department. This

could be explained by the provisional nature of both the technology and the data it yields, arguably being outwith the core set expertise of this group. It could also be seen in terms of this relevant social group's response to the technology; using their normative processes and conventions as robust platform from which to make their case for not engaging with the technology.

In view of most departments' policy of not seeing people simply to explain a DTCGT, I wondered if referrals had not been processed beyond the initial enquiry. But most of the clinicians suggested that referrals were never made, rather than being refused and returned to the GP. Most clinicians expressed surprise, as they had anticipated a deluge of referrals to interpret DTCGT following the publicity and publications associated with the launches of the high media profile personal genomics companies. One doctor equated the lack of referrals to a lack of uptake of DTCGT by the public, though it was not clear to me how she reached this conclusion. This could be interpreted as her projection of her own understanding of the lack of utility of DTCGT onto the public.

The interesting thing is it seems that the public have already realised it's not that useful. We're not being inundated with people having had it at all. (Doctor CP11)

Of the 16 clinicians interviewed, half had counselled someone in relation to some type of DTCGT and 2 had additionally discussed enquiries about DTCGT results from GPs¹¹. Some of these referrals were made because the person also had a family history of cancer and was concerned about their own risk of developing a malignancy, or they were anxious about their health in relation to another long-term condition of which there was a history in the family. In the consultations that were described, the clinicians generally established the patient's concerns and proceeded to discuss the relative risks to the patient on the basis of the knowledge provided by the test in question and their family history. Further testing was offered as indicated. Given that

¹¹ Most of these consultations were for referrals agreed to either because the consultand fell into the category of acceptable referral due to family history of genetic disease as well as having bought DTCGT, or because they were referred to one of the departments that had no agreed referral criteria. 7 consultations were for DTC SNP genotyping for common complex disease risk. CP5's DTCGT consultands were all journalists and their consultations were non-NHS.

patients had been referred to the Clinical Genetics or Oncogenetics Department (in one doctor's case), it is understandable that clinicians described these specific consultations in the context of their usual genetic counselling consultations, as illustrated below.

I explained at the beginning of the consultation we were mostly going to be focusing on his family history of cancer because that was his reason for referral and that was the reason we were seeing him in clinic, but that I was willing to look into and try and interpret his 23andMe analysis if he'd like me to. What he was most anxious about, was his 23andMe analysis had told him that he was a significantly increased risk of diffuse [site] cancer. Now that rings alarm bells for us because there is an inherited pre-disposition to diffuse [site] cancer and it's a horrible condition, that was quite a long way down probably about half way down the page but the marker wasn't in the gene we look at and he had no family history [of that site of cancer]. But the things that were at the top of his list of risks he completely ignored. One was osteoarthritis and the other was non-cancer related, they were clearly things he wasn't worried about and they were the only things that he was a significant risk for. The next things on the list were [site] cancer and [site] cancer but their increased risks were something like 0.2 percent to 0.26 percent and that was an increased risk! I think that numbers are really hard. We come across this all the time when you're trying to deliver information to somebody about a risk in a way that's meaningful to them. If it's at the top of a list and it's in bold and it's got an up arrow saying "increased risk" that all means far more than the numbers that say 0.2-0.63. (Scientist CP3)

This scientist described the consultand's reaction to his test results being driven by his anxiety about developing a specific cancer (of which there was a history in his family, though it was not the type of cancer referred to above). She was able to allay his concerns and explain the meaning of the percentages that the report presented, giving additional interpretation to the

report, shifting the consultant's understanding and thus his embodiment of his DNA. She was interested in his fixation on one particular result in the report, which, with his family history, led to him embodying his DNA in a more deterministic manner. But she also pointed out the difficulty people have in understanding risk, an opinion voiced by several of the clinicians.

One senior nurse recounted her experience of counselling a journalist who bought a DTCGT for the purpose of making a documentary. In contrast with the previous example, this consultation appears to have facilitated the consultant's embodiment of his DNA in a more considered manner, having adopted a blithe approach initially.

I think it's actually a really complex area because there are a lot of things that get confused. In clinical care we have an idea of what a 'test' is and it's about solving a clinical question and you know at a more public health level we have an idea of what screening is but again that's very bound by conditions and what we can do about it. It seems to me with DTCGT the sort of susceptibility testing I think is what everyone got very engaged with and everyone has got excited about. [One of the journalists] he had a 23andMe test but on discussion with him it also emerged he had a positive family history of breast cancer and was Jewish. And he presented a documentary about this. The documentary was a very personal documentary about his genes but he said, he did get it [understand the significance of genetic information]. He got it because actually once his family history became apparent because of his ethnicity and actually he found out there was a real genetic test he could have that might explain his family history and the fact he lost [his mother], he got the power of that highly predictive genetic information and [it's] not necessarily something you want just to receive on a computer screen without context. So he really did get it and he said to me that he'd gone into it thinking this was all a bit of a laugh and it was all completely meaningless and it suddenly wasn't. (Nurse CP5)

It appears from this nurse's explanation that two things influenced this consultand's views about genetic testing. First the timing of his DTCGT in its coinciding with his mother's illness and subsequent death from breast cancer brought the potential of genetic information into a sharper focus than it might otherwise have done. Second, his meeting with this nurse led to the offer of a "real genetic test", that is, one that is more predictive of breast cancer, which would potentially influence his and his family's genetic identity more than his DTCGT. However, the nurse describes how giving him more information and interacting with him personally helped him to view his and his family's situation differently. She demonstrates this by emphasising how his understanding and thus his embodying shifted, illustrating the power of clinicians' group protocols in conventional genetic counselling.

Two doctors, CP4 and CP6, had both seen siblings of people who had bought DTCGT and were concerned about the results. CP6 had seen the sibling of someone whose family had a history of a long-term condition and offered this view on the basis of her experience in the consultation.

I think its going to be one of those heart-sink ones whenever they come up because we would still go back to first principles when anyone came in with a direct-to-consumer test. Well what is the family history, is there something that's going on? And try and work through it that way because that would then try and put a clinical picture to what this piece of paper is saying. So it's an almost back-to-front consultation in a way; coming with a result and then trying to work it out.
Doctor CP6

This doctor's use of the term "heart-sink ones" illustrates the difficulties she and other participants identified in trying to help consultands embody their DNA through SNP genotyping. It represents the tension between the two relevant social groups in this study. Users of DTCGT approach genetic information and its embodiment through DTCGT but this approach causes consternation for clinicians because theirs is different and founded on socially shaped, agreed and accepted counselling approaches. Whether her concern is due to the lack of control over the process because the patient has instigated testing, as indicated by saying "it's a back-to-front consultation", or the lack of

utility of the data generated, or both, is unclear. However, in common with other clinicians, she demonstrates the use of tried and proven techniques in clinical genetics (in contrast to indeterminate SNP genotyping), including recording a family history to establish the indication of familial genetic disease. This doctor and other clinicians used this approach together with genetic counselling principles to help consultands embody the genetic aspects of their identities.

As well as users learning from their extension of their identities into the genomic, some of the clinicians described corresponding learning and thinking about DNA. This was from their experiences of either counselling people about DTCGT or using SNP data in future clinical work.

Two clinicians referred to taking the opportunity to learn about how commercial genomics companies present information about genetics and SNP genotyping as well as their presentation of results and the basis for data analysis. Having studied genomics company websites, they felt that there were examples of good practice that could be learned from. In particular, the scientist who saw the man concerned about his risk of diffuse [site] cancer stated

The results themselves, I actually have to say I quite like the way they're presented. I think we could learn something from the way they are presented; nice slick swishy website with lots of information and graphs and things. They use a nice pictogram of a hundred people and then they colour them in to show what the risk is; and so this one, which was 0.23 percent or whatever, one little man had his feet coloured in ((laughs)) and I think that's a really good way of showing risk. (Scientist CP3)

The graphics and presentation on 23andMe's website appealed to her and gave food for thought about alternative approaches to demonstrating risk in consultations, which could be drawn on in the future. She suggested that these approaches to presenting information provided effective means for communicating test information in a fashion which individuals could make sense of.

One clinician was eager to explain to me how SNP data is being used in oncology research, a demonstration of how SNP genotyping is being mobilised differently in different networks. This doctor's research work uses SNP data in a research project where people with a family history of a specific cancer will have SNP genotyping. Particular variants are thought to be linked to an increased susceptibility to this cancer and are being researched as a potential method of screening for increased risk.

In my work, a lot of what we've found now is in SNP profiles; so of the common cancers it's yielded the most SNPs so we've now got 78 SNPs [in this study] what we do is offer primary biopsy and then retrospectively SNP profile them. As you know at the moment there is no screening ... so it's in the research setting we're looking at the role of SNP profiling.
(Doctor CP11)

This demonstrates the doctor's engagement with SNP genotyping technology in her work. Her enthusiasm for its application arguably shows her work to enrol support for this emerging novel application of SNP genotyping although this is still within her social group's context as a clinician working in the NHS rather than in a commercial DTC context.

Conclusion

Novas and Rose might describe DTCGT as engaging in "practices of the self" as participants work at imagining and embodying DNA through their own or others' thoughts and experiences (Novas and Rose 2000:503). Although writing before the advent of DTCGT in relation to single gene disorders, they propose that individuals who work to uncover their genetic selves are incorporating new identities into their existing identity and self-actualisation. This is arguably more directly relevant in individuals with single gene mutations linked to disease than varying risks of common complex diseases, as Novas and Rose suggest. However, the possibility of establishing a new genetic identity characterised by inheritance of SNPs associated with their ancestry, traits, disease risk or carrier status can be interpreted as identity practice, as demonstrated in this chapter by exploring how the participants think about and embody DNA individually. Clinicians' contributions provide an extension to that

practice whilst simultaneously providing opportunities for their own learning and reflexivity in relation to their clinical genetic practice and identities.

This chapter and the one preceding have explored participants' experiences in relation to DNA, both social and personal. In the third theme from the data, participants' experiences associate the implications of knowledge about DNA from DTCGT to the wider context of healthcare provision, specifically the UK's NHS. In the next chapter my analysis indicates stark contrasts between the two relevant social groups of participants as they align themselves to either personalised or collective medicine. This demonstrates most clearly the importance of power relations between these groups and the influence of the wider social context of the study in the NHS in contributing to the on-going nature of the disputes about commercial SNP genotyping technology in the UK.

Chapter 6 : Testing the NHS

The importance of the NHS in the national identity of the UK was exemplified by its inclusion in the opening ceremony of the 2012 Olympic games in London (and this coincided with the data collection for this study). More recently a Commonwealth Fund report and a national UK survey both suggested that the NHS rates more highly than other developed world healthcare models in its provision of health care and in the UK population's support for it (YouGov 2014). Indeed, public support for the NHS has been unstinting since its inception in 1948 (Klein 2010). However, whilst this is perhaps predictable, it is arguably a superficial and simplistic representation of an organisation in which so many have vested interests, with those interests resulting in different visions of the NHS obligations and how they can be met. The NHS represents the wider social context shaping DTCGT in the UK. The participants in this study generally presented diametrically opposing views of commonly held beliefs about the NHS when talking to me about personalised genomics in the context of the NHS. Unsurprisingly, this took the form of users' support for personalised medicine, (though not necessarily at the expense of the NHS), while the clinicians support collective medicine and seek to protect the NHS. The data illustrate these conflicting views and the tensions that arise when expectations of both personal and collective medicine are applied to the NHS and it is subsequently found to be lacking. Such tensions raise questions about what the NHS's obligations to the UK public are, particularly in the face of increasingly neoliberal national and international policy, public expectation for more individualised health care and the on-going challenge of funding a contemporary public health service. In the data these tensions focus around three themes related to responsibility, empowerment or expertise, and regulation, each of which I shall examine in turn.

Responsibility

The beginnings of a more personalised focus for health care in the UK can be associated with policy responses to criticisms of a culture of dependency that was thought to result from welfare services provided by the state after the Second World War. In addition to political moves towards neoliberalism since

the 1970s and the encouragement of self-sufficiency, a focus on individuals being more involved and responsible for their health and wellbeing has gradually become more evident (Nuffield Council on Bioethics 2010). Evidence of this in health policy can be found in the Thatcher government's white paper "Working for Patients" in its aim to "give patients greater choice" (Klein 2010:153). The concomitant shift in health policy from sickness treatment to health promotion and subsequent governments' and policy writers' repeated emphasis on "personalisation" have resulted in expectations that individuals will exercise autonomy and responsibility in relation to their health and exercise choice in relation to intervention or treatment decision-making (Arribas-Ayllon et al 2011b). DTCGT companies' marketing strategies capitalise on all four of the P4 medicine attributes - Participation, Personalisation, Prediction, Prevention (Hood and Friend 2011) - and this has increasingly been used as a model for public health since the completion of the HGP (Groves and Tutton 2013).

However, responsibility is not solely a feature of personalised medicine. It is also fundamental to collective medicine. Responsibilities in collective medicine fall to individuals' duty to the population and the common good, including engaging with vaccination programmes or preventing the spread of communicable diseases, while the health service has a duty of care to patients by managing resources and rationing services (Klein 2010, Dickenson 2013). These features are shown in the data, with some of the users talking about personal responsibility to gain information to manage health and disease risk and how SNP genotyping can provide information that facilitates this. The clinicians were more focused on their responsibility for providing genetic counselling to consultands and protecting the NHS more widely from the potential drain on scarce resources that privately initiated testing could cause.

Some of the users made few, if any, references to responsibility for health and conveyed the impression that they viewed testing as providing personal rather than medical information. This corresponded with a suggestion from one clinician who was explaining why she thinks referrals to interpret DTCGT have not been made to clinical genetics clinics.

There are some certain aspects of public perception that see genetic testing as somewhat different; not a medical

investigation but as an identity investigation or something like that so I'm aware that, that that's where the appeal for over-the-counter genetic testing comes from. (Doctor CP4)

This doctor contrasts the professional view of genetic testing as providing potentially troubling medical information about illness that may require intervention with what she thinks the lay understanding of genetic testing is. An "identity investigation" implies that genetic information (specifically SNP genotyping) is trivial, rather than providing serious health information to be acted upon and indeed demonstrates her interpretation of the public personalising DNA.

The users who did refer to the health information in DTCGT in relation to responsibility and health care referred to three issues in relation to this: the role of DTCGT in providing information to help people take more responsibility for their health, the lack of involvement of a healthcare professional in discussing DTCGT results and concerns about NHS staff's lack of engagement with personal genomics. In the data there is evidence of tension between the personalisation inherent in users' expectations and the belief that the NHS cannot or will not engage with personalised medicine, as demonstrated by Nurse CP11's earlier cited extract "we're not touching it with a bargepole". An interesting feature of these users' interviews was their use of tropes including the importance of knowledge and the future promise of genomic knowledge in relation to personalised health care. In his writing on "Forceful Futures", Van Lente suggests the futures of technologies are fixed in specific languages, based on McGee's proposal that collective publics use an "ideograph", a vocabulary that guides their behaviour and beliefs in acceptable ways. In Van Lente's argument, as in personal genomics, this enables early adopters to assume a collective belief in the future promise of the technology thus strengthening their technological frame (McGee 1980, Van Lente 2000, Klein and Kleinman 2002). I feel that this is evident in the user participants' discourse of personalised medicine.

Along with Fiona (a geneticist), Carol and Christine (public health academics) both referred to the potential for genotyping to arm people with information that could enable them to take responsibility for their health and prevent future disease, thus reducing the burden on the NHS. Two of the genealogists spoke

in a similar vein; Barbara indicated that, although she had no background in genetics or health care, she too thought that genotyping could be valuable for preventative health care.

But it [DTCGT] would have a benefit because it's come leaps and bounds, if it means you can prevent something happening. What concerns me is we should be adopting preventive health care but we're not; we're waiting until someone becomes very ill before we're even treating them ...[it's] worse with all the cutbacks. This is where I see genetics things coming in...have a programme mapped out for you to avoid getting ill and dying which would cost the NHS a lot, so it would have a preventative role ... there's a lot we don't know which will be coming through in the next few years ... but it could have a preventative role. (Barbara UP3)

Barbara's suggestion is that the beleaguered NHS could benefit financially from using genomic information to prevent illness and lower the costs of intervention and terminal care that result. She seems to view the NHS as currently being reactive rather than proactive, as providing treatment for sickness rather than health promotion. By suggesting a shift towards prevention, she is demonstrating the tension between personalised and collective medicine, in addition to reproducing sentiments proposed by commercial genomics companies' marketing material. I should note that none of the participants claimed to have acted on their DTCGT in terms of making lasting changes to their lifestyle for the purposes of future disease risk management, whether or not they referred to the potential value of personal genomics in doing so.

The majority of the participants did not seek a consultation with any healthcare professional about their DTCGT. This is similar to earlier US-based research findings about early adopters of DTCGT (McGowan et al 2010). It does not support concerns that DTCGT will result in overburdening of healthcare services with consultations and subsequent investigations for the "worried well", as mentioned by some clinicians and which is a concern repeatedly forecast in the ELSI literature (Goldsmith et al 2012). However, three participants told

their GP about their test results at an appointment arranged for another reason but felt their GP was not interested in their test results. There was a general perception that the NHS is under pressure and that it is not realistic to expect hard-pressed doctors to know everything and respond appropriately or to expect that genomics is currently relevant to general practice. Thus, people did not bother to tell their GPs about their test, as it was not immediately relevant to their consultation or was not seen as directly related to the collective medicine the NHS provides. Christine viewed this from a public health perspective, first acknowledging the demands on primary care in the NHS, then suggesting how personalised genomics could help.

I could talk about this forever and if I went to see a GP I think the biggest fear for them is they've only got 5 minutes and you know more than they do ... the worried well...I think that's who the tests could end up being attractive to and I don't think that's a bad thing either. They may stay away from your [doctor's] door actually. (Christine UP5)

Christine observes that GP appointments are not compatible with her sharing her interests in health care and personal genomics (she had stated earlier in the interview that she did not feel there was anything specific that needed to be discussed with her GP). She anticipates the GP's response as being one of anxiety in the face of a patient with expertise, although she relates this to managing time-limited appointments rather than exploring the issue of professional versus patient expertise. She suggests that the information DTCGT contains could relieve pressure on primary care by alleviating anxiety in those who take up time with unfounded health concerns, a view that contradicts the ethical arguments against DTCGT on the basis of increasing anxiety among testers.

Laura and the two couples I interviewed (David and Elizabeth and Jane and Ian) all expressed concerns that in their experience the NHS was not sufficiently engaged in genomics. They all felt that the NHS should take more of an active role in utilising genomics for health promotion rather than leaving it to individuals to pursue genomics independently. Jane and Ian were strongly in favour of public health services, as well as genomics; Jane did not see them as incompatible, as she shows here.

It should be available to everybody, they should choose whether or not to have it, the GP would explain what it was about and they would make a decision to test on that basis ... I do think it should be supported by the NHS definitely, it should go through the NHS and it would save them a lot of money. (Jane UP11)

Jane is explaining her vision for how the perceived problems with commercial genomics, such as the lack of counselling, could be avoided if genotyping were adopted by the NHS for health screening. Her suggestion to offer genotyping to everyone through the NHS represents her vision of the choice to have individually tailored health care provided free at the point of delivery by the NHS, seemingly blending personalised and collective health care and merging individual responsibility with collective provision. The tensions between personalised and collective medicine, particularly in respect of funding, do not appear to concern her, given the net saving.

Some participants referred to obtaining genomic test information in stronger terms. They felt that having their genomic data was their right. Two of these were Helen and Laura, who had originally sought DTCGT to shed light on their health problems. Their expression of “rights” seemed linked to their need to exercise their autonomy following difficulties with healthcare professionals who appeared to act in a paternalistic manner. Ann, the self-professed DNA enthusiast and genealogist, took this entitlement further. She echoes Helen and Laura and the democratisation of genomic information argument that 23andMe rely on, which is echoed by some bloggers on genomesunzipped and demonstrated in McGowan et al’s research on early adopters of DTCGT in the USA (McGowan et al 2010).

People should have the right to their own genetic information, not via a doctor or anyone else. (Ann UP2)

This asserts the personalisation argument in much stronger terms. Ann supports the view that genomic information is the individual’s personal information rather than privileged knowledge to be held and interpreted by

medical institutions in the manner of Foucault's interpretation of professionals' clinical gaze over patients (Foucault 1994).

However, democratisation does not only apply to genomic information. The NHS is a social democratic organisation, set up by the state and run as a public service to provide collective medical care for the nation. Although they appeared to place as much and perhaps more, value on the institution of the NHS as the users, the clinicians' impression of responsibility was centred on their service provision. The other side of the democracy coin is represented by the clinicians' opinions about rationing to protect scarce NHS resources from profligate spending on privately generated health information.

The Nuffield Council on Bioethics report on personalised health care discusses the possible implications of increasing access to health information by individual UK consumers (specifically body scanning and genotyping). In addition to considering the possible benefits and harms of these sources of individualised health information, the report outlines the potential impacts on the NHS both financially and ethically. Increased uptake of NHS services to investigate the sequelae of personally accessed testing or scanning represents a drain on NHS resources, a loss of solidarity gained from combining risk and distributing services to encompass the vulnerable and disadvantaged as well as provoking debates about the fair and equitable use of public resources (Nuffield Council on Bioethics 2010). Several of the clinicians in the study referred to the first of these impacts by describing the collective responsibility to ration health services. This is to protect resources in order to provide a service to those genuinely in need of genetic counselling, though it was not clear on what basis these judgements are made.

Many participants, despite lack of evidence of it existing, expressed the obvious tension between DTCGT and NHS Clinical Genetics. They were concerned about the potential for scarce resources to be taken up by counselling the public who had bought DTCGT independently and then been referred to NHS genetics services for help, though most would not be seen at the departments whose staff I interviewed unless additional criteria applied. Clinicians with administrative responsibilities in their departments expressed concerns about service provision and set DTCGT as 'other' or out-with the usual clinical genetics service provision. One referred to the tests "we want to

do”, inferring that DTCGT is outside the remit of clinical genetics, framing it in paternalistic terms. Another with departmental managerial responsibilities outlined her concerns along similar lines, though she also describes being at a loss for solutions to this challenge, exemplifying the on-going dispute about DTCGT in the context of the NHS.

If people ... want to get their 23andMe, I'm not bothered. I am bothered if they then use up healthcare resources to interpret findings that are meaningless for a test that wouldn't be offered on the healthcare system. And you know I don't know how we deal with that. I mean I don't think this just applies to genetic tests. It applies to body scanning, it applies to lots of things. (Nurse CP5)

She uses the word “bothered” to stress her understanding of the conflict being presented by personalised medicine to NHS resources, rather than the challenge it may present to individuals who avail themselves of direct-to-consumer health information services. This understanding would appear to encompass the idea that her genetic counselling service would refuse to see people to discuss DTCGT alone. However, beyond understanding the conflict of rationing services in her own department, she suggests that the conflict remains so far unresolved providing evidence of lack of closure of the debates about DTCGT in the UK.

This doctor demonstrates a tension between using expensive resources for unplanned consultations and the moral imperative of providing succour to those in need of healthcare services, although her department was alone in providing counselling to anyone who requested it. The explanation for this department's approach appears to excuse their lack of rationing and possibly their lack of engagement with a need to ration services by falling back on the NHS principle of free health care to all at the point of access.

Well one of the things I find difficult is that we get, when these things come up the people are very, very upset and they want to be dealt with on an urgent basis and so that's really expensive time, that kind of emergency time. And I find it really hard to justify taking up emergency time but I don't

want to see the patient suffering either; if they are very upset about it I really struggle with abandoning them. And they are all patients of the NHS so we end up doing it. (Doctor CP2)

An experienced doctor with academic and commercial responsibilities in addition to his clinical role expressed a more democratic view, in the sense that he presented arguments for both personalised and collective medicine perspectives.

It comes down to whether you want to take a public health approach or an individual healthcare view of it and we all oscillate between those positions. There's clearly a danger that unregulated, genetic testing causes a lot of people a lot of distress, exposes unrecognised non-paternities, makes people anxious who are already anxious which is why they did the test, doesn't resolve their anxiety, it just makes them go to the doctor and pester him who then has an opportunistic cost because he's so busy seeing that person that he doesn't see the next one and you can portray a very negative perspective. On the other hand you can also say that if you empower the individuals to care for themselves and make those tests available in a controlled way, quality control them well, we could massively expand genetic testing and people's appreciation and understanding of it because it'll touch more and more of their lives and you'll just make it more and more part of routine health care which is where it's now capable of moving. So I kind of sit in the optimistic let's have a go camp. (Doctor CP10)

This extract demonstrates the participant's overarching view of the debate between personalised and collective medicine, no doubt informed by his more strategic and wide-ranging role as a Consultant. Whilst he outlines the issues that are used in arguments against DTCGT, he then goes on to propose how increasing use of genetic testing could be taken up as part of "routine health care". This is interesting because this doctor uses the terms that are characteristic of both perspectives, such as anxiety on the one hand and empowerment on the other. Rather than presenting similar views to the other

clinicians by defending the NHS and protecting the public from harm, he appears to be supportive of widening access to genetic information and uses the tropes of personalisation by suggesting this could “empower the individuals to care for themselves”. This is more congruent with the views of the users who talked about the potential for personal genomics to facilitate increased individual responsibility for health. It also suggests that genomics may indeed become integral to health care in the UK. However, this may not be in the individualised way that some users envisage, given the contingent and complex nature of genomic information that is based on population studies and appears currently to simply reinforce established public health advice or stratify people into treatment groups, rather than provide truly individually tailored health care (Hedgecoe and Martin 2003, Kraft and Hunter 2009).

Empowering individuals suggests equipping people with the authority and confidence to manage their lives. This requires knowledge and understanding of their genomic information. As has been demonstrated in Chapters Four and Five, this study's users of DTCGT have developed knowledge and understanding of genetics and genomics by virtue of their prior knowledge and experience of DTCGT, giving them a level of expertise in personalised genomics. The issue of how this equates to expertise commensurate with that of genetics clinicians presents challenges for the NHS and clinical genetics services in particular, as I shall explore in the next section.

Empowerment or expertise

The concept of patient empowerment has been adopted in Western society as an approach to engage citizens in health promotion and preceded genomics by several decades in healthcare policy in the UK (Petersen and Lupton 1996). This has partly contributed to the shift in the relationships between the public and healthcare professionals, along with challenges to medical authority from a more informed and critical public as well as alterations to funding streams and management structures in the NHS (Freidson 1988, Bury 1997, Elston 2002). Because NHS policy has highlighted the importance of patient engagement and neoliberal policies have promoted a focus on individual autonomy and empowerment, the pursuit of DTCGT to gain health information is arguably unsurprising. Some of those who have bought DTCGT join the informed public who can be critical of healthcare professionals. About half of

the users in this study fall into this category and voiced strong opinions about what they perceived to be the deficits in NHS staff knowledge and service provision. Clinicians were more united in their views; they have different ideas about the value of SNP genotyping, as shown in Chapter Four, and a wider perspective of what NHS genetic service should prioritise. In this section I shall explore how the concept of empowerment and users' and clinicians' knowledge of SNP genotyping relate to expertise and how these different interpretations result in conflict. Empowerment is specifically relevant here because it is, as the word suggests, the concept of power that is central to this aspect of the dispute about DTCGT. The credentialed expertise of clinicians who are employed by the NHS, the authority at the centre of the wider social context of the dispute about DTCGT technology, provides a cohesive and influential position from which they can contest the claims of those who advocate for DTCGT. Correspondingly empowered users of DTCGT, who are also increasingly empowered NHS patients, have a different experiential view of the technology and use their informed position to contest clinicians' (and others') claims against DTCGT.

It is no surprise that some of the users of DTCGT in this study appeared to equate their ability to gain health-related information with a sense of autonomy and empowerment, given the emphasis on personalisation used by commercial genomics companies. In doing so they compare their knowledge to that of the healthcare professionals they come into contact with, finding the latter to be wanting. Whilst very few participants stated that they knew more about genomics than healthcare professionals, several were critical of either GPs or the NHS because of their perceived lack of engagement with, or ignorance about, genomics. Being in a position to critique healthcare professionals would indicate that these people feel they are knowledgeable about genomics and health and feel empowered to challenge NHS staff who do not place the same emphasis on personal genomics as they do. I feel it is important to examine how this relates to expertise, as both groups of participants imply that they have knowledge and understanding that could be equated with different understandings of expertise.

Prior refers to experiential or partial knowledge as constituting lay-expertise in health care rather than the more broadly based knowledge and expertise of qualified experts (Prior 2003). In this study it would appear that users may

have expertise in DTCGT by virtue of their experience, while clinicians have skills and expertise in clinical genetics rather than SNP genotyping *per se*. However, as Collins and Evans assert the term “lay-expertise” is an oxymoron and would be more likely to support my suggestion that DTCGT users are experiential experts (Collins and Evans 2002). Evans and Plows pointed out that the use of the terms “scientist” and “public” in the debate about public engagement with science are too reductionist to be helpful, assuming the term “public” equates to the lay-person (Evans and Plows 2007). Users of DTCGT, who are experiential experts in personal genomics, might be members of the public but, as I demonstrated in Table 3.1, most of this study’s users also have varied experience of genetics, genetic genealogy, public health, illness and personal genotyping. Thus, they cannot be considered to be in the same category as members of the public who have no understanding or experience of genetic or genomic testing, with the exception of Jane and Ian. So to equate DTCGT users with the public, whom Evans and Plows describe as those with “no particular expertise in the topic other than that acquired in everyday life”, appears to be an incongruous association (Evans and Plows 2007:828).

Using focus groups, Kerr et al explored the public’s understanding and lay expertise about the new genetics. Whilst none of the participants in my study explicitly claimed expertise in SNP genotyping and its application to health risk, I would suggest that the users are similar to participants described in Kerr et al’s study, by virtue of being experts in their own lives with a good grasp of ethical and social issues related to genetics. This level of knowledge was relevant and applicable for them and their kin, if not detailed in relation to the technical aspects of testing and interpretation of results (Kerr et al 1998). Whether due to their experiential expertise of SNP genotyping and their self-knowledge, or because of earlier difficulties with healthcare services, it is clear that several of the users expressed concerns about NHS doctors’ ignorance of or lack of interest in genomics. For some this was related to personal experiences of health care unrelated to genomics. Helen, Laura and Nicola all expressed criticism of NHS doctors, which was informed by their negative experiences of trying to get help for illnesses that conventional NHS and private healthcare providers had been unable to diagnose or treat satisfactorily. They felt disempowered as individuals in the NHS and felt that there was often little, if any, appreciation of their own knowledge or views in their interactions

with healthcare professionals. Laura describes her difficulties in the extract below and has similar suggestions to Barbara in relation to prevention.

The health service seems to work with doctors being in charge with the patient being a kind of vessel that they look at and treat and examine; I rather baulk at that, I like to be involved in it and I do meet doctors who are like that, it's a kind of partnership as to how you deal with the various problems, but I have also met other people who tell you what to do. I find it really frustrating and difficult to deal with, so I like having control over it and I'd rather skip the doctors giving me a pile of information I already know and sometimes don't agree with and I like to have the information there and be able to mull it over in my own time. I do find doctors' appointments are really pressured as well, you get 10 minutes to talk to a Consultant sometimes and you have to say everything in that 10 minutes, coherently; and you don't get another appointment for months for instance and it's just a horrible, horrible experience ... I think the medical profession ought to get with it, make use of it [genomics]. I think they are pretty stuck in the past. I think it's appalling the lack of uptake of new technologies in the NHS. It's just a huge big sluggish monster and they are barely doing any sequencing of people as far as I can tell. I don't think they are any good at preventative or diagnostic medicine, it's all reactive what the NHS does and I wish they'd get more proactive and do things ahead of time rather than letting people get ill and then put a sticking plaster over the top of it. (Laura UP15)

Laura describes finding short, time-pressured NHS appointments unhelpful for her condition, particularly as she feels she seldom has any control or any contributory role in the management of her problem. The implication is that SNP genotyping has given her personal health information at her own instigation and which she can access at any time, resulting in a new sense of autonomy and empowerment. This is an example of Juengst et al's suggestion that empowerment could be used to reverse the control of medical paternalism,

as people like Laura (and Helen and Nicola) gain knowledge that is no longer privileged and confined to medical professionals (Juengst et al 2012). She appears to contrast this more positive experience with her experiences in the NHS and associate her negative impressions with a perception that the NHS is failing in relation to new genomic technologies and preventative medicine.

Others were sceptical about doctors' knowledge of genomics, although most did not refer to specific incidents. However, Ann, the DNA enthusiast with an encyclopaedic knowledge of genetic genealogy, was explicitly critical about healthcare professionals' knowledge of genomics. The collective "we" in this extract refers to genetic genealogists who are members of ISOGG.

The amateurs seem to know more than the experts and certainly for these tests we've found that to be the case. I mean things like doctors don't get training in genomics. I can't imagine there are many people in the health service who know the first thing about how genomics works, so if I wanted to know, if there was anything of concern in my results I would go to the people within ISOGG. We've got people who actually are doctors as well and they're the ones who really, their opinion I would trust more than anyone else because they've got the medical background but they've got expertise from using all the different tests [themselves]. In fact a lot, I mean even the Human Genetics Commission, they've put some misleading stuff out on their website about genetics tests and I wrote to them about it and they never changed it. (Ann UP2)

It was not clear to me on what basis Ann made her assertions about NHS doctors' lack of training or knowledge of genomics. However, she clearly feels that her knowledge of genomics and that of fellow enthusiasts in her biosocial community is greater than that of qualified healthcare professionals and this gives her a platform from which to dismiss them. Her assertion that she would refer to a doctor in her genetic genealogy organisation if she had any questions suggests that, although she uses a deficit model to view NHS staff in relation to personal genomics, she may be more comfortable discussing genomic information in a community she identifies with, rather than with

outsiders whose views of personal genomics do not support her own. This is indicated by her criticism of the Human Genetics Commission information.

Maria, the young genetics researcher, felt that some doctors were unlikely to know much about genomics.

I don't know how up to date they are kept with all these things. I can imagine some GPs in some parts of the country if you turn up with a 23andMe result, they wouldn't know what to do with it. (Maria UP15)

Her assumption appears to be that, as a researcher, she is working at the forefront of genetics and genomics. Having bought a DTCGT, she has additional personal experience that could be interpreted as expertise, which she imagines places her in a more knowledgeable position than that of many GPs. The tentative nature of her remark, as expressed in her use of the phrase "I imagine", could indicate she lacks experience on which to base it.

Two of the clinicians, an experienced nurse and the oncologist, both felt that GPs were ideally placed to handle and interpret enquiries about DTCGT results because of their experience of risk assessment in very short time frames. It is not clear if this perception was based on their experience or was a plausible suggestion given their knowledge of the nature of consultations in general practice. However, it was not confirmed by either the users' experiences or other clinicians' views.

Almost half of the clinicians expressed similar doubts about GPs' knowledge and understanding about genetics or SNP genotyping, as Maria and the other users referred to above. Some suggested that they need to have topical updates to familiarise them with developments in the field or that they had too little time to deal with genetic issues in 10-minute appointment slots and should refer patients on.

In general practice they are more likely to refer. I think that's partly because GPs just don't have time and partly because they accept that they send a lot of patients on to specialist services for whatever reason and a lot of clinicians don't

understand a lot about genetics anyway. Probably less than you'd expect actually. (Doctor CP15)

This doctor, a trainee in clinical genetics, assumes that specialist expertise is the preserve of hospital-based clinical specialists rather than GPs. He describes their reliance on referring patients to geneticists, as do other specialists in hospitals. His comment about lack of understanding about genetics by non-specialist colleagues suggests that he is surprised by how little genetics knowledge his colleagues have retained or developed, not being involved in the subject on a daily basis as he is. This view is more aligned with those of the users, who use a counter-deficit model in their views about GPs' understanding and engagement.

Users' views about doctors' ignorance of, or lack of engagement with genomics cannot necessarily be attributed to their relative expertise in the field, as respective understandings about the meaning of genomic data can be divergent. Whilst the users described above can be seen to be adopting the utopian vision of personalised medicine and criticising the NHS for not endorsing this view, the clinicians, in supporting the collective public health service, have a different perspective of the utility of genomics. As I have already described in Chapters Three and Four, the data show that many of the clinicians' technological frame supports the notion that the contingent nature of SNP genotyping data rendered its application to relevant health information useless and were concerned that the public might not interpret the risk data appropriately. These differences are supported by Parsons's and Atkinson's description of the fundamental differences between professional and lay interpretation of risk in a study of women with first-degree relatives affected by Duchenne muscular dystrophy (Parsons and Atkinson 1992). Joanna Latimer's analysis of medical dominance in the genetics clinic also suggests why these two perspectives are in conflict. Whilst she suggests that, on the one hand new genomic technologies can be seen as empowering and enabling, on the other they are also problematic. This is because their contingent nature undermines the prestige that medical knowledge derives from being associated with science and truth (Latimer 2013).

Medical expertise, which is often described as being based on privileged knowledge, endows practitioners with power and authority over those who are

not part of the profession (Freidson 1988, Elston 2002). Thus, it is exclusive and arguably undemocratic. Experiential expertise in new technologies is similarly undemocratic, excluding those who are not part of that group of early adopters. This results in experiential experts in the public speaking *to* those who have no experience rather than *for* them, despite their thoughts to the contrary, as Epstein emphasised in his work with HIV/AIDS activists (Epstein 1995). Personalised genomics, in common with other new medical technologies, is specifically undemocratic because it excludes those who cannot afford it or who are otherwise vulnerable, as well as challenging expert authority (despite the democratising arguments for personal genomics). This disparity is at the heart of the conflict between this study's participants' opposing views and between personalised versus collective medicine. The NHS and the clinicians who work in it are essentially providing a public health service for the collective good, which inevitably requires rationing, and many of them imply that this includes protection from unanticipated demands such as those that result from privately generated, uncertain health information (Juengst et al 2012).

In exploring how new medical technologies are experienced, negotiated and challenged in the clinic, Brown and Webster noted the impact of these actions on the trust relationship between patients and healthcare professionals (Brown and Webster 2004). They suggest that healthcare practice is unfeasible when trust fails; whilst it would be an exaggeration to claim a breakdown of trust between users and healthcare professionals, these data suggest a shift in the nature of some of the users' relationships with the doctors they encounter that may be partly informed by their engagement with personal genomics. The threat to the relationship between practitioner and patient has been partly responsible for many calls for regulation of personalised genomics, the topic I shall go on to next to complete this analysis of the data.

Regulation

This section builds on the detailed discussion about Regulation of genetic testing in Chapter Two, which I will not repeat here. This landscape has changed somewhat during the course of this study, owing to the changes in the market of DTCGT described in Chapter Two. However, these changes post-dated the interviews. Discussions I had with participants in the interviews

elicited strong views about regulation that shed further light on participants' thinking about personalised medicine and the NHS and I will go on to examine these next.

As I argued in Chapter Two, moral order in society relies on people sharing understanding of matters important to that community; the absence of consensus about DTCGT leaves room for uncertainty. This ambiguity may partially explain why many clinicians, ethicists and lawyers interested in the debate about the availability of health information technologies direct-to-consumer call for regulation. Motivations for doing so differ whether they are driven by hopes for public protection, paternalism, boundary work in protecting different groups' identity and practice, or similar, it appears to be the fall-back position for these concerned professionals and academics. Others disagree with regulation; this may be because they are enactors of this emerging technology seeking to recruit selectors to invest in or make use of DTCGT as described in the section on Expectations in Chapter Three. Alternatively, they may be selectors (here I equate this to early adopters) whose expectations have been built up by the enactors and who do not wish to consider the potential uncertainties or difficulties that the new technology is associated with (Groves and Tutton 2013). Finally, there are those who are more familiar with the scientific aspects of DTCGT and its uncertainties, those who arguably have more expertise in the field and who feel that onerous regulation would be unwieldy and excessive given the nascent and non-deterministic nature of SNP genotyping information (Hennen et al 2008). All these groups' agendas contribute to the contestations about DTCGT and the lack of stability of the technology.

I asked participants about their views on regulation of DTCGT and their reactions to suggestions that DTCGT should not be allowed. Users' were mostly strongly in favour of maintenance of the status quo in terms of availability of DTCGT, though for various reasons. These included it being too early in the developments to regulate it, it being too late as it is already freely available, and the more popular argument about autonomy and the rights to one's own genetic information. Clinicians were generally in favour of some kind of regulation to ensure transparency in marketing materials and quality assurance which could be interpreted as an extension of the professional protocols that give their group its influence in censoring DTCGT.

The users who championed personalised medicine and SNP genotyping were likely to comment on the relationship of regulation to medical hegemony. The geneticist Fiona observes that

The rhetoric that's coming out at the moment from the medical profession over direct-to-consumer testing does sound a bit like 'but we've always done this bit, we want to carry on doing this bit' rather than actual real genuine concern. (Fiona UP8)

Fiona conveys the perception that the medical profession wish to maintain professional authority over genetic testing. The phrase "we've always done this bit, we want to carry on doing this bit" suggests that she interprets this as related to medical professional authority and boundary work. She appears to think that, like her, the medical profession does not necessarily view DTCGT as harmful, but rather that the prospect of individuals autonomously obtaining their own health information pushes the boundaries between professional authority and informed patients too far.

Some users qualified their views about regulation in relation to the lack of utility or harm from DTCGT, suggesting that there was no point in regulating something that simply provided personal information. Whilst she supported this view, the genetics statistician Kirsten acknowledges a different perspective in the debate over who should have access to genomic information.

Yeah, it seems really odd to me that you want to regulate a test that has so little utility. It just seems a complete waste of time and using a sledgehammer to crack a nut sort of thing, so I'm aware of some of this and have been on the websites like some of the genetics associations in the States who say "You should not do a direct-to-consumer genetic test – that's it". And it's like "What? Hang on, no conflict of interest here of course!" But I think the level of regulation should reflect the value of the information that they are providing. So I would expect a much higher level of vigilance for, you know, sequencing for *BRCA1* variants for example which could

have huge downstream implications for mammography and oophorectomy or whatever, than compared to a company that's telling me that my type 2 diabetes risk genetically is slightly above average. So yeah, it's not something that concerns me. And I think that there's the whole other side of this isn't it, where genomesunzipped is coming from, that this is our DNA that we have a right to find out what is in that and the medical community should not stand between us and our DNA, empowerment rather than endangerment and I think that's slightly disingenuous. (Kirsten UP14)

Kirsten expresses her view about the pointlessness of regulation for relatively non-deterministic information by using the metaphor "using a sledgehammer to crack a nut". She emphasises this by critiquing medical professionals' boundary work, which is implicit in the policy documents that she has found online. She uses the example of *BRCA1* sequencing as a contrast, by way of emphasising the non-deterministic nature of SNP genotyping. Her views seem to be pragmatic and balanced and similar to those expressed by Hennen et al who proposed that regulation need only be proportional to risk (Hennen et al 2008). She also directly criticises those who subscribe to the democratisation of genomic data for reasons of empowerment, implying that this view is no more appropriate than a paternalistic one.

Few of the users appeared to have considered either the problematic nature of interpreting SNP genotyping data or the confidentiality of their data or samples in the present or future, should the genomics companies cease trading in their present form. As a result, discussions with users about regulation seldom encompassed issues related to quality assurance or requirements for sample and data storage or destruction. The same cannot be said of the clinicians' views, which were more focused on these issues.

The majority of clinicians thought that DTCGT should be regulated. This advocacy for regulatory control could be interpreted as being aligned with this relevant social group's professional power and the reassertion of boundaries between medicine and the public. However, nationally (or internationally) applied regulation would require the engagement of, and negotiation between, different groups of actors, which appears to have been avoided in relation to

DTCGT, resulting in its lack of stabilisation as a new technology. Suggestions about regulation were mostly grounded in participants' direct experience of their professional working directives, including stringent quality standards for genetic testing laboratories, as in the NHS and professional requirements for Genetic Counsellor registration that are required for NHS clinical genetics services. Some participants expressed their suggestions from a moral standpoint, suggesting that NHS Clinical Genetics services provide the normative framework on which other provisions should be based, as this participant suggests.

They should just provide the correct service. If you could guarantee that any company out there was going to make sure that people were given a good service and had all the relevant information in an appropriate way to start with so they weren't going into something with their eyes shut then no but I don't know how so I have to say yeah, I think they should be regulated. Just to guarantee that people are going to get the right service. (Nurse CP9)

What is the "right service"? (Teresa)

The "right service" is preparation and understanding and an education about what they're going in for. I suppose for me it's proper informed consent to do the test. And proper informed consent is understanding what the consequences are of this test so you can make up your mind if it's something you want to go ahead with. If you have had that and after you had that information you still think that is the right test for you then that is the right test for you and that's absolutely fine. But it's having that information and at the right level for that person. (Nurse CP9)

This nurse supports the need for regulation and emphasises this in relation to informed consent. She uses the words "correct" and "right", giving emphasis to her support for regulation in relation to the principles that genetic counselling is based on. She does not appear to consider the different types of information that genetic testing and genotyping generate or the respective implications for

the consultand; she appears to assume that any kind of genetic information should be accessed through the genetic counselling framework.

This nurse was not alone in suggesting that regulation was required for public protection. Like these other participants, she appears to be basing her suggestions on the ethical principles underpinning genetic counselling which have been developed with monogenic disorders inherited in a Mendelian pattern. As other clinicians pointed out, applying the frameworks for regulation that apply to inheritance patterns and implications of monogenic disorders is not appropriate for genomic data based on GWAS (Arribas-Ayllon 2011). Those participants who suggested this were very experienced in their field, with wider responsibilities than the clinicians who supported regulation for the purposes of public protection. The experienced doctor with a broad portfolio of responsibilities expressed it thus.

But in a sense this conversation is slightly being distracted, as is always the case because really when we think about over-the-counter-testing we always get hung up on the really high penetrance conditions. But there's a whole other massive market that's not high penetrance disease and I'm weary about talking about HD because it always comes up you know "What about HD?" HD is an annoyance in a way because we crack the exception before we go to the rules. The vast majority of genetic testing is done to benefit the health care of the individual and the family and does so in a much more direct way than simply predicting they've got an incurable disease. So, for the high-penetrance autosomal dominant conditions for which we have a treatment it really is a no-brainer that we should be doing more of it. And if you look at conditions like FH which is the one I always throw back at the HD model, [it's] about 20 times more common and causes vast numbers of young people to drop down dead avoidably. The thing is that when we do cholesterol measurement we're doing a genetic test, we're just measuring the gene product rather than the gene itself. It is outrageous to be honest that we are still not routinely testing everybody for FH. Vastly common, totally treatable, people

dying in their 40s and 50s that could have been prevented if they'd been picked up and put on statins. But you know the NHS can't afford to roll it out and they don't like putting it out over-the-counter because it's genetics. (Doctor CP10)

The interesting point in this extract is that this doctor dismisses HD as the basis for regulation that should be applied to generating and communicating any genetic information. He illustrates this by contrasting HD with familial hypercholesterolaemia (FH), a very common genetic disorder. Although it is inherited in the same pattern as HD and has potentially lethal consequences, it is easily treatable. Despite these similarities, marketing a genetic test for FH over-the-counter has failed to be approved, owing to concerns about DTC access to genetic testing in the UK, because it is a genetic test rather than because of any potential harm. The doctor is clearly frustrated by this barrier to helping people potentially at risk of avoidable disease because of funding constraints. These blocks to selling testing DTC could be interpreted as representing a reluctance of the collective provision of health care in the NHS to move beyond the deterministic model of genetic testing and acknowledge the different types of genetic information and testing that new medical technologies are producing. Equally it could be interpreted as representing as their adherence to a particular technological frame for genetic testing in general, rather than one specific to DTCGT owing to the challenges that institutional and professional opposition present for new medical technologies (Green 1991).

Other suggestions for regulation included adherence to advertising standards for which requirements are already in place and enforcement of legal requirements in international law to control the global reach of online marketing. A few clinicians and one of the users referred to the concept of self-regulation as being an appropriate mechanism by which to achieve acceptable test quality and ethical standards without any impact on public services or costs. It appeared that the foundation for all these suggestions was based in the boundary work discussed in Chapter Four and was simultaneously aimed to protect the NHS from opportunistic demands on resources from early adopters.

Several authors have noted that grounds for regulation rely on perceptions of boundaries that no longer exist, such as those between authoritarian

healthcare professionals and ignorant patients or between expert scientists or researchers and the uninformed public. The undermining of these boundaries indicates that, even if regulation in personalised medicine is attempted beyond that of the FDA's Cease and Desist notices, it is unlikely to be effective and may not be an appropriate response (Boddington 2009, Juengst et al 2012, Prainsack et al 2008).

Common ground

If SNP genotyping for common complex diseases has the potential to disrupt the trust on which patient-practitioner relationships have historically been based, the data in this study suggest that one aspect of genomics on which members of both the study's relevant social groups concur is pharmacogenomics. This is perhaps fortunate, given that one of the areas of health care in which patients (in the UK) continue to be dependent on qualified healthcare professionals is in the prescribing of medicines. However, the scientific uncertainties that apply to disease-risk testing are equally applicable in pharmacogenomics but arguably have more serious implications. Establishing someone's genotype for drug metabolism will lead to prescription of drug treatment along with others of similar genotype, there being no individually designed medicines or market for their development (Prainsack and Wollinsky 2010). In addition, pharmacogenomics can also marginalise those who cannot access testing or exclude from treatment those whose genotype is not catered for (Juengst et al 2012). Despite these concerns, the majority of participants in both users' and clinicians' groups spoke positively about the potential of pharmacogenomics to improve health care. For users, this was typically couched in discourse related to empowerment, whilst clinicians referred to SNP genotyping for drug prescribing in favourable terms, unlike their views on testing for disease risk. The following two extracts demonstrate examples of this.

Carol anticipated a future in which she would negotiate her drug regimen with her GP.

On my genetic thing it says there's a sensitivity to Metformin. I obviously get it from my mother and Metformin is such a typical drug for giving to Type II diabetic patients so it's going

to be interesting if I ever get to the point of having a GP say he's going to put me on Metformin, [I wonder] how much notice he's going to take when I say "I don't think so". (Carol UP4)

Carol's description of how she will handle communicating her pharmacogenomics data in future is arguably presented in rather confrontational terms. She envisages the GP will prescribe the drug in question without any discussion and that she will refuse to have it, thus anticipating on-going challenges by informed patients to authoritarian doctors in an effort to personalised medical treatment.

The doctor who led a regional genetics department and who referred to DTCGT as "snake oil" has a different view in relation to pharmacogenomics as this extract illustrates.

I can see the value in these types of things; you know Warfarin, Warfarin's a black art. When I worked on the wards as a junior doctor, I was on a cardiology ward right. I was told, "Start this guy on Warfarin." "Right, how do you do that?" "Prescribe him 10 [mg] today, 10 tomorrow and 5 the day after that and test his INR." With the wee old women you'd maybe go 10, 5, 5 or something like that but it was a bit of a guess. And you'd test their INR and some guys INR hadn't shifted from baseline so you'd whack them another 10 [mg] and some little old woman who was ready to bleed out all over the ward because her blood was so thin, it was like dilute orangeade! So it was, it was a guess, but now we're getting a handle on that. A little bedside SNP [would do it]. They come in and get full Us and Es, you'd type the SNP so we'd know how to prescribe and then start them on Warfarin. Patients with rheumatoid [arthritis] or Methotrexate, type them so we know [how much to give them]. It just seems to me sensible to get that kind of information. But that's not predicting a genetic disease; we need this information to look at how to dose you. (Doctor CP13)

The doctor's vivid narrative explains his positive view about SNP genotyping for pharmacogenomics, as being related to better prescribing (and thus reduced side-effects), as an encouraging prospect. He explores the positive potential of SNP genotyping amidst medical jargon related to prescribing anticoagulants, which requires titration to the patient's coagulation time¹². His exploration of this possibility suggests that he envisages possible applications for SNP genotyping in the NHS in future. However, studies have not demonstrated that genotyping patients for Warfarin metabolism and dosing them accordingly makes any significant difference to the efficacy or safety of this difficult and potentially dangerous treatment. Neither the efficacy of patients' anticoagulant therapy, in terms of the time that their clotting is within the target range during treatment, nor the side-effects of inappropriate dosing, including haemorrhage or thromboembolism, are significantly different in those whose dosing is guided by genotype data rather than by clinical variables alone (Kimmel et al 2013, Stergiopoulos and Brown 2014). This casts doubt on the possible advantages of point-of-care genotyping in Warfarin dosing; in addition there are difficulties with the adoption of other pharmacogenetic tests, related to lack of evidence of efficacy or clinical validity, cost considerations, lack of clinicians' knowledge and inconsistent regulation in the EU (UK Pharmacogenetics Study Group 2006).

The participants' support for pharmacogenomics could represent a space in which to negotiate the socially acceptable aspects of genomic technology in the NHS in future, an opportunity for stabilisation of the technology as Hedgecoe and Martin argued in relation to pharmacogenomics over a decade ago (Hedgecoe and Martin 2003).

The challenges in reconciling personalised and collective medicine within a public health service are significant and may be impossible to overcome,

¹² Warfarin dosing is titrated according to the patient's clotting times as measured by their Prothrombin Time (PT) and aims to achieve a time within the International Normalised Ratio (INR). Warfarin has a narrow therapeutic index (established by an INR of between 2.0-3.0 or 3.5 depending on the indication for anticoagulation); its metabolism varies considerably between individuals, and is affected by diet and other medications as well as individual (genetic) differences in metabolism. Side effects of haemorrhage or thromboembolism are not uncommon and can be fatal which is why titration of dose to INR is important (Keeling et al 2011). The genetic basis for individual differences in Warfarin metabolism includes variants in the gene *CYP2C9*. This encodes the enzyme Cytochrome P450 2C9 that plays a major role in oxidation of various compounds (including Warfarin) in the liver, but it is not the only genetic influence on drug metabolism (Kniffin 2010).

particularly as the future of the NHS is increasingly the topic of much public, media and political wrangling. However, having discussed their views about DTCGT and the challenges it represents for health care, some of the participants (users *and* clinicians) were keen to highlight the importance of the NHS for the country and the public, as I did at the beginning of this chapter. They referred to examples from their practice or research, or the media references to the NHS as a national emblem, which were ubiquitous at the time some of the interviews were conducted, to illustrate the UK public's attachment to the NHS and the preference for its services over seeking healthcare information independently. This excerpt represents the overall sentiment about the public and the NHS.

They are actually preferring to go for the tried and tested NHS, because people know what they're doing so let's trust them [NHS staff] and maybe that is the motivation for people going through that somewhat tortuous route [genetic counselling], maybe people are kind of seeing the benefits of that. It is an issue of trust that they're not clicking in droves and sending their 99 pounds, or dollars even. (Scientist CP7)

This scientist, having earlier reflected on why people might choose DTCGT, concluded that the relationship of trust between the NHS and its patients is intact. Her use of the phrase "tried and tested" suggests that this might be related to their familiarity with this model, rather than pursuing the personalised medicine options for obtaining genetic information, despite the cumbersome nature of clinical genetics.

Maria, the genetics researcher who tested because of her "narcissistic" curiosity, saw distinct differences between the NHS responsibilities and her interest in DTCGT.

The NHS doesn't have the resources to do these things for people who are just interested or curious, here people really generally trust the NHS and I think most British people have huge affection for the NHS and there's a lot of respect for it. It's something we are really proud of and so we tend to trust the NHS, even though it's not perfect. (Maria UP16)

Again, the importance of trust is evident here as in previous extracts and Maria uses it repeatedly. She describes the public's view (which is presumably founded on her own), using emotive words such as "trust", "affection" and "respect", suggesting that the model of collective medicine on which the NHS stands is positive and beneficial overall.

Conclusion

I would argue that the data analysed in this chapter are representative of the prevailing tension between personalised and collective medicine in the UK. Whilst DTCGT is the point of focus for the conflict between the two positions, I would argue that it is not the cause; rather, it is a vehicle that this study's participants have used to express their views in relation to this emerging challenge. It is important to re-iterate that, whilst support for personalised medicine was voiced by quite a few of the users, it was not a position universally held by that group, although all the clinicians supported the NHS collective model of health care, as one would anticipate.

As the data have demonstrated, expectations that healthcare services could be overwhelmed by demands for consultations from people worried about DTCGT results have not yet materialised. The NHS is not being tested in that sense – in clinical genetics departments at least. However, the NHS is being tested by the concepts of personalisation and consumerism and the associated challenges of public responsibility and expertise, which are related consequences of personalisation and an increasingly informed public. Arguably, this should come as no surprise, particularly to the NHS and its staff. Healthcare policy in the UK has been increasingly orientated towards individual choice and health promotion since Thatcher's first radical changes and shift from the paternalistic, universal model towards a more consumer, choice-focused one, in rhetoric at least (Klein 2010). In light of these changes, the uptake of opportunities to avail oneself of health information is not unreasonable. The reportedly dismissive response by healthcare professionals could be interpreted as understandably frustrating and is the basis for the Nuffield Council on Bioethics recommendation that healthcare professionals be trained in genomic technologies (Nuffield Council on Bioethics 2010). Conversely, professional frustrations are equally understandable, given the lack of transparency in commercial health information marketing materials and

the unremitting and significant restraints within which clinicians are expected to provide services to patients who have increasingly high expectations of the NHS. In addition, the apparent lack of any changes to users' lifestyles in the light of their test results perhaps indicates a lack of genuine engagement with responsibility for their health and negates the argument for empowerment.

When examining the concept of "personalisation", the Nuffield Council on Bioethics suggested it encompassed four aspects, including individualised care, holistic care, commodification of care and responsibility and autonomy in managing one's own care (Nuffield Council on Bioethics 2010). With the exception of commodification, these aspects are not new to the fundamental basis of professional health care (theoretically at least) and I would argue that health care has always been personalised to some extent. A recent medical paper even cunningly invented the term "personomics" to revisit the importance of individualised care within increasingly 'omic'-focused medicine (Ziegelstien 2015). The shift to personalised health care is thus not the point here, an argument lent weight in the case of DTCGT by the impersonal, population studies GWAS data are based on. The point is who should take responsibility for individuals' health; the dissonance of views between the public (as represented by some of the users in this study) and the NHS (represented by the clinicians) demonstrates the ambiguous position health care in the UK currently finds itself in. We are between what Klein refers to as Church (paternalistic, collectivised medicine) and Garage (consumerist, individualised medicine) (Klein 2010:282). New health technologies do offer the potential for people to be more involved in their health management rather than simply being passive recipients of healthcare decisions. This could facilitate disease-risk management as a cooperative venture, although the caveats that marketing information should be more transparent and healthcare professionals should be trained in genomics both apply in order for a relationship of trust between professionals and the public to be maintained.

In the next and final chapter of this thesis I shall pull together the three themes I have explored in Chapters Four, Five and Six. In doing so, I shall discuss the shaping of discourse about personal genomics technology in the UK in relation to biosociality, before concluding with suggestions for future study in this area.

Chapter 7 : An unresolved dispute and future possibilities

It remains to draw conclusions about how users and genetics clinicians are shaping the technology of personal genomics in the UK and discuss the implications of the findings of this study. To do so, I will summarise the current state of the debate about DTCGT and then discuss the important aspects of SCOT in this study and the three principal data themes. Then I shall broadly relate theoretical work on biosociality to the empirical findings of this study. Finally, I shall complete the thesis with a discussion about the possibilities for future work in this area that could either follow the disputes over new genomic technologies into WGS (which is where I see this particular dispute shifting) or engage in theory development.

Developments in the commercial sector following the FDA's letter to 23andMe in November 2013 show how this company, at least, is willing to both circumvent some of the obstacles to selling personal genotyping directly to the public and simultaneously work with regulators to address others, thus keeping their business active and their options open. In 2014 23andMe launched SNP genotyping tests that include common complex disease risk testing in both Canada and the UK, as neither country has regulation in place to prevent them doing so (Gibbs 2014, Pickard 2014). More recently the company received authorisation from the FDA to market a test for Bloom's syndrome carrier status. But as Cecile Janssens points out, this is for a specific carrier test and not for 23andMe's more broad SNP genotype service including common complex disease susceptibility testing (Janssens 2015). Obtaining approval to market testing for Bloom's syndrome is an interesting move on both the company's and the FDA's parts. This test is relatively harmless and uncontentious; using it as a foundation from which to negotiate with regulators for future approvals could be interpreted as symbolic of future potential for widening public discussion and knowledge about commercial genomics.

Whilst the academic ELSI community is arguably less focused on the disputes about DTCGT now than in the latter half of the last decade, there is no indication from the literature or this research that this is due to any settlement or closure of the points of debate. I would suggest that this is due to two other factors. First, as several researchers and the data from this study suggest, there is little evidence of users of DTCGT coming to any harm (Heschka et al 2008, McGowan et al 2010, Kaphingst et al 2012). Second, focus has shifted to WGS, as exemplified by the announcements from David Cameron and Barack Obama about the UK's and the US's respective national initiatives to complete WGS projects in healthcare research and their attendant promissory expectations (Genomics England n.d., Brice 2015, Collins and Varmus 2015). However, whilst the focus of media and scholarly attention may have moved on to a new problem, the points of contention about DTCGT in the UK remain unresolved and this concluding chapter will summarise these outstanding issues next.

The findings

Claims and counter-claims about selling SNP genotyping based on contingent science directly to the public have resulted in the disputes around DTCGT that this study has sought to illuminate. Whilst purchasing health-related tests in the context of insurance-based private health care such as the US model may not seem controversial, in the UK's publicly funded NHS system it was not clear how users or clinicians would be affected by this commercially-offered genomic technology or influence its development. Interviews with early adopters of personal genomic testing and genetics clinicians who may have a role in supporting users, or at least a professional interest in the debate about DTCGT, have shed light on both groups' views and experiences. In this section I shall discuss the use of SCOT for this study and each of the three data themes as they relate to my conclusions about this research.

Social construction of technology

The use of the SCOT framework for this study has demonstrated its value in illuminating the black box of DTCGT. Here I will summarise the factors

in each of the components of SCOT that are noteworthy for their specific bearing on this study.

Relevant Social Groups

Paying attention to the relevant social groups' understandings about DTCGT, and to the groups within those groups, has shed light on contrasting experiences, groups' influence and power, their different technological frames and the subsequent impact on people's understanding of DTCGT. Genetic genealogists encountering DTCGT with health data for the first time appear to maintain their enthusiasm for personal genomics. This is demonstrated through their strong networks and their eagerness to share their experiences and support for the technology with me. Those who work in genetics research directly or in similar areas conveyed curiosity about the technology and that they were sharing this with colleagues in professional networks; those with health concerns seemed to view it as a means to gain personal information that they had agency over and share with family. The couple who were outliers appeared to have a different experience; this could be seen as contributing an experience more representative of the wider population without the other users' arguably activist attributes. However, whether they are more widely representative or not, collectively the users in this study are arguably illustrative of a diverse group. The challenge of having diverse relevant social groups in analysing SCOT is that some powerful voices may maintain that they speak for the whole group when in fact sub-group's experiences and technological frames may differ (Russell 1986), and that may well be the explanation for the outliers' different experiences.

The genetics clinicians' influence is aligned to their professional duty to individuals and the NHS. The moral boundary work they engage in informs much of their understanding and discourse about DTCGT though the professional sub-groups of doctors, scientist counsellors and nurse counsellors demonstrate variations in the emphasis they place on this in relation to DTCGT. This groups' collective professional power and their generally sceptical views of DTCGT based on its lack of clinical utility, present a strong counter influence to the users' enthusiasm for DTCGT.

Wider social context

The context of the NHS in this study is key to understanding how the relevant social groups both conceptualise DTCGT in relation to responsibility for health and the influence of these views on DTCGT. The NHS represents a powerful construct in that it is widely supported by citizens and its employees in the UK (Klein 2010), and participants from both groups in this study had positive views of it. However their different expectations of its provision, is one of the principal points of conflict in relation to DTCGT. Users suggest that a more individualised consideration of people's health needs through genomics is important, in line with commercial rhetoric in personal genomics. But this is in opposition to the conservative approach clinicians represent. They defend the collective model of health care that they envisage may be threatened by those needing help with interpreting privately acquired screening information, although this eventuality has not so far materialised.

Interpretative flexibility

The different meanings and interpretations of DTCGT by users and genetics clinicians in the UK essentially align to one of two positions. Users appear to support its personal utility and relevance for its individually orientated information although there was very little evidence of this influencing people's behaviour. Its direct availability is key to their view and they would not countenance regulation. They view this as unnecessary and paternalistic. Genetics clinicians are mainly sceptical of the value of SNP genotyping, owing to the flawed science it is based on, its misleading marketing and concerns about potential harm to individuals who are receiving genetic information without conventional support.

The participants also demonstrated their interpretative flexibility *within* their individual accounts of what they understand DTCGT to represent. This illustrates the relevance of Potter's and Mulkey's experience of scientists' interpretative flexibility in understanding the variation in their views and how their understandings are re-worked according to the specific context under consideration (Potter and Mulkey 1985). A scientist recognised that disease-risk results should not lead her to make unhealthy lifestyle choices, but pharmacogenomics results meant she

would not agree to being prescribed certain oral hypoglycaemics in the future. A doctor thought DTCGT was akin to snake oil but that SNP genotyping for warfarin metabolism could be a boon for both patients requiring anticoagulation therapy and the junior doctors who need to prescribe for them. Participants seeking genomic information to shed light on chronic ill health shifted their hope on to the future promise of their genomic data when the tests failed to fulfil their expectations.

Stability and closure

These changes and swings in understanding DTCGT illustrate and support the lack of stability of this technology in the context of the UK and the NHS, despite pharmacogenomics providing a focal point for converging views between some users and some clinicians. Stabilisation is described as being relative for different groups and needs to be achieved in the eyes of each of the relevant social groups (Pinch and Bijker 1984). Whether DTCGT will ever achieve closure or not is currently moot, but I suspect that it is unlikely given the irreducible nature of the differences between the two groups in this study. I suggest that it will be stabilised by virtue of being superseded by new problems, as I shall consider in the final section of this chapter. Having established the significance of interpretative flexibility in terms of participants' accounts, I shall go on to summarise the three main themes from the study.

Networks and expectations

Perhaps surprisingly, given the personalised basis for marketing DTCGT and its association with the increasingly neoliberal culture that the participants in this study live in, social networking and sharing with others who have common interests were principal features of the participants' accounts. Both groups' networks and their attendant social practices were in existence *before* the emergence of personal genomics. As demonstrated in Chapter Four, participants found out about DTCGT through their social networks, whether recreational, family, or professional. Networks and membership of them are crucial because enrolment to and socialising within them informs and reinforces the participants' values in respect of DNA. This is achieved by shaping expectations in relation to new technology, as Brown and Michael suggest, or bolstering professional

authority by alignments within professional networks through what Callon might refer to as networks of associations (Callon 1986, Brown and Michael 2003). As a result, norms can be adopted that provide principles for behaviour within the actors' respective networks. This is supported by the distinctly different experience of the two users who were not directly involved in these networks but were bought DTCGT by a relative who was.

In trying to establish users' motivations for testing, it appears that most people's expectations and engagement were not about clinical utility. They were more aligned to the concept of personal utility, as demonstrated by references to their interest and curiosity and their extension of their personal identities through this newly acquired genomic information. This supports the observation that personal utility is important to DTCGT users, whether as a moral justification for engaging with it or as an alternative criterion for judging its utility (Khoury et al 2009, Vayena et al 2012, Bunnik et al 2014). For the three participants specifically interested in health-related data their expectations for useful data from their DTCGT were not reified. None seemed particularly disappointed by the characteristically unfulfilled promise of personal genomics to date (Brown 2003), but translated utility of their data into future hope, in a fashion similar to Brown's "regime of hope" (Brown 2005:333).

Clinicians were orientated to the importance of clinical utility and their professional network appears to be important in facilitating their shared moral stance against DTCGT on the basis of its lack of clinical utility and companies' misleading marketing. Their networks also circulated the expectation that their services would be inundated with requests for consultations to explain DTCGT results. This has not been reified, any more than have those users' hopes for clinically useful information about their health, as I established through the difficulties recruiting clinicians to the study and from the accounts of their experiences from those who did volunteer. So the impact of DTCGT on NHS genetics services has proved to be negligible. This last point is also related to the issue of expertise, in that it would appear to indicate that credentialed expertise in clinical genetics is not required for DTCGT interpretation, despite the early concerns about genetic tests being sold directly to the public without

involvement of clinicians (Wolfberg 2006, Matloff and Caplan 2008, Marietta and McGuire 2009).

Participants' expectations of the NHS represent a point of tension around responsibility and the groups' different understandings of it as I will discuss shortly in the section on Individualised or collective medicine

Individuals and identity work

Participants' expectations about DNA on a personal level constituted their thinking about DNA and the information they gleaned about it, incorporating this into their self-knowledge or aligning it into their genetic counselling practice. This personal and internal reflection relies on pre-existing knowledge and, for users, facilitates the extension of individuals' identities in many facets of their life, including their health and families. For clinicians, it facilitates imagining what users' motivations and experiences of genetic testing might be and how this might extend the possibilities for public engagement with genetics or conflict with their genetic counselling approach and lead to anxiety and a possible drain on precious NHS resources.

The user participants in this study appear to be building on their pre-existing knowledge of genetics in deciding to acquire additional information about themselves with their DTCGT,¹³ in keeping with many early adopters of new technologies (McGowan et al 2010, Vayena et al 2012). Users incorporated this new information into their personal identities, as O'Riordan suggests, forming new personal and social identities (Atkinson and Glasner 2007, Kelly 2007, O'Riordan 2010, Prainsack 2014a). Arguably, this knowledge also provides a foundation for imagining their future health care on a more individual plane. This could be described as extending their own self-knowledge as well as their knowledge of genomics, particularly given that all the users claimed to understand their DTCGT results. Apparently negative responses from GPs with whom some users shared their test results appear to have bolstered users' views that the NHS is out-dated in terms of its care

¹³ This is with the exception of Jane, one of the two exceptional cases whose knowledge of genetics was minimal.

provision. This aligns to Novas's and Rose's "somatic individual", for whom the outcomes of extending one's identity into the genetic, influences relationships with others including clinicians (Novas and Rose 2000:487).

Clinicians' individual thoughts about the possible impact of commercial genotyping varies from being actively encouraging to expressing concerns about possible harm to individuals and their kin through rehearsal of the discourse of genetic counselling. Clinicians' cautious attitude to DTCGT appears to be based on the post-eugenics anxiety that the informed consent, non-directive counselling model of clinical genetics has been built on (Fox Keller 1992). But their moral boundary work here is based on the model of single-gene mutation or chromosomal abnormality that clinical genetics services commonly deal with, rather than imprecise SNP genotyping for common complex disease susceptibility. However, their concerns are arguably unfounded, given that the anticipated onslaught of requests for consultation from the worried-well, with their electronic device-access to their SNP genotype results, does not appear to have materialised, leaving clinicians concerns largely imaginary so far.

These individual expectations, which are in a dynamic relationship with the socially shared aspects of personal genotyping, both influence and converge in the final theme in the data, the NHS, which I shall complete this section by summarising.

Individualised or collective medicine

The central focus of the dispute about DTCGT in the UK context is between the models of personalised versus collective medicine. As I outlined in Chapters One and Two, an increasingly neoliberal emphasis on globalisation has supported the commercialisation of health care and the impetus for individuals to take responsibility for their health. This has been capitalised on by personal genomics companies and those who campaign for democratisation of health information; many companies have deliberately marketed their products by emphasising the personal or individual in their choice of company name (23andMe) and their marketing focuses on providing information to facilitate autonomy and empowerment

in relation to lifestyle choices for health. Marketed largely on the Internet, which is awash with the use of similar rhetorical devices, personal genomics purports to empower responsible individuals with access to their valuable personal genomics information via their iPhones, iPads, iMacs or similar devices (Dickenson 2013).

In parallel with the published debates about DTCGT, this specific issue figures as a central point of tension for participants in this study, with responsibility being the feature on which the tension between users and clinicians turns. Users' adoption of new genomic technology represents a more responsible approach to their view of themselves or their life strategies, particularly in relation to health. Genetics clinicians' counselling work is centred on helping consultands to develop responsibility in terms of health behaviours and relationships with family, whether actual or potential. So whilst both seek to achieve the same end, their concerns about how it will be achieved are at odds.

Users appear to have aligned themselves with the tropes of personalisation that commercial genomics companies use to market their products, regardless of their original motivation for engaging with SNP genotyping. Clinicians, on the other hand, express concerns about the scientific basis for SNP genotyping in order to illustrate its irrelevance in terms of the provision of clinical genetics services in the NHS. Whilst nurses and some doctors also rehearse the moral arguments for maintaining genetic testing within their professional service, several clinicians show a more balanced view, acknowledging some of the possible advantages to obtaining genomic information without having to endure the rigours of navigating the NHS to do so. However, the point clinicians seem agreed on is their protection of the collective NHS model of health care; their dismissal of DTCGT appears to be as much about their concern to protect the NHS from being drained of its resources by the worried well pursuing genomic tests privately (or indeed other screening options) (Henrikson et al 2009, Nuffield Council on Bioethics 2010).

As Richard Tutton shows, personalised medicine is not an entirely novel concept. Individualised medicine has figured in health care for centuries,

with a focus on the individual featuring in medical practice before the development of hospitals in the 19th century (Foucault 1994) and being increasingly central to healthcare practice and policy since the latter half of the 20th century (Klein 2010, Tutton 2012). The alignment of personalised medicine to the HGP enabled researchers and scientists to raise expectations and funding for their pharmacogenomics research (Hedgecoe 2003). This useful rhetorical tool, which is in keeping with the neoliberal agenda of the times, disguises the fact that pharmaceutical developments are not being individualised in a literal sense. Rather, populations are being categorised into groups who respond differently physiologically to particular drugs (Hedgecoe and Martin 2003, Tutton and Jamie 2013). Individualised drug treatment is highly unlikely to be reified, given the lack of financial incentive to pharmaceutical companies to invest in what could be termed niche products.

It is ironic to note that whilst the tropes of personalisation, individual autonomy and democracy are used to market DTCGT and are being subscribed to by users of personal genomics, the data that customers' SNPs are analysed against are large-scale data used for GWAS (Khoury et al 2010, Prainsack et al 2014). What is more, the basis on which personal genomics companies' business plans are developed, that is, to sell their customers' health and genomic data on to biotechnology and pharmaceutical companies, is anything but personal. This aspect of commercial biotechnology was alluded to by one of this study's clinician participants, along with others with an interest in the field and subsequently announced by 23andMe early in 2015 (Hamilton 2008, Herper 2015).

The two moral standpoints, of personal autonomy and responsibility on the one hand and informed consent and protection from harm on the other, align themselves to the two opposing models of health care, personalised and collective medicine respectively, while both illustrate different aspects of responsibility. The data show how users' and clinicians' views, being linked to personalised health care in the case of the users and collective health care in the case of the clinicians, may thus seem diametrically opposed in their interpretation of the relevance of genomics to the NHS. The shared views about pharmacogenomics, rather than being a basis for

resolution of the debates about DTCGT, are instead of concern. This hope for the future promise of genomics on the part of some clinicians and many users indicates a lack of understanding of the capabilities of pharmacogenomics, which has so far been unable either to show better routes to effective prescribing of many common drugs (including anticoagulant therapy¹⁴) or to be translated effectively into the clinic (Voorra 2011).

Participants' interpretative flexibility and the tensions and agreements between users and clinicians in relation to the three main themes emerging from the data demonstrate the on-going and complex nature of the debate about personalised medicine, particularly in the UK's context of collective health provision in the NHS. This is further complicated by the apparent lack of categorical alignments of this study's users and clinicians into groups that support or oppose personal genomics or behave in particular ways depending on their experience of it. However, from the indeterminate positions of the participants' views, empirical evidence has emerged related to previous theoretical work on biosociality. I shall discuss this in the following section before concluding with suggestions for future work on the basis of this study and the ensuing discussion.

Biosociality

The debates around personalised genomics have not only so far failed to resolve but now appear more opaque than originally envisaged at the start of this study. Whilst it is neither possible nor necessarily appropriate to resolve that opacity here, it warrants discussion in relation to the implications of the findings for current theory.

The central tension between personal and collective medicine revealed in this study is a focal point for exploring the relationship between genomics and biosociality. The concept of biosociality in this study is in contrast to Rabinow's original use of the term that he suggested related to groups formed by those with common genetic anomalies after testing (Rabinow

¹⁴The drug most participants referred to in discussing pharmacogenomics was Warfarin, a commonly prescribed but potentially dangerous anticoagulant. See Common ground in Chapter Six for detail.

1996). However, my study demonstrates the importance of sociality in genomics, whether pre- or post-testing, supporting Gibbon's and Novas's suggestion that biosociality is useful for thinking about emergent identity practices associated with changing understanding of disease and diagnosis (Gibbon and Novas 2008). Following on from Rabinow's work on biosociality, Nikolas Rose has suggested that people's social groupings and the formation of new associations and communities around genetic conditions are part of the identity work that results from their genetic testing¹⁵ (Rose 2007). Subsequent to developing these social groups and networks, sustained as they are by the expectations they circulate (Brown 2005), members develop their individual knowledge and build on this to develop a level of expertise, in this case about genetics and, for users, genotyping technologies and commercial testing. Indeed this could be equated to Brekke and Sirnes examination of biosociality in which users are seen as actively engaged in using and analysing biomedical developments (Brekke and Sirnes 2011:349). For both users and clinicians this knowledge, or what Jenny Reardon refers to as "learnedness", appears to emerge as a result of their membership of networks (whether recreational, occupational or professional) and informs their identity, both individual and social (Reardon 2011:97). Importantly in this study, networks also inform participants' decisions either to test or to be wary of commercial testing, although the clinicians were not unified in condemning DTCGT.

The problem of the clash between individual and collective medicine highlighted by the data is bound up with tensions. Rose's work on biomedical politics and genomics points to individual responsibility, showing how people are encouraged to optimise their future health by undertaking self-management of their somatic existence. This carries an ethical dimension by placing value on bodily health, which could be seen to influence people's decision to engage with genomics for susceptibility testing to avoid future disease. Furthermore, public health initiatives seek to provide biological education of citizens in order for them to take responsibility for and improve their health (Hood and Friend 2011). On this

¹⁵ It is important to note that Rose sets this in the context of individuals having complex fields of identity practices of which their genetic identity is part, rather than individual identity being a singular notion.

basis it is then arguably unsurprising that, when an opportunity to engage with health information at a molecular genomic level is available, interested people avail themselves of it. In this context, biological citizens can be conflated with highly discriminating consumers, who are often seen as a powerful resource for informing and driving others' decisions through shared networks, including the Internet. Consumers of DTCGT are thus likely to consider their test results in the context of the health and illness discourse that they have been exposed to and urged to engage with as responsible citizens, particularly as this discourse is reinforced by the rhetorical style of personal genomics companies' information (Rose 2007). Motivations for testing by the users in this study were not intrinsically related to the concerns for their future health¹⁶. However, having tested, they did appear to engage with the health-related test results in a manner informed by both the commercial genomics market's rhetoric of personalisation and public health discourses around responsibility for health.

In light of this it is understandable that people might feel that it would be appropriate to share this information with healthcare professionals they consult with, usually GPs. Indeed, Rose suggests that, in developing a molecular genetic aspect to their identity and behaving as responsible citizens in respect of their health, people are able to become more active participants in their health and can work in alliance with, rather than as subservient to, healthcare professionals (Rose 2007). Discussing the incorporation of genomics into identity practices, O'Riordan similarly suggests that medical issues (as exemplified by genomics) are no longer confined to the boundaries of professional medicine (O'Riordan 2013). It is therefore predictable that the users' reported experiences of either dismissal or disinterest (or in one case referral of their concern to a hospital consultant) seemed to cause or reinforce feelings of frustration and dissonance between the public and professional interest in genomics. It is possibly emblematic of the inequalities that Plows and Boddington

¹⁶ Three users were motivated to test by current health problems. Two other users tested with family health problems in mind. Barbara was interested in her family history of cancer, which had informed her initial engagement with genealogy that led into genetic genealogy, but her motivation for testing was not solely related to her concerns about developing cancer in the future. Christine's parents were both affected by cancer but she also had a wider interest in testing informed by her work as a public health scientist.

argue persist, but are underplayed by Rose and Novas' work on biocitizenship, here in respect of professional and institutional power over individual citizens or patients (Plows and Boddington 2006). This would explain users' calls for the NHS to adopt a more contemporary, collaborative approach to health care generally and to the inclusion of genomics in particular.

The relationship of biosociality to either engagement with emerging genomic technologies or moral boundary work associated with distancing them is clearly important, as shown by this study and its substantiation of earlier theoretical work in this area. Having developed learnedness through biosociality, claims are made for what might be termed expertise about personal genomics by both early adopters and healthcare professionals, but this aspect of the findings is complex and thus difficult to evaluate satisfactorily here.

Expectations and future directions

From the data collected and my analysis of them, I do not think that rhetorical closure of the debate around DTCGT has been, or will be, achieved (Pinch and Bijker 1987). Despite regulatory changes such as the FDA's letter to 23andMe and regulatory suggestions from the joint European Academies of Science Advisory Council and the Federation of European Academies of Medicine report, companies are working round these challenges to renew their commercial ventures; personal genomics appears to present a moving target rather than a point for resolution of the technology (EASAC FEAM 2012, Brice 2013, Cohn and Surofchy 2014, Prainsack 2014b).

What is more likely is that the expectations of WGS being circulated and invested in at government level in the UK and the USA will shift both public attention and the definition of the problem on which the disputes about genomics are focused. Both of these large projects have, like the HGP and subsequent commercial DTCGT, been initially funded by public investment on the basis of their promissory potential for personalised diagnosis and treatment of disease in the future, with private commercial investment being incorporated for their future sustainability (Genomics

England n.d., Brice 2015). The irony of the use of public money to fund projects that are then capitalised on for profit by private commerce in true neoliberal fashion is notable. But funding issues aside, the debates about genomic technology such as the accuracy of sequencing data, disclosure of incidental findings, who gets WGS, how their data are used and by whom, are likely to distract from and collapse the current disputes around DTCGT (Caulfield et al 2008, Allyse and Michie 2013).

Genetic testing in general and consumer genomics, in particular, continues to provide cases for study that are troubling and complex. I have shown through the data chapters and the discussion in this chapter that the concept of biosociality is fundamental in developing an understanding of people's engagement with emerging genomic technologies and of the disputes that result, particularly in the UK context. This indicates the potential for further work in this area from two perspectives. First, it will be important to examine people's relationships to genomic technology as it develops and is more widely adopted. Second, there is a need for further theory development in respect of biosociality in genomics, but particularly in the concept of citizenship and its relationship to people's engagement with genomics.

Bibliography

23andMe (2012a) *23andMe's Parkinson's Research Featured on NPR*. 23andMe Research. [Online] <http://blog.23andme.com/23andme-research/23andmes-parkinsons-research-featured-on-npr/> accessed 15.i.2013

23andMe (2012b) *23andMe Raises More than \$50 Million in New Financing*. 23andMe Press Release [Online] <http://mediacenter.23andme.com/press-releases/23andme-raises-more-than-50-million-in-new-financing/> accessed 14.i.2013

23andMe (2013) *23andWe*. 23andMe research webpage [Online] <https://www.23andme.com/research/> accessed 3.ii.2013

23andMe (2015) *23andMe's Personal Genome Service is Now Available for Purchase at Superdrug*. 23andMeMedia [Online] <http://mediacenter.23andme.com/en-eu/blog/2015/03/30/superdrug/> accessed 4.v.2015

ACMG (2008) *Policy on Direct-to-Consumer Genetic Testing*. American College of Medical Genetics, Bethesda MD. [Online] <http://www.acmg.net/AM/Template.cfm?Section=Search2&Template=/Search/SearchDisplay.cfm> accessed 15.ii.2010

Allyse M, Michie M (2013) "Not-so-incidental findings;" the ACMG recommendations on the reporting of incidental findings in clinical whole genome and whole exome sequencing. *Trends in Biotechnology* 31(8): 439-441

Annes JP, Giovanni MA, Murray MF (2011) Risks of pre-symptomatic direct-to-consumer genetic testing. *New England Journal of Medicine* 363(120): 1100-1101

Armstrong D (1995) The rise of surveillance medicine. *Sociology of Health and Illness* 17(3): 393-404

Armstrong N, Eborall H (2012) The sociology of medical screening: past, present and future. *Sociology of Health and Illness* 34(2): 161-176

Armstrong D, Michie S, Marteau T (1998) Revealed identity: a study of the process of genetic counselling. *Social Science and Medicine* 47(11): 1653-1658

Arribas-Ayllon M (2011). The ethics of disclosing genetic diagnosis for Alzheimer's disease: do we need a new paradigm? *British Medical Bulletin* 100(1): 7-21

Arribas-Ayllon M, Bartlett A, Featherstone K (2010) Complexity and accountability: the witches brew of psychiatric genetics. *Social Studies of Science*. 40(4): 499-524

Arribas-Ayllon M, Sarangi S, Clarke A (2011a) Promissory accounts of personalisation in the commercialisation of genomic knowledge. *Communication and Medicine* 8(1): 53-66

- Arribas-Ayllon M, Sarangi S, Clarke A (2011b) *Genetic Testing: Accounts of Autonomy, Responsibility and Blame*. Routledge, Abingdon
- Ashmore M (1989) *The Reflexive Thesis: Wrighting Sociology of Scientific Knowledge*. University of Chicago Press, London
- Atkinson P (1995) *Medical Talk and Medical Work: The Liturgy of the Clinic*. Sage, London
- Atkinson P, Coffey A (2002) Revisiting the relationship between participant observation and interviewing. In Gubrium JA, Holstein JF (Eds) *Handbook of Interview Research: Context and Method*. Sage, Thousand Oaks, CA.
- Atkinson P, Delamont S (2006) Rescuing narrative from qualitative research. *Narrative Inquiry* 16(1): 164-172
- Atkinson R, Flint J (2001) Accessing hidden and hard-to-reach populations: snowball research strategies. *Social Research Update* 33: [Online] <http://sru.soc.surrey.ac.uk/SRU33.html> accessed 28.x.2014
- Atkinson P, Glasner P (2007) New genetic identities. IN Atkinson P, Glasner P, Greenslade H (Eds) *New Genetics, New Identities*. Routledge, Abingdon
- Barnes B (1982) The science and technology relationship: a model and a query. *Social Studies of Science* 12(1): 166-172
- Bartley M (1990) Do we need a strong programme in medical sociology? *Sociology of Health and Illness* 12(4): 371-390
- BBC (2000) *Scientists Crack Human Code*. BBC News Science/Nature [Online] <http://news.bbc.co.uk/1/hi/sci/tech/805803.stm> accessed 10.i.2013
- BBC (2005) *US Chat Show Host Could be a Zulu*. BBC News [Online] <http://news.bbc.co.uk/1/hi/world/africa/4096706.stm> accessed 15.i.2013
- Bhatanacharoen P, Greatbatch D, Clark T (2004) *The Tipping Point of the "Tipping Point" Metaphor: Agency and Process for Waves of Change*. Durham University Business School. [Online] <http://www.dur.ac.uk/resources/ihrr/tippingpoints/OLKCPaperforIHRR.pdf> accessed 7.ix.2013
- Bijker WE (1997) *Of Bicycles, Bakelites and Bulbs: Toward a Theory of Social Change*. Massachusetts Institute of Technology Press, Cambridge, MA.
- Bijker WE (2010) How is technology made? That is the question! *Cambridge Journal of Economics* 34: 63-76
- Bijker WE, Hughes TP, Pinch TJ (1987) *The Social Construction of Technological Systems: New Directions in the Sociology and History of Technology*. Massachusetts Institute of Technology Press, Cambridge MA.
- Bloor D (1991) *Knowledge and Social Imagery* (2nd Edn). University of Chicago Press, Chicago

- Bloss CS, Schork N, Topol EJ (2011) Effect of direct-to-consumer genome wide profiling to assess disease risk. *New England Journal of Medicine* 364(6): 524-534
- Boddington P (2009) The ethics and regulation of direct-to-consumer genetic testing. *GenomeMedicine* 1(71): 1-2
- Borry P (2008) Europe to ban direct-to-consumer genetic tests? *Nature Biotechnology* 26(7): 736-737
- Borry P, Cornel MC, Howard HC (2010) Where are you going, where have you been? A recent history of the direct-to-consumer genetic testing market. *Journal of Community Genetics* 1(3): 101-106 [Online] <http://www.springerlink.com/content/1uk1w43425251412/> accessed 15.vi.2012
- Borry P, Howard HC, Senecal K, Avard D (2009) Direct-to-consumer genome scanning services: also for children? *Nature Review Genetics* 10(1): 8
- Borup M, Brown N, Konrad K, Van Lente H (2006) The sociology of expectations in science and technology. *Technology Analysis and Strategic Management* 18(3/4): 285-298
- Bosk CL (1992) *All God's Mistakes: Genetic Counseling in a Pediatric Hospital*. Chicago University Press, Chicago
- Bourdieu P, Wacquant LJD (1992) *An Invitation to Reflexive Sociology*. University of Chicago Press, Chicago
- Brekke OA, Sirnes T (2011) Biosociality, biocitizenship and the new regime of hope and despair: interpreting "Portraits of Hope" and the "Mehmet Case". *New Genetics and Society* 30(4): 347-374
- Brett GR, Metcalfe SA, Amor DJ, Halliday JL (2012) An exploration of genetic health professionals' experience with direct-to-consumer genetic testing in their clinical practice. *European Journal of Human Genetics* 20: 825-830
- Brice P (2013) FDA warns 23andMe to cease DTC genetic testing. *PHG Foundation* [Online] <http://www.phgfoundation.org/news/15045/> accessed 10.xii.2013
- Brice P (2014) 23andMe engages with FDA on health-related genetic tests. *PHG Foundation* [Online] <http://www.phgfoundation.org/blog/16172/> accessed 23.vi.2014
- Brice P (2015) Obama's precision medicine Initiative: the future of healthcare? *PHG Foundation*. <http://www.phgfoundation.org/blog/16551/> accessed 20.ii.2015
- Brown N (2003) Hope against hype – accountability in biopasts, presents and futures. *Science Studies* 16(2): 3-21
- Brown N (2005) Shifting tenses: reconnecting regimes of truth and hope. *Configurations* 13(3): 331-355

- Brown N, Michael M (2003) A sociology of expectations: retrospectively prospecting and prospectively retrospectively. *Technology Analysis and Strategic Management* 15(1): 3-18
- Brown N, Rappert B, Webster A (2000) *Contested Futures: a Sociology of Prospective Technoscience*. Ashgate, Aldershot
- Brown N, Webster A. (2004) *New Medical Technologies and Society: Re-ordering Life*. Polity Press, Cambridge
- BSGM (no date) *British Society for Genetic Medicine*. [Online] <http://www.bsgm.org.uk/information-education/for-patients/what-happens-at-a-medical-genetics-appointment/> accessed 10.vi.2014
- Bunnik EM, Janssens JW, Schermer MHN (2014) Personal utility in genomic testing: is there such a thing? *Journal of Medical Ethics* 41(4): 322-326
- Bunnik EM, Schermer MHN, Janssens JW (2011) Personal genome testing: test characteristics to clarify the discourse on ethical, legal and societal issues. *BiomedCentral Medical Ethics* 12(11): [Online] <http://www.biomedcentral.com/1472-6939/12/11> accessed 13.v.2012
- Burton A (2015) Are we ready for direct-to-consumer genetic testing? *The Lancet* 14:138-139 [Online] <http://www.thelancet.com/pdfs/journals/laneur/PIIS1474-4422%2815%2970003-7.pdf> accessed 13.i.2015
- Bury M (1997) *Health and Illness in a Changing Society*. Routledge, Abingdon
- Callon M (1986) Some elements of a sociology of translation: domestication of the scallops and fisherman of St Brieuc Bay. IN Law J (Ed) *Power, Action and Belief: A New Sociology of Knowledge*. Routledge, London
- Caplan A (2007) *Women Should be Wary of Genetic Risks Ads*. Breaking Bioethics on NBC NEWS.com [Online] http://www.msnbc.msn.com/id/20761643/ns/health-health_care/t/women-should-be-wary-genetic-risk-ads/#.UPQzNrbf1ck accessed 14.i.2013
- Caulfield T, Ries NM, Ray PN, Shuman C, Wilson B (2009) Direct-to-consumer genetic testing: good, bad or benign? *Clinical Genetics* 77: 101-105
- Chadwick R (1999) Genetics, choice and responsibility. *Health, Risk and Society* 1(3): 293-300
- Chapman M (2010) *Report on the Direct-to-Consumer Genetic Testing Industry*. Cesagen ESRC, Cardiff
- Charmaz K (2000) Grounded theory: objectivist and constructivist methods. IN Denzin NK, Lincoln YS (Eds) *Handbook of Qualitative Research (2nd Edn)*. Sage, Thousand Oaks CA.
- Cherkas L, Harris J, Levinson E, Spector T, Prainsack B (2010) A survey of UK public interest in internet-based personal genome testing. *PLOS One* 5(10): <http://journals.plos.org/plosone/article?id=10.1371/journal.pone.0013473> accessed 8.x.2010

Chilibeck G, Lock M, Sehdev M (2011) Postgenomics, uncertain futures and the familiarization of susceptibility genes. *Social Science and Medicine* 72: 1768-1775

Clarke AE (2003) Situational analyses: grounded theory mapping after the postmodern turn. *Symbolic Interaction* 26(4): 553-576

Coffey A, Atkinson P (1996) *Making Sense of Qualitative Data: Complementary Research Strategies*. Sage, Thousand Oaks, CA.

Cohn B, Surofchy D (2014) Regulating direct-to-consumer genetic testing vs. the right to know. *Synapse Online*
<http://synapse.ucsf.edu/articles/2014/05/22/regulating-direct-consumer-genetic-testing-vs-right-know> accessed 22.v.2014

Collins H (1981) Stages in the Empirical Programme of Relativism. *Social Studies of Science, Special Issue: Knowledge and Controversy: Studies of Modern Natural Science* 11(1): 3-10

Collins H, Evans R (2002) The third wave of science studies. *Social Studies of Science* 32(2): 235-296

Collins FS, Varmus H (2015) A new initiative on precision medicine. *The New England Journal of Medicine* [Online]
<http://www.nejm.org/doi/full/10.1056/NEJMp1500523> accessed 20.ii.2015

Collins RE, Wright AJ, Marteau TM (2011) Impact of communicating personalized risk information on perceived control over the risk: a systematic review. *Genetics in Medicine* 13(4): 273-277

Conley J (2013a) If 23andMe falls in the forest, and there's no one there... *Genomics Law Report* [Online]
<http://www.genomicslawreport.com/index.php/2013/12/03/if-23andme-falls-in-the-forest-and-theres-no-one-there/#more-13198> accessed 8.v.2014

Conley J (2013b) The revolt of the C's: class action filed against 23andMe. *Genomics Law Report* [Online]
<http://www.genomicslawreport.com/index.php/2013/12/05/the-revolt-of-the-cs-class-action-filed-against-23-and-me/> accessed 4.v.2015

Conley J, Vorhaus D, Cook-Deegan R (2011) How will Myriad respond to the next generation of BRCA testing? *The Genomics Law Report* [Online]
www.genomicslawreport.com/index.php/2011/03/01/how-will-myriad-respond-to-the-next-generation-of-brca-testing/ accessed 14.i.2013

Conrad D (2009) Welcoming GINA into the workplace. *The Genomics Law Report* [Online]
<http://www.genomicslawreport.com/index.php/2009/11/24/welcoming-gina-into-the-workplace/> accessed 7.v.2012

Conrad P, Gabe J (1999) Sociological perspectives on the new genetics: an overview. *Sociology of Health and Illness* 21(5): 505-516

Corden A, Sainsbury R (2006) *Using Verbatim Quotations in Reporting Qualitative Social Research: Researchers' Views*. University of York, York

Council of Europe (2008) *Additional Protocol on The Convention of Human Rights and Biomedicine, Concerning Genetic Testing for Health Purposes*. CoE, Strasbourg [Online] <http://conventions.coe.int/Treaty/EN/Treaties/html/203.htm> accessed 18.v.2010

Cox SM, Starzomski RC (2004) Genes and geneticization? The social construction of autosomal polycystic kidney disease. *New Genetics and Society* 23(2): 137-166

Creswell JW (2013) *Qualitative Inquiry and Research Design: Choosing among Five Approaches*. Sage, Los Angeles, CA.

Davies W (2014) A bibliographic review of neoliberalism. *Theory, Culture and Society* [Online] http://theoryculturesociety.org/william-davies-a-bibliographic-review-of-neoliberalism/?utm_content=buffer33bf3&utm_medium=social&utm_source=twitter.com&utm_campaign=buffer accessed 11.vi.2015

Davis JE (2008) Moral order. *Culture* [Online] www.iasc-culture.org/eNews/2008.../LastWord_CultureSpring08.pdf accessed 19.i.2013

Delanty G (2002) Constructivism, sociology and the new genetics. *New Genetics and Society* 21(3): 279-289

Dickenson D (2013) *Me Medicine vs. We Medicine: Reclaiming Biotechnology for The Common Good*. Columbia University Press, New York

Dickson D (2005) The case for a 'deficit model' of science communication. *SciDev.Net* [Online] <http://www.scidev.net/global/communication/editorials/the-case-for-a-deficit-model-of-science-communic.html> accessed 20.v.2015

Dimond R, Bartlett A, Lewis J (2015) What binds biosociality? The collective effervescence of the parent conference. *Social Science and Medicine* 12(6): 1-8

DoH (1991a) *The Patient's Charter*. Department of Health, London, HMSO

DoH (1991b) *The Health of the Nation*. Department of Health, London, HMSO

DoH (2010) *Equity and Excellence: Liberating the NHS*. Department of Health, London [Online] https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/213823/dh_117794.pdf accessed 26.v.2014

Dreyer M, Erdmann J, Rehmann-Sutter C (forthcoming) *Genetic Transparency? Ethical and Social Implications of Next Generation Human Genomics and Genetic Medicine*, Brill Rodopi, Amsterdam.

Durant J, Hansen A, Bauer M (1996) Public understanding of the new genetics. IN Marteau T, Richards M (Eds) *The Troubled Helix: Social and Psychological*

Implications of the New Human Genetics. Cambridge University Press, Cambridge

EASAC, FEAM (2012) *Direct-to-Consumer Genetic Testing*. European Academies of Science Advisory Council, Federation of European Academies of Medicine. [Online]

http://www.easac.eu/fileadmin/PDF_s/reports_statements/Easac_12_DTCGT-Lay_Web.pdf accessed 29.i.2013

Editorial NG (2002) What's brewing in genetic testing? *Nature Genetics* 32(4): 553-554

Edleman E, Eng C (2009) A practical guide to interpretation and clinical application of personal genomic screening. *British Medical Journal* 339 (1 November 2009): 1136-1140

Elston MA (2002) The politics of professional power IN Gabe J, Calnan M, Bury M (Eds) *The Sociology of the Health Service*. Routledge, London

Epstein S (1995) The construction of lay expertise: AIDS activism and the forging of credibility in the reform of clinical trials. *Science, Technology and Human Values* 20(4): 408-437

etc Group (2008) *Direct-to-Consumer DNA Testing and The Myth of Personalised Medicine: Spit Kits, SNP Chips and Human Genomics. Special Report on Human Genomics I. Erosion, Technology and Concentration Group* (etc Group) [Online] <http://www.etcgroup.org/node/675> accessed 20.vi.2012

Evans JP, Dale DC, Fomous C (2011) Preparing for a consumer-driven age. *New England Journal of Medicine* 363(12): 1099-1103

Evans JP, Green RC (2009) Direct-to-consumer genetic testing: avoiding a culture war. *Genetics in Medicine* 11(8): 568-569

Evans R, Plows A (2007) Listening without prejudice? Rediscovering the value of the disinterested citizen. *Social Studies of Science* 37(6): 827-853

Evans R, Plows A, Welsh I (2007) Towards an anatomy of public engagement with medical genetics IN Atkinson P, Glasner P, Greenslade H (Eds) *New Genetics, New Identities*. Routledge, Abingdon

Farkas DH, Holland CA (2009) Direct-to-consumer genetic testing two sides of the coin. *Journal of Molecular Diagnostics* 11(4): 263-265

Farrer LA, Cupples LA, Haines JL, Hyman B, Kukull WA, Mayeaux RH, Myers MA, Pericak-Vance MA, Risch N, Van Duijn CM (1997) Effects of age, sex and ethnicity on the association between lipoprotein E genotype and Alzheimer's disease: a meta-analysis. *Journal of the American Medical Association* 278: 1349-1356

FDA (2010) *Medical Devices: Letters to Industry*. Food and Drug Administration. [Online]

<http://www.fda.gov/MedicalDevices/ResourcesforYou/Industry/ucm111104.htm> accessed 15.vi.2012

- Featherstone K, Atkinson P (2012) *Creating Conditions*. Routledge, London
- Ferris KO (2007) The sociology of celebrity. *Sociology Compass* 1(1): 371-384
- Foster MW, Sharp RR (2008) Ethics watch: out of sequence: how consumer genomics could displace clinical genetics. *Nature Reviews Genetics* 9(6): 419
- Foucault M (1994) *The Birth of the Clinic: An Archaeology of Medical Perception*. Vintage Books, New York
- Fox Keller E (1992) Nature, nurture and the Human Genome Project. IN Kevles DJ, Hood L (Eds) *The Code of Codes: Scientific and Social Issues in the Human Genome Project*. Harvard University Press, Cambridge, MA.
- Fraker M, Mazza AM (2010) Direct-to-consumer genetic testing: summary of a workshop. *National Academies Press Online*. [Online] http://www.nap.edu/openbook.php?record_id=13021&page=R1 accessed 21.vi.2012
- Franke U (2010) On the bumpy road towards personalized medicine. *EMBO Molecular Medicine* 2: 1-2
- Freidson E (1988) *Profession of Medicine: A Study of the Sociology of Applied Knowledge*. Chicago University Press, Chicago
- Garfinkel H (1967) *Studies in Ethnomethodology*. Prentice Hall, Englewood Cliffs, NJ.
- GeneticHealth (2005) *GeneticHealth Website* [Online] <http://www.genetic-health.co.uk/index.asp> accessed 2.iv.2015
- GeneWatch UK (2002) *The Body Shop and Sciona*. GeneWatch UK [Online] <http://www.genewatch.org/sub-425647> accessed 15.i.2013
- GeneWatch UK (2004) *Genetic Tests and Health*. GeneWatch UK [Online] [http://www.genewatch.org/pub.shtml?als\[cid\]=492775](http://www.genewatch.org/pub.shtml?als[cid]=492775) accessed 15.vi.2012
- GeneWatch UK (2015) *Shoppers Warned not to Buy Gene Tests from Superdrug*. GeneWatch UK [Online] [http://www.genewatch.org/article.shtml?als\[cid\]=492860&als\[itemid\]=575612](http://www.genewatch.org/article.shtml?als[cid]=492860&als[itemid]=575612) accessed 4.v.2015
- Genomics England (n.d.) *Genomics England*, Department of Health. [Online] <http://www.genomicsengland.co.uk/> accessed 20.ii.2015
- Gibbon S, Novas C (2008) *Biosocialities, Genetics and the Social Sciences: Making Biologies and Identities*. Routledge, London
- Gibbs S (2014) DNA-screening Test 23andMe launches in UK after US ban. *The Guardian*, 2nd December 2014. [Online] <http://www.theguardian.com/technology/2014/dec/02/google-genetic-testing-23andme-uk-launch> accessed 2.xii.2014

- Gieryn TF (1983) Boundary work and the demarcation of science from non-science: strains and interests in professional ideologies of scientists. *American Sociological Review* 48 (December): 781-795
- Giovanni MA, Fickie MR, Lehmann LS, Green RS, Meckley LM, Veenstra D Murray MF (2010) Health-care referrals from direct-to-consumer genetic testing. *Genetic Testing and Molecular Biomarkers* 14(6): 817-819
- Gladwell M (2000) *The Tipping Point: How Little Things Can Make a Big Difference*. Abacus, London
- Glaser BG (1999) The future of grounded theory. *Qualitative Health Research* 9(6): 836-845
- Goffman E (1989) On fieldwork. *Journal of Contemporary Ethnography* 18(2): 123-132
- Goldsmith L, Jackson L, O'Connor A, Skirton H (2012) Direct-to-consumer genomic testing: systematic review of the literature on user perspectives. *European Journal of Human Genetics* 20(8): 811-816
- Gollust SE, Chandros Hull SC, Wilfond BS (2002) Limitations of direct-to-consumer advertising for clinical genetic testing. *Journal of the American Medical Association*, 228(14); 1762-1767
- Gordon ES, Griffin G, Wawak HP, Gollust SE, Bernhardt BA (2012) "It's not like judgment day": public understanding of and reactions to personalized genomic risk information. *Journal of Genetic Counseling* 12(3): 423-432
- Gray SW, O'Grady C, Karp L, Smith D, Sanford Schwartz J, Hornick RC, Armstrong K (2009) Risk information exposure and direct-to-consumer genetic testing for *BRCA* mutations among women with a personal or family history of breast or ovarian cancer. *Cancer Epidemiology, Biomarkers and Prevention* 18(3): 1303
- Green K (1991) Shaping technologies and shaping markets: creating demand for biotechnology. *Technology Analysis & Strategic Management* 3(1): 57-76
- Grodzins M (1957) Metropolitan segregation. *Scientific American* 197(4): 33-41
- Groves C, Tutton R (2013) Walking the tightrope: expectations and standards in personal genomics. *BioSocieties* 8(2): 181-204
- Gurwitz D, Bregman-Eschet Y (2009) Personal genomics services: whose genomes? *European Journal of Human Genetics* 17(7): 883-889
- Habermas J (1970) *Towards a Rational Society: Student Protest, Science and Politics*. Beacon, Boston, MA.
- Hacking I (1999) *The Social Construction of What?* Harvard University Press, Cambridge, MA.
- Hallowell N (1999) Doing the right thing: genetic risk and responsibility. *Sociology of Health and Illness* 21(5): 597-621

Hallowell N, Ardern-Jones A, Eeles R, Foster C, Lucassen A, Moynihan C, Watson M (2006) Guilt, blame and responsibility: men's understanding of their role in the transmission of *BRCA1/2* mutations within their family. *Sociology of Health and Illness* 28(7): 969-988

Hallowell N, Foster C, Eeles R, Ardern-Jones A, Murray V, Watson M (2003) Balancing autonomy and responsibility: the ethics of generating and disclosing genetic information. *Journal of Medical Ethics* 29: 74-79

Hallowell N, Foster C, Eeles R, Ardern-Jones A, Watson M (2004) Accommodating risk: responses to *BRCA1/2* genetic testing of women who have had cancer. *Social Science and Medicine* 59: 553-565

Hamilton DP (2008) 23andMe's price cut: the end of commercial personal genomics? *BNET Industries* [Online] <http://www.cbsnews.com/news/23andmes-price-cut-the-end-of-personal-genomics/> accessed 18.iii.2012

Hammersley M, Atkinson P (1995) *Ethnography: Principles in Practice* (2nd Edn). Routledge, Abingdon

Harris A, Wyatt S, Kelly SE (2013) The gift of spit (and the obligation to return it): how consumers of online genetic testing services participate in research. *Information, Communication and Society* 16(2): 236-257

Harris A, Wyatt S, Kelly SE (2014) Autobiologies on YouTube: narratives of direct-to-consumer genetics testing. *New Genetics and Society* 33(1): 60-78

Harvey A (2011) From lab to lifestyle: translating genomics into healthcare practices. *New Genetics and Society* 30(4): 309-327

Hauskeller C (2011) Direct-to-consumer genetic testing: regulations cannot guarantee responsible use; an international industry certificate is needed. *British Medical Journal* 342: 1163-1164

Hedgecoe AM (2003) Terminology and the construction of scientific disciplines: the case of pharmacogenomics. *Science, Technology and Human Values* 28: 513-537

Hedgecoe AM (2004) *The Politics of Personalised Medicine: Pharmacogenetics in the Clinic*. Cambridge University Press, Cambridge

Hedgecoe A (2008) From resistance to usefulness: sociology and the clinical use of genetic tests. *BioSocieties* 3: 183-194

Hedgecoe AM (2012) Trust and regulatory organisations: the role of local knowledge and facework in research ethics review. *Social Studies of Science* 42(5): 662-683

Hedgecoe AM and Martin PA (2003) The drugs don't work: expectations and the shaping of pharmacogenetics. *Social Studies of Science* 33(3): 327-364

Henrikson NB, Bowen D, Burke W (2009) Does genomic risk information motivate people to change their behavior? *Genome Medicine* 1(4): 37.1-37.3 [Online] <http://genomemedicine.com/content/1/4/37> accessed 4.v.2010

- Hennen L, Sauter A, Van den Cruyce E (2008) *Direct-to-Consumer Genetic Testing Study*. European Parliament, Brussels [Online]
http://www.europarl.europa.eu/stoa/default_en.htm accessed 5.iii.2010
- Herper M (2012) *With deCODE Deal Amgen Aims to Discover Drugs Like We Meant to in 1999*. Forbes Business [Online]
<http://www.genomicslawreport.com/index.php/2012/12/10/implications-of-amgendecode-deal-for-genetic-testing-consumers/> accessed 14.i.2013
- Herper M (2015) *Surprise! With \$60 Million Genentech Deal, 23andMe Has a Business Plan*. Forbes, Pharma and Healthcare. [Online]
<http://www.forbes.com/sites/matthewherper/2015/01/06/surprise-with-60-million-genentech-deal-23andme-has-a-business-plan/> accessed 6.i.2015
- Heschka JT, Palleschi C, Howley H, Wilson B, Wells PS (2008) A systematic review of perceived risks, psychological and behavioural impacts of genetic testing. *Genetics in Medicine* 10(1): 19-32
- HGC (2003) *Genes Direct: Ensuring the Effective Oversight of Genetic Tests Supplied Directly to the Public*. Human Genetics Commission, London. [Online]
<http://www.hgc.gov.uk/Client/document.asp?DocId=34&CAtegorYId=10>
 accessed 25.ii.2010
- HGC (2007) *More Genes Direct: A Report on Developments in the Availability, Marketing and Regulation of Genetics Tests Supplied Directly to the Public*. Human Genetics Commission, London. [Online]
<http://www.hgc.gov.uk/Client/document.asp?DocId=139&CAtegorYId=10>
 accessed 25.ii.2010
- HGC (2010) *The Common Framework of Principles for Direct-to-Consumer Genetic Testing Services: Principles and Consultation Questions*. Human Genetics Commission, London. [Online]
<http://www.hgc.gov.uk/Client/Content.asp?ContentId=816> accessed 25.ii.2010
- Hildenbrand W (2014) *Market Demand: Theory and Empirical Evidence*. Princeton University Press, Princeton, NJ.
- Hogarth S, Javitt G, Melzer D (2008) The current landscape for direct-to-consumer genetic testing: legal ethical and policy issues. *The Annual Review of Genomics and Human Genetics* 9: 161-182
- Holstein JA, Gubrium JF (2004) The active interview IN Silverman D (Ed) *Qualitative Research: Theory, Method and Practice (2nd Edn)*. Sage, London
- Holtzman NA (1999) Promoting safe and effective genetic tests in the United States: work of the task force on genetic testing. *Clinical Chemistry* 45(5): 732-738
- Hood L, Friend SH (2011) Predictive, personalized, preventive, participatory (P4) cancer medicine. *Nature Reviews: Clinical Oncology* 8(3): 184–187
- House of Lords (2009) *Genomic Medicine, Science and Technology Committee, 2nd Report of Session 2008-2009. Volume 1*

Howard HC, Borry P (2013) Survey of European clinical geneticists on awareness, experiences and attitudes towards direct-to-consumer genetic testing. *Genome Medicine* 5(5): 45 [Online]

<http://genomemedicine.com/content/pdf/gm449.pdf> accessed 10.v.2015

HRA (n.d.) *National Research Ethics Service*, Health Research Authority, NHS [Online] <http://www.hra.nhs.uk/about-the-hra/our-committees/nres/> accessed 21.x.2014

Huang AJ (2000) The rise of direct-to-consumer advertising of prescription drugs in the United States. *Journal of the American Medical Association* 284(17): 2240

Hull SC, Prasad K (2001) Reading between the lines: direct-to-consumer advertising of genetic testing. *Hastings Center Report* 31(3): 33-35

International Biosciences (2015) *Genetic Predisposition DNA Testing*. [Online] <http://www.ibdna.com/regions/UK/EN/?page=genetic-predisposition-dna-testing> accessed 3.v.2015

International Human Genome Sequencing Consortium (IHGSC) (2004) Finishing the euchromatic sequence of the human genome. *Nature* 431:931-945. [Online]

<http://www.nature.com/nature/journal/v431/n7011/full/nature03001.html>
accessed 20.vi.2012

James KM, Cowl CT, Tiburt JC, Sinicrope PS, Robinson ME, Frimansdottir KR, Tiedje K, Koenig BA (2011) Impact of direct-to-consumer genomic testing on risk perception and worry on patients receiving routine care in a preventive health clinic. *Mayo Clinic Proceedings* 86(10): 933-940 [Online]

<http://www.mayoclinicproceedings.org/article/S0025-6196%2811%2965234-8/abstract> accessed 13.i.2013

Janssens C (2015) *FDA Approves 23andMe's DNA Test, but not The One It Halted Earlier*. Huffington Post, 23rd February 2015 [Online]

http://www.huffingtonpost.com/cecile-janssens/23andme_b_6723328.html
accessed 17.iii.2015

Janssens ACJW, Gwinn M, Bradley LA, Oostra BA, van Duijn CM, Khoury MJ (2008) A critical appraisal of the scientific basis of commercial genome profiles used to assess health risks and personalize health interventions. *American Journal of Human Genetics* 82: 593-599

Jerolmack C, Khan S (2014) Talk is cheap: ethnography and the attitudinal fallacy. *Social Methods and Research* 43(2): 178-209

Jeske D (1996) Perfection, happiness and duties to self. *American Philosophical Quarterly* 33(3): 263-276

Jordens CFC, Kerridge IH, Samuel GN (2009) Direct-to-consumer personal genome testing: the problem is not ignorance – it is market failure. *American Journal of Bioethics* 9(6-7): 13-15

Jostins L, MacArthur D (2011) DTC genetic testing and the FDA: is there an end insight to regulatory uncertainty? *genomesunzipped*. [Online] <http://www.genomesunzipped.org/category/law-and-regulation> accessed 25.vi.2012

Juengst ET, Flatt MA, Settersten RA (2012) Personalized genomic medicine and the rhetoric of empowerment. *Hastings Centre Report* 42(5): 34-40

Kalf RR, Mihaescu R, Kundu S, de Knijff P, Green RC, Janssens AC (2013) Variations in predicted risks in personal genome testing for common complex diseases. *Genetics in Medicine* [Online] http://www.mdffoundation.com.au/resources/MDResearchNews/july_2013/Issue_137_MD_Research_News%2017_13.pdf accessed 9.v.2015

Kaphingst KA, McBride CM, Wade C, Alford SH, Reid R, Larson E, Baxeavanis AD, Brody LC (2012) Patients' understanding of and responses to multiplex genetic susceptibility test results. *Genetics in Medicine* 14: 681-687

Katsanis SH, Javitt G, Hudson K (2008) Public health. A case study of personalized medicine. *Science* 320: 53-54

Kaufman DJ, Bollinger JM, Dvoskin RL, Scott JA (2012) Risky business: risk perception and the use of medical services among customers of DTC personal genetic testing. *Journal of Genetic Counseling* 21(3): 413-422

Kaye J (2008) The regulation of direct-to-consumer genetic tests. *Human Molecular Genetics* 17(R2): R180-183

Kaye J, Boddington P, de Vries J, Hawkins N, Melham K (2009) Ethical implications of use of whole genome methods in medical research. *European Journal of Human Genetics*. 18: 398-403

Keeling D, Baglin T, Tait C, Watson H, Perry D, Baglin C, Kitchen S, Makris M (2011) Guidelines on oral anticoagulation with warfarin – fourth edition. *British Journal of Haematology* [Online] http://www.bcsghguidelines.com/documents/warfarin_4th_ed.pdf accessed 19.iii.2015

Kelly SE (2007) From 'scraps and fragments' to 'whole organisms': molecular biology, clinical research and post-genomic bodies. IN Atkinson P, Glasner P, Greenslade H (Eds) *New Genetics, New Identities*. Routledge, Abingdon

Kerr A (2004) *Genetics and Society: A Sociology of Disease*. Routledge, London

Kerr A, Cunningham-Burley S, Amos A (1997) The new genetics: professionals' discursive boundaries. *The Sociological Review* 45(2): 279-303

Kerr A, Cunningham-Burley S, Amos A (1998) The new genetics and health: mobilizing lay expertise. *Public Understanding of Science* 7: 41-60

Khoury MJ, McBride CM, Shully SD, Ioannidis JPA, Feero WG, Janssens ACJW, Gwinn M, Simons-Morton DG, Bernhardt JM, Cargill M, Chanock SJ, Church GM, Coates RJ, Collins FS, Croyle RT, Davis BR, Downing GJ, DuRoss A, Friedman S, Gail MH, Ginsburg GS, Green RC, Greene MH,

Greenland P, Gulcher JR, Hsu A, Hudson KL, Kardia SLR, Kimmel PL, Lauer MS, Miller AM, Offit K, Ransohoff DF, Roberts JS, Rasooly RS, Stefansson K, Terry SF, Teutsch SM, Trepanier A, Wanke KL, Witte JS, Xu J (2009) The scientific foundation for personal genomics: recommendations from a National Institutes of Health - Centers for Disease Control and Prevention multidisciplinary workshop. *Genetics in Medicine* 11(8): 559-567

Kimmel SE, French B, Kasner SE, Johnson JA, Anderson JL, Gage BF, Rosenberg YD, Eby CS, Madigan RA, McBane RD, Abdel-Rahman SZ, Stevens SM, Yale S, Mohler SM III, Fang MC, Shah V, Horenstein RB, Limdi NA, Muldowney JAS, Gujral J, Delafontaine P, Desnick RJ, Ortel TL, Billett HH, Pendleton RC, Geller NL, Halperin JL, Goldhaber SZ, Caldwell MD, Califf RM, Ellenberg JH (2013) A pharmacogenetic versus a clinical algorithm for warfarin dosing. *New England Journal of Medicine* 369(24): 2283-2293

Klein HK, Kleinman DL (2002) The social construction of technology: structural considerations. *Science, Technology and Human Values* 27(1): 28-52

Klein R (2010) *The New Politics of the NHS: from Creation to Reinvention*. Radcliffe Publishing, Abingdon

Kniffin CL (2010) *Coumarin Resistance*. Online Mendelian Inheritance in Man (OMIM) [Online]

<http://www.omim.org/entry/122700?search=warfarin&highlight=warfarin>

accessed 19.iii.2015

Kraft P, Hunter DJ (2009) Genetic risk prediction – are we there yet? *New England Journal of Medicine* 360(17): 1701-1703

Kuehn BM (2008) Risks and benefits of direct-to-consumer genetic testing remain unclear. *Journal of American Medical Association* 300(13): 1503-1505

Kutz GD (2010) *Direct-to-Consumer Genetic Tests: Misleading Test Results are Further Complicated by Deceptive Marketing and Other Questionable Practices*. United States Government Accountability Office GAO-10-847T [Online] <http://www.gao.gov/assets/130/125079.pdf> accessed 23.iv.2013

Kvale S, Brinkmann S (2009) *InterViews: Learning the Craft of Qualitative Research Interviewing (2nd Edn)*. Sage, Thousand Oaks, CA.

Latimer J (2007) Creating text, analysing text: a note on ethnography, writing and power. *Cardiff University School of Social Sciences Working Paper*. 106

Latimer J (2013) *The Gene, the Clinic and the Family: Diagnosing Dysmorphology, Reviving Medical Dominance*. Routledge, Abingdon

Latour B (2005) *Reassembling the Social: An Introduction to Actor-Network Theory*. Oxford University Press, Oxford

Leachman SA, MacArthur DG, Angrist M, Gray SW, Bradbury AR, Vorhaus DB (2011) Direct-to-consumer genetic testing: personalized medicine in evolution. *ASCO Education Book*, 34-40

- Leighton JW, Valverde K, Bernhardt BA (2012) The general public's understanding and perception of direct-to-consumer genetic test results. *Public Health Genomics* 15(1): 11-21
- Lenzer J, Brownlee S (2008) Knowing me, knowing you. *British Medical Journal* 336(7649): 858-860
- Lessa I (2006) Discursive struggles with social welfare: Restaging teen motherhood. *British Journal of Social Work* 36(2): 283-298
- Lloyd AJ (2001) The extent of patients' understanding of the risk of treatments. *Quality in Health Care* 10(Suppl 1): i14-i18
- Lock M, Freeman J, Chilibeck G, Beveridge B, Padolsky M (2007) Susceptibility genes and the question of embodied identity. *Medical Anthropology Quarterly* 21(3): 256-276
- Lynch M, Cole SA, McNally R, Jordan K (2010) *Truth Machine: The Contentious History of DNA Fingerprinting*. University of Chicago Press, Chicago
- MacKenzie D (1990) *Inventing Accuracy: A Historical Sociology of Nuclear Missile Guidance*. Massachusetts Institute of Technology Press, Cambridge, MA
- MacKenzie D, Wajcman J (1999) *The Social Shaping of Technology* (2nd Edn). Open University Press, Maidenhead
- Mardis E (2010) The \$1,000 genome, the \$100,000 analysis? *Genome Medicine* 2:84 [Online] <http://www.biomedcentral.com/content/pdf/gm205.pdf> accessed 19.ii.2015
- Marietta C, McGuire AL (2009) Direct-to-consumer genetic testing: is it the practice of medicine? *Journal of Law, Medicine and Ethics* 37(2): 369-374
- Marteau T, Bekker H (1992) The development of a six-item short-form of the state scale of the Spielberger state-trait anxiety inventory (STAI). *British Journal of Clinical Psychology* 31: 301-306
- Marteau TM, French DP, Griffin SJ, Prevost AT, Sutton S, Watkinson C, Attwood S, Hollands GJ (2010) Effects of communicating DNA-based disease risk estimates on risk-reducing behaviours. *The Cochrane Library* [Online] <http://onlinelibrary.wiley.com/doi/10.1002/14651858.CD007275.pub2/full> accessed 10.v.2015
- Marteau TM, Roberts S, LaRusse S, Green RC (2005) Predictive genetic testing for Alzheimer's disease: impact upon risk perception. *Risk Analysis* 25(2): 397-404
- Matloff E, Caplan A (2008) Direct to confusion: lessons learned from marketing BRCA testing. *American Journal of Bioethics* 8(6): 5-8
- Mavroidopoulou V, Xera E, Mollaki V (2015) Awareness, attitudes and perspectives of direct-t-consumer genetic testing in Greece: a survey of

potential consumers. *Journal of Human Genetics* [Online] doi:10.1038/jhg.2015.58 accessed 11.vi.2015

McBride CM, Alford SH, Reid RJ, Larson EB, Baxevanis AD, Brody LC (2009) Characteristics of users of online personalized genomic risk assessments: implications for physician-patient interactions. *Genetics in Medicine* 11(8): 582-587

McBride CM, Wade CH, Kaphingst KA (2010) Consumers' views of direct-to-consumer genetic information. *Annual Review of Genomics and Human Genetics* 11: 427-446

McGee MC (1980) The ideograph: a link between rhetoric and ideology. *The Quarterly Journal of Speech*. 66(February): 1-16

McGowan ML, Fishman JR (2008) Using lessons learned from BRCA testing and marketing: what lies ahead for whole genome scanning services? *American Journal of Bioethics* 8(6): 18-20

McGowan ML, Fishman JR, Lambrix MA (2010) Personal genomics and individual identities: motivations and moral imperatives of early users. *New Genetics and Society* 29(3): 261-290

McGowan ML, Fishman JR, Settersten RA, Lambrix MA, Juengst ET (2014) Gatekeepers or intermediaries: The role of Clinical commercial genomic testing. *PLOS ONE* 9(9): [Online] <http://journals.plos.org/plosone/article?id=10.1371/journal.pone.0108484> accessed 10.v.2015

McGuire AL, Burke W (2008) An unwelcome side effect of direct-to-consumer personal genetic testing: raiding the medical commons. *Journal of the American Medical Association* 300(22): 2669-2671

McGuire AL, Diaz CM, Wang T, Hilsenbeck SG (2009) Social networkers' attitudes to direct-to-consumer personal genome testing. *American Journal of Bioethics* 9(6-7): 3-10

McKinlay JB, Marceau LD (2002) The end of the golden age of doctoring. *International Journal of Health Services* 32(2): 379-416

Meek J (2002) Public misled by gene test hype. *The Guardian* 12th March 2002 [Online] <http://www.theguardian.com/uk/2002/mar/12/medicalsecience.genetics> accessed 8.v.2015

Meikle J (2015) Superdrug criticised by doctors for stocking genetic self-testing kits. *The Guardian* 31st March 2015 [Online] <http://www.theguardian.com/science/2015/mar/31/superdrug-criticised-doctors-genetic-self-testing-kits> accessed 4.v.2015

Michie S, Marteau TM, Bobrow M (1997a) Genetic counselling: the psychological impact of meeting patients' expectations. *Journal of Medical Genetics* 34: 237-241

- Michie S, Bobrow M, Marteau TM (2001) Predictive genetic testing in children and adults: a study of emotional impact. *Journal of Medical Genetics* 38: 519-526
- Michie S, Bron F, Bobrow M, Marteau TM (1997b) Non-directiveness in genetic counselling: an empirical study. *American Journal of Human Genetics* 60: 40-47
- Michie S, Weinman J, Miller J, Collins V, Halliday J, Marteau TM (2002) Predictive genetic testing: high risk expectations in the face of low risk information. *Journal of Behavioural Medicine* 25(1): 33-50
- Mokyr J (1991) Evolutionary biology, technological change and economic history. *Bulletin of Economic Research* 43(2): 127-149
- Nash C (2004) Genetic kinship. *Cultural Studies*. 18(1): 1-33
- Navigenics (2013) *Navigenics Website* [Online] <http://www.navigenics.com/> accessed 14.i.2013
- Newton G (2001) *Human Genome Project History 10: Press Views of the first draft.* The Wellcome Trust. [Online] http://genome.wellcome.ac.uk/doc_WTD022315.html accessed 20.vi.2012
- NHS (2011) *NHS Choices: Your Health, Your Choices.* [Online] <http://www.nhs.uk/Pages/HomePage.aspx> accessed 10.i.2013
- NICE (2012) *Medicines and Prescribing Support from NICE.* The National Institute for Health and Clinical Excellence. [Online] <http://www.nice.org/mpc/index.jsp> accessed 18.vi.2012
- Nicholson M, McLaughlin C (1987) Social constructionism and medical sociology: a reply to M.R. Bury. *Sociology of Health and Illness* 9(2): 107-126
- Ng PC, Murray SS, Levy S Venter JC (2009) An agenda for personalised medicine. *Nature* 461(8): 724-726
- Nordgren A, Juengst ET (2009) Can genomics tell me who I am? Essentialist rhetoric and direct-to-consumer testing. *New Genetics and Society* 28(2): 157-172
- Novas C, Rose N (2000) Genetic risk and the birth of the somatic individual. *Economy and Society* 29(4): 485-513
- Nuffield Council on Bioethics (2010) *Medical Profiling and Online Medicine: The Ethics of Personalised Healthcare in a Consumer Age.* Nuffield Centre for Bioethics, London. [Online] <http://www.nuffieldbioethics.org/personalised-healthcare-0> accessed 23.vi.2012
- O'Neill FK (2007) Fashioning flesh: inclusion, exclusivity and the potential of genomics. IN Atkinson P, Glasner P, Greenslade H (Eds) *New Genetics, New Identities.* Routledge, Abingdon
- O'Reilly T (2009) *What is Web 2.0?* O'Reilly Media Inc. [Online] http://books.google.co.uk/books?hl=en&lr=&id=NpEk_WFCMdiC&oi=fnd&pg=

[PT1&dq=web+2.0&ots=OXQGP2jEI_&sig=gS7Tq4yQ7k3xWT0r3P7DGyS51lo](#)
accessed 16.i.2013

O’Riordan K (2010) *The Genome Incorporated: Constructing Biodigital Identity. Theory, Technology and Society*. Ashgate, London

O’Riordan K (2013) Biodigital publics: personal genomes as digital media artefacts. *Science as Culture* 22(4): 516-539

Ormond KE (2009) What is the role of non-geneticist physicians, and are they prepared for it? *Virtual Mentor* 11(9): 678-682

Oudshoorn N, Pinch T (2005) *How Users Matter: The Co-construction of Users and Technology*. Massachusetts Institute of Technology Press, Cambridge, MA.

Paling J (2003) Strategies to help patients understand risks. *British Medical Journal* 327 (7417): 745-748. [Online]
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC200818/>
accessed 15.v.2014

Parsons E, Atkinson P (1992) Lay construction of genetic risk. *Sociology of Health and Illness* 14(4): 437-455

Patch C, Sequeiros J, Cornel MC (2009) Genetic horoscopes: is it all in the genes? Points for regulatory control of direct-to-consumer genetic testing. *European Journal of Human Genetics* 17: 857-859

Petersen A (1996) Risk and the regulated self: the discourse of health promotion as politics of uncertainty. *Australian and New Zealand Journal of Sociology* 31(1): 44-57

Petersen A, Bunton R (2002) *The New Genetics and the Public’s Health*. Routledge, London

Petersen A, Lupton D (1996) *The New Public Health: Health and Self in the Age of Risk*. Allen and Unwin, Australia

Picard A (2014) *Controversial Genetic Self-test Kits Coming to Canada*. Business News Network, 2nd October 2014 [Online]
<http://www.bnn.ca/News/2014/10/2/Controversial-genetic-self-testing-kits-coming-to-Canada.aspx> accessed 2.x.2014

Pinch TJ, Bijker WE (1987) The social construction of facts and artifacts: or how the sociology of science and the sociology of technology might benefit each other. IN Bijker WE, Hughes TP, Pinch TJ (Eds) *The Social Construction of Technological Systems: New Directions in the Sociology and History of Technology*. Massachusetts Institute of Technology Press, Cambridge MA.

Pinker S (2009) My genome, my self. *New York Times Magazine*. [Online]
http://www.nytimes.com/2009/01/11/magazine/11Genome-t.html?pagewanted=all&_r=0 accessed 15.i.2013

Plows A (2011) *Debating Human Genetics: Contemporary Issues in Public Policy and Ethics*. Routledge, Abingdon

- Plows A, Boddington P (2006) Troubles with biocitizenship? *Genomics, Society and Policy* 2(3): 115-135
- Potter RJ, Mulkay M (1985) Scientists' interview talk: interviews as a technique for revealing participants' interpretative practices IN Brenner M, Brown J, Canter D (Eds) *The Research Interview: Uses and Approaches*. Academic Press, London
- Prainsack B (2014a) The powers of participatory medicine. *PLOS Biology* 12(14): e1001837 [Online]
<http://www.plosbiology.org/article/info:doi%2F10.1371%2Fjournal.pbio.1001837> accessed 23.viii.2014
- Prainsack B (2014b) Understanding participation: the 'citizen science' of genetics. IN Prainsack B, Schicktanz G, Werner-Felmayer G (Eds) *Genetics as Social Practice*. Ashgate, Farnham
- Prainsack B, Reardon J, Hindmarsh R, Gottweis H, Naue U, Lunshof JE (2008) Misdirected precaution: personal genome tests are blurring the boundary between experts and lay-people. *Nature* 456: 34-36
- Prainsack B, Schicktanz G, Werner-Felmayer G (2014) *Genetics as Social Practice*. Ashgate, Farnham
- Prainsack B, Wolinsky H (2010) Direct-to-consumer genome testing: opportunities for pharmacogenomics research? *Pharmacogenomics* 11(5): 651-655
- Prior L (2003) Belief, knowledge and expertise: the emergence of the lay expert in medical sociology. *Sociology of Health and Illness* 25(3): 41-57
- Rabinow P (1996) *Essays on the Anthropology of Reason*. Princeton University Press, Princeton, NJ.
- Rabinow P (2007) *Reflections on Fieldwork in Morocco (30th Anniversary Edn)*. University of California Press
- Rabinow P, Rose N (2006) Biopower today. *Biosocieties* 1: 195-217
- Raman S, Tutton R (2010) Life, science and biopower. *Science, Technology and Human Values* 35(5): 711-734
- Rapp R (2000) *Testing Women, Testing the Fetus: The Social Impact of Amniocentesis in America*. Routledge, New York
- Ray T (2011) *GAO Stands by its 2010 Report on DTC Genomics Firms: Lead Investigator Assigned to New Position*. Pharmacogenomics Reporter. [Online]
<http://www.genomeweb.com/dxpgx/gao-stands-its-2010-report-dtc-genomics-firms-lead-investigator-assigned-new-pos> accessed 12.vi.2012
- Reardon J (2011) The 'persons' and 'genomics' of personal genomics. *Personalized Medicine* 8(1): 95-107

Richards M (1996) Families, kinship and genetics. IN Marteau T, Richards M (Eds) *"The Troubled Helix." Social and Psychological Implications of the New Human Genetics*. Cambridge University Press, Cambridge

Richards M (2010) Reading the runes of my genome: a personal exploration of retail genetics. *New Genetics and Society* 29(3): 291-310

Roberts JS, Cupples LA, Relkin NR, Whitehouse PJ, Green RC (2005) Genetic risk assessment for adult children of people with Alzheimer's disease: the risk evaluation and education for Alzheimer's disease (REVEAL) study. *Journal of Geriatric Psychiatry and Neurology* 18(4): 250-255

Roberts C, Sarangi S (2005) Theme-oriented discourse analysis of medical encounters. *Medical Education* 39(6): 632-640

Robinson JC (2005) Managed consumerism in health. *Health Affairs* 24(6): 1478-1489

Rose N (2007) *The Politics of Life Itself: Biomedicine, Power, and Subjectivity in the Twenty-First Century*. Princeton University Press, Princeton, NJ.

Rosen P (1993). The social construction of mountain bikes: technology and postmodernity in the cycle industry. *Social Studies of Science* 23(3): 479–513

Rothwell PM (2005) External validity of randomised controlled trials: "to whom do the results of this trial apply?" *The Lancet* 365(9453): 82-93

Russell S (1986) The social construction of artifacts: a response to Pinch and Bijker. *Social Studies of Science* 16: 331-346

Russo G (2006) Home health tests are 'genetic horoscopes'. *Nature* 442(3): 497

Ryan GW, Bernard HR (2000) Data management and analysis methods. IN Denzin NK and Lincoln YS (Eds) *Handbook of Qualitative Research (2nd Edn)*. Sage, Thousand Oaks, CA.

Sander C (2000) Genomic medicine and the future of healthcare. *Science* 287(5460): 1977-1978

Saukko PM, Richards SH, Shepherd MH, Campbell JL (2006) Are genetic tests exceptional? Lessons from a qualitative study on thrombophilia. *Social Science and Medicine* 63: 1947-1959

Saunders B, Kitzinger J, Kitzinger C (2014) Anonymising interview data: challenges and compromise in practice. *Qualitative Research* [Online] <http://qrj.sagepub.com/content/early/2014/09/23/1468794114550439.full.pdf+html> accessed 29.ix.2014

Scott MB, Lyman SM (1968) Accounts. *American Sociological Review* 33(1): 46-62

Scott J, Marshall G (2009) *Oxford Dictionary of Sociology (3rd Edn)*. Oxford University Press, Oxford

- Seidel J (1991) Method and madness in the application of computer technology to qualitative data analysis. IN Fielding NG, Lee RM (Eds) *Using Computers in Qualitative Research*. Sage, London
- Seidman I (2013) *Interviewing as Qualitative Research: A Guide for Researchers in Education and the Social Sciences (4th Edn)*. Teachers College Press, New York
- Silverman D (2000) *Doing Qualitative Research: A Practical Handbook*. Sage, London
- Silverman PH (1995) Commerce and genetic diagnostics. *Hastings Center Report* 25(3): Special Supplement S15-18
- Skirton H, Goldsmith L, Jackson L, O'Connor A (2012) Direct-to-consumer genetic testing: a systematic review of position statements, policies and recommendations. *Clinical Genetics* 82(3): 210-218
- Skirton H, Kerzin-Storrar L, Barnes C, Hall G, Longmuir M, Patch C, Scott G, Walford-Moore J (2013) Building the Genetic Counsellor profession in the United Kingdom: two decades of growth and development. *Journal of Genetic Counseling* 22(6): 902-906
- Stergiopoulos K, Brown DL (2014) Genotype-guided vs clinical dosing of warfarin and its analogues: meta-analysis of randomized clinical trials. *Journal of the American Medical Association Internal Medicine* 174(8): 1330-1338
- Strauss AL (1987) *Qualitative Analysis for Social Scientists*. Cambridge University Press, Cambridge
- Sturgis P, Cooper H, Fife-Schaw C, Shepherd R (2004) Genomic science: emerging public opinion. IN Park A, Curtice J, Thomson K, Bromley C, Phillips M (Eds) *British Social Attitudes: The 21st Report*. Sage Publications, London
- Temple EC, Brown RF (2011) A comparison of internet-based participant recruitment methods: engaging the hidden population of cannabis users in research. *Journal of Research Practice* 7(2): article D2 [Online] <http://jrp.icaap.org/index.php/jrp/article/view/288/247> accessed 24.x.2014
- Tutton R (2012) Personalizing medicine: futures present and past. *Social Science and Medicine* 75(10): 1721-1728
- Tutton R (2014) *Genomics and the Reimagining of Personalized Medicine*. Ashgate, Farnham.
- Tutton R, Jamie K (2013) Personalized medicine in context: social science perspectives. *Drug Discovery Today: Therapeutic Strategies*. [Online] <http://dx.doi.org/10.1016/j.ddstr.2013.05.003> accessed 10.iii.2015
- Tutton R, Prainsack B (2011) Enterprising or altruistic selves? Making up research subjects in genetic research. *Sociology of Health and Illness* 33(7): 1081-1095
- UK Pharmacogenetics Study Group (2006) *Policy Issues in Pharmacogenetics: Policy Briefing*. [Online]

<http://www.york.ac.uk/media/satsu/res-pgx/PGxpolicyissues2006.pdf>
accessed 2.ii.2015

Urban Dictionary (2013) *Urban Dictionary* [Online]
<http://www.urbandictionary.com/define.php?term=interesting>
accessed 4.ix.2013

Van Lente H (2000) Forceful futures: from promise to requirement IN Brown N, Rappert B, Webster A (Eds) *Contested Futures: A Sociology of Prospective Technoscience*. Ashgate, Aldershot

Van Ommen GJB, Cornel MC (2008) Recreational genomics? *European Journal of Human Genetics* 16: 403-404

Vayena E, Gouna E, Streuli J, Hafen E, Prainsack B (2012) Experiences of early users of direct-to-consumer genomics in Switzerland: an exploratory study. *Public Health Genomics* 15: 352-362

Vayena E, Prainsack B (2013) The challenge of personal genomics in Germany. *Nature Biotechnology* 31(1): 16

Ventola CL (2011) Direct-to-consumer pharmaceutical advertising: therapeutic or toxic? *Pharmacy and Therapeutics* 36(10): 669-674, 681-684

Vineis P, Christiani DC (2004) Genetic testing for sale. *Epidemiology* 15(1): 3-5

Voora D (2011) Pharmacogenomics of Warfarin: clinical implications. *Medscape Genomic Medicine*. [Online]
<http://www.medscape.com/viewarticle/744039> accessed 20.iii.2015

Vorhaus D (2010a) Meet the new decode, same as the old decode? *Genomics Law Report*. [Online]
<http://www.genomicslawreport.com/index.php/2010/01/25/meet-the-new-decode-same-as-the-old-decode/> accessed 26.vi.2012

Vorhaus D (2010b) The past, present and future of DTC genetic testing regulation. *Genomics Law Report*. [Online]
<http://www.genomicslawreport.com/index.php/2010/08/05/the-past-present-and-future-of-dtc-genetic-testing-regulation/> accessed 12.viii.2011

Vorhaus D (2011) Surreptitious genetic testing: a new bill in Texas and the Iowa straw poll. *Genomics Law Report*. [Online]
<http://www.genomicslawreport.com/index.php/2011/08/12/surreptitious-genetic-testing-a-new-bill-in-texas-and-the-iowa-straw-poll/>
accessed 26.vi.2012

Vorhaus D (2012) Implications of Amgen/deCODE deal for genetic testing consumers. *Genomics Law Report*. [Online]
<http://www.genomicslawreport.com/index.php/2012/12/10/implications-of-amgendecode-deal-for-genetic-testing-consumers/> accessed 14.i.2013

Vorhaus D, MacArthur D (2010) Consumer genetics needs more transparency not excessive regulation. *Xconomy*. [Online]

<http://www.xconomy.com/national/2010/06/23/consumer-genetics-needs-more-transparency-not-excessive-regulation/> accessed 25.vi.2012

Wade CH, Wilfond BS (2006) Ethical and clinical practice considerations of genetic counselors related to direct-to-consumer marketing of genetic tests. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics* 142C(4): 284-292

Wagner JK (2013) Looking back at Myriad. *Genomics Law Report* [Online] <http://www.genomicslawreport.com/index.php/2013/06/19/looking-back-at-myriad-a-users-guide/> accessed 7.v.2015

Walford G (2005) Research ethical guidelines and anonymity 1. *International Journal of Research and Method in Education* 28 (1): 83-93

Wallace H (2008) Most gene test sales are misleading. *Nature Biotechnology* 26: 1221

Ward M (2006) *How the web went world wide*. BBC News [Online] <http://news.bbc.co.uk/1/hi/sci/tech/5242252.stm> accessed 15.vi.2012

Wayback Machine (2013) *Sciona Partners with the Body Shop to Bring Leading Genetic Services to the High Street*. Sciona 2001 archived by Wayback Machine [Online] http://web.archive.org/web/20020210153557/http://www.sciona.com/public/news/261101_news.htm accessed 3.ii.2013

Wellcome Trust Sanger Institute (2014) *Genome Reference Consortium*. [Online] <http://www.sanger.ac.uk/research/areas/bioinformatics/grc/> accessed 19.ii.2015

Williams R, Edge D (1996) The social shaping of technology. *Research Policy*, 25: 865 899

Wolcott HF (2001) *Writing up Qualitative Research (2nd Edn)*. Sage, London

Wright C, Burton H, Hall A, Moorthie S, Pokorska-Bocci A, Sagoo G, Sanderson S, Skinner R (2011a) *Next Steps in the Sequence: The Implications of Whole Genome Sequencing for Health in the UK*. PHG Foundation, Cambridge

Wright CF, Gregory-Jones S (2010) Size of the direct-to-consumer genomic testing market. *Genetics in Medicine* 12: 594

Wright CF, Hall A, Zimmern RL (2011b) Regulating direct-to-consumer genetic tests: what's all the fuss about? *Genetics in Medicine* 13(4): 295-300

Wolfberg AJ (2006) Genes on the web: direct to consumer marketing of genetic testing. *New England Journal of Medicine*, 6(355): 543-545

Wyatt S (2008) Technological determinism is dead; long live technological determinism. IN Hackett I, Amsterdamska O, Lynch M, Wajcman J (Eds) *The Handbook of Science and Technology Studies*. Massachusetts Institute of Technology Press, Cambridge, MA.

Wynne B (1992) Misunderstood misunderstanding: social identities and public uptake of science. *Public Understanding of Science* 1: 281-304

YouGov (2014) *NHS One of The Best in The World*. YouGov [Online] <http://yougov.co.uk/news/2014/06/20/nhs-one-best-developed-world/> accessed 18.viii.2014

Ziegelstein RC (2015) Personomics. *Journal of the American Medical Association Internal Medicine* [Online] <http://archinte.jamanetwork.com/article.aspx?articleid=2247166> accessed 5.v.2015

Appendices

Appendix 1: School of Social Sciences REC Application

Cardiff School of Social Sciences

Ethical Approval Papers

MPhil/PhD Doctorate Research Projects

SREC/826

Principal Investigator (PI):

Teresa M. D. Finlay, ESRC +3 PhD Student, Socsi/Cesagen,
finlaytm@cardiff.ac.uk

3 Alexandra Road, Oxford, OX2 0DD

The research:

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Direct-to-consumer genetic testing (DTCGT) for disease risk is now widely available to purchase online. Those in favour of DTCGT champion the rights of individuals to access their genetic information, while many medical professionals think a vulnerable public needs to be protected by regulation. Neither of these views is evidence-based, but they reflect changes in society that the growth in personalised medicine is driving. The paucity of published research data indicates that little is known about DTCGT generally and its uptake in the UK in particular. This PhD research project thus aims to:-

- i. explore UK users’ motivations and expectations of DTCGT for disease risk and its impact on them
- ii. establish how people make sense of complex disease risk information
- iii. explore clinical genetics professionals’ perceptions of DTCGT’s impact on users, and their involvement with helping users of DTCGT
- iv. draw out the implications of DTCGT for users, professionals and the NHS.

The research will answer the following questions:

- i. What do people know about DTCGT and what are their sources of information?

- ii. What are people's experiences of DTCGT and to whom do they turn for further help?
- iii. What are clinical genetics professionals' views of DTCGT?
- iv. What are clinical genetics professionals' experiences of supporting users of DTCGT?
- v. What are the likely implications of DTCGT for users and professionals in the NHS?

Funding:

The PI has an ESRC funded Social Science PhD studentship with support from Cesagen.

Participant details:

- 1) Up to 20 DTCGT users will be purposively recruited online, the medium in which DTCGT is traded. Advertisements will be placed on Internet search engine pages linked to searches for DTCGT companies, related weblogs and User Group websites inviting people who have had a test to visit the research project website for information and then to email the PI to express interest in being interviewed about their experiences. Participants (Users) will be UK residents, able to read and converse fluently in English, 18 years or older, who have had a DTCGT and are competent to give consent.
 - People who are not UK residents will not be eligible as they will not have automatic access to the NHS.
 - People who are not able to read or converse fluently in English will not be recruited as it will undermine the quality of the interview data and participants' confidentiality to use translators for interviews, as well as being beyond the scope of this project.
 - People must be 18 years or older to buy a DTCGT and thus to participate in this research.

On receipt of emails from interested Users the PI will reply attaching a copy of the Participant Information sheet (Users) (Appendix 6) and Consent Form (Appendix 8) and request a telephone contact number to screen the person for suitability to participate and arrange an interview. Understanding of the project and competence to consent will be assessed in the screening call prior to interview.

- 2) Up to 20 registered clinical genetics professionals will be purposively recruited using snowball sampling, starting with clinical geneticists known to the PI and supervisors. An email (Appendix 5) will be sent to known clinicians inviting expressions of interest from those who have been consulted by users about their DTCGT results (not necessarily the Users recruited to participate in this study). These clinicians will be

asked to forward the invitation to colleagues elsewhere in the UK who have similar clinical expertise. Participants (Clinicians) will be UK residents, registered and practising as clinical geneticists or genetics counsellors in the NHS. These participants will be participating in their professional capacity and as such it is assumed they will be fluent in English and competent to consent by virtue of their professional registration and practice in the UK.

Methodology and data handling:

Methods: Participants will agree to an interview, preferably conducted in person but possibly on the telephone if a meeting is difficult to arrange. Interviews with Users and Clinicians will last no longer than 1.5 hours and will be audio-recorded. The PI will record participants' consent at the beginning of the interview either in writing or on the telephone, depending on the mode of the interview. Interviews will be semi-structured and will aim to cover the topics required to answer the research questions and meet the research aims and objectives. The PI will keep a diary recording non-verbal communication and other related observations during the interviews. Interview recordings will subsequently be transcribed by the PI. Interview data will be analysed using a theme-orientated approach.

Storage: The PI will be responsible for the research data, all of which will be stored in the form of electronic files including participants identifying information, voice recordings, interview transcripts and written analysis and discussion of the findings. These will be stored in password-protected files on encrypted devices. Any paper copies of data will be stored in a locked filing cabinet on University premises. Raw data pertaining to the project will be viewed by the PI (and possibly supervisors) only; no other independent contractors (e.g. research assistants or transcribers) will be involved. Electronic data will be stored for 10 years following completion of the project after which time it will be destroyed. Data will be by deletion from electronic devices or shredding.

Anonymity: Participants will each be assigned a code and their data subsequently handled using their individual code. Participants could be identified by their code should there be a need to contact them for further information following initial data analysis. Participants' information and codes will be stored separately from their data. While all possible steps will be taken to anonymise data in the written report and publications, it may not be possible to completely anonymise participants' contributions due to the relatively small numbers of people involved. Participants will be made aware of this in the Participant Information Sheets (Appendices 6 and 7).

Dissemination: Findings from the study will be shared with participants, colleagues and peers at local, national and international specialist meetings and published in the professional literature. Copies of the completed PhD thesis will be stored in academic libraries.

Shaded box (ethical) considerations:

Possible Harm: At the beginning of interviews when consent is obtained, the PI will re-iterate that all interviews are in confidence and that participants can withdraw at any time without penalty, should they feel unable to continue. Should they choose to do this all data pertaining to that interview will be destroyed and will not be included in the research.

Some participants may experience strong emotions when discussing issues related to disease risk and/or the perceived impact of genetic information on family members. The interviews will be conducted sensitively by the PI who is a registered nurse and lecturer with considerable experience of facilitating challenging, emotional discussions. Should a participant become distressed, the interview will be suspended and they will be reminded that they may withdraw from the study if they wish. Should emotional support be required over and above that which can be immediately provided, the PI will recommend that the participant seek help from an appropriate external source such as their General Practitioner surgery or similar agency.

Information and consent:

In the recruitment advertisements prospective participants (Users) will be directed to the study website for information about the study including the Participant Information Sheet (Users) (Appendix 6) and Consent Form (Appendix 8). The website will ask potential participants to email the PI within one week with their expression of interest in participating in an interview. A reply will be sent requesting a telephone contact number with a copy of the Participant Information Sheet (Users) and Consent Form attached in case these have not been accessed on the website already. If a reply with the contact telephone number is not received within one week, one email reminder will be sent.

Prospective participants (Clinicians) will be sent a Participant Information Sheet (Appendix 7) and Consent Form (Appendix 9) with the email inviting expressions of interest in participating in the research by clinicians. On receiving an email expressing interest, a reply will be sent requesting a telephone contact number with a copy of the Participant Information Sheet (Clinicians) and Consent Form attached in case these have not been accessed from the previous email. If a reply with the contact telephone number is not received within one week, one email reminder will be sent.

The time and venue for interviews will be arranged by phone call at the participants' convenience, taking precautions to minimise risks to the PI as per the Code of Practice for the Safety of Social Researchers.

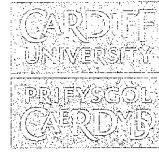
Consent to be interviewed will be obtained from participants by the PI prior to the interview either in writing on the form or verbally on the telephone with the researcher completing the form depending on the mode of the interview (Appendices 8 and 9).

It is unlikely that any of the participants will be known to the PI or in a dependent relationship with her. No payments or incentives will be offered for participation and participants will be reminded that their contribution to the

research is based on altruism rather than any personal benefit; consent should therefore be given without any pressure (real or implied) to do so.

Appendix 2: School of Social Sciences REC Approval

Cardiff School of Social Sciences
Director Professor Malcolm Williams
Ysgol Gwyddonau Cymdeithasol Caerdydd
Cyfarwyddwr Yr Athro Malcolm Williams



Cardiff University
Glenorgan Building
King Edward VII Avenue
Cardiff CF10 3WT
Wales UK
Tel: Ffôn: +44(0)129 2087 5179
Fax: Ffôn: +44(0)129 2087 4175
www.cardiff.ac.uk
Prifysgol Caerdydd
Ardalied Morgannwg
Ardalio Bwrda Eborac VII
Caerdydd CF10 3WT
Cymru Y Breytas Gyfnewid

19th January 2012

Our ref: SREC/826

Teresa Finlay
PhD Programme
SOCSI

Dear Teresa

Your project entitled "*Direct-to-consumer genetic testing: Users' experiences of testing for disease risk and genetics clinicians' perceptions of its impact on NHS genetics services*" has now been approved by the School of Social Sciences Research Ethics Committee of Cardiff University following its meeting on 8th December 2011 and you can now commence the project.

If you make any substantial changes with ethical implications to the project as it progresses you need to inform the SREC about the nature of these changes. Such changes could be: 1) changes in the type of participants recruited (e.g. inclusion of a group of potentially vulnerable participants), 2) changes to questionnaires, interview guides etc. (e.g. including new questions on sensitive issues), 3) changes to the way data are handled (e.g. sharing of non-anonymised data with other researchers).

All ongoing projects will be monitored every 12 months and it is a condition of continued approval that you complete the monitoring form.

Please inform the SREC when the project has ended.

Please use the SREC's project reference number above in any future correspondence.

Yours sincerely

A handwritten signature in black ink, appearing to be 'Tom Horlick-Jones', written over a horizontal line.

Professor Tom Horlick-Jones
Chair of the School of Social Sciences Research Ethics Committee

cc: E Renton
Supervisor: M Arribas-Ayllon

Cardiff University is a registered charity, no. 1136255
Mae Prifysgol Caerdydd yn caiswr cofrestrig, rhif 1136255

Appendix 3: Cardiff and Vale University Health Board, R & D Committee Governance Review Approval

GIG | Bwrdd Iechyd Prifysgol
CYMRU | Caerdydd a'r Fro
NHS | Cardiff and Vale
WALLES | University Health Board

Ysbyty Athrofaol Gymru
University Hospital of Wales
 Health Park,
 Cardiff, CF14 4XW
 Gwynedd, CF14 4XW
 Fax: 029 2074 3838
 Mifffon: 029 2074 3832

Peri Yr Amodol Sghdan,
Gwynedd, CF14 4XW
 Fax: 029 2074 3838
 Mifffon: 029 2074 3832

Ench yffwrdd
 1872
 Wladfa Iechyd: Telephone Network
 Direct (local) line: unioynfydd

From: Professor JI Elison
 R&D Director
 R&D Office, 2nd Floor TB2
 University Hospital of Wales
 Cardiff
 CF14 4XW

Tel: 029 20746986
Fax: 029 20745311
CAV_Research.Development@wales.nhs.uk

20 August 2012

Professor Angus John Clarke
 Institute of Medical Genetics
 Health Park
 Cardiff
 CF14 4XW

Dear Professor Clarke

Cardiff and Vale UHB Ref: 120TD/6485 : Direct-To-Consumer Genetic Testing: Clinical Genetics Professionals's Experiences And Perceptions Of Its Impacts On The NHS.

NISCHR PCU Ref: 963331

The above project was forwarded to Cardiff and Vale University Health Board R&D Office by the NISCHR Permissons Coordinating Unit. A Governance Review has now been completed on the project.

Documents approved for use in this study are:

Document	Version	Date Approved
Protocol	2	31/07/12
Participant Info Sheet: Genetics Clinicians	2	31/07/12
Participant (UHB/MS) Recruitment Email	2	31/07/12
Consent Form	2	31/07/12

I am pleased to inform you that the UHB has no objection to your proposal. Please accept this letter as confirmation of permission for the project to begin within this UHB.

Version 1.0, 06/06/10

Page 1 of 3

Bwrdd Iechyd Prifysgol Gwynedd a'r Fro www.gwyneddibwrddiechydprifysgol.wales.nhs.uk
 Cardiff and the Vale University Health Board's Governance Review Unit, Cardiff, Wales, University Health Board



You have informed us that Cardiff University is willing to act as Sponsor under the Research Governance Framework for Health and Social Care.

Your study may be eligible for adoption by the NISCHR Clinical Research Portfolio (CRP). You should now apply to have this trial adopted in order for the UHB to receive funding to support this study. If your study is adopted onto the NISCHR CRP, it will be a condition of this NHS research permission that you will be required to regularly upload recruitment data onto the portfolio database.

To apply for adoption onto the NISCHR CRP, please go to:
<http://www.wales.nhs.uk/sites2/areas/clinical/crp/crp201208231979>. Once accepted, NISCHR CRP studies may be eligible for additional support through the NISCHR Clinical Research Centre, but you must not apply for funding at:
<http://www.wales.nhs.uk/sites2/areas/clinical/crp/crp201208231979>

If your study is adopted into the portfolio, please inform NISCHR-PCU and the R&D Office of your portfolio ID number.

To upload recruitment data, please follow this link:
http://www.crcrc.nhr.ac.uk/about_us/accesses/portfolio/recruitment_uploading
 recruitment data will enable NISCHR to monitor research activity within NHS organisations, leading to NHS R&D allocations which are activity driven. Uploading of recruitment data will be monitored by your colleagues in the R&D office. If you need any support in uploading this data, please contact
CAV_research.development@wales.nhs.uk

May I take this opportunity to wish you success with the project and remind you that as Principal Investigator you are required to:

- Inform the R&D Office if this project has not opened within 12 months of the date that the funding is awarded
- Inform NISCHR PCU and the UHB R&D Office if any external or additional funding is awarded for this project in the future
- Submit any substantial amendments relating to the study to NISCHR PCU in order that they can be reviewed and approved prior to implementation
- Ensure NISCHR PCU is notified of the study's closure
- Ensure that the study is conducted in accordance with all relevant policies, procedures and legislation
- Provide information on the project to the UHB R&D Office as requested from time to time to include participant recruitment figures

Yours sincerely,

J. Elison
 Professor Jonathan I Elison
 R&D Director

CC: R&D Lead Prof Dhavendra Kumar
 Sponsor contact Chris Shaw, RACD Cardiff University

Version 1.0, 06/06/10

Page 2 of 3

C.I. TF
 ASOP DELAH
 Acsop OR MAN

Appendix 4: Email to Recruit Clinicians



Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

|Participant (Clinicians) recruitment letter.

To who it may concern,

I am undertaking research into DTC genetic testing bought online by the public for disease risk. This is an ESRC-funded PhD study being undertaken in the School of Social Sciences at Cardiff University.

I am interested in interviewing genetics clinicians (doctors, nurses, counsellors) who are, or could be involved in counselling people who have bought these tests, about the results. If you are interested in participating please would you read the information sheet and consent form attached to this email for your information. If, after reading the attached documents you think the study is relevant to you and you would like to participate, then please email me expressing your interest and I will contact you to arrange an interview.

If you would be so kind as to forward this email to any NHS colleagues to whom you think it might be relevant, I would be most grateful.

Yours faithfully

TMD Finlay RN, BSc(Hons), MSc, PGDipEd

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk
Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Appendix 5: Information Sheet for Users



Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 Version 2

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Appendix B

Participant Information Sheet – DNA Test Users

Study Title

Online DNA tests: a study into the experiences of people who buy DNA tests on the internet to find out their risk of disease, and the experiences of the NHS genetics services personnel who help them with the results.

Invitation paragraph

You are being invited to take part in this research study for a PhD student project. Before you decide, it is important for you to understand why the research is being done and what it would involve. Please take time to read the following information carefully and discuss it with friends or relatives, and your GP, Geneticist or Genetics Counsellor if you wish. Ask if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part. Thank you for reading this.

What is the purpose of the study?

DNA tests can be bought on the Internet to find out ancestry, paternity, nutritional needs, how people respond to some drugs, and some diseases they may be at risk of developing. There are different views about the benefits and risks of these tests but these views are not yet supported by enough research into what people actually experience. Also, it is likely that many people who buy these tests, and possibly their GPs, do not really understand the results. This PhD research project aims to develop knowledge about UK residents’ experiences of buying a DNA test online and any effect on NHS genetics services.

Why have I been chosen?

You have been chosen because you have bought an online DNA test to see what diseases you may be at risk of developing, and you responded to a request to participate in the research.

Do I have to take part?

It is up to you to decide whether or not to take part. You can keep a copy of this information sheet and the consent form for future reference. If you decide to take part you will be asked to give your consent but will still be free to withdraw at any time and without giving a reason. Your participation is not linked to any DNA test company, your GP or a genetics clinic, and would not affect the standard of care you receive.

What would happen to me if I take part?

If you do decide to take part please email the PhD student researcher at finlaytm@cardiff.ac.uk including a telephone number she can call you on, and information about when it is best for her to call you. She would then telephone you to go through the research project, and arrange an interview. The interview would be arranged at a time and

1

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 Version 2

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Appendix B

place to suit you; if a meeting is difficult to arrange, the interview could be conducted on the telephone. The interview will cover why you decided to buy a DNA test online, how you felt about it before and after getting the results, whether you understood the results and who you asked for help if not. It will also cover how you feel about sharing the results with your family and how what you learned affected your relationships with them, if that is relevant. It should last no longer than an hour and a half, and probably be shorter. At the beginning of the interview you will be asked to consent to being a participant in the research study. The interview would be audio-recorded.

What would I have to do?

You would have to have a telephone conversation with the researcher to ensure it is appropriate for you to participate, and to agree a time and place for the interview. At the time of the interview you will be asked to consent to being involved; if the interview is to be conducted face to face, you would be asked to read and sign a consent form if you agree with it. If the interview is to be conducted on the telephone the consent form will be read to you over the telephone and you will be asked if you agree to the statements read to you. This will be written on a consent form by the researcher. The interview would then go ahead immediately and be audio-recorded. If you felt you did not want to continue with the interview at any point you would be free to ask to stop without giving a reason, and without affecting any care you may receive from the NHS. The interview recording and any notes about it up to that point would be destroyed and not included in the study.

What are the possible benefits of taking part?

There is no direct benefit to you or your family from taking part in this study. Some people may find it helpful to talk about their experiences, but any benefit would be to future users of online DNA testing, NHS Genetics Services and possible future regulation of this kind of test.

Would my taking part in this study be kept confidential?

All information which is collected about you during the course of the research will be kept strictly confidential. Any information about you contained in the research will have a code assigned to it; your name and contact details will not be included so that you could not be identified from it. The researcher will be the only person who knows what participants’ codes are. That information will be stored in a locked filing cabinet or password protected files on the researcher’s computer at Cardiff University, separate from the research interview data. Only the researcher will have access to these.

It is possible that what you say in the interview would be quoted in the research report and you may recognise your words should you choose to read the report, but they would not have your name or identifying information attached. The consent form will ask for permission to quote you.

2

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk



Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 Version 2

"Direct-to-consumer genetic testing: users' experiences of testing for disease risk and genetics clinicians' perceptions of its impact on NHS genetics services."

Appendix B

On completion of the study data will be stored for up to 10 years in locked filing cabinets or on encrypted computers in password protected files at the researcher's home and/or on Cardiff University premises. At the end of this period the data will be destroyed either by shredding hard copies or deleting electronic files and backup copies.

What would happen to the results of the research study?

The results of the research will be written up in a PhD thesis which will be kept in Cardiff University Library; they will be presented at conferences and published in professional journals. They may also be used to inform policy relating to regulating DNA tests. If you would like a copy of the results this could be sent to you when the research is complete.

Who has reviewed the study?

Cardiff University School of Social Sciences Research Ethics Committee has reviewed and approved the research project. It is also supported by the Economic and Social Research Council.

Contact for further information

If you have any further questions please do not hesitate to contact me, the researcher, at the following email address finlaytm@cardiff.ac.uk

Once again – thank you for taking the time to read this.

Appendix 6: Information Sheet for Clinicians



Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Participant Information Sheet – Genetics Clinicians

Study Title

Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.

Invitation paragraph

You are being invited to take part in this research study for a PhD student project. Before you decide, it is important for you to understand why the research is being done and what it would involve. Please take time to read the following information carefully and discuss it with colleagues and friends if you wish. Ask if there is anything that is not clear or if you would like more information. Take time to decide whether or not you wish to take part. Thank you for reading this.

What is the purpose of the study?

DNA tests can be bought on the Internet to establish ancestry, paternity, nutritional needs, pharmacogenetics, and common disease risk. There are different views about the benefits and risks of these tests but these views are not currently supported by a robust evidence-base. Also, it is likely that many people who buy these tests, and possibly their GPs, do not really understand the results. This PhD research project aims to develop knowledge about UK residents buying a DNA test online and the effect on NHS genetics services.

Why have I been chosen?

You have been chosen because you work in clinical genetics, have counselled one or more people who have bought a DNA test for disease risk online or are interested in doing so, and you responded to a request to participate in the research.

Do I have to take part?

It is up to you to decide whether or not to take part. You can keep a copy of this information sheet and the consent form for future reference. If you decide to take part you would be asked to give your consent but would be free to withdraw at any time and without giving a reason. Your participation is not linked to any DNA test company, genetics service or your employing NHS Trust, and would not affect your employment.

What would happen to me if I take part?

If you do decide to take part please email the PhD student researcher at finlaytm@cardiff.ac.uk including a telephone number she can call you on, and information about when it is best for her to call you. She would then telephone you to go through the research project to ensure it is appropriate for you to participate, and arrange an interview. The interview would be arranged at a time and place to suit you. If a meeting is difficult to arrange, the interview could be conducted on the telephone. The interview will cover your experiences of counselling and supporting people who have bought a DNA test online or

1

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

your interest in doing so, your views about these tests including their current and future impact on NHS genetics services, and how you feel about the public taking such testing into their own hands. It should last no longer than an hour and a half, and probably be shorter. At the beginning of the interview you will be asked to consent to being a participant in the research study. The interview would be audio-recorded.

What would I have to do?

You would have to have a telephone conversation with the researcher to ensure it is appropriate for you to participate, and to agree a time and place for the interview. At the time of the interview you will be asked to consent to being involved; if the interview is to be conducted face to face, you will be asked to read and sign a consent form if you agree with it. If the interview is to be conducted on the telephone the consent form will be read to you over the telephone and you will be asked if you agree to the statements read to you. This will be written on a consent form by the researcher. The interview would then go ahead immediately and be audio-recorded. If you felt you did not want to continue with the interview at any point you would be free to ask to stop without giving a reason.

What are the possible benefits of taking part?

There is no direct benefit to you or your service from taking part in this study. Some people may find it helpful to talk about their experiences, but any benefit would be to future users of online DNA testing, NHS Genetics Services and possible future regulation of this kind of test.

Would my taking part in this study be kept confidential?

All information that is collected about you during the course of the research will be kept strictly confidential. Any information about you contained in the research will have a code assigned to it; your name and contact details will not be included so that you cannot be identified from it. The researcher will be the only person who knows what participants’ codes are and that information will be stored in a locked filing cabinet or password protected files on the researcher’s computer at Cardiff University. Only the researcher will have access to these.

It is possible that what you say in the interview would be quoted in the research report and you may recognise your words should you choose to read the report, but they would not have your name or identifying information attached. The consent form asks for permission to quote you.

On completion of the study data will be stored for up to 10 years in locked filing cabinets or on encrypted computers in password protected files at the researcher’s home and/or on Cardiff University premises. At the end of this period the data will be destroyed either by shredding hard copies or deleting electronic files and backup copies.

2

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk



Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

What would happen to the results of the research study?

The results of the research will be written up in a PhD thesis, which will be kept in Cardiff University Library; they will be presented at conferences and published in professional journals. They may also be used to inform policy relating to regulating DNA tests. If you would like a copy of the results this could be sent to you when the research is complete.

Who has reviewed the study?

Cardiff University School of Social Sciences Research Ethics Committee has reviewed and approved the research project. It is also supported by the Economic and Social Research Council.

Contact for further information

If you have any further questions please do not hesitate to contact me, the PhD student researcher, at the following email address finlaytm@cardiff.ac.uk

Once again – thank you for taking the time to read this.

3

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Appendix 7: Consent Form for Users



Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 Version 2

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Appendix C

Participant identification number

Researcher’s contact details:

Teresa Finlay

6 Museum Place

Cardiff, CF10 3BG

finlaytm@cardiff.ac.uk

Please initial boxes

1. I confirm that I have read and understand the information sheet dated ...3/01/12., version.2.. for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving a reason and without my medical care being affected.

3. I agree to the use of anonymous verbatim quotes in the presentation of the research.

4. I understand that the interview will be audio taped.

5. I agree to take part in the above study.

Name of Participant _____ Date _____ Signature of Participant _____

Name of Researcher _____ Date _____ Signature of Researcher _____

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk
Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk
Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Appendix 8: Consent form for Clinicians



Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

"Direct-to-consumer genetic testing: users' experiences of testing for disease risk and genetics clinicians' perceptions of its impact on NHS genetics services.

Participant identification number

Researcher's contact details:

Teresa Finlay

6 Museum Place

Cardiff, CF10 3BG

finlaytm@cardiff.ac.uk

Please initial boxes

1. I confirm that I have read and understand the information sheet dated 3/01/2012, version 2 for the above study. I have had the opportunity to consider the information, ask questions and have had these answered satisfactorily.

2. I understand that my participation is voluntary and that I am free to withdraw at any time, without giving a reason and without my legal rights being affected.

3. I agree to the use of anonymous verbatim quotes in the presentation of the research.

4. I understand that the interview will be audio recorded.

5. I agree to take part in the above study.

Name of Participant _____ Date _____ Signature of Participant _____

Name of Researcher _____ Date _____ Signature of Researcher _____

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk
Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk
Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Appendix 9: Interview Schedule for Users



Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 (this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Users’ interview prompts

Interview Schedule – DNA Test Users

- Meet and greet –
introduce self, PhD student researcher at Cesagen, Cardiff University
- Thank you for agreeing to participate in the study
Can I just check you have read the information sheet?
the consent form?
are happy for the interview to go ahead?
- Please may I ask you to initial the boxes on the consent form if you are happy to and then sign at the bottom, I will also sign. We need to do two copies, one for you and one for me.
(If skype/phone “I will complete, copy and send you one to keep”)
- OK – now we can get started. If at any point you want the interview to stop please just let me know. Just a reminder that I am recording this interview and am switching the recorder on
- SWITCH RECORDER ON
- Happy to start? – OK. Please could you tell me about yourself
your age
your ethnicity
your occupation (present/former)
education (level/subject)
social status/family (children/siblings)
- Where did you find out about this research study?
- Where/how did you find out about personal genomics tests bought online?

1

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 (this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Users’ interview prompts

- When did you have your personal genomics test?
- Why did you decide to buy a test?
- Which company did you use?
- What test did you buy?
- Why did you choose that one?

- Tell me about your experience of testing
- How did you experience getting the results?
- How did the results make you feel?
- How do you feel now?
- Do you still get updates? What’s that like?

- Did you consult your GP or other healthcare team about them?
- What was that like?

- What has been the impact for you and your family?

- What are your thoughts about informed consent?
(Were you adequately informed beforehand? Do you think it should be more/less rigorous etc)

2

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee,

Study Number SREC/826 Date 3/01/2012 (this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Users’ interview prompts

- What are your thoughts about counselling in relation to the results of your/these tests? (Would you have benefitted from it/should it be provided? In what form etc)
- What are your thoughts about confidentiality in relation to your genetic information/genetic testing generally
- What are your thoughts about regulation of genetic tests? (Should they be regulated, to what extent, if so by whom/banned altogether?)
- (If a 23andMe customer) ask opinion about the patenting issue
- Do you have a sense that uptake is different in the UK from elsewhere? Why do you think that is?
- Is there anything else you would like to tell me in relation to your experience of your test and its results?
- Is there anything I haven’t asked you about that you were expecting?

Would you like to be sent a copy of the summary of the results when the study is complete?

If yes - where shall I send them?

Thank you so much for your help with the research and your participation in this interview – I really appreciate it.

Appendix 10: Interview Schedule for Clinicians



Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

(this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Clinicians’ interview prompts

Interview Schedule – Genetics Clinicians

- Meet and greet.
Introduce self, PhD student researcher at CESAGen, Cardiff University
- Thank you for agreeing to participate in the study
Can I just check you have read the information sheet?
The consent form?
Are you happy for the interview to go ahead?
- Please may I ask you to initial the boxes on the consent form if you are happy to and then sign at the bottom, I will also sign. We need to do two copies, one for you and one for me.
(If skype/phone “I will complete, copy and send you one to keep”)
- OK – now we can get started. If at any point you want the interview to stop please just let me know. Just a reminder that I am recording this interview and am switching the recorder on now.
- SWITCH RECORDER ON
- Happy to start? – OK. Please could you tell me about yourself
your profession and occupational role,
how long have you been working in this role?
- How did you find out about personal genomics tests bought by the public online?
- Tell me something about your understanding of these tests and your initial thoughts about them.

1

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

(this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Clinicians’ interview prompts

How many patients have you seen about their DTCGT? Over what sort of timeframe, when was the first, and the most recent?

- Why were they referred to your service?
- Tell me about your experience of counselling people who have tested.
- Did/do you feel adequately prepared for counselling this group of people?
- In your view, did they seem to understand the information the tests actually convey?
- What was your assessment of the impact on the individual and their family of DTCGT?
Positive, negative?
- DTCGT could be viewed as providing a different model of DNA testing from clinical genetics services. What are your views about the differences?

What are your thoughts about informed consent in relation to DTCGT?

- What are your thoughts about the lack of counselling in relation to DTCGT, thinking about those who are not referred for counselling, or don’t take it up.
- What are your thoughts about confidentiality in relation to DTCGT and individuals’ data and samples?
- What are your thoughts about regulation of DTCGT?
(Should it be regulated, to what extent, if so by whom, or should it be banned altogether?)

2

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk

Cardiff University, School of Social Sciences Research Ethics Committee, Cardiff and Vale University Health Board, and National Institute for Social Care and Health Research

Study Number SREC/826 Date 3/01/2012 Version 2; CVUHB 12/DTD/5455, NISCHR PCU/96331 Date 20/08/2012

(this doc not submitted - post approval)

“Direct-to-consumer genetic testing: users’ experiences of testing for disease risk and genetics clinicians’ perceptions of its impact on NHS genetics services.”

Clinicians’ interview prompts

- What are your views about patenting in genetics? For example the recent 23andMe patent for analysing SNPs associated with PD?
- Do you have a sense that uptake of DTCGT is different in the UK from elsewhere? Why do you think that is?
- What are your thoughts or impressions about the implications of DTCGT for users (in the UK)
- What are your thoughts about the potential impact or burden of counselling this group might be for the NHS?
- What are your views about primary care HCP’s understanding of, and preparation for, DTCGT in particular and genomics in general?
- Is there anything else you would like to tell me in relation to your experiences of DTCGT?
- Is there anything I haven’t asked you about that you were expecting?

Would you like to be sent a copy of the summary of the results when the study is complete?

If yes - where shall I send them?

Thank you so much for your help with the research and your participation in this interview – I really appreciate it.

3

Principal Investigator: Teresa Finlay RN, MSc; PhD Student; finlaytm@cardiff.ac.uk

Supervisors: Prof A Hedgecoe hedgecoeam@cardiff.ac.uk

Dr M Arribas-Ayllon arribas-ayllonm@cardiff.ac.uk