Testing the NHS: the tensions between personalised and collective medicine produced by personal genomics in the UK

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Key words

DTCGT, NHS, genomics, personalised medicine.

Abstract

This study examines direct-to-consumer genetic testing (DTCGT) in the UK using the Social Construction of Technology (SCOT) framework to draw conclusions about how commercial genotyping is being shaped by principal groups involved with the technology. Different tests are available including single nucleotide polymorphism (SNP) genotyping for ancestry and health information, the latter being the focus of this study. I conducted interviews with DTCGT users and genetics clinicians who had either been consulted about DTCGT or who were willing to discuss their views. This paper is about one of the study's three themes, the NHS. DTCGT seemed to provide a point of focus for conflict between personalised and collective medicine in the UK, rather than being its cause. This suggests that DTCGT is more likely to achieve stability by virtue of being superseded by new technologies, rather than by achieving closure.

Introduction

DTCGT has been available for purchase on the Internet for ten years. The DTCGT market offers numerous types of test; the focus for this project is the testing that claims to provide information about individual risk for common complex diseases. These tests are often sold as part of a genome-wide scan

of hundreds of thousands of single nucleotide polymorphisms (SNPs), also known as SNP genotyping, that provide information on a range of traits including ancestry, physical traits, genetic disease, carrier status and pharmacogenomics as well as common complex disease risk (Leachman et al 2011).

Facilitated by convergent developments in biotechnology, information technology and the Internet (Wright et al 2011), DTCGT capitalises on neoliberal policies that emphasise individual consumerism. The effect of these policies on health care has been to devolve responsibility for health to individuals resulting in a group of autonomous consumers of healthcare services. Individuals are being encouraged to exercise accountability by managing their health and the risks to it, which DTCGT marketing capitalises on (Tutton and Prainsack 2011). Knowledge that was previously the preserve of genetics clinicians, imparted in controlled clinical environments, has been democratised and made available to the public without recourse to the medical establishment (Borry et al 2010). Companies' marketing tactics employ tropes of personalisation to emphasise the advantages of information for making life-style and health choices in an autonomous, confidential manner (Arribas-Ayllon et al 2011). However, despite companies' astute marketing there are powerful counterclaims about the accuracy of the science on which genotyping is based (Janssens et al 2008). There are also concerns about the public's ability to understand the risk information they are sold without genetic counselling (Collins et al 2011). In addition health professionals' accountability to patients is challenged by their misgivings about the utility and potential harm that the information from commercial SNP genotyping could cause (Kraft and Hunter 2009). DTCGT is particularly problematic in this regard because of the capacity of genetic knowledge to cause harm to the individual and also to their relatives. So whilst the privacy and opportunity to exercise autonomy associated with DTCGT are presented

as positive attributes by some (Juengst et al 2012), the potential consequences of any breach of confidentiality could be severe (Finlay et al 2016). These potentially divergent views about DTCGT represent the kind of challenge to society's moral order that formed the basis of the calls for regulation of commercial genomics and its lack of stabilisation as a technology (Evans and Green 2009). Thus DTCGT can be seen as a contested technology, which this study sought to examine in the UK context.

Background

Early research into DTCGT mostly comprised analyses of website material or surveys as to who *might* engage with DTCGT or how they might interpret results, rather than directly investigating the impact of DTCGT *per se*. Website analyses by Lachance et al (2010) and Singleton et al (2012) found a predictable bias towards positioning the benefits of testing, rather than a balanced presentation of advantages and disadvantages or limitations of testing. Content was variable within and across different sites, which could appear confusing and difficult to understand for some. Saukko et al (2010) undertook a qualitative analysis of nutrigenomic testing websites. Nutrigenomics companies sell supplements on the basis of genotyping a more limited range of SNPs thought to relate to metabolism rather than common complex disease risk, but this study's findings appear applicable to the wider commercial genomics market. The study is noteworthy because the authors suggested that the boundary work performed by regulators and researchers to separate medical testing from commercial genomics is circumvented by companies who legitimise their products by positioning them as lifestyle products. Whilst companies selling SNP genotyping for common complex disease risk, ancestry and physical characteristics are not using testing to sell other products, their tropes of empowerment to enable

positive lifestyle choices seem similarly designed. This is highlighted in an analysis of the discourse on websites for the three main personal genomics companies in 2007-2010, deCODEme, Navigenics and 23andMe. Whilst there were differences between all three websites, consumer empowerment was a common trope, suggesting that personalised health information could facilitate healthier lifestyle choices and thus reduce disease risk. The authors suggest that marketing rhetoric promising greater self-knowledge to alleviate future risk manages to obscure the uncertainty associated with SNP genotyping data (Arribas Ayllon et al 2011).

Early survey research investigated what motivated people to consider testing, how they might interpret test results and whether information about the risks and limitations of testing would influence their decisions. Women with a history or family history of breast or ovarian cancer were found to choose testing less frequently when the risks of testing were included in mock websites. These findings suggest that the positive framing of DTCGT websites, as illustrated by the website analysis studies cited above, may attract the unwary with a concern about their health history (Gray et al 2009). McGuire et al (2009) surveyed Facebook users in the United States of America (USA) (though not through Facebook) about DTCGT. Almost half of the cohort was aware of DTCGT, although fewer than 10% had tested. However, two thirds of respondents suggested that they would consider testing and a third indicated that they equated testing with diagnostic information. Whilst an indication does not equate to action, these results prompted the authors to conclude that healthcare services needed to prepare to be consulted by DTCGT users seeking advice about test results. Adding to concerns about this increased demand on health services, a UK-based survey of people on the TwinsUK register established that, while a

much smaller proportion of survey respondents in the UK claimed to be aware of DTCGT, enough would consider testing (depending on the price) to prompt similar warnings about the impact on healthcare providers (Cherkas et al 2010). A survey by Leighton et al (2012), which compared Facebook users' interpretation of mock test results with those of genetics counsellors in the USA, found that whilst the Facebook users found the results easy to understand, they interpreted their meaning differently from the genetics counsellors. The authors suggested that people require support to understand the nature of testing information.

The first published empirical work involving users of DTCGT was McGowan et al's (2010) study in the USA. They found that these early adopters were curious about their genomes, knowledgeable about genetics and did not appear to suffer harm as a result of their test results nor make any significant changes to their lifestyle. These findings were confirmed by later studies of genotyping test users including a mixed methods study of early adopters in Switzerland (Vayena et al 2012). More recently the longitudinal PGen study conducted by George Church's group in Boston surveyed users of DTCGT (Carere et al 2016). They found that this cohort's understanding of genetics was also relatively high before and after testing but their confidence in applying their genetics knowledge was reduced after testing, possibly as a result of their experience of receiving results. However, in line with studies outlined above, users did not report any increased anxiety after testing.

However, despite the considerable literature about DTCGT published by scientific genetics, medical, ethics, law and sociology communities, at the time of commencement of this study the experience of DTCGT in the United

Kingdom (UK) represented a metaphorical “black box” (Lynch et al 2010, Bijker et al 2012). Accordingly I chose to investigate DTCGT in the UK context using Pinch and Bijker’s Social Construction of Technology (SCOT) model. The choice of framework is deliberate, as my intention is to shed light on the arguments and contested nature of DTCGT, specifically examining relevant social actors’ experiences within the wider context of the UK’s National Health Service, as I shall explain.

Using SCOT to examine DTCGT in the UK healthcare context

Rather than simply viewing SNP genotyping as a laboratory test, the competing claims and discourses it provokes indicate that DTCGT warrants analysis as an assemblage that is being socially constructed and shaped by the discourses and actions of the actors involved with it. In this context discourses are understood as practices of sharing and developing beliefs and understanding through discussion, evaluation and interpretation in order to produce knowledge about the technology (Lessa 2006). In SCOT these principles are applied to

- relevant social groups of actors, in this case test users resident in the UK and NHS genetics clinicians
- the wider social context, in this case the NHS
- actors’ interpretative flexibility
- the stabilisation and closure of disputes surrounding the technology (Bijker et al 2012).

In their critical review of SCOT, Klein and Kleinman argue that assuming relevant social groups are homogeneous misses the potential influence that individuals may have on technology owing to their political, economic or professional influence (Klein and Kleinman 2002). They advocate for more

detailed analysis of the constituent members of relevant social groups whilst also paying attention to the wider context for a more comprehensive analysis. Accordingly I shall describe the characteristics of participants in the users' and genetics clinicians' groups in the methods section.

The use of SCOT illustrates the influence of human intervention on SNP genotyping in the UK, which is crucial to challenge deterministic views. A technologically deterministic position could absolve people from responsibility in their engagement with the technology (Wyatt 2008), while a bioethical one could assume a paternalistic interpretation, which risks interpreting findings in a non-symmetrical manner. Given the potential impact of genetic information on people and their social groups, an exploration of the social construction of this technology seems to provide a germane approach to understanding the impact of DTCGT in the UK.

Thus the wider social context of the UK and, more specifically, the NHS is central. From its inception in 1948 the NHS has provided care free at the point of access to UK citizens on the basis of a collective medical model. The medical profession has influenced NHS patients' access to health interventions relying on patients to trust that this is decided in their best interests. This has increasingly translated into rationing and policy shifts associated with economising and the public taking responsibility for their health to discourage dependency on the NHS (Department of Health 2010, Klein 2013). Healthcare professionals' gatekeeping role and formerly sacrosanct expertise is arguably challenged by people's direct access to health information, altering trust-based professional-patient relationship (Tutton 2014). The original collective medical model is shifting from what

Klein calls the paternalistic “church”, where care is dispensed to a trusting public, to the consumerist “garage” to which people drop in and take what they want (Klein 2013: 306). Thus the NHS’ iconic status in British culture, representing democracy and the welfare state (Busby and Martin 2006), is increasingly challenged by demands from medical innovation, societal expectation for personalisation and reduced funding owing to political incentives for austerity (Bambra 2013).

Genomics exemplifies the individual, particularly in relation to human genomics, which has provided DTCGT companies with the basis for focusing their marketing on personalisation and individual responsibility. This aligns with the simultaneous globalisation of neoliberal ideas including emphasis on individual choice and consumerism, tropes that DTCGT companies foreground (Arribas Ayllon et al 2011). This polarisation of collective and individual is considered by Donna Dickenson. She suggests the DTCGT companies’ tropes of personalisation in the name of empowerment and individual responsibility for health undermines collective medicine in an increasingly commercial domain (Dickenson 2013). The contradiction inherent in DTCGT companies’ marketing personalised health information on the basis of population-derived genomic data is central to her argument that commercial genomics is simply profiteering. This is despite the collapse or takeover of many DTCGT companies which suggests failure in that regard otherwise (Vorhaus 2012).

These interdependent but conflicting aspects of collective and individualised medicine are emblematic of this study. Thus the study focuses on users and genetics clinicians as the two relevant social groups influencing DTCGT

within the social context of the NHS. Interpretative flexibility is key for illuminating the influence of different views of technology presented to different audiences, at different times with different intentions and outcomes (Bijker et al 2012). Whilst Williams and Edge (1996) argue that the infinite possibilities that could be proposed are unhelpful in analysing how closure is achieved, analysis of variations in accounting can be particularly valuable when claims are repeatedly disputed (Potter and Mulkay 1985). The concept is thus important for this study, as inconsistent and contradictory views are evident in the data.

In order to understand the impact of DTCGT in the UK I collected and analysed the stories, experiences and thoughts of two relevant social groups involved with the technology in the UK. The findings discussed in this paper focus on the tension between personalised and collective medicine that DTCGT creates, one of the three themes evident in the study's data.

Methods

DTCGT was relatively novel and un-researched in the UK context at the time this study commenced. The SCOT framework was chosen to illuminate how the relevant social groups of test users and genetic clinicians engage with DTCGT in different and competing ways for social, economic, professional and regulatory reasons. It was obvious to involve test users resident in the UK as one of the relevant social groups, owing to their direct experiential knowledge of DTCGT and the potential for them to seek help from NHS genetics services. Literature related to DTCGT suggested that genetics clinicians would have influential views and shared understandings about DTCGT owing to their clinical specialism. These were likely to contrast with

views of users' or producers' of the technology so these practitioners were involved as the other relevant social group for the study. In addition, with their status as experts in clinical genetics, they would be most likely to be called upon to interpret results and provide counselling in the event of any concern. Both groups have interests in DTCGT technology and each shares experience and knowledge of DTCGT, which Bijker refers to as a "technological frame" (Bijker 2010:69). But their conflicting interpretations and knowledge claims provide a suitable starting point for examining the influence of their interests on the technology (Bartley 1990).

The other group of healthcare professionals that could be relevant in the UK context is General Practitioners (GPs); their role as gatekeepers for specialist consultations in the UK (Louden 2008) indicates that they are likely to be users' first port of call for help with interpretation of health test information. However, as the literature indicates lack of understanding of genomics by most doctors, genetics clinicians presented a more relevant group, given their experience of advising and supporting people in relation to genetic conditions (Edleman and Eng 2009, Salari 2009).

With approvals from Research Ethics and Research and Development committees in place, I sought participants for the study. To be eligible to participate, users had to be an adult UK resident and have purchased DTCGT that included health information. This purposeful sampling was intended to facilitate discussion about their engagement with the technology and their views on the relationship of the health-related aspects to their health care in the NHS context. I purposely used the Internet to recruit users. At the time of the study personal genotyping was largely marketed and sold

online; I hoped that test users would be conversant with finding information about genomics online and would come across invitations to be involved in research. However, unlike genetics clinicians whose geographical locations could be easily established, DTCGT users were people who Brown and Webster describe as being without embodiment (Brown and Webster 2004). This is due to their identities as DTCGT users being linked to their online personas rather than their physical, embodied ones. I thus anticipated having to use several approaches to recruit participants to this group.

Recruitment of users was surprisingly easy however. A webpage describing the study (now removed) and a blog post on “genomesunzipped”, a website providing independent, expert commentary on commercial genomics requested volunteers (http://www.genomesunzipped.org/ n.d.). These Internet sites were quickly located by most of the user participants. Members of the UK branch of the International Society of Genetic Genealogists (ISOGG) discovered the study website within hours of it going live. Some volunteered for the study or informed others about it. Other participants were either biosciences researchers who read the blog post on genomesunzipped or people known to participants, as snowball sampling was also used for recruitment (Atkinson and Flint 2001). Consequently, most of the group of user participants had similar characteristics to the early adopters described in McGowan et al’s study of DTCGT users in the USA (McGowan et al 2010). The early adopter profile of this relevant social group suggests that they are likely to be more informed about the technology and to enthuse others. This was evident from the over-representation of participants who were working in genetic research and curious about DTCGT, or who were avid genetic genealogists. All the users had tested with 23andMe, having purchased a test from the company prior to changes made as a result of

FDA intervention in 2013 (Conley 2013). Some had also tested with other companies for genetic genealogy information (see Table 1 for details). Seventeen interviews were conducted over four months in 2012, two on the telephone and fifteen face-to-face at the participants' choice of location.

Despite their known geographical locations, clinicians were more difficult to access. A number of strategies were used, including emails to British Society of Genetic Medicine Members (BSGM), leafleting delegates at a BSGM conference and contacting regional Genetics Services in the UK to raise awareness of the study. Recruitment initially targeted clinicians who had counselled patients about DTCGT, but was very slow. Subsequently it emerged that this was likely to be due to few referrals for DTCGT, despite concerns that clinical genetics services could be overwhelmed by requests for consultations with the worried well who struggled to interpret their genotyping report (Hogarth et al 2008). So the inclusion criteria were broadened to include clinicians who were willing to discuss their views about DTCGT, whether they had encountered patients who had sought counselling or not. This resulted in 16 interviews with genetics clinicians of various professions over eleven months between 2012 and 2013 at the participants' choice of location. One was on the telephone, fifteen were face-to-face (see Table 2 for a summary of Genetics Clinicians' Information).

Interviews were semi-structured and in-depth, mixing specific and open questions as indicated by the participant's experiences. Questions and prompts covered demographic information, types of testing used or encountered, referrals to discuss testing, people's experiences of testing or counselling testers as well as general exploration of their views about

DTCGT. Interviews were audio-recorded and I transcribed them to familiarise myself with data from the interviews onwards. Interviews commenced after participants had consented to participation including the use of verbatim, de-identified excerpts. To maintain confidentiality, data extracts in this paper are attributed to user participants using synonyms and occupation, but to clinicians using an assigned code that identifies their place in the sequence of clinician participant (CP) interviews and their profession. These different approaches also maintain a distinction between the two groups in the data analysis.

The transcript data were analysed with the principles of impartiality and symmetry in mind. This was deliberate in order to avoid attributing my own views to the participants and to enable me to adopt an objective and symmetrical approach to the data from the two relevant social groups studied here in relation to DTCGT specifically. An iterative approach was taken to the data analysis to elicit the themes within the texts of the interview conversations (Creswell 2013). Repeated reading of the text, making notes about patterns and disparities, and using reflective notes about the interviews informed my thinking with the data. The data were coded inductively with in vivo codes that emerged from the participants' discourse (Ryan and Bernard 2000). The data were then re-read alongside the literature and deductively collected into categories. The final stage of analysis commenced by writing and connecting the categories and literature in a more direct manner. From this perspective I used further reading and writing to interact with the data and explore its categories alongside my thoughts (Coffey and Atkinson 1996). From this iterative process three overarching themes became evident across the participants' data. These were

1. Socialising DNA: the influence of social networks on the participants' technological frame and discourse about DTCGT
2. Personalising DNA: participants' individual ideas about DNA in the context of SNP genotyping and their incorporation into participants' genetic identities or clinical practice
3. Testing the NHS: the problem of expectations for personalised medicine in the context of collective medicine provision

The third theme of personalised medicine and its challenge for the NHS is the focus of this paper.

Findings – Testing the NHS

Public support for the NHS has been unstinting since its inception (Klein 2013). Whilst not surprising, the organisation that the public champions is arguably more simplistic than the complex one in which so many have vested interests and for which there are different visions of its obligations and how to meet them. The two groups in this study volunteered opposing beliefs about personal genomics in the NHS context, illustrating the contested nature of commercial genotyping in this context. There was predictable support for personalised medicine (though not necessarily at NHS expense) from some DTCGT users, while clinicians described support for collective medicine and the need to protect the NHS' scarce resources. Tensions between these two groups focused on responsibility, either for individual health or service provision, and the concepts of empowerment of the public versus professional expertise. Areas of agreement focused on pharmacogenomics and a shared support for the NHS, though these are unlikely to indicate imminent stabilisation of the technology. I shall discuss

each of these sub-themes in turn, using excerpts from the data to illustrate the points made.

Responsibility

Discussion from some test users about personal information and responsibility for health suggests uptake of policy moves towards individuals taking more responsibility for their own health (Department of Health 2010). However, they also indicate adoption of companies' tropes of personalisation and empowerment, which Van Lente suggests is due to technologies becoming fixed in specific language that directs beliefs and behaviour (Van Lente 2000).

Users who referred to the health information from testing in relation to responsibility drew attention to the opportunity for making lifestyle alterations or seeking health screening based on the health information provided by DTCGT. One user had sought medical advice, as her test results suggested an increased risk of cancer (supporting her family history). Similarly, Ian, who does not have a background in genetics or biotechnology, stresses the need for cancer screening.

> "I must get my PSA count done, at my age I should be doing it anyway. I haven't got any symptoms but I should and having got the information [from DTCGT] it's really highlighted it to me that I should. I'm very aware of that but I haven't done anything about it yet …" Ian (retired).

This illustrates personal responsibility for health monitoring, not least perhaps because there is no effective screening programme for prostate cancer. However the value of PSA testing is unclear bringing Ian's assertion into question (Public Health England 2016). Ian stresses his personal responsibility repeatedly. However, his lack of action is in common with other users none of whom described making lasting lifestyle changes or having interventions for disease risk management in light of test results. This supports findings in the study of early DTCGT adopters in the USA and the REVEAL study (McGowan et al 2010, Chilibeck et al 2011). It also refutes concerns about the impact on the NHS of concerned citizens seeking interventions following DTCGT that Dickenson forecasted (Dickenson 2013).

Clinicians' concerns in respect of responsibility were focused on providing genetic counselling and protecting the NHS from the repeatedly cited potential drain on scarce resources by privately-initiated testing (Nuffield Council of Bioethics 2010, Dickenson 2013). The clinicians who had counselled people following DTCGT voiced concerns.

> "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." CP2 (Doctor)

This represents a unique reference to counselling someone in distress following DTCGT in this study. Patients' distress is emphasised, as is their

expectation for counselling, to highlight the moral challenge of juggling finite resources. This is framed in a manner that suggests this occurs more often than evidence suggests, possibly in a similar adoption of prevailing discourse about anticipated pressure on Genetics Services from DTCGT as users adoption of tropes of personalisation and empowerment. Concerns about use of scarce resources being taken up by counselling people who had initiated testing privately were more widely expressed despite little evidence of it. Most inferred that DTCGT was outside the remit of clinical genetics. This nurse defends the NHS responsibility for rationing, pointing to an impasse in balancing personalised and collective medicine.

> "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." CP5 (Nurse)

An experienced doctor, with academic responsibilities in addition to a clinical role, expressed a more equivocal view. Arguments for both collective and personalised medicine are presented, possibly due to a strategic view of genomic technologies' potential diagnostically and for NHS revenue generation through developing commercial biotechnology opportunities.

> "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 healthcare which is where it's now capable of moving. So I kind of sit in the optimistic let's have a go camp." CP10 (Doctor)

The explicit references to both sides of the responsibility debate and the interpretative flexibility demonstrated in recognising that they are not mutually exclusive suggests a vision that supports genomic technologies generally rather than dismissing DTCGT. This locates DTCGT within the wider context of genomic technologies including Genome England's 100,000 Genomes Project and increasing use of genome sequencing in "mainstream medicine", which are being forecast to shift genetics clinicians roles towards more educational and advisory responsibilities (www.genomicsengland.co.uk n.d., AGNC 2014:1, Clayton-Smith et al 2015).

The suggestion that genomic information could be empowering depends on understanding genomics and its role in contributing to people's health

information. This could be interpreted as having expertise in genomics; the issue of how user and clinician participants equate this to clinical genetics expertise is explored next.

Empowerment or Expertise

Those of the study's group of users with a primary interest in health results and genealogy expressed support for personalised health information commensurate with what Bunnik et al refer to as the personal utility of DTCGT (Bunnik et al 2015). Given NHS policies highlighting the importance of patient engagement and empowerment, people's pursuit of health information from personal genomics is perhaps unsurprising. However, this has fuelled criticisms of healthcare professionals dismissive of patients' efforts, owing to their paternalistic approach and ignorance of contemporary genotyping technologies (Jordens et al 2009, Bunnik et al 2015). Several users in this study were among them, voicing strong opinions about their perceptions of deficits in NHS staff knowledge and service provision. Their technological frame of DTCGT is that it provides important personal health information. It is worth remembering that at least half the group of users in the study have occupational experience of genetics, whilst those who are genealogists have experiential expertise in genotyping, having purchased more than one test. Whilst the group of clinicians all have the credentialed clinical genetics expertise that Collins and Evans describe, their experiential expertise of SNP genotyping was arguably not comparable to the users' (Collins and Evans 2007). However their technological frame of DTCGT is of technology based on contingent science, one referring to it as "snake oil". Not all were disparaging, but their view of DTCGT as providing meaningless information sets clinicians in opposition to those users whose technological frame of DTCGT saw it as empowering.

Several users voiced scepticism about doctors' understanding of DTCGT, describing dismissive attitudes. This supports assertions that the democratisation of genomic information could erode trust between patients and healthcare professionals (McGuire and Burke 2008, Tutton 2014). The following excerpt is from a user who works in bioinformatics and had tested partly due to her curiosity about genomics as well as personal and familial health issues that she wished to investigate. This seems to have been driven by her need for autonomy so that she could access and consider her health information independently.

> "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 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 (Biologist)

Despite her self-knowledge and understanding of biology, Laura's emphatic statement suggests that medical hegemony leaves her disempowered. The promise of personalisation and empowerment offered by DTCGT is attractive partly because it circumvents conventional medical power and gatekeeping, bolstering her sense of agency. However, her observation about medicine's lack of integration of genomics to explain the NHS' inability to help her appears to justify her use of DTCGT. Here, her flexible interpretation of genomics arguably equates DTCGT with sequencing and invests genomics with diagnostic potential that has yet to be realised (Tutton 2014).

In contrast, clinicians questioned the value of SNP genotyping and some view public understanding of genetics with a deficit model compared to their clinical expertise. One genetic counsellor and a doctor implicitly position clinicians as having expertise.

> "People access tests for very different reasons and unfortunately I think the public do think it's CSI." CP1 (Scientist)

This counsellor's experience of patients' understanding of genomics is equated with a popular television crime drama. The suggestion is that fictional representations of genetics are absorbed and repositioned as truth by people consulting clinical genetics services. Here the contingent nature of

the scientific basis for DTCGT could be equated to fictional representations of science implying that clinical genetics expertise is needed to explain genetic information. Another clinician's experiences also suggested that people view genetics deterministically.

> "I think that people often start off in a position where genetics is really something very important and very deterministic. Umm, 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" CP4 (Doctor)

This view invests the majority of British society with deterministic understanding of genetics on the basis of this doctor's experience, the implication being that DTCGT users could be misled and caused needless harm by DTCGT results. Indeed Harris et al's argument that deterministic interpretation of genetic information is supported when accessed through the Internet strengthens this concern (Harris et al 2016). However, neither of these extracts indicate consideration for people represented by some users in this study who do have genomics knowledge and who felt they were able to understand their test results, nor the finding that none of the users, whatever their interpretation of DTCGT, reported experiencing significant or lasting concerns from testing.

While some of these users' views suggest a utopian view of personalised medicine that they criticise doctors for shunning, the clinicians' support for collective medicine necessitates a different view of the utility of personal

genomics. Juxtaposing these views results in on-going conflict between these relevant groups about the technological frame of DTCGT as well as having repercussions for the relationship of trust between patients and healthcare professionals as predicted (McGuire and Burke 2008, Wyatt et al 2013). Brown and Webster suggest that where trust fails, healthcare practice is unfeasible and, whilst this point does not appear to have been reached by these participants, these excerpts do point to a shift in the clinician-patient relationship to a more distrusting position or at least a less readily assumed trust (Brown and Webster 2004). However, while SNP genotyping for common complex diseases has the potential to disrupt trust between patients and practitioners and fuel the on-going disputes about personal genomic technology, there are aspects on which participants in both groups shared common opinions, which I shall complete the discussion with.

Common ground

The data in this study suggested that some of the participants in both groups shared similar views about two areas relating to DTCGT in the NHS context. Firstly, there were shared understandings about the value and significance of the NHS both as the country's principal healthcare provision and as an institution strongly associated with the UK's national identity, endorsing Klein's assertion that the NHS is a "national treasure" (Klein 2013:vi). Secondly, there were similar views among some participants in both groups about DTCGT's pharmacogenomics information.

Support for the NHS was evident in some participants' references to examples from their practice or experience as users of the NHS to illustrate a shared perception of people's preferences for its services over seeking

healthcare information independently. Clinicians referred to people's faith in the NHS based on their experiences as healthcare professionals. Whilst this may seem to be contradictory to the earlier suggestion that DTCGT users' trust in the NHS is being challenged by personalised medicine, not all users expressed these views; some users were more equable. One genetics researcher was eager to separate her self-professed "narcissistic curiosity" in pursuing DTCGT from the NHS' responsibilities.

> ""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 (Geneticist)

The commonly described recreational aspect of personal genomics is often a motivation for testing, driven by people's curiosity (Prainsack et al 2008). As with many other geneticists and genealogist users in this study, Maria viewed her test as satisfying her curiosity about her DNA rather than related to health care or substituting any NHS responsibility for her health. Her reference to the NHS' lack of perfection suggests an acknowledgement that, despite its dissonance with the consumerism of personalised medicine, the collective medical model is acceptable and widely supported by the public because of its centrality to British national identity (Gershlick et al 2015).

The second aspect of shared views about SNP genotyping was more surprising, being about pharmacogenomics, the information from SNPs that

suggest variance in pharmacodynamics or kinetics (the physiological processes affected by drugs or those which metabolise them respectively). This could be seen as fortunate, given the dependence of patients on qualified healthcare professionals for prescribing of medicines. However, the contingent nature of the science underpinning disease-risk information from SNP genotyping is equally applicable to pharmacogenomics and arguably has more serious implications if prescribing decisions are made on the basis of its assumed utility.

Richard Tutton's exploration of the historical context of personalised medicine refers to the rhetorical devices used to promote the compelling idea that pharmacogenomics will result in drug treatments designed for individuals rather than populations (Tutton 2012). The expectation that genomics will deliver individually designed drug regimens in a new molecular iteration of personalised medicine has not been realised. It has resulted in the concept being modified along more achievable lines, such as stratified medicine where people are assigned to groups of treatment according to genetic differences (Tutton 2014). In light of this it is significant that some participants in both the relevant social groups being studied spoke positively about the potential of pharmacogenomics to improve health care. The following extracts from one user of DTCGT and one clinician illustrate this point. The first is from a researcher in public health who envisages negotiating her future drug therapy with her GP.

> "On my genetic thing it says there's a sensitivity to Metformin. I obviously get it from my mother [who reacted badly to it] 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 (Public Health researcher)

Carol's suggestion about her future treatment envisages a confrontational consultation and prescription without any discussion or consideration of concordance. Her refusal of it based on her pharmacogenetics SNP analysis illustrates her use of interpretative flexibility in relation to DTCGT. She had earlier dismissed results relating to disease risk because of the inconclusive nature of SNP genotyping but seems to be more deterministic about the pharmacogenetics information. Carol's justification for this on the basis of her DTCGT results ignores her family history of intolerance of this drug. Family history is thought to be more reliable than SNP genotyping in health information and is probably contributing to her thinking in this regard, though here she chooses to focus on her DTCGT results.

Some clinicians also demonstrated interpretative flexibility in referring to pharmacogenomics positively, having dismissed its utility for common complex disease risk information. One doctor gave a colourful vision of his ideas for anticoagulant prescribing on the basis of SNP genotyping.

> "I can see the value in these types of things; you know Warfarin, [prescribing] Warfarin's a black art. When I worked on the wards I was told, "Start this guy on Warfarin. Prescribe him 10 [mg] today, 10 tomorrow and 5 the day after that and test his INR [blood clotting time]." And you'd test their INR and some guy's INR hadn't shifted from baseline so you'd whack them another 10 and some little old

> woman was ready to bleed out all over the ward because her blood was so thin, it was like dilute orangeade. So it was a guess, but now we're getting a handle on that. A little bedside SNP so we'd know how to prescribe, and then start them on Warfarin. But that's not predicting a genetic disease; we need this information to look at how to dose you." CP13 (Doctor)

Titrating this risky anticoagulant medication within the narrow margins of safe efficacy indicates the clinical challenge involved and its associated stress. Using interpretative flexibility this doctor imagines a solution to that challenge. However, this potential clinical application for SNP genotyping in the NHS is not so far supported by research. Genotyping patients for Warfarin metabolism and prescribing dosage accordingly does not appear to make any significant difference to the efficacy or safety of this difficult and potentially dangerous treatment (Stergiopoulos and Brown 2014). This casts doubt on the efficacy of point-of-care SNP genotyping for Warfarin dosing, supporting concerns about clinicians' lack of knowledge in this regard (UK Pharmacogenetics Study Group 2006).

Participants' mutual support for pharmacogenomics could represent a point from which the relevant social groups involved in DTCGT in the UK could negotiate socially acceptable aspects of genomics technology in the NHS in future. This could provide an opportunity for stabilisation of the technology, as Hedgecoe and Martin (2003) argued in relation to pharmacogenomics. However, the likelihood of this is small and its foundations are tenuous in the extreme, given the contested basis of personalised medicine in the context of SNP genotyping in particular, as these findings show.

Conclusion

Findings from this study suggest that DTCGT is a less disruptive technology in the UK context than early concerns forecast. This is evidenced firstly by the study's group of users' lack of concern or uptake of interventions on the basis of test results. Secondly, the lack of impact on NHS clinical genetics services is demonstrated by difficulties recruiting genetics clinicians with experience of DTCGT to the study and participants' reports of few enquiries about post-DTCGT counselling. However, this should not obscure the conflict between some of the participants in relation to the NHS context, which focused on the tension between personalised and collective medicine, responsibility and expertise.

The issue of who should take responsibility for individuals' health is being exposed by DTCGT and, as Klein suggests, the NHS is currently in an ambiguous position in this regard (Klein 2013). The concepts of personalisation and consumerism and the associated expectation of public responsibility are particularly challenging for the NHS in times of austerity. Arguably this is the predictable outcome of health policy that has increasingly emphasised health promotion, individual responsibility and choice, rhetorically at least (Klein 2013). Thus it is not unreasonable of people to avail themselves of opportunities to obtain health information and to feel aggrieved when their efforts are dismissed by healthcare professionals. Equally understandable are healthcare professionals' frustrations about the lack of transparency of commercial health information marketing and the significant restraints within which they are expected to provide services to an increasingly demanding public, some of whom appear to show little genuine engagement with

responsibility for their health. This lack of engagement is despite adopting the language of commercial genomics companies in relation to DTCGT and using interpretative flexibility to assume belief in the promise of DTCGT technology to provide personalised health information.

Users' experiential expertise is important in considering their level of engagement and understanding of what their genomic data is communicating. Most users in this study have knowledge of genetics from personal research or education, and have spent considerable time immersed in learning about personal genomics either independently or through their occupation. In addition their networking activities have facilitated the dissemination and development of what has been referred to as "technical know-how" (Evans and Plows 2007:836). Users' experiences of DTCGT could be referred to as lay-expertise, in a manner similar to studies by Kerr et al about the public and their knowledge of genetics, Prior's work on lay-expertise in medical sociology and Epstein's work with HIV-AIDS activists (Epstein 1995, Kerr et al 1998, Prior 2003). However, as these authors all indicate, 'lay-expertise' belies the complexity inherent in who lay actors are and what constitutes their expertise, as exemplified by the heterogeneous nature of this study's group of users. In addition, investing users with expertise could be questioned in light of the interpretative flexibility demonstrated by some when extolling the virtues of their DTCGT information. Whilst their expertise in clinical genetics both unites the clinicians and informed their inclusion as a relevant social group for this study, their expertise in personal genomics is also questionable. Although credentialed, contributory experts in clinical genetics, most clinicians' experience and expertise relates to chromosomal abnormalities and genetic mutations

rather than SNP genotyping. Some are well versed in the literature relating to genomics and are able to cut through users' apparent experiential expertise by critiquing the scientific basis of testing, the panels used for SNP analysis and the lack of inclusion of wider genetic anomalies related to specific conditions. However, they are mostly not engaged with the public who are buying DTCGT. Whether expert or not, understanding of personal genomics influences participants' technological frames of DTCGT, setting many of them apart into those who support the personal utility of DTCGT and those who dismiss it as lacking utility and a threat to collective medicine.

Analysing the views of users and genetic clinicians about DTCGT and the challenges for the NHS suggests that the potential for SNP genotyping technology to achieve stability in a healthcare context in the UK is probably remote. While the relevant social groups in this study may suggest that DTCGT is a focus for the on-going disputes around personalised versus collective medicine in the UK, I would argue that DTCGT *per se* is not the cause of that dispute. Rather it is the vehicle for participants' expressions of concern as they are confronted with challenges to their respective perceptions of responsibility for health. Shared appreciation of the NHS was evident in both groups and some clinicians' interpretative flexibility in relation to pharmacogenomics led to common visions with some users for its future potential. However, I would argue that these aspects of common ground are unlikely in themselves to lead to a resolution of the disputes about DTCGT in the near future, given the generally irreducible nature of the differences between these two groups' views about responsibility and empowerment. It is more likely that the technology will be stabilised by virtue of being

superseded by new problems such as whole genome sequencing and expectations of the information it will yield.

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References

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

AGNC (2014) *The Future Role of Genetic Counsellors in Genomic Healthcare.* Association of Genetic Nurses and Counsellors. [Online] http://www.agnc.org.uk/media/925792/agnc_vision_statement_final.pdf accessed 13/04/2017

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 4/04/2016

Bambra C (2013) All in it together? Health inequalities, austerity and the 'great recession' IN Wood C (Ed) *Health in Austerity*. Demos, London [Online] https://www.demos.co.uk/files/DEMOS_Health_in_Austerity_-_web.pdf?1379898927 accessed 4/04/2017

Bartley M (1990) Do we need a strong programme in medical sociology? *Sociology of Health and Illness* 12(4): 371-390

Bijker WE (2010) How is technology made? That is the question! *Cambridge Journal of Economics* 34: 63-76

Bijker WE, Hughes TP, Pinch T (2012) *The Social Construction of Technological Systems: New Directions in the Sociology and History of Technology* (Anniversary Edn). MIT Press, Cambridge MA.

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 4/09/2016

Brown N, Webster A. (2004) *New Medical Technologies and Society: Re-ordering Life*. Polity Press, Cambridge.

Bunnik EM, Janssens JW, Schermer MHN (2015) Personal utility in genomic testing: is there such a thing? *Journal of Medical Ethics* 41(4): 322-326. doi:10.1136/medethics-2013-101887

Carere DA, Kraft P, Kapphingst KA, Roberts JS, Green RC (2016) Consumers report lower confidence in their genetics knowledge following direct-to-consumer personal genomic testing. *Genetics in Medicine* 18(1): 65-72. doi:10.1038/gim.2015.34

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): e13473. doi:10.1371/journal.pone.0013473

Chilibeck G, Lock M, Sehdev M (2011) Postgenomics, uncertain futures and the familiarization of susceptibility genes. *Social Science and Medicine* 72: 1768-1775. doi: 10.1016/j.socscimed.2010.01.053

Clayton-Smith J, Newbury-Ecob R, Cook J, Greenhalgh L (2015) *The Evolving Role of the Clinical Geneticist*. Clinical Genetics Society. [Online] http://www.bsgm.org.uk/media/956131/theevolvingroleoftheclinicalgeneticist_29.07.15.pdf accessed 13/04/2017

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

Collins H, Evans R (2007) *Rethinking Expertise*. University of Chicago Press, London.

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. doi:10.1097/GIM.0b013e3181f710ca

Conley J (2013) 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 4/09/2016

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

Department of Health (2010) *Healthy Lives, Healthy People: Our Strategy for Public Health in England*. Department of Health, The Stationery Office. [Online] https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/216096/dh_127424.pdf accessed 15/03/2017

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

Edleman E and 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

Evans JP and Green RC (2009) Direct-to-consumer genetic testing: avoiding a culture war. *Genetics in Medicine* 11(8): 568-569. doi:10.1097/GIM.0b013e3181afbaed

Evans R, Plows A (2007) Listening without prejudice? Rediscovering the value of the disinterested citizen. *Social Studies of Science* 37(6): 827-853

Finlay T, Gibson S, Koch L, Tocchetti S (2016) Personal genomics: transparent to whom? IN Dreyer M, Erdmann J, Rehmann-Sutter C (Eds) *Genetic Transparency: Ethical and Social Implications of Next Generation Human Genomics and Genetic Medicine*. Brill Rodopi, Amsterdam.

Freidson E (1988) *Profession of Medicine: A Study of the Sociology of Applied Knowledge*. Chigaco University Press, Chicago

genomesunzipped (no date) *genomesunzipped: public personal genomics* [Online] http://genomesunzipped.org/about 13/04/2014

Genomics England (no date) *The 100,000 Genomes Project*. [Online] https://www.genomicsengland.co.uk/ accessed 13/04/2017

Gershlick B, Charlesworth A, Taylor E (2015) Public Attitudes to the NHS. The Health Foundation, London. [Online] http://www.health.org.uk/sites/health/files/PublicAttitudesToTheNHS.pdf accessed 30/03/2017

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): 1303-1311. doi:10.1158/1055-9965.EPI-08-0825

Harris A, Kelly S, Wyatt S (2016) *Cybergenetics: Health Genetics and New Media*. Routledge, Abingdon.

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

Hogarth S, Javitt G, Melzer D (2008) the current landscape for direct-to-consumer genetic testing: legal, ethical and policy issues. *Annual Review of Genomics and Human Genetics* 9: 161-182 doi:10.1146/annurev.genom.9.081307.164319

Hood L, Friend SH (2011) Predictive, personalized, preventive, participatory (P4) cancer medicine. *Nature Reviews: Clinical Oncology* 8(3): 184–187 doi:10.1038/nrclinonc.2010.227

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. doi: 10.1016/j.ajhg.2007.12.020

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. doi: 10.1080/15265160902874411

Juengst ET, Flatt MA, Settersten RA (2012) Personalized genomic medicine and the rhetoric of empowerment. *Hastings Centre Report* 42(5): 34-40. doi: 10.1002/hast.65

Kerr A, Cunningham-Burley S, Amos A (1998) The new genetics and health: mobilizing lay expertise. *Public Understanding of Science* 7: 41-60. doi: 10.1177/096366259800700104

Klein HK, Kleinman DL (2002) The social construction of technology: structural considerations. *Science, Technology and Human Values* 27(1): 28-52

Klein R (2013) *The New Politics of the NHS: from Creation to Reinvention* (7th Edition). Radcliffe Publishing, Abingdon

Kraft P, Hunter DJ (2009) Genetic risk prediction – are we there yet? *New England Journal of Medicine* 360(17): 1701-1703.

Lachance CR, Erby LAH, Ford BM, Allen VC, Kaphingst KA (2010) Informational content, literacy demands, and usability of websites offering health-related genetic tests directly to consumers. *Genetics in Medicine* 12(5): 304-312. doi: 10.1097/GIM.0b013e3181dbd8b2

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

Lessa I (2006) Discursive struggles with social welfare: Restaging teen motherhood. *British Journal of Social Work* 36(2): 283-298. doi: 10.1093/bjsw/bch256

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. doi: 10.1159/000327159

Louden I (2008) The principle of referral: the gatekeeping role of the GP. *British Journal of General Practice* 58(547): 128-130. doi: 10.3399/bjgp08X277113

Lynch M, Cole SA, McNally R, Jordan K (2010) *Truth Machine: the Contentious History of DNA Fingerprinting*. University of Chicago Press, Chicago

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. doi: 10.1080/14636778.2010.507485

McGuire AL and 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. doi: doi:10.1001/jama.2008.803

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. doi: 10.1080/15265160902928209

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 4/04/2017

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, Reardon J, Hindmarsh R, Gottweis H, Naue U, Lunshof JE (2008) Personal genomes: Misdirected precaution. *Nature* 456: 34-35 doi:10.1038/456034a

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. doi: 10.1111/1467-9566.00339

Public Health England (2016) *Prostate Cancer Risk Management: Programme Benefits and Risks of PSA Testing*. Public Health England Gov.UK [Online] https://www.gov.uk/government/publications/prostate-cancer-risk-management-programme-psa-test-benefits-and-risks/prostate-cancer-risk-management-programme-pcrmp-benefits-and-risks-of-psa-testing#references accessed 15/04/2017

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

Salari K (2009) The dawning era of personalised medicine exposes a gap in medical education. *PLoS Medicine* 6(8) [Online] http://www.plosmedicine.org/article/info%3Adoi%2F10.1371%2Fjournal.pmed.1000138 accessed 4/04/2017

Saukko PM, Reed M, Britten N, Hogarth S (2010) Negotiating the boundary between medicine and consumer culture: online marketing of nutrigenomic tests. *Social Science and Medicine* 70(5): 744-753. doi:10.1016/j.socscimed.2009.10.066

Singleton A, Erby LH, Foisie KV, Kaphingst KA (2012) Informed choice in direct-to-consumer genetic tests (DTCGT) websites: a content analysis of benefits, risks and limitations. *Journal of Genetic Counseling* 21: 433-439 doi: 10.1007/s10897-011-9474-6

Stergiopoulos K, Brown DL (2014) Genotype-guided versus 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. doi: 10.1001/jamainternmed.2014.2368

Tutton R (2012) Personalizing medicine: futures present and past. *Social Science and Medicine* 75(10): 1721-1728. doi: 10.1016/j.socscimed.2012.07.031

Tutton R (2014) *Genomics and the Reimagining of Personalized Medicine.* Ashgate, Farnham.

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. doi: 10.1111/j.1467-9566.2011.01348.x

UK Pharmacogenetics Study Group (2006) *Policy Issues in Pharmacogenetics: Policy Briefing*. [Online] http://www.york.ac.uk/media/satsu/res-pgx/PGxpolicyissues2006.pdf accessed 17/04/2017

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

Vayena E, Gourna 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. doi.org/10.1159/000343792

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 17/04/2017

Williams R, Edge D (1996) The social shaping of technology. *Research Policy* 25: 865-899. doi: 10.1016/0048-7333(96)00885-2

Wright C, Burton H, Hall A, Moorthie S, Pokorska-Bocci A, Sagoo G, Sanderson S, Skinner R (2011) *Next Steps in the Sequence: The Implications of Whole Genome Sequencing for Health in the UK*. PHG Foundation Cambridge. [Online] http://www.phgfoundation.org/file/10363/ accessed 4/09/2016

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* (3rd Edn). MIT Press, Cambridge MA.

Wyatt S, Harris A, Adams S, Kelly S (2013) Illness online: self-reported data and questions of trust in medical and social research. *Theory, Culture and Society* 30(4): 131-150. 10.1177/0263276413485900

Tables

Table 1: User Participants' Demographic and DTCGT Information

Age (years)	**Range**	**27 – 70**
	Mean	**52**
	Median	**53.3**
Sex	**Women**	**12**
	Men	**5**
Recruitment source	**ISOGG**	**5**
	Genomesunzipped	**8**
	Known to participant	**4**
Ethnicity	**White British**	**13**
	Irish	**2**
	Ashkenazi Jew British	**1**
	Italian Australian	**1**
Highest education level	**Unknown**	**1**
	Further education level	**2**
	Degree level	**6**
	Masters level	**3**
	Doctoral level	**5**
Occupation	**Genetics research**	**5**
	Public health research	**2**
	Other non-genetics/biological science occupations	**10**
DTCGT type	**23andMe**	**17**
	FTDNA	**5**
	Sorenson Y DNA	**2**
	Sorenson Surname DNA	**1**
	Autosomal DNA	**1**
	Oxford Ancestry	**1**
	Whole exome sequence	**1**
Number of tests each	**5 tests**	**1**
	3 tests	**1**
	2 tests	**5**
	Only tested with 23andMe	**10**

Table 2: Genetics Clinicians' Professional Information and Consultations for DTCGT

Profession	**Doctors**	**8**
	Nurses	**4**
	Scientists	**4**
Consultations for DTCGT	**Doctors**	**5**
	Nurses	**5**
	Scientists	**1**
Types of DTCGT consulted about	**SNP genotyping including health risk data**	**7**
	SNP genotyping ancestry	**1**
	Single gene sequence	**2**
	DIY research lab tumour genome sequence	**1**
People consulting about DTCGT (NHS unless specified otherwise)	**Own test**	**6**
	Relatives' test	**2**
	Own test (private consultations for journalists investigating DTCGT)	**3**