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Personal genomics and individual identities: motivations and moral imperatives of early users

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Abstract

Since 2007, consumer genomics companies have marketed personal genome scanning services to assess users' genetic predispositions to a variety of complex diseases and traits. This study investigates early users' reasons for utilizing personal genome services, their evaluation of the technology, how they interpret the results, and how they incorporate the results into health-related decision-making. The analysis contextualizes early users' relationships to the technology, the knowledge generated by it, and how it mediates their relationship to their own health and to biomedicine more broadly. The results reveal that early users approach personal genome scanning with both optimism for genomic research and scepticism about the technology's current capabilities, which runs contrary to concerns that consumers may be ill equipped to interpret and understand genome scan results. These findings provide important qualitative insight into early users' conceptualizations of personal genomic risk assessment and illuminate their involvement in configuring this technology in the making.

Keywords

personal genome scanning; direct-to-consumer services; genomic risk assessment

Introduction

A primary goal of genomic research is to translate findings into personalized healthcare services. The myriad efforts that make up what is currently referred to as personalized genomic medicine (PGM) generate a wide range of interpretations of what it means for medicine to be individualized, predictive, and preventive. Since late 2007 and early 2008, the commercial entities 23andMe, deCODEme, and Navigenics have offered personal genome scanning services for purchase over the Internet. For fees ranging from \$399 to \$2000, these companies analyze over a million single nucleotide polymorphisms across the genome to assess their customers' individual genetic predispositions to a variety of complex diseases and traits. Consumer genomics companies utilize the rhetoric of personalized genomic medicine in advertising their services, suggesting that they can fulfil the promises of PGM by providing individualized risk assessments and allow consumers to take personal responsibility for their unique health risks (Nordgren and Juengst 2009). Personal genomics enthusiasts such as Steven Pinker (2009), one of the first individuals to have his entire genome sequenced through the Personal Genome Project, have characterized the advent of these personal genome scans as a first step towards the instantiation of clinical PGM, concomitant with the promise of information that can improve disease prevention. However, some clinicians and scientists have

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raised concerns about the clinical utility of genome scanning given the current state of translational genomic research (Hunter, Khoury and Drazen 2008, Janssens et al. 2008, Offit 2008).

In order to explore this technology in the making we present findings from our research study designed to assess the emergence of direct-to-consumer (DTC) personal genome services from the perspective of individuals who used these services within the first two years of their availability on the market. As the potential precursor to a “new” practice of medicine, this study investigates these early users’ reasons for utilizing personal genome services, their evaluation of the technology in its current incarnation, how they interpret the results, and how they incorporate them into their health-related decision-making to explore if and how personal genome scans enact an altered moral imperative to care for the self, either through biomedical channels or apart from them. The analysis further explores early users’ relationships to the new technology, the knowledge generated by it, and how it mediates their relationship to their own health and to biomedicine more broadly. Our emphasis here is not on the scans’ scientific veracity, but rather on the cultural and social context within which they sit, and the potential implications of their use and availability.

Interviews with this group of key informants offer a window into the worldviews and moral frameworks of early users of personal genome scans. Counter to medical, scientific and bioethical speculation that consumers may not be equipped to interpret results and may understand personal genome scan results as health informative or diagnostic (Cho 2009, Hunter et al. 2008, McGuire et al. 2009), our findings reveal that these early users approach personal genome scanning technology with both optimism and personal interest in the burgeoning field of genomic research and scepticism about the genomic technology’s current capabilities. These findings provide important empirical insight into early users’ evaluations of personal genomic risk assessment and illuminate their involvement in configuring a technology in the making. They may also be instrumental in understanding the implications of conceptualizing health, healthcare and personal identity in genomic terms, and provide new insight into the largely informational rather than medical value that early users attribute to personal genome scanning technology in its current state.

Background

Individuals’ predispositions to a range of genetic conditions conveyed by personal genome scanning services are both an outgrowth of and dependent on the results of population-level genomic research studies called genome wide association studies. According to the United States Department of Health and Human Services, “genome-wide association studies (GWAS) are defined as studies of genetic variation across the entire human genome that are designed to identify genetic associations with observable traits (such as blood pressure or weight), or the presence or absence of a disease or condition” (National Institutes of Health 2009). GWAS seek to identify the single nucleotide polymorphisms (SNPs) that are common to the human genome and to determine how these polymorphisms are distributed across different populations.

To carry out GWAS, researchers use two groups of participants: people with the disease being studied (cases) and similar people without the disease (controls). Researchers obtain DNA from each participant and place it on sequencing chips which analyze each participant's genome for strategically selected markers of genetic variation, or SNPs. If certain genetic variations are found to be significantly more frequent in people with the disease compared to people without disease, the variations are said to be “associated” with the disease. The associated genetic variations can serve as pointers to the region of the human genome where the disease-causing

problem resides, and depending on the strength of the association, these numbers are converted into disease susceptibility risk figures that are communicated via the DTC genome scan results.

Subsequent to the rapid proliferation and publication of GWAS results, three companies emerged as competitors in the DTC personal genome scanning market: 23andMe, deCODEme and Navigenics. When these services first became available in late 2007 and early 2008 the costs ranged from \$999 to \$2500, but by summer 2010 the prices had dropped to a range of \$399 to \$2000. All of these services offer consumers the possibility of learning their individual genetic risk factors for a range of diseases, although each company puts different emphases on using genome scanning for making health and lifestyle-related recommendations, genetic ancestry tracing, heritability patterns, and social networking around genetic similarities and differences, thus configuring the ideal user of each service somewhat differently (Woolgar 1991, Saukko et al. 2010).

Currently, 23andMe provides genotype information for about 600,000 SNPs and consumers with data on a “growing list of more than 155 diseases, conditions, and traits” by using a customized chip “to answer specific questions about our customers' health, ancestry and other traits” (23andMe 2009a). deCODEme claims to offer the “most comprehensive genetic health scan available”, as its scan is based on a chip that analyzes roughly 1.2 million SNPs associated with 50 diseases (deCODEme 2009a). And finally, Navigenics describes its scan as using the gold standard Affymetrix GeneChip® microarray to analyze customer's DNA (Navigenics 2009a). This technology provides consumers with a complex analysis of “featuring 1.8 million genetic markers, including more than 906,600 single nucleotide polymorphisms (SNPs) and more than 946,000 probes for the detection of copy number variation” (Affymetrix 2009) associated with 40 diseases, conditions and traits.

Recent analyses of the marketing rhetoric used to advertise DTC genome scanning services have characterized the companies as empowering users to become responsible consumers of genomic risk susceptibility information – individuals who are knowledgeable of their own personal genomic information and have the capacity to use it to promote their health (Harvey 2010, Saukko et al. 2010). For instance, the Navigenics (2009b) website touts:

There's DNA. And then there's what you do with it. Navigenics is the leading personalized genetic testing company. We use the latest science and technology to give you a view into your DNA, revealing your genetic predisposition for important health conditions and empowering you with knowledge to help you take control of your health future.

23andMe (2008) claims to “believe that your genetic information should be controlled by you,” invoking a neoliberal, rights-based approach to direct access to personal genomic information. Finally, deCODEme's website suggests that “getting to know your personal genome will empower you and provide you with a road map to improve your health. In many cases, early detection of risk allows you to prevent a disease from ever taking hold” (deCODEme 2009b). These companies have also emphasized the ideology of individual responsibility for health management to catalyze consumers. For example, the Navigenics website states that “When you understand your DNA and how it affects your health, you have valuable information to help you catch health conditions early, reduce their effects or even prevent them entirely. Because life is not just about living long, it's about living well” (Navigenics 2009c).

Experts' responses to DTC genome scans

However, as DTC personal genome scanning entered the market these services provoked controversy which has continued up to this day. This has been particularly apparent within the “expert” discourses of scientific, medical, and bioethics literatures, where concerns have been raised about the regulation and oversight of DTC genome services, consumer protections, and

physician preparedness for the influx of patients with DTC genome scan results. These for-profit entities marketing genome scans disrupt the traditional trajectory for the development and dissemination of genetic tests recommended by genetics professional organizations by offering genetic services directly to consumers instead of through a physician. This complicates regulatory challenges because these companies are not required (and may not have any incentive) to ensure the accuracy, clinical validity, reliability, and clinical utility of the services they offer (Offit 2008). Echoing this concern, some physicians and scientists have argued that DTC genome scan results lack clinical utility as well as analytical and clinical validity due to the ongoing and incomplete translation of GWAS upon which DTC companies rely for generating risk susceptibilities (Hunter, Khoury and Drazen 2008, Janssens et al. 2008, Offit 2008).

This has been coupled with concerns that imagined consumers or “virtual users” (Saetan 2000, p. 16) may lack the “genetic literacy” required to interpret the complex medico-scientific information contained in genome scan reports which could result in unnecessarily dramatic responses to or hollow assurances regarding reported risk susceptibilities (Cho 2009, Hunter et al. 2008, McGowan & Fishman 2008). McGuire and colleagues (2009) have hypothesized that consumers of personal genome scanning may be likely to interpret the results of these scans as informative for healthcare decision-making and possibly even diagnostic, even though these services are currently marketed with the caveat of being informative rather than medical products. However, these claims have been made in the absence of examining the users themselves to understand how they can come to be “experts” in their own right once they employ and “take-up” new technologies.

The existing empirical literature on the uptake and use of personal genome scanning is limited, and to date has focused on potential consumers’ attitudes regarding personal genome scanning, their interest in using these services, and whether and how they genome scanning results would inform their healthcare decisions (McGuire et al. 2009). This study reported that the majority of prospective users of personal genome scanning anticipated that they would want a physician to help interpret the results of the scan, despite the fact that these services are offered directly to consumers without medical mediation.

The impetus for the current study is to examine whether and how the motivations and moral imperatives of early users of personal genome scans resonate with and diverge from expert discourse around personal genome scanning as it has emerged in the commercial, scientific, medical and bioethical literature. To our knowledge this is the first study to qualitatively examine the perspectives of users of DTC personal genome scans to assess their motivations and experiences of utilizing these services. Given the rapid expansion of genomic services directly to consumers, the perspectives of users of personal genome scans are imperative for assessing the social and cultural construction of this emergent technology and the implications of understanding personal identity in genomic terms.

Early users as lay experts

For this effort we were most interested in understanding how early users of a novel genomic technology conceptualized its value and meaning in their own lives. This interest is framed by an understanding that early users of novel technologies are uniquely situated as “lay experts” and co-constructors of these techniques.

Deborah Heath, Rayna Rapp and Karen-Sue Taussig (2004, p. 159) have argued that as “thinking genetically” has increased in society the landscape of genetic science, biotechnology and the public sphere has changed, and in turn has blurred the distinction between experts and lay people in participation in the governance of genetic technoscience. Through the somaticization of genetics and incorporation of biomedical language and technologies into

everyday life, what once may have been the discourse of science is now also within the purview of lay experts (Novas and Rose 2000), and is a facet of the increasing biomedicalization of conceptualizations of health, illness and self-care (Clarke et al. 2003). Expert discourse is replete with characterizations of the lay public's deficit knowledge of genetics and genomics (Wynne 2005). However, science and technology studies scholars have argued that it is important to recognize that public understanding of science varies across social groups, social location, social context and the perceived relevancy of the science to individuals' own lives, and that lay people can contribute significantly to broad understandings of scientific practice and cultural meanings of science (Wynne 1991, 1992, Irwin and Wynne 1996, Kerr et al. 1998). Specifically, Angrist (2009) refers to early users of DTC scans as utilizing "citizen science," a practice with a long history in the environmental health movement and elsewhere. Hence, it is our stance that the experiential knowledge of users of DTC genome scanning technology can provide insight into the range of meanings of this emergent technology in everyday life (Lie and Sorenson 1996), its benefits and its shortcomings in an "up close" way that scientific and bioethical experts observing from a distance cannot (Franklin and Roberts 2006).

As an emergent technology that brings together bioscience, culture, society and the market, DTC personal genome scanning has "interpretive flexibility" in that it can mean different things to developers, users and a range of other constituencies (Bijker 1992, Clarke and Montini 1993, Garrety 1997, Pinch and Bijker 1984). In technology development interpretative flexibility means that "social interests shape technologies throughout their design process and usage and, in turn, technologies shape the activation of different social constituencies" (Timmermans and Berg 2003, p. 103). Steve Woolgar (1991) has argued that technologies are developed with specific types of consumers in mind, and that the ideal consumer is inscribed into the process of technology development. While "built-in" technology design inevitably impacts and constrains the ways in which consumers make use of it (Akrich 1992, Rohrer 2005) and the intended "script" may typically be the way in which consumers use and make sense of a new technology, the uses and meanings of an emergent technology are not necessarily fixed and are likely open to reinterpretation, re-invention, resistance and even co-design once in the hands of users (Berg 1994, Bijker 1992, Clarke and Fujimura 1992, Clarke et al. 2003, Cowan 1987, Lie and Sorenson 1996, Mackay et al. 2000, Nichter and Thompson 2006, Rohrer 2005). Brian Wynne (2005) has argued that public science imposes meaning on genomics by casting it in the reductionist terms of discourses of risk, however at this point in time little is known about the meanings and uses consumers are ascribing to DTC genome scanning and whether and how the meanings and implications of genome scanning that have crowded the public forum impact the interpretations of other constituencies. By focusing on user perspectives in this study we recognize the co-constitution of users and technoscience in the social life of the emergent technology of DTC personal genome scanning.

Furthermore, our interest in early users' involvement in the social and cultural construction of personal genome scanning technology grew in tandem with the commercialization of these services and marketing directly to consumers via the Internet. The consumer genomics market and development of Internet networks marks the historically specific emergence of forms of affect mediated through electronic communication technologies (Anagnost 2004, Ong 2006), and science and technology studies theorists have argued that the Internet serves as a host of new forms of genetic subjectivity and knowledge building by allowing individuals "translocal engagements" around shared genetic and biomedical experiences (Clarke et al. 2003, Heath et al. 2004, Novas and Rose 2000, Saukko 2004). Since DTC personal genome scanning presents as a risk assessment and self-surveillance technology born of the genomics and Internet age, we were also interested to explore how early users of the technique made use of the Internet as a tool to purchase the product but also to commentate on it.

In sum, we chose to study early users as they represent the users most able to understand the science and contribute to the implications of using scanning services. Focusing on this population is useful in a number of ways: 1) they represent the “ideal” consumer group of Internet savvy individuals with engaged participation and awareness of how the companies would like the scans to be used; 2) they serve as informants (rather than merely interview participants) to teach and dialogue with us about their understandings of these scans; 3) they are likely the most interested and invested consumers, most prone to find ingenious and creative ways of using this information; and 4) early users are thoughtful and deliberate about their use of the technology, and can provide clues about the wider value of this technology of the self (Foucault 1988) and its applicability in conceptualizing life-optimization strategies (Bumiller 2009). Instead of assuming that these users represent all current and future users’ perspectives, we approach the distribution of power amongst actors within social-technical networks as an empirical question to be explored (Lie and Sorenson 1996, Oudshoorn and Pinch 2003) and look to our informants to educate us about the potential benefits and hazards of this technology and its mode of delivery, and whether and how personal genome scanning generates new forms of technoscientific identities (Clarke et al. 2003, Novas and Rose 2000).

Methodology

We began our recruitment efforts by using a targeted sampling technique whereby we identified and recruited participants by using the Google search engine; we entered key search terms, including “23andMe blog/user”, “deCODEme blog/user”, “Navigenics blog/user” and from the results identified individuals who wrote weblog (blog) entries about their experiences using these scans. Nineteen bloggers were identified in this manner, and recruitment emails were sent to these individuals. We also sent a recruitment email to one personal contact whom we had met at a professional meeting in Boston in 2008. Seven bloggers and one personal connection agreed to participate in the interview. These seven interviews were conducted between January and February 2009, and from these interviews we sought additional participants by using a snowball sampling approach. This involved asking each individual we interviewed if they would be willing to pass our information along to anyone else they knew of who had also used a DTC genome scan, and if they had a personal blog we asked if they would be willing to post a virtual ad for our study. Six interviewees responded to virtual advertisements for our study that were posted on blogs, and three participants were referred to us by another interviewee. Lastly, in March 2009 we sent recruitment emails to fourteen individuals who had published their personal genome information on <http://www.SNPedia.com>, a website that serves as a database of over 7000 SNPs from the genomics literature and the effects of variations in DNA that individuals can use to interpret their personal genome scan results. From these fourteen recruitment emails we had eight people respond in the affirmative.

Our research team conducted twenty-four semi-structured interviews with early users of DTC personal genome scan services by phone between January and March of 2009. All interviews were audio-recorded, fully transcribed, quality checked and imported into Atlas.ti, a software program for qualitative data analysis. Due to poor sound quality during one recorded interview one interview was lost, and we were only able to transcribe and analyze 23 interviews for our final sample.

Using both deductive and inductive qualitative analysis, initial codes were developed by the research team by drawing on existing scholarship and through notation of themes drawn from initial review of the interview transcripts. Once a basic list of codes was established, the interview transcripts were divided among the team and first-level coding was completed, and similar first-level codes were collapsed into higher-order interpretive codes (Huberman and Miles 2002). Both types of codes were then applied to the entire interview. Using a grounded

theory approach (Charmaz 2006), codes were further sorted into broader conceptual categories and incorporated into the theoretical foundation of this project, and verified through an iterative process to ensure that our codes captured the richness of the themes within the data.

Based on this strategy a coding manual was developed by the research team to provide a definition for each code, illustrative quotes, and special instructions for applying them. Our overall coding strategy was based on a thematic grounded theory approach rather than a quantification of themes. To enhance construct validity and reliability of the conceptual domains identified, a common set of interviews was initially coded by all three coders to establish reliability. The entire set of coded transcripts was also reviewed and discussed by the research team for thematic analysis.

Demographics

Our analysis focuses on in-depth interviews with 23 consumers of DTC genome scans who had used these services within the first year and a half of their availability in the market (November 2007 – March 2009). Sixteen participants were male (70%) and 22 (96%) self-identified as Caucasian. Ages of participants ranged from 23 to 80 with a mean age of 42. Twenty (87%) participants resided in the United States, and the others lived in Australia, Canada, and Hungary. Participant professions that were most well-represented in the sample included the fields of information technology (6) and law (4), but also ranged considerably from stay-at-home mom to army sergeant to professional genealogist.

Participants in this study used the personal genome scanning services offered by 23andMe (20), deCODEme (6) and Navigenics (3). Eighteen participants used the services of a single company: 23andme (15), deCODEme (2) and Navigenics (1). Three participants had used the services of both 23andMe and deCODEme, and one participant used both 23andme and Navigenics. One participant had used the services of all three companies.

Of the 18 participants who paid for the 23andme service, 10 paid the original price of \$1,000; two paid \$499; and six paid \$399. This reflects 23andme's marketing decision to drop the price of the scan over the course of the first two years of its availability. The six deCODEme users paid the full \$1000 fee, and the one paying Navigenics customer paid \$2500 for the service. One participant received the services of 23andMe for free, as did one Navigenics user, and one participant in our study received free test kits from both 23andMe and Navigenics.

Reasons for personal genome scan

Using open-ended questions and structured probes, we queried our participants about their motivations for seeking a personal genome scan at its earliest stage of availability on the market. Participants gave a range of reasons for seeking these services which were not mutually exclusive. Given our research focus on DTC genome scanning as a potential precursor to a “new” practice of medicine, this study investigates respondents' reasons for utilizing personal genome services as it relates specifically to themes that were raised within the interviews related to attaining health and genetic risk information and the value of using the technology in its earliest stage of availability.[†]

The two most commonly cited reasons for having a personal genome scan were 1) to gain health-related information and 2) to learn about individual genetic risk factors. Nearly every participant mentioned at least one, if not both of these as motivating factors for using a personal

[†]While some respondents also gave reasons for seeking personal genome scanning to glean ancestry information; to satisfy a personal curiosity; and for social networking possibilities through personal genome scanning service websites, these reasons fall outside the scope of our analysis here and will be addressed in our future work.

genome service. The dominance of these themes in participants interviews is of interest not only because of how commonly these reasons were cited, but for the striking similarity between our respondents' and the personal genome testing companies' rhetoric of empowering consumers to prevent the manifestation of disease through knowledge of personal health information in their marketing materials. These reasons for using genome scanning technology mirror the intended use of the technology by consumers as set forth by the companies, suggesting that these early users' reasons map on to what Woolgar (1991) would characterize as the developers' ideal consumers.

Other motivating factors that participants' described spoke more to their personal characteristics: professional and personal interests in being on the vanguard of adopting and testing the capabilities of new technologies. Nearly half of the participants explicitly stated that they worked in a field that had either a direct or indirect relationship to DTC genomics, for example, bioinformatics, information technology, medicine, academia, law, genealogy, and science journalism. These participants reported having developed a strong professional interest in having their personal genome scanned in order to gain first hand experience about the product. However, it is critical to note that despite their professional interest in these services and their potential personal investment in their success, these users were not as a group more likely to tout the benefits of these scans without acknowledging their current limitations.

Just under half of the participants characterized themselves as "early adopters" of new technologies, and that this was a key reason that they wanted to use these services. Given our recruitment strategy, this was, in many ways, to be expected. In sociological theory, "early adopters" are traditionally characterized by their early uptake of a new technology but also by their desire and willingness to promote, shape, and tweak the technology for their own personal use (Rogers 2003). Our participants were all chronologically early users (as evidenced by the willingness of the majority to pay for the product at its original price), but moreover their investment as early adopters was demonstrated through how we found them: through their public pronouncements about their own uptake of these services, via weblogs or other internet sources. Some respondents also explained that they wanted access to their own personal genomic raw data to run it on the free bioinformatics platform called Promethease that mines the raw generated SNP chips to learn more about one's genotype than the individual commercial services provide (Cariaso and Lennon 2009). Although there was not complete overlap between the group that called themselves early adopters and the group who wanted to make use of the raw data online, we think that this only lends credence to our theory that all of these users fall into a particular category of consumers. Rather than taking up the technology with its intended "script", this subset of users seem especially prone to trying new technologies soon after their public availability and are interested in manipulating, adapting, augmenting, and finding new uses for a new technology and therefore also in part responsible for its broader dissemination and for shaping it for future users (Rogers 2003).

Desire for personalized risk assessment

Many participants expressed a desire for getting a DTC genome scan for a personalized risk assessment. Many participants indicated that they wanted to assess their personal risk susceptibilities in order to have the information needed to take personal responsibility for preventive health measures. While others wanted a scan in order to compare their personal genetic risk factors against their family history of disease.

The following quote illustrates the first perspective of the perceived value of getting a personal genome scan for personal risk assessment:

My thought process was, 'Okay, this is a new technology that's come along where I might, I hopefully can, actually use some of this information to actually get a better

assessment about what my risk of various diseases are above just what the standard White male, non-smoker, with a certain you know weight that I've got and everything,' and you know rather than that, my hope was that I'd actually should be able to get a more personalized assessment of my risk. (User 1)

The hope expressed here actually echoes much of the discourse on these companies' websites about the enhanced value of these scans over a simple medical assessment and also mimics the scientific discourse about the potential benefits of the results of the Human Genome Project more generally (e.g., Guyer and Collins 1993, Willard, Angrist and Ginsburg 2005). Although their assessment of the utility these scans may not live up to their hopes, it seems that amongst our respondents the marketing of these tests is effective. Early users seemed attracted to these scans for the very reasons that the companies posited. These users were very much willing and able to "think genetically" (Heath et al. 2004).

Another participant spoke of the potential and hoped for added benefit of a DTC genome scan over known family history risk assessments:

You know I looked at my family history and there are a couple of susceptibilities, looking at what my great grandparents died of and my grandparents died of, and you know knowing my parents and my siblings, and I was curious what markers would be there. But on the other hand I've got a fairly realistic view, I think ... I guess some genetics is destiny. You know if you're blue-eyed, you're going to be blue-eyed... you know, but other than that, I've got a pretty clear-headed assessment of okay, if I've got a slightly higher propensity for Type II Diabetes say, you know the genetics of that might be a two or three percent risk change, and diet and lifestyle factors might be a ten or fifteen percent change, you know... I know some things about myself. I know my own proclivities and propensities and I was curious to see how those played out, and I was also interested you know, do I have the risk factors for the thing that killed my grandfather? You know but there's some curiosity there. (User 2)

Although this participant indicates that there may indeed be some added benefit to the scan over what he already knows about his health risks, he is fairly careful to temper this optimism with his knowledge that the augmented knowledge, at this point anyway, is likely to be minimal and ultimately adds up to "some curiosity" about it rather than the belief that this knowledge will be either necessary or life-altering.

Value of DTC genome scan

Through coding the data, our inductive analysis revealed that our informants assessed the "value" of DTC genome scanning results in a variety of ways. Four main themes emerged through which participants assessed the value of having their personal genome scanned: the applicability of genome scanning results in the clinic context; the predictive power of the results; the usefulness of genome scanning as a personal risk assessment tool; and the reliability and validity of genomic research and its applications. This is not to say that all or even most of our informants unquestioningly accepted that there is added value in having their genome scanned by one of these companies. Rather, these were the main categories that they used to evaluate whether these DTC scans were of any use to them now or if they may be in the future.

Clinical applicability of genome scan results

One key theme that emerged through our data is how these early users of this technology characterize the clinical applicability of their personal genome scan results. In general, informants evaluated the applicability of genome scans to the clinical context in the following three, not mutually exclusive, ways: that the clinical applicability of the results is currently very limited, that the applicability of DTC genome scanning results is apparent in the arena of

pharmacogenomics only, and that in the future the clinical applicability of DTC genome scanning will increase substantially.

In describing the current limitations of the clinical applicability of DTC genome scanning, nearly half of our participants felt that the results of the scan are not medically actionable. As one participant put it:

There is very little that I can do about the disease associations I have because most of those diseases, we don't have any scientific literature yet which tells me what treatments I should pursue. It merely tells me I'm absolutely higher or lower risk, but there's no information about how I should respond to this. (User 8)

This response has less to do with the scan results themselves, than the uncertainty about how one can and should act upon the results. More pointedly, some participants argued that a lack of physician knowledge of personalized genomics limits the clinical acceptance and hence clinical applicability of DTC genome scanning results. As one participant described her experience:

I printed out my results and I took it to my doctor at my annual check-up and my doctor sort of glanced at it and said, 'Well I have no idea what to do with this,' and so I felt like you know the traditional medical profession is completely unable to help with genomic scanning, that the consumer has to really do their own research. (User 13)

Finally, other participants cautioned that at this point in time it would not be practical or advisable to use DTC genome scan results in place of medical evaluation or diagnostic genetic testing. For instance, one participant argued that:

As long as they're [the companies selling DTC genome scanning services are] clear that they're making it as a novelty service, I have no problem... Nobody should act on this as a medical test, because who knows for the price you're paying if you're getting the same quality control as a medical test... with perhaps other checks or more highly trained people. I don't know, maybe they're very good. I can't say, but I'm just saying that if I had a very negative [DTC genome scan] result, I would get retested to make sure, you know, by a person who is certified for this. (User 19)

This respondent is speaking less about the scientific veracity of genome scanning in general and more about the reliability of the generated by the DTC company's lab, and in so doing reinforces the companies disclaimers that they are not providing medical information to consumers.

In terms of the current clinical applicability of the results of these services, some respondents postulated that if there is current clinical applicability for personal genome scanning it is in the nascent field of pharmacogenomics, which can help determine if a particular drug will be useful or harmful to an individual based on their genomic profile. In reference to the medical studies that have associated one's specific genetic makeup with the effectiveness of cholesterol-lowering statin drugs found on company websites (23andme 2009b, deCODEme 2009c) one respondent explained that in a conversation about genome scanning with his physician:

We talked specifically about that and the possibility that there may be medicines in the future that I may need, and I may be able to tell by looking at those [genome] scans... I didn't need any statins, but my father had been on statins for many years, so there is a good chance that I might need them at some time in the future, and that DNA scan provided information that in my case so far it has said that if I needed statins that they would not be a danger for me and that they would probably work... I know when a doctor prescribes a medicine for a particular condition, they've never

had any indication in the past of in most cases of whether the patient may be a person that that is going to be a safe and effective medicine... and now with patients who do have the DNA scan, that gives them at least something extra to go on, even though the science is still fairly new. (User 17)

While most of the participants in our study characterized the current clinical utility of DTC genome scanning in limited terms, an optimistic yet guarded perspective on genome scanning was a recurrent theme in our interviews. This was especially apparent in our respondents' projections for the future, as the scanning technologies themselves advance and presumably make their way more into the mainstream. Despite current scientific limitations many participants believed there would ultimately be broader acceptance and adoption of genomic applications in the clinical context. The following quote captures this perspective:

I have this [DTC genome scan results] available, not so much the interest in diseases, but there's a lot of information now on which drugs will work well for my own genomic profile, and I want... I don't expect them [clinicians] to come back and really have concrete information about how I should make my own decisions. I think I've done pretty good homework on this, but I would like my doctor to understand this is available. I think long-term, perhaps five, ten years away this will be more and more part of what's considered standard of care, but for the moment I accept that I'm probably a little bit ahead of my own doctor with regard to the genomic aspects, and I focus on them for things where I need a specific treatment, I require a prescription, or in some other way there is something that they can genuinely provide me and they're a gatekeeper for some set of resources. (User 8)

This futurist orientation towards the clinical applicability of personal genome scanning raises the question as to what threshold early users of the technology are setting for the transition towards widespread adoption and clinical uptake of results for diagnostic and therapeutic purposes. While our respondents did not convey uniform projections of the future clinical applicability of personal genome scanning, factors that they believed would contribute to broader acceptance and adoption of this new technology in the clinical context include lower costs of scanning, increased clinician awareness of its capacity, and the expansion of research studies and their ability to detect population-wide genome variations for disease genes. If these tests do become more mainstream, it will be up to users themselves to decide how to make use of these tests—on their own, in consultation with a physician, or maybe another type of healthcare professional. The interpretive flexibility of these scans is evident here as users contemplate the tests' utility for the future. And it is clear that different users would use these tests (if they use them at all) in different ways.

Many of these early users of DTC genome scanning hoped that the results would provide genetic risk information that would enable them to make health related decisions, what Kristin Bumiller (2009) has characterized as employing life-optimization strategies, but they felt that the medical actionability of their results was limited both by the state of the technology itself and the clinicians' lack of awareness of the technology. In the former case, DTC genome scanning resembles many other forms of genetic testing that are available in the clinical context in that genetic tests can provide information about risk susceptibility for specific diseases but few clinical options exist to help patients determine how they should act in response to a genetic test result. Gene therapy and pharmacogenomics have not developed on pace with genetic and genomic research, thus it is not surprising that many of our respondents noted the gap between the availability of genome scan results and corresponding medical responses. While the DTC companies have taken the stance that having access to one's personal genomic risk information can help individuals to take control of their genetic future, early users of these services expressed some scepticism that control would be attained through medical means in the immediate sense.

Similarly, the recognition by some participants in this study that they were “ahead” of their clinicians in terms of their awareness of genomics indicates that many of these early users conceptualize themselves as sophisticated in their genetic literacy and personally responsible for knowledge of their genomic profile and its attendant risk susceptibilities regardless of clinician awareness of the technology. Many of these users of personal genome scanning characterized themselves as lay experts on the capabilities of genome scanning (without using the terminology of lay expertise) and embraced the notion of the democratization of genomic information and the move away from clinicians as gatekeepers of personally relevant information. The notion that users themselves can be experts and that they can help democratize the use of a new technology echoes both other studies of “lay experts” (e.g., Irwin and Wynne 1996, Kerr et al. 1998) and perhaps more importantly the burgeoning, dominant neoliberal ideology of Internet technology, which focuses on open access to knowledge, transparency, and a breakdown of the expert-lay divide.

Predictive power

As was mentioned above, many respondents believed that disease associations reported in genome scan results do not indicate a specific course of treatment or therapeutic action. Irrespective of its immediate clinical applicability, participants also characterized the value of DTC genome scanning in terms of its predictive power—the scientific capability of these tests to calculate an individual’s risk of developing a specific disease. Our respondents presented two qualified interpretations of the predictive power of genome scanning in its current form: that DTC services have low predictive power and that the predictive power of DTC genome scan results is strongest when taken into consideration with other predictors of health risks.

Taking the first stance on the predictive power of this technology, more than half of the participants argued that the power of personal genome scanning to predict the manifestation of health risks is quite low given the current state of genomic science and the technology used to interpret population-level genomic research. In describing his attitudes toward the newness of the technology, one participant captured the dominant sentiment on a genome scan’s power for predicting health risks:

Maybe in five or ten years it will be a good help for clinicians and patients when they work together on the patient’s health, but right now, as I said, these are just numbers, no more. As long as we don’t know all of the SNPs we can know about, no one can say they can predict diseases, because they cannot, and I don’t think they will ever be able to predict medical conditions properly, but as long as we don’t know millions of SNPs, not just two million, but five, six or ten millions of SNPs as they thought there are that many SNPs, as long as we don’t know all of these SNPs, I don’t think anyone can predict medical conditions properly. (User 16)

This perspective on the predictive power of personal genome scanning is characterized by our informants’ commonly repeated sentiment that the statistical power of genome scanning technology will remain low until more is known about the genome itself. However, this idea rests on the assumption that the genome can at some point be understood to the extent that predictive power to assess disease risk can be realized. So while these informants expressed doubt regarding the current capacity of this technology to assess whether they themselves would develop diseases in the future, there was a high level of acceptance of the promissory potential for the predictive capacity of this technology to be realized that has been forwarded by DTC genomics companies. These users’ conceptualization of the future realization of genome scanning’s goals as a predictive and diagnostic tool echoes the popular rhetoric of PGM as the future of medicine as a whole. Finally, a number of participants in this study indicated that they had professional interests in the field of genomics, which may explain the level of implicit buy-in from this group of users. Certainly consumer investment in and uptake

of this rhetoric is important for the survival of these companies, but as discussed below, these users' views of the realization of these goals may not be predicated on genome scanning technology alone but on other factors of scientific and technological development.

Despite the above-mentioned criticism of the low predictive power of genome scanning, an alternative perspective that a few participants shared was that when taken alone the predictive power of genome scanning is low, but as one tool in what we call a "wellness toolkit" along with family history, environmental factors, and lifestyle factors such as diet and exercise they believed that genomic information could be useful for predicting health risks. For instance:

I think right now it's not very predictive, but I see it as another piece of information to be considered in conjunction with family history and diet and lifestyle. So you know my feeling is, if you have access to the information at low cost and can say anything about it and view it in this larger context, this holistic context, then why not? (User 4)

This assessment does not attribute a strong predictive power of disease risk to genome scan results, but it attributes some predictive power to these tests by taking a more expansive approach to personal responsibility for managing health risks. Taken as one component of a wellness toolkit, these respondents perceive a capacity for hereditary, environmental and lifestyle information to predict the possibility of disease manifestation when analyzed together. This assessment of the value of DTC personal genome scanning represents a neoliberal individualist approach to health management which presumes that individuals can and should do everything that they can in order to learn about their own risk susceptibilities and to manage them accordingly. In fact, the uptake of DTC genome scanning may itself represent a move away from clinical interpretation and management of one's health to a self-management approach. Not unlike the companies' claims that knowledge of one's health risks can allow one to take action to prevent or manage the manifestation of disease, the respondents who championed this approach to the DTC genome scan results characterized personal health not as destiny but as something that can and should be managed to achieve the desired personal outcomes. These, arguably, are the ideal consumer reactions inscribed into the technology design and marketing.

Personal risk assessment

While some participants characterized the value of DTC genome scans as one component of their overall health profile, others assessed the value of their genome scan results in terms of the capability of the service for providing personalized risk assessment. Several participants provided a retrospective assessment of the kind of health-related information that the personal risk assessment tool provided and evaluated the personal risk assessment tool by comparing one's results to family history and/or phenotype. These respondents expressed both acceptance of and disappointment in the kind of health-related personal risk information that their DTC genome scan provided. For instance, on the more accepting side of the spectrum one respondent said:

Okay, you know the 23andMe profile, it says 'You are such and such a risk factor as compared to the average of the population,' and sort of comparing that to what I know about my family's medical history and saying like, 'Oh, interesting. I didn't know that we had you know higher risk, so that makes sense. It's something that I remember my grandmother dealing with,' and just kind of comparing it like in a more, you know, like a real hard and fast sort of medical explanation, or you know something on paper, something that's sort of scientifically backed up that says, 'Okay, this risk factor, this is you know what you can expect to potentially deal with and not deal with,' and then seeing how that compares to just sort of the stories that I know from my family, 'cause

we don't, my family doesn't talk about medical stuff a lot, so it's interesting to see like some of that on paper. (User 18)

On the other hand, some respondents expressed disappointment in the kind of health-related personal risk information that DTC genome scanning provides, remarking that they perceived that the companies were conservative in their approach to reporting only well-established genome associations and the results reported were predictable or self-evident. As one respondent conveyed:

And you know they told me that I might have a, you know, slightly increased chance of heart disease. Well sure, my dad had a, you know, angioplasty a few years ago. I mean part of all this also that I learned is that most of the genetic stuff or the predispositions that science will tell you about, you can figure out by looking at your family right now without any of the [GWAS] studies. (User 5)

However, some respondents characterized the predictability of the results as a positive thing in the sense that the *lack* of surprises presented in their results conferred confidence in the capability of personal risk assessment tools themselves. Representing this viewpoint, one respondent explained:

My lipid numbers are not particularly good and you know I was considered sort of a borderline risk for Metabolic Syndrome, and so when I got my Navigenics data and you know it said that I was at risk for obesity, for heart disease and Type II Diabetes, I wasn't surprised and you know in a way I was sort of reassured in the sense that my genes seem to be in sync with my phenotype. (User 4)

Given the similarities between the genome scanning services that our respondents utilized, the range of evaluations of personal risk assessment tools demonstrates that early users of DTC genome scanning do not necessarily accept the results uncritically, and that they are thoughtful in their interpretation of the results and how they relate to their own personal situation. For some of the more critical respondents, their scepticism may also be related to an already high level of genetic literacy and familiarity with genetics and genomics at the time when they underwent personal genome testing. While some respondents did convey that they learned of heightened risk susceptibilities for some serious chronic diseases from their genome scan results, interestingly none of our respondents expressed great surprise at any health-related risk factors that DTC results conveyed. Perhaps this is because of knowledge of genetics, family history or phenotypic evidence of these risk susceptibilities that they already possessed, and in fact our respondents did often mention these already known risk associations. Had there been surprising or unexpected health-related risk susceptibilities revealed to these early users of genome scanning perhaps their reactions about the value of DTC genome scanning as a personal risk assessment tool would have differed.

It is also noteworthy that these respondents expressed a degree of acceptance of the validity of health-related risk associations once they received their results. So while DTC genome scans merely report risk susceptibilities for specific diseases, which could be interpreted as uncertain (as they have been in the scientific literature), respondents in our study accepted these risk associations with some level of certainty when compared against family history and/or phenotype. Perhaps this is an acknowledgement that similarly to the assessment that the predictive power of genome scanning is best characterized in relation to other measures of wellness, so too is the evaluation of the "truthfulness" of health-related risk factors reported by genome scanning. However, attributing predictive power to DTC genome scanning results and conceptualizing these tools as valuable, disappointing or reassuring for personal risk assessment relies on users' acceptance of the validity and reliability of the genomic research that is applied to genome scanning technology. It is to this issue that we now turn.

Validity and reliability of genomic research and applications

The final way in which our informants evaluated the overall value of DTC genome scans is by assessing the reliability and validity of the scans. Of critical importance to them was the validity of these tests over time and their external validity in terms of the results' relationship to participants' "real" disease risks. The basis of these evaluations rests in large part on participants' assessment and understanding of the GWAS research that undergirds the results from the DTC genome scans. The promise of GWAS is that the genetic variations, when combined with clinical and other phenotype data, offer the potential for increased understanding of basic biological processes affecting human health and illness. However, our respondents did not take GWAS results to be absolute predictors of disease due to the state of the science and the role of environmental factors in complex disease development.

Some participants trusted the validity of the findings of GWAS that were applied in DTC genome scanning tools, while simultaneously recognizing the limitation that the findings are only as reliable and valid as the current state of the research. As one informant put it:

I guess I trust that for the markers that they're [companies providing DTC genome scanning services are] reviewing it will be accurate... like I trust fairly well that the technology is well enough developed to not give a false-positive or a false-negative. It seems like, I mean, there's just so much, there's so many markers and you know the genetic code of a human being is so complex that it's clear that it's only picking at you know a couple of little bits and pieces here and there. So I think it's a very incomplete picture, but I guess I would say it's an accurate but very incomplete picture. (User 18)

Other respondents expressed their trust in the validity of the genomic research utilized in genome scanning services, even though they don't think there is much clinical utility to it yet because of its questionable reliability—that the interpretation of their genomic information is likely to change rapidly as more studies are completed. For example:

The technology is moving so fast, so it was expensive for the value that was delivered, but yet I wanted to be an early adopter, so I probably can't really complain about that. And I think it will keep getting more useful, so every time I log in to 23andMe they post more conditions and more research. So I'm pretty happy with how the service is progressing. And I know it's interesting to... I mean it's still early days, and so it's interesting to me to see still all of the things that we need you know from a science perspective to be able to really understand the genome. (User 13)

However, she goes on to say that "the information is still not actionable...we're also starting to also realize how important the environmental component is and the toxins we're exposed to in our environment, and our personal behavior and how that could be improving, and so it's a complex problem that I think the genome is one of the easiest ways to start getting real data to look at" (User 13). Here this participant is less critical of the early stage of the scientific data collection and more reflective on the inherent complexity of disease causation as a combination of genetic, environmental, and behavioral factors.

Alternately, two individuals expressed the perspective that the DTC scan results are unreliable and not entirely valid because the genomic research applied in DTC genome scanning services is questionable due to the fact that GWAS are too new and have not been firmly established in the scientific literature. Therefore, in their minds, little stake should be put in the results of the genome scans. As one informant characterized his scepticism:

The most data you get is when you download these [the raw data generated by the genome scan] and put them through a program that reads all the SNPs and links it to known studies. The research with these SNPs, generally a lot of them aren't very

good, and the problem with this area is it really, I think more than even other scientific studies, these studies need to be replicated and I'm not seeing that very much, which you know is not a good thing. (User 20)

This participant's perspective demonstrates a belief in the veracity of the scientific method whereby research is deemed "good" based on systematic studies where replication leads to increased reliability and eventually to inherent validity. Along this same trajectory, some respondents explained how they understood the future of genomic research and applications to include the expansion of GWAS in that the more information about the genome that is available, the better users of genome scanning will be able to interpret the results, and the more useful the results will become for users, since the current science of the field can be difficult to interpret. While some rearticulated the position that more research will help to improve the current value of genomic applications, others characterized the future value of genomic research and its applications in uncertain terms. As one participant remarked:

I think everybody has to understand, as I understood going into it, that it is an extremely new science and there's a lot of uncertainty there, and that's one thing that I have always emphasized in science in general, that most people tend to think of science as a collection of facts, and it's not really a collection of facts. It's things that are known with some level of probability at first, and then we get a higher and higher level of certainty, but that never becomes fully certain, and science is always subject to change.... I've always seen that in new developments in particular, and even sometimes in old developments in science, that they're always subject to being overturned as we discover new information, so that when you have a new science like genomics, the information is always very tentative. (User 17)

This early user expresses an epistemology of genomic science which understands scientific knowledge as situated, partial, and temporally constrained, which is a far cry from the "knowledge deficit" that critics of DTC genome scanning have attributed to virtual users. This nuanced assessment of the value of genomic science demonstrates critical engagement with the scientific data presented in applications like genome scan results with striking similarity to the scientific assessment of these tools as expert critics themselves.

A final theme discussed by participants was that more genomic research will yield new, more affordable genomic technologies like whole genome sequencing that will in time outpace the first wave of genome scanning technologies. The following quote captures both of these perspectives on the future of genome scanning:

In the long-term, I think the standard will probably be whole genome sequencing, and so at that point a scan... would be less informative. So... I guess I would say until sort of the shift until the affordable whole genome sequencing, I feel like the information is only going to be more valuable over time. After that shift, it will be sort of a, you know we'll have far less information in a comparable whole genome sequencing. (User 7)

The characterization of personal genome services as the "early days" of personalized applications of genomic research reinforces confidence in the future of the field. However, given the overall sentiment about the current value of the DTC scans (based on questionable and/or incomplete scientific data) it is not surprising that few of our informants are using the data in order to inform their health and behavioral decision-making. However, how our participants are incorporating this data into their overall picture of their health status and health futures is a topic that was much discussed.

Personal health impact of genome scan results

One of the unknowns in the realm of DTC genome scanning is how users apply these results to their everyday lives, if at all. Much of the controversy in the expert discourse has centered on the speculation that consumers will interpret their genome scan results as diagnostic and take preventive health action based on their results (e.g., McGuire et al. 2009). Despite the companies' marketing rhetoric of empowering consumers to prevent the manifestation of disease through knowledge of personal health information, these companies all state that their services do not provide medical information, and are not meant to influence consumers' health behaviors. Given these ambiguities, we were interested in exploring whether and how early users of DTC genome scanning felt that their results influenced their health behavior both within and outside of the biomedical context.

The vast majority (19) of participants reported that receiving their results did not influence them to take any sort of immediate preventive health action based on their results. Ten in this group reported that their results contributed nothing to the way they thought about or approached their health in the immediate sense. For example, one participant summed up this sentiment as follows: "I don't think they've affected it at all. <laughter>I know what I should do and I don't always. I should get more exercise and not sit in front of the computer so long. <laughter>" (User 21). So while Harvey (2010, p. 371) has argued that "genetic susceptibility testing provides information that the 'genetic entrepreneur' can use not just to protect but to generate 'vital capital', attaining a state of optimal wellness specific to their genetic constitution," these respondents typically acknowledged "health" as an important value, but did not go so far as to argue that new information about themselves had inspired them into health-seeking action.

Nine other participants reported that while they did not take any immediate action from having received their results, they still characterized the results as a motivator to either maintain or increase their current amount of exercise and improve their diets. For example, one respondent said:

You know it's kind of different to know that you're not getting enough exercise and stuff and kind of know that on an abstract level, and even though it [the DTC genome scan results] is loaded up with caveats, just to see it all measured in a kind of quasi-tangible way, for me personally made me sit up and take it a little more seriously. So that's been like the biggest you know kind of reaction I've had is 'Ah, you know there's Diabetes Type II. I'd better start paying attention. I've got to start getting exercise and watch what I'm eating,' all that kind of stuff... (User 9)

This finding that genetic risk information had little to no impact on the majority of respondents' health behaviors is in line with recent reports that individual risk susceptibility information for common genetic variants does not change individuals' health behaviors (Henrikson, Bowen & Burke 2009, McBride et al. 2010)..

However, this same body of literature has indicated that some types of genetic risk susceptibility information *does* prompt individuals to change their health behaviors (Henrikson, Bowen & Burke 2009, McBride et al. 2010), which was also borne out in the respondents for this study as the four remaining participants reported that they had taken some preventive health action when presented with their genomic risk assessment results. For instance, upon receiving his genome scan results one participant reported that having knowledge that he had an elevated risk for thrombophilia motivated him to learn more about the condition in order to best manage it and to take certain preventive health measures to reduce the incidence of developing deep vein thrombosis (DVT) while travelling:

So I guess because having known that and with all the travel that I've done, I've been really careful ever since not to drink [alcohol] on flights and not to go to sleep on flights and to get up and keep moving around. So the feeling that I get from that is that armed with the knowledge, I mean I actually take pretty what is really not a lot of hassle to actually really eliminate that as a risk, and not knowing, if I'd not known this, I mean I could've actually had a DVT by now... So I think that's an example of the power of actually knowing about your personalized risk profile. (User 1)

While the preventive measures that this participant took did not necessarily indicate a medical consultation, this individual characterized the knowledge that he gleaned from receiving genome scan results to be personally empowering in the arena of preventive health. In fact, his language mirrors the rhetoric of the marketing material of the DTC companies regarding the value of these services.

Taking preventive action to the clinical context, three individuals reported consulting a physician for advice regarding preventive health measures to avoid the manifestation of specific genetic risk susceptibilities for male-pattern baldness, macular degeneration, and male-pattern osteoporosis. However, although they reported their genetic scan results as the catalyst to seek medical advice, these three informants acknowledged having been previously aware of the prospect of having inherited their specific genetic risk factor based on their family histories.

For example, a participant in his early thirties who explained that his results confirmed his familial history of susceptibility to male pattern baldness characterized how these scan results helped push him to seek out the advice and treatment he had otherwise been avoiding:

The one thing that I did do was look at medical or rather pharmaceutical treatments to preventing male baldness, but again that wasn't something that I discussed with my doctor in the context of 'I have a genetic scan. This is the result.' It was more a case of, 'This is something that I would rather avoid, if it's possible, and what can we do about it'.... And I guess the context of that was it's always something that I've been paranoid of. I have long hair and I like having long hair and I am vain about my hair, and I've always had a low level of paranoia about looming male pattern baldness... I was reluctant to turn to the pharmaceuticals because they're... I'm just not a big fan of the notion of daily medical supplements, but given that the genetic testing is relatively unequivocal that you know there was nothing that was going to save me from male pattern baldness in the SNPs that were sampled. I just felt that I may as well bite the bullet sooner rather than later. (User 10)

Another participant provided a cogent example of how he used personal genome scan data to confirm an inherited risk factor with unknown genetic origins. He used his genome scan results to take personal risk assessment a step further to take responsibility for individualizing genetic risk-related research using the Promethease database. In his own words:

By far the main thing I was interested in was the preventive medicine aspect and finding out things that may be directions instead of just taking a standard American male of my age and doing all the things that a standard American male is supposed to do of my age to stay healthy. I wanted to really correlate this [family history of disease] with my DNA for the things that I was at risk for, and also get, perhaps get some clues; because for one thing, one thing that I particularly thought might be interesting, and I didn't expect to see this as fast as I did, but due to the fact that my father and all of his brothers had this osteoporosis problem, I thought that out of the three billion base pairs on your DNA, there was probably one that was causing this particular problem, and I thought I'd begin the process of trying to find that one base

pair out of the three billion that caused this problem, which was quite a bad one. (User 17)

For these individuals, personal genome scan results confirmed that they had inherited a familial risk factor, and thus concretized the risk as reality. Taking the veracity of genomic risk susceptibility results at face value, each of these individuals interpreted their reported inherited genetic risk factor as “relatively unequivocal” (to use the words of User 10), which indicated to them that preventive health action was warranted. Based on these results we postulate that for individuals with family histories of specific diseases that are reported by genome scan services there may be an increased acceptance of the science and clinical applicability of results if they themselves receive information of their personal genomic risk susceptibilities for these diseases that both verify their familial history and which can translate into an impetus for preventive health action. This claim challenges the companies’ stance that they are not providing medical information to consumers. However, given the small sample size of this study additional research is indicated to assess whether those who receive results conveying elevated risk factors for specific diseases would characterize themselves as “genetically at risk” or “the worried well” more than users who did not receive eye-catching risk susceptibility results.

These respondents offer a window into the ways in which early users of this technology characterize the value of genome scan results for informing health behaviors. Our research suggests the majority of them do not interpret their results as deterministic, and most conveyed an understanding that their scans are not going to provide them with any certainty, but rather additional information that can be used in conjunction with other health-related information in order to make better informed lifestyle decisions. While a small number of our respondents took targeted preventive health-related measures upon receiving their genomic risk susceptibility reports, these individuals may be further characterized as having been armed with familial knowledge of disease manifestation that prompted their health-related actions.

Aside from these individual cases our data does not substantiate bioethical and medical experts’ concern that consumers of DTC genome scans will seek medical interpretation of their results without additional knowledge of the implications of heritable risk factors (e.g., McGuire and Burke 2008, Offit 2008). This is not to suggest that these experts’ concerns may not be realized if genome scanning is more widely adopted outside of the circle of thoughtfully invested early users. Perhaps though the information that DTC genome scan results provide are not so dissimilar from the other types of personal health information that are publically available in the contemporary socio-political context. The availability of health and wellness information online to lay users has grown exponentially over the last few decades, thus DTC genome scan results may not be so different from the other types of information that patients present to their clinicians based on their own personal research. When taking into consideration the vast array of health related products, technologies and information available to the lay public, the concern that these particular tests may result in a ‘raid on the medical commons’ (McGuire and Burke 2008) may be overstated. Having said that, it’s equally important to note that these users also understand that the technologies they are using are in their infancy, and are a part of a larger field, still early in development. Thus, with continued research and advances in genomics, these technologies may develop into more sophisticated tools that users may very well characterize in terms of being more soundly predictive and more medically actionable.

Conclusion

Interviews with this group of informants in the shift toward conceptualizing health and healthcare in terms of individual genetic susceptibility offer a window into the worldviews and moral frameworks of early users of personal genome scans, which may be instrumental in understanding the promise and perils of PGM. These informants have provided important

insights into early users' relationships to the new technology, what they perceive to be the knowledge generated by it, and how it mediates their relationship to their own health and to biomedicine more broadly.

While respondents took up varied stances on the stability of scientific knowledge, the types of research that will contribute to deepening our understanding of the human genome, and the eventual applications that genomic research can have in the diagnostic and therapeutic realms, they often characterized the future of genomic research in positive terms that echoed the scientific propaganda of PGM (e.g., Guyer and Collins 1993, Willard, Angrist and Ginsburg 2005). While certainly optimistic, participants also clearly conveyed scepticism in terms of their expectations not only for the immediate applicability of their individual genome scans, but also for the future of the nascent field of consumer genomics. Furthermore, most of the respondents indicated that they did little or nothing to alter their health-related behaviors after having received their individual genomic risk assessment. Taken together these findings are significant, as they run contrary to speculations raised in medical, scientific and bioethics literature that consumers' would approach DTC personal genome scans with inadequate genetic literacy and interpret results as medical information and/or diagnostic (Cho 2009, Hunter et al. 2008, McGuire and Burke 2008, McGuire et al. 2009). Given these findings, early users of personal genome scanning can be characterized as lay experts on the value of these technologies

Furthermore, it is our stance that companies marketing these services and their early users are in the process of co-constituting the meaning of personal genome scanning technology. Given the ways in which DTC personal genome scanning companies have inscribed specific uses for the technology into their marketing and service delivery, the demographics of our targeted sample of early users of personal genome scans, the fact that many of these individuals held some professional stakes in the field of genomics and the reality that all had enough interest in this application of genomic research to get a scan (and usually pay for it) in its earliest days of availability, it is not surprising that the participants were "thinking genomically" (to borrow from Heath et al. 2004) and well-versed in the contemporary discourse of the field of genomics. While the study size and demographics of our respondents may be cast as a limitation of the study, we believe it is also important to characterize respondents as lay experts finding innovative uses and explanations of the current state and future of the field.

Preliminary findings such as these can help us proactively think through potential benefits and challenges presented and generated by the availability of personal genome scanning technology. In addition, exploring users' attitudes toward the current value of the field of genomics is important for situating the context of our actual findings. Their perspectives today are critical in shaping the future of the field tomorrow. So while they certainly acknowledged specific limitations of the actual genome scan itself, as well as the field of genomics, they held overwhelmingly positive and optimistic views for the future of genomics as a field. However, at this juncture we cannot make a generalizable claim that this new technology enacts a uniform moral imperative or unique ethic of self-care for users of the technology.

Rather, we propose that it may be more productive to think about DTC genome scanning services less as a new medical technology and more as a new information technology being co-constituted by the developers and early users. The increasing use of bioinformatics to coalesce, analyze, and interpret large amounts of scientific, and especially genomic, data should force us to rethink the relationship between the medico-scientific technologies (and how we study them) and other emergent information technologies. DTC genome scanning services represents the comingling of these technologies, and more importantly for the scope of this paper, the comingling of the ideologies of the "new genetics" with the "democratic" user-driven ethos of the Internet and its developers (perhaps best exemplified by Google, a significant

sponsor of 23andme). Companies like 23andme, deCODEme and Navigenics are trying to capitalize early on the potential of bioinformatics for encouraging users to embrace self-directed learning, which Nordgren and Juengst (2009, p. 164) have characterized as an illustrative enactment of “the populist “open-source” ethos of the Internet and postmodern suspicion of authority and paternalistic expertise.” This allows users to understand and interpret large amounts of data, either through the visualization techniques and links to scientific papers provided through company websites and access the raw data, if they so choose. Despite the inscribed “open-access” technology design, the companies inevitably constrain the extent to which consumers can make autonomous use of the technology through the ways in which they package and portray information on their websites. This company-facilitated access, when combined with ideologies of neoliberalism, open access, and a trust-based system for information analysis, may help us understand the somewhat circumscribed perspectives of our early users. Thus it may be more likely that early users may make use and make sense of this emerging information technology in new and innovative ways by taking raw data outside of the companies’ web portals. Moreover, it opens up the possibility to explore the new subjectivities and engagements that these users embody and will embody in the future.

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Biographies

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Marcie Lambrix is a doctoral student in the Department of Sociology at Case Western Reserve University. Her sociological research focuses on medical sociology and research methods, in which she holds specializations from Case. Ms. Lambrix is currently working on her dissertation regarding the introduction of DTC genome scans into the clinic, including how doctors and patients make meaning of these scans in their interactions, as well as examining the potential broader issues for the medical profession (e.g. issues of dominance and

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