

Online Genetics for Health: An Empirical Investigation of How Users Matter

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Abstract

This thesis is an empirical study of why and how users do online genetics for health. Research on these users is needed to inform current academic debates and upcoming policy decisions on this emergent technical practice. Since 2017, online genetics companies have intensified their efforts to legitimize themselves as scientific sources of health information. In this context, I apply a sociological “users matter” lens to understand how users play a role in interpreting this technical practice. Specifically, I pose two research questions: (1) Why do people do online genetics for health? (2) How do users engage with and interpret health information from online genetics? I interviewed ten (10) users in total. In terms of motives, participants pursue this practice largely for curiosity and identity formation. Some also have specific health questions about which they are curious. To a lesser extent, some users are interested in perceived social benefits, like challenging experts and contributing to research. In terms of interpretation: The users I interviewed see themselves as maintaining a “scientific” engagement with online genetics for health, and they do not find truth or meaning in their genetic results. This suggests that, at least for this group of users, online genetics is not as “transformative” as some critics and proponents have assumed. This study demonstrates how users (and their social identities) matter: despite recent efforts by online genetics companies to construct themselves as medically and scientifically valid sources of health information, these users interpret online genetics in different ways, largely pursuing it for identity development and often questioning its medical and scientific legitimacy because they identify as scientific and skeptical users.

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Chapter 1: Introduction

1.1 Research Context and Objectives

The Human Genome Project (HGP) was completed in 2003, when the full human genome was mapped, which initiated a so-called “genomic revolution” (Phelan et al., 2013; Carter, 2007). One of the driving forces behind the HGP was the belief that it would lead to advances in personalized medicine (Collins, 2010; Lipkin & Luoma, 2016). It is in this context that the online genetics industry emerged and since 2003 the industry and market for online genetics has grown tremendously. Now, with an Internet connection, a couple hundred dollars, and a donation of spit, anyone can order an online genetic test for health information. Users will receive a testing kit, instructions to spit in a tube and mail their kit back to testing companies. Companies interpret users’ genetic data and provide them with health reports. Users can also download their “raw” (uninterpreted) genetic data and upload the file into third-party interpretation sites, for additional health reports (Allyse et al., 2018; Badalato et al., 2017). This study consists of empirical research on how and why online genetics for health is done, in practice, by users. Studying these users will inform current academic debates and upcoming policy decisions on this emergent technical practice, as well as contribute to the wider discussion on how individuals make sense of online information without the mediation of so-called experts (i.e. medical professionals) (Dommett & Pearce, 2019; Nichols, 2017; Policy Horizons Canada, 2018).

Some commentators on online genetics worry that users lack the technical literacy to understand online genetic test results, while companies are making grand claims that people are empowered from doing online genetics (Fishman & McGowan, 2014). Social scientists know little about how and why people do online genetics for health. Existing research on these users is

mostly quantitative research from the health sciences, which has reported on trends in online genetics use, with contradictory findings on what people seek from online genetics and how they respond to it (Frieser et al., 2018; Koeller et al., 2017; Wang et al., 2018; Nielsen et al., 2014). Quantitative research has examined users without contextualizing their use of online genetics, by focusing on the impact of results on users in binary terms (e.g. whether they changed health behaviours). Qualitative social science research on current users participating in this evolving technical practice is needed to understand how users interpret online genetics services for health, and how they incorporate this information into their everyday lives and daily practices.

The small body of social science research that does exist on users of online genetics for health mostly consists of “grand theory” and little empirical work. This study intends to build on the existing social science of online genetics by leveraging a unique theoretical perspective. The two dominant social science approaches used to study online genetics users have thus far employed a “transformational” lens (Hedgecoe & Martin, 2008), which rests on the assumption that genetic technologies transform people (Lippman, 1994; Nelkin & Lindee, 2004; Rabinow, 1996; Rose, 2008). Taking up a call made by Fishman and McGowan (2014) that there is not enough empirical research to support such “grand theorizing” about online genetics users; I apply a non-transformational lens in this study instead, taking a science and technology studies perspective which recognizes that “users matter” (Oudshoorn & Pinch, 2003). This approach recognizes that technologies are designed with “scripts” which encourage certain user responses, beliefs, and behaviours (Akrich & Latour, 1992); yet, users may read and respond to technologies in unexpected ways and this happens within a wider cultural context (Bijker, 1992; Jasanoff, 2004; Lie & Sorenson, 1996; Cowan, 1987). In line with the users matter approach, my study begins from the recognition that genetics is symbolic and social, which also fits with recent

studies highlighting that genetic information may not be purely scientific and may need to be annotated and interpreted for people to make sense of it (Nelson, 2016; O’Riordan, 2011).

In taking the users matter approach, this study endeavours to empirically investigate current users of online genetics for health. Most of the existing qualitative empirical work on users has analyzed users indirectly, by analyzing company products, platforms, marketing, and stakeholder statements or social media posts by users (Nordgren & Juengst, 2009; Nelson & Fullerton, 2018; Hogarth, 2017; Harris et al., 2016). A few qualitative studies have actually studied users directly through interviews, but these were studies of early users of online genetics for health (Ruckenstein, 2017; McGowan et al., 2010). Importantly, online genetics has changed considerably since 2017, with significant industry efforts to legitimize online genetics as a valid source of health information (see chapter 2). In this context, it is imperative to study current users’ perspectives to determine how they perceive online genetics for health, why they do it, and how they interpret online genetic health information. To address these objectives, I pose two research questions: (1) Why do people do online genetics for health? (2) How do users engage with and interpret health information from online genetics?

1.2 Thesis Outline

I present my research in six chapters: introduction (chapter 1), literature review (chapter 2), theoretical and methodological approach (chapter 3), findings (chapter 4), discussion (chapter 5), and conclusion (chapter 6). In the subsequent chapter 2, the literature review, I begin by providing key background information to contextualize this study, explaining what online genetics is and tracing the evolution of online genetic testing for health to its current form. I then review empirical findings from existing quantitative and qualitative literature related to users of

online genetics for health, indicating a gap in qualitative interview research on current users of online genetics for health. My research addresses this gap.

In chapter 3, I discuss the theoretical and methodological approach of this study. To start, I draw on concepts and theoretical insights from several authors to develop my theoretical framework. This framework consists of a “users matter” approach, which suggests that users play a role in interpreting technical practices and urges an empirical investigation of how users do online genetics in practice. In line with this theoretical approach, I recognize the symbolic value which has been culturally attributed to genetic information and techniques. After constructing my theoretical approach, I present the methodology of this study: a qualitative research design, data collection through interviews, and an abductive data analysis approach.

In chapter 4, I present the key findings of my study, grouped under two broad themes constructed from interview data: motives and interpretation. In the first section, I cover users’ motives for doing online genetics. These findings indicate that my participants do online genetics out of curiosity, even though some also have more specific health questions about which they are curious; rather than health, participants use online genetics for self-knowledge and identity development. My participants believe in the power of the data from online genetics to tell them something insightful about themselves. In the second section, I present findings related to interpretation. These findings demonstrate that my participants, most of whom claim to be more knowledgeable than other users, did not find meaningful or insightful results through doing online genetics for health. Most of my participants convey that they hold a critical engagement with online genetics.

In chapter 5, the discussion, I interpret these findings in light of my research questions. To answer the first question on user motives, I explain that my participants do online genetics for

health because of curiosity about identities and a desire for self-knowledge. They perceive the health information from online genetics as just one more source of self-insight. In addition to this main motive, some also have specific health questions they want to answer. Additionally, some of my participants also have hope that online genetics may yield potential social benefits, like challenging expert control of knowledge and contributing to novel medical research. However, these motives are not important for them, and they are skeptical about how online genetics companies can contribute to such social goals. In response to my second research question, on interpretation: I explain that the users I interviewed construct themselves as maintaining a critical, “scientific” engagement with online genetics for health. Consequently, they do not feel that they have found meaningful results through this practice. This suggests that, at least for this group of users, online genetics does not have as “transformative” an effect as some of its critics and proponents have assumed. These findings validate the users matter theoretical approach taken in this study, demonstrating how users of online genetics matter. Finally, in chapter 6, I conclude my thesis by reiterating the implications of my study, noting its limitations, and underlining important points for further research.

Chapter 2: Literature Review

In this literature review, I begin with a background section, explaining what online genetics is and tracing the evolution of online genetic testing for health to its current form, in order to contextualize this study. Tracing the development of online genetics for health highlights how companies have gone from marketing their services as recreational to medical and scientific. Next, empirical findings from existing quantitative and qualitative literature related to users of online genetics for health are reviewed in order to highlight a gap in qualitative interview research on current users of online genetics for health. This is the gap I address in this study.

2.1 Background

2.1.1 Online genetic testing

Borry, Cornel, and Howard (2010) explain that while companies offering direct-to-consumer genetic tests on the Internet have been around since 2003, the number of companies offering the test was not significant until 2007 (p. 102). They attribute the growth of online genetics to progress in genetic sequencing and genotyping, which “has changed DNA analysis from an intensive, burdensome, and expensive process to a relatively cheap and easy one” (Borry et al., 2010, p. 102). Writing for *MIT Technology Review*, Regalado (2018) says the online genetics market “blew up” in 2017: “The number of people who have had their DNA analyzed with direct-to-consumer genetic genealogy tests more than doubled during 2017 and now exceeds 12 million, according to industry estimates” (para. 1).

Online genetic tests can be ordered over the Internet directly by users, who receive a testing kit to collect their saliva sample and mail it back to the testing company. The company collects users’ genetic information from the saliva sample and uses proprietary computational

and human analysis techniques to interpret the data in order to provide users with ancestry and health reports (Allyse et al., 2018). Tests by the most popular testing company 23andMe rely on genotyping techniques, which connect certain genetic variants found in DNA to ancestry and health traits. This information is then packaged to the user in an ancestral report which gives a percentage breakdown of ethnicity, along with a health report indicating whether a user has gene variants linked to a variety of diseases and health traits, including: breast cancer, prostate cancer, celiac disease, Alzheimer's disease, Parkinson's disease, and more (23andMe, 2019a). Spector-Bagdady, Fakihi, Krenz, Marsh, and Roberts (2019) observe that "23andMe, with over 10 million consumers, controls one of the largest genetic and phenotypic databanks in the world" (p. 1). Kirkpatrick and Rashkin (2017) provide an overview of the three key industry players among online genetic testing companies: Family Tree DNA, 23andMe, and AncestryDNA. Family Tree DNA launched first, in 2000; 23andMe in 2007; and AncestryDNA in 2012 (Kirkpatrick & Rashkin, 2017, p. 7). Of the three, only 23andMe offers health reports to users; the other two only offer ancestry reports. However, all three company services allow users to download their raw (uninterpreted) data files for uploading these data to third-party sites (Kirkpatrick & Rashkin, 2017).

2.1.2 Third-party interpretation sites

Thus, the topic under study, online genetics for health, refers not only to the main genetic testing companies like 23andMe, but also companies offering third-party interpretation tools. The growth of these sites is the latest development in online genetics for health. These websites allow users to upload their raw (uninterpreted) genetic data files from testing sites (like 23andMe and AncestryDNA) to the interpretation tool, to receive additional health information. The format of the information users receive on these sites is often quite distinct from testing sites.

Promethease, which seems to be the most popular third-party site, analyzes users' raw data file and links it to scientific articles relating to the genetic variants found in the file; the articles are compiled in a database called SNPedia (Badalato, Kalokairinou, & Borry, 2017). SNPedia describes itself as "a wiki investigating human genetics. We share information about the effects of variations in DNA, citing peer-reviewed scientific publications. It is used by Promethease to create a personal report linking your DNA variations to the information published about them" (SNPedia, 2017, para 1). As Wang et al. (2018) emphasize, the "landscape of direct-to-consumer (DTC) genomics is continuously evolving" (p. 35). They explain what led to the development of third-party sites. In late 2013, 23andMe received a "cease and desist" letter from the US regulator the Food and Drug Administration (FDA) demanding the company stop marketing its reports as health information, in the absence of sufficient evidence to support such health claims (Wang et al., 2018, p. 36). As a result, 23andMe decided to stop providing users health reports. Users interested in health information thus sought out third-party interpretation sites. However, Wang et al. note, 23andMe later received FDA authorization to offer consumers some of its health reports again in spring of 2017. Regardless, analysts predict that third-party interpretation sites will continue to grow in popularity (Wang et al., 2018, p. 36). Indeed, the sites are increasingly popular and represent the latest major development in online genetics. The emergence of third-party sites which connect users to academic articles reflects growing efforts in the online genetics industry to legitimize itself as scientifically valid.

2.1.3 "Regulatory hacking"

Efforts at legitimization in the online genetics industry have also taken on other forms. As noted, in the last few years, 23andMe has received regulatory authorization to market its reports as health products. In fact, Burfield and Harrison (2018) highlight that when faced with

regulatory confrontation, 23andMe was strategic in its decisions to work with the FDA rather than against it. Burfield and Harrison call this strategy “regulatory hacking,” using it as an exemplary case study in their so-called “playbook for startups”. These strategically cooperative efforts proved advantageous for the company in the long run: in March 2018, 23andMe achieved what Anne Wojcicki called “a milestone in consumer health empowerment” by gaining FDA authorization to provide users with breast cancer risk reports (Gill, Obley, & Prasad, 2018, p. 2377). In an interview with Kara Swisher for *Recode Decode*, Wojcicki said that going forward 23andMe will focus on expanding the “health side” of their business even more, and discussed the company’s relationship with the FDA:

It’s interesting to note that of all the sort of more direct-to-consumer companies out there, we’re the only ones who’ve gone through the FDA process [...] we’re really proud of pioneering a direct-to-consumer path, and we’re really proud of actually, we’re the only ones out there that don’t require a physician’s oversight or a prescription (Johnson, 2018).

2.1.4 Research partnerships

In addition to “regulatory hacking,” online genetics companies have entered into research partnerships with both private stakeholders and academic institutions, in further efforts toward scientific validation. Australia’s chief scientist Alan Finkel (2019) argues it is important to recognize that online genetics companies have financial incentives in trying to legitimize their activities as scientific, when analyzing their interests in research partnerships (p. 5). Finkel elaborates that the significance of these financial stakes was demonstrated in July 2018: “when the pharmaceutical giant GlaxoSmithKline acquired a \$300 million stake in the genetic testing company 23andMe. In exchange, 23andMe handed over exclusive rights to its customer database for GlaxoSmithKline to use for drug discovery” (p. 5).

In addition to private partnerships, Richterich (2018) notes that biotechnology companies are using ties to public funding agencies and universities in order to legitimize their work.

Richterich suggests that corporations often rely on public funds to do company research and develop products (p. 61). As example, Sharon (2016) calls attention to 23andMe receiving a “US\$1.4 million research grant from the [National Institutes of Health] to expand its database, and with recent National Cancer Institute funding of Google and Amazon run genome clouds” (p. 569). Richterich (2018) stresses that funds often flow in the opposite direction as well. This is further explored by Spector-Bagdady et al. (2019), who illuminate the growing number of partnerships made between the online genetics industry and academic researchers and institutions. They assert that while more attention has been given to the deal between GlaxoSmithKline and 23andMe, there are emerging and unstudied unions between industry and academia as well. Spector-Bagdady et al. assessed scientific articles published on PubMed between 2011 and 2017 which cited data from online genetics company databanks and report that:

(1) The number of publications using private genetic data is increasing over time (from 4 in 2011 to 57 in 2017); (2) there are two main models of data-sharing, including researchers using existing private data held by industry (n = 172) or researchers sending in new samples for analysis (n = 6); (3) 45% of the publications were supported at least in part by the National Institutes of Health (Spector-Bagdady et al., 2019, p. 1).

The growing number of research partnerships made by online genetics companies with private and public stakeholders are part of ongoing industry attempts at acquiring scientific legitimacy. This is the current context in which my study of online genetics users is situated where there is a lingering academic need for scholarship which explores how current users of online genetics engage with this practice.

2.2 Quantitative literature

A review of the empirical findings from existing literature related to users of online genetics for health reveals empirical gaps in the literature which I address in my study. Most of

the existing research related to online genetics users comes consists of survey research done in the disciplines of medicine, health sciences, genetics, and health psychology. These studies mainly focus on the impact of genetic test results on peoples' health perceptions and behavior, with conflicting results. A few of these studies have also surveyed users on their motivations for testing. A meta-review of these quantitative studies was conducted by Frieser, Wilson, and Vrieze (2018) who studied the behavioural impact of genetic test risk results for complex diseases and found that genetic risk results do not “strongly impact self-reported negative behavior or psychological function of at-risk individuals” (p. 1134). However, they found that “return of results does appear to moderately increase self-reported healthy behavior in carriers” (p. 1134).

Among the survey studies there are mixed findings on user responses to genetic test results. Almeling and Gadarian (2014) conducted a survey experiment on a US nationally representative sample of 2100 “healthy people” in order to ask them how they would react to hypothetical genetic risk information for common diseases (including colon cancer, heart disease, and Alzheimer's disease). They found that, overall, “healthy people in the general population react to hypothetical genetic risk information by wanting to take action” (Almeling & Gadarian, 2014, p. 488). Similarly, Li, Ye, Whelan, and Truby (2016) reviewed studies in nutrition science to determine: “whether genetic risk communication affects motivation and actual behaviour change towards preventative lifestyle modification” (p. 924) for cardiometabolic diseases. However unlike Almeling and Gadarian (2014), these authors indicate that there is no or inconsistent evidence that receiving genetic test results leads people to higher motivation to change their lifestyle or to actual behavioural changes related to diet or physical activity which may lower the risk of cardiometabolic disease (p. 924). Moreover, they also stress

that of the thirteen studies they reviewed “eight were at high or unclear risk of bias” (Li et al., 2016, p. 924). Also addressing the aftermath of testing, Koeller et al. (2017) “analyzed survey data pre- and post-PGT from 1026 23andMe and Pathway Genomics customers”, and found that after receiving their online genetic test results, 4% percent of users scheduled an appointment with a genetic counsellor, and 38% would have done so if genetic counselling had been available to them (p. 1275). Under half of the respondents felt motivated to change their health behaviours after receiving test results (Koeller et al., 2017, p. 1276). Similarly, van der Wouden et al. (2016) found that while 63% of their 1026 survey respondents had a desire to share their online genetic test results with a healthcare professional, only 35% reported actually doing so after six months (pp. 515-516).

Green et al. (2009) report on the effects of disclosure of genetic risk of Alzheimer’s on individuals’ sense of wellbeing. They divided 162 people who had a parent with Alzheimer’s disease (but were not themselves symptomatic) into two groups. The first received genetic results confirming a genotypic link to the genetic variant linked to Alzheimer’s, and the other did not receive these results. Green et al. then measured “anxiety, depression, and test-related distress 6 weeks, 6 months, and 1 year [later]” (p. 245). Overall, the participants who had their higher risk of Alzheimer’s revealed to them did not experience “significant short-term psychological risks” (p. 245). Those who were told they did not have the mutation reported less distress, but those who had high levels of distress before taking the genetic test were “more likely to have emotional difficulties after disclosure” (Green et al., 2009, p. 245). Similarly, Egglestone, Morris, and O’Brien (2013) surveyed 189 consumers and 86 potential consumers of online genetic tests to determine the effect of test results on respondents’ health behaviour but also their health anxiety. They found that only 27.3% of users claimed to have changed their health

behavior after receiving genetic test results (p. 565). Additionally, only 24.6% of the users reported a change in their level of health anxiety (mostly a reduction in anxiety) (p. 565). The study suggests that only a minority of users made health behaviour changes; the vast majority (about three quarters) of the users surveyed did not.

Sherman, Shaw, Champion, Caldeira, and McCaskill (2015) assessed both how participants responded to hypothetical genetic disease risk information and participants' resulting interest in subsequent clinical versus online genetic testing. They found that for online genetic testing only, "participants allocated to the high risk condition (75%) had greater testing interest than those in the low (25%) category" (p. 706). Additionally, they conclude: "DTC testing is perceived as a viable, but less preferred, option compared with clinic-based testing" (p. 706). This study suggests that users may still value clinical tests and advice from health professionals, compared to information from direct-to-consumer online genetics, after doing online genetic tests.

Some of the quantitative literature related to online genetics users is concerned with why certain users may be less responsive to genetic test results compared to other users. Koeller et al. (2017) found that online genetics users who are more confident in their genetic literacy are less likely to be responsive to results from online genetics. This is in line with survey research by Gollust et al. (2017), who found that users with elevated genetic risk for common diseases are less likely to support widespread availability of direct-to-consumer, online genetic testing. In research on users of clinical genetics, Kaphingst et al. (2015) found that those with low or limited health literacy are more interested in genetic testing than those with self-reported "adequate" health literacy (p. 105). Like Koeller et al. (2017), they also found that respondents who claim to have higher health literacy are less likely to change their health behaviours after

receiving genetic test results (Kaphingst et al., 2015, p. 105). Yet, other survey research by Chapman et al. (2019) contradicts some of these findings. Their study focuses on levels of genetic knowledge and personal engagement with genetics in the general public (rather than solely people who have taken genetic tests). Chapman et al. tested 5404 respondents from Russia, the UK, and the USA on their genetic literacy, and measured their attitudes toward genetics, and found that: “those with greater genetic knowledge were on average more willing to use genetic knowledge for their personal health management” (2019, p. 9).

While the majority of these survey studies have focused on the impact and effects of genetic testing on user health behaviour, a minority have considered user motivations for doing the testing. Wang et al. (2018) recruited 321 users of third-party interpretation websites to survey their motives for using these sites. They highlight that participants are “motivated to explore raw DNA for ancestral information (67%), individual health implications (62%), or both (40%)” (Wang et al., 2018, p. 35). In line with previous studies, only a minority of participants (30%) followed up on their results with a doctor (p. 35). Moreover, Wang et al. extrapolate that while using third-party sites, their respondents “face challenges in understanding the results and may seek out clinical assistance in interpreting their raw DNA results” (2018, p. 35). Another survey study by Haeusermann et al. (2017) explores what leads certain users to share their genetic data online, on public sites, after obtaining their genetic test results. While they primarily focused on motives for sharing data, their study also surveyed users about motives for testing and reactions following testing. When they asked respondents to rank three motives for getting tested: “46.41% gave ancestry as their first option, followed by health (41.56%) whereas contribution to research trailed third (14.35%)” (p. 4). Additionally, Haeusermann et al. asked respondents if information from online genetics had improved their health or lifestyle: “around a third checked

not at all (33.20%) or to a small extent (38.49%) while 19.14% recorded a moderate and 9.16% a large improvement” (p. p. 4).

The vast majority of existing empirical research on online genetics users and health consists of survey research, which has primarily focused on measuring the effects of genetic results on users’ health behaviour. While this research offers some generalizability and interesting trends to further explore, I explore in this study the complex and possibly contradictory experiences and perceptions that users may associate with online genetics for health. Qualitative research allows space for subjectivity and ambiguity (Silverman, 2013), which is important since the mixed findings from survey research imply that the motives, perspectives, experiences, and forms of engagement among online genetics users may be complex. Qualitative interviews are a method for exploring contradictions and messiness in depth. Sociologists agree that qualitative research is preferable to quantitative when examining new social phenomena (Weiss, 1994). While the popularity of online genetics is growing rapidly, it is still an emergent technical practice, with limited research on users. As Harris et al. (2016) have observed, online genetics is in a state of flux. Given the evolving nature of online genetics, qualitative research, which remains grounded in and aware of the flexible and changing terrain it operates in, is a good approach to studying users.

2.3 Qualitative literature

2.3.1 Clinical genetics users

A minority of the qualitative research on clinical/offline genetics users suggests that they may experience negative or anxious beliefs related to health after using genetic tests. Based on interviews, Brunstrom et al. (2016) found that young women who have been tested for breast and ovarian cancers might feel more pressured to make life decisions earlier in their lives than their

peers who have not been tested. Hamilton et al. (2009) also used interviews to study women who took genetic tests for breast and ovarian cancers, with similar results: participants faced certain challenges which they attributed to the genetic testing experience, such as increased self-surveillance related to health.

Most of the existing qualitative studies indicate that people are not significantly impacted emotionally by genetic test results. Meisel and Wardle (2014) examine how genetic test results for weight gain risk psychologically impact recipients: upon receiving results, most interviewees said they psychologically oriented themselves toward long-term weight management goals by modifying their health and lifestyle behaviours to prevent weight gain (p. 182). Meisel and Wardle note that these interviewees are not fatalistic about weight, often shifting their attitude about weight from one of self-blame and negative emotional thinking to a more detached “objective” attitude, by attributing weight partly to genetics (p. 183). Similarly, Gordon et al. (2011) interviewed sixty participants who were given genetic disease risk results by a medical research organization. They highlight that these participants, who had received risk information for eight diseases, were not reactive or “emotional” toward genetic test results (p. 426). Notably, despite the majority of participants indicating they want to improve their health, a minority of them actually act in response to their genetic results (Gordon et al., 2011, pp. 427-428).

An early study of clinical genetics users by Lock, Freeman, Sharples, and Lloyd (2006) relied on interviews with first-degree relatives of people diagnosed with late-onset Alzheimer’s disease. Most of the interviewees “embrace the idea of complexity in connection with Alzheimer’s disease causation and [many] draw on a concept of ‘blended inheritance’ with respect to the disease that ‘runs’ in their family” (Lock et al., 2006, p. 277). Thus, Lock et al. argue, people often incorporate genetic test results about disease into existing knowledge of their

family health history, rather than let genetic test results override existing knowledge. In another study, Lock (2011) once again analyzed interview data from people who had taken a genetic test for Alzheimer's disease (AD). Lock explains that the interviews "not only made it clear that many individuals were not able to accurately recall what they had been taught about their supposed personalized risk of AD, but it was also evident that many continued to hold beliefs that they already held prior to entering the study about who in their family is at particular risk of AD, based on physical appearance or purported personality types" (2011, p. 698).

Some of the qualitative studies complement the survey scholarship, shedding light on uneven use of genetic results. Research by Dingel, Ostergren, Heaney, Koenig, and McCormick (2017) investigates whether and how people with addictions use genetic knowledge to make sense of their addiction. They asked 63 interviewees in addiction treatment programs "how they make meaning of a genetic understanding of addiction," and how they refer to genetic knowledge "in dealing with the stigma of addiction" (Dingel et al., 2017, p. 568). Two-thirds of the interviewees read their results through a belief that genetic explanations for addiction are not "useful" for their treatment and recovery efforts, and that the causes of addiction are "irrelevant to their daily struggle to remain abstinent" (Dingel et al., 2017, p. 568). At the same time, one-third of participants stated that notions of genetic predisposition alleviate the stigma and shame of addiction for them personally, and the majority believed that genetic explanations for addiction are helpful for reducing stigma around addiction in general (p. 568). The participants thus drew on genetic knowledge in varying ways, in part depending on the meaning they put in genetic determination for health. In another paper based on the same individuals, Dingel, Ostergren, Koenig, and McCormick (2019) further reveals that: "Most who had a family history of addiction subscribed to a notion that addiction 'runs in families,' while most who lacked a

family history of addiction used this fact to reject the notion of genetic inheritance of addiction” (p. 53). Dingel et al. close their article with the position that health information from genetic testing “affects peoples’ perceptions of addiction in small but important ways” (2019, p. 53).

2.3.2 Online genetics users

Finally, I turn to qualitative studies which look at online genetics users. Many of these empirical studies focus their analysis on the companies, including their websites and marketing materials, and then discuss the implications of these materials on users. An early analysis of online genetics company marketing by Nordgren and Juengst (2009) highlights genetic essentialism (the view that identity is genetic) in company marketing and offers conclusions about how users may be affected by it. The authors attribute the fast-growing popularity of online genetic tests to three cultural trends: “the pre-modern search for a naturalistic understanding of identity, the modern enthusiasm for science, and the post-modern emphasis on radical individual self-determination” (Nordgren & Juengst, 2009, p. 157). They express concern that users’ identities will be negatively impacted through online genetics, with unreliable and misleading test results (p. 157). The argument by Nordgren and Juengst (2009) relies on a textual analysis of websites for online genetics companies, as well as online testimonials by “satisfied” users featured on company websites.

Hogarth (2017) also focused his analysis of online genetics on company platforms and executives’ comments, particularly those of 23andMe. He details how the company’s executives have strategically aligned themselves with morals related to so-called “disruptive innovation” and consumer empowerment, key indicators of success among Silicon Valley start-up companies. He writes:

23andme wanted to transform healthcare by refocusing it on the goal of disease prevention to radically extend life expectancy. [...] For 23andme, this emphasis

on preventive healthcare meant that genetic risk assessment was at the heart of its technoscientific vision (Hogarth, 2017, p. 259).

In addition to the disruptive innovator image that 23andMe's executives have identified themselves with (Hogarth, 2017), analysis of the website itself reveals how the platform constructs this narrative. Saukko (2018) highlights the platform's efforts at cultivating a "participatory air" for users, by keeping the site interactive and inviting users to contribute their data to company research (p. 1320). Harris et al. (2016) also discuss how 23andMe may appeal to users with notions of empowerment, by conveying an interest in participatory research and demonstrating a willingness to use self-reported data for research, something not done in conventional medical research. However, Harris et al. also note that 23andMe has presented its services as more recreational rather than medical, in an effort to avoid regulatory scrutiny (p. 35). Of course, as highlighted in section 2.1 (Background), there have been many changes in the industry since then, which is one of the limitations of their analysis.

The existing research on third-party interpretation sites, such as Promethease and SNPedia, also illuminates how online genetics websites may appeal to ideals of user empowerment. An analysis of these sites by McGowan et al. (2017) notes that they portray themselves as liberators of science and knowledge from the closed doors of academia and the traditional research industry. This is done in an attempt to align themselves with the "citizen science" movement, which aims for inclusion of "'lay scientists' in research design and funding, and in collecting, analyzing, applying, and disseminating data" (McGowan et al., 2017, p. 497). One of their featured interviewees is Greg Lennon of SNPedia, whose quote summarizes the appeal to citizen science values that online genetics companies use in their marketing to users:

We ought to be doing more for ourselves and the world than just publishing things in journals that only other scientists read. [...] There has to be some way so that we can discuss and make available to at least those who are interested to non-scientists, as well as scientists [...] Well it seems that if you put billions of dollars

into sequencing and trying to understand the point of the genome, then there ought to be numerous ways (SNPedia is one of them) to make it matter to people (p. 504).

Similarly, Nelson and Fullerton (2018) explore how third-party interpretation website developers, in claiming to empower users, also differentiate their platforms from more popular user-friendly sites like 23andMe. The developers claim that, compared to 23andMe, third-party interpretation sites help users better understand the complexity behind genetics, provide a more transparent online genetics service, give users more options about contributing their data, and provide users with educational scientific tools (Nelson & Fullerton, 2018, p. 777). These findings on third-party interpretation sites imply that users may be enticed in part by marketing promises of knowledge empowerment. A paper on third-party interpretation sites by Badalato, Kalokairinou, and Borry (2017) expresses ethical concerns related to third-party interpretation websites. While they rate third-party sites as less problematic for users than main genetic testing sites, Badalato et al. believe that, like testing sites, third-party tools also have issues related to “inadequate informed consent, questionable clinical validity and utility, and lack of medical supervision” (2017, p. 1189). However, like most of these studies, this one collected data from websites rather than directly engaging participants in interviews.

A similar analysis of online genetics and user empowerment is found in Crawford, Bailey and Briggs (2019), who focus on the implications of big genomics projects which promise users to return their whole genome sequencing results. Crawford et al. examined the types of data usually returned to participants through genetic testing and research. Their analysis highlights issues with company goals of user empowerment, noting that:

[The] inventory of resources required to receive, process, and interpret return of research results exposes the potential for access disparities and warns the scientific community to mind the gap so that all participants have equal access and understanding of the benefits of human genetic research (Crawford et al., 2019, p. 691).

Thus, much of the existing qualitative empirical work on online genetics users has come to conclusions about users based on analysis of company products, platforms, marketing, designers, and stakeholders.

There is a small but growing body of qualitative empirical literature which draws on data from users of online genetics for health. Some of these studies rely on more indirect access to user perspectives. For instance, research by Harris et al. (2016) analyzed YouTube videos posted by 17 users of 23andMe, in which users “unboxed” their results for viewers. Assessing users, Harris et al. explain that they do not come across as “patients”: “The users were not unwell or suffering. They were curious and rarely connect to specific disease categories or conditions” (p. 35). The users behave as though online genetic testing is just another form of “playful” self-expression for them (p. 47). However, they note that this could be due to the nature of YouTube, which may invite more playful performance of user narratives (p. 50). Harris et al. analyze how users understand health information from online genetics. They demonstrate that:

[Users] contributed their own interpretations following wayfaring lines, accepting and refuting findings according to their own understandings of illness, often using the results to explain what they think they already knew (Harris et al., 2016, p. 46).

While Harris et al. offer some interesting insights into users, it is important to note the limitations of their work. First, their study relies on data collected in 2012, effectively making it a study of early adopters of online genetic testing practice. As highlighted in section 2.1, the online genetics industry has changed a lot since 2017 (Regalado, 2018). Second, relying on YouTube posts in order to understand users is limited because it is not a direct account of user experiences and perceptions. Given the public nature of YouTube posts, it is possible that users filtered their performance and did not feel comfortable about discussing more serious health concerns, a limitation which Harris et al. themselves allude to in their publication (p. 50). To

elaborate, it could be possible that users who were more health-concerned were less likely to post their experiences online. Finally, it is also difficult to determine whether 23andMe sponsored these YouTube posts as product placement. Kragh-Furbo and Tutton (2017) also looked at YouTube unboxing videos of 23andMe genetic testing kits in order to study users. They argue that, in the eyes of users, doing online genetics gives them perceptions of empowerment. They quote one of the YouTubers: “I always thought genetic testing would be complicated and expensive, where you had to go to some lab with hooded scientists, but we live in 2016 and it’s actually as simple as a little box being shipped to your door” (Kragh-Furbo & Tutton, 2017, p. 176). Again, the same limitations of Harris et al. (2016) also apply to Kragh-Furbo and Tutton (2017): understanding users requires going beyond public (and possibly even company-sponsored) posts online.

Similarly, additional qualitative studies have drawn on social media posts by users. Chow-White et al. (2018) did a content analysis of Twitter posts (tweets) mentioning online genetics. They found that discourse by users on online genetics (and particularly on 23andMe) is mostly positive. In analyzing tweets, they coded many (32%) of them as “unassigned” (p. 459), which categorized tweets as neither negative nor positive. In addition to categorizing tweets as positive or negative, Chow-White et al. also analyzed prevalent themes across the tweets and found that users are “enthusiastic” and “curious” about online genetics, and also tend to speak against government regulation of companies (p. 459). However, this study does not offer a deeper understanding of user opinions or experiences. The code of “unassigned” tweets hints that a variety of complex and potentially contradictory user perceptions exist. Additionally, Chow-White et al. did not interview users themselves, but relied on Twitter posts, which come with word count limitations. Again, the public nature of social media posts is also a limitation.

Finally, there are a few qualitative studies of users which rely on more direct access to user perspectives, by including interviews and discussions with users in the study design. However, as all of these studies rely on data collected up until 2015, they again constitute research on early adopters of this technique. For example, Lee (2014) conducted a four-year ethnography on direct-to-consumer genomic testing. Her paper argues that “the boundaries separating the categories of patients, consumers and research participants” in personal genomics have become “porous” (p. 136). Her paper primarily focuses on how “consumption and production become entangled, as individual biomaterial is transformed or rendered into biocapital” (Lee, 2014, p. 136). However, Lee’s research does not go in depth into user perspectives on why and how they use online genetics. At the same time, this study is insightful for understanding the user experience in the context of the early stages of the online genetics industry, especially as it relies on ethnographic data collected prior to 2014 (p. 136).

A study by Ruckenstein (2017) focuses on how users make sense of information from online genetic tests, examining: “how knowledge from various sources is coordinated and translated, delivered to people, accepted, refuted, shared, explored, and played with” (p. 1025). Based on discussions with participants, Ruckenstein states that most of the users saw test results from 23andMe as “reliable” or “somewhat reliable” (p. 1029). However, users did not consider them to be determinative of health outcomes in the form of “predictions, prognoses, or generators of further action” (Ruckenstein, 2017, p. 1029). Some of Ruckenstein’s participants also appreciated the availability of raw data files from 23andMe, but most of them seemed to believe they were not in a position—given limitations in their medical knowledge and skill with data—to understand the raw data on their own (pp. 1033-1034). Although the participants were largely positive about online genetics, they agreed with experts that “regulatory oversight is

needed to ensure the quality and safety of [direct-to-consumer] testing services” (Ruckenstein, 2017, p. 1035). Most of Ruckenstein’s participants had taken their genetic test in 2013 and she collected data in 2015 (p. 1036). Hence, this is also a study of early users. The study is also limited in that it does not explore user motives, and thus does not make connections between motives and the way people make sense of online genetics. Such an analysis is needed to explore how peoples’ intentions, health concerns, and other motives may relate to how they interpret online genetics results.

One interview study of early users of online genetics was conducted by McGowan, Fishman, and Lambrix (2010) and did investigate why early users are motivated to do online genetics, how they evaluate the technology, and how they respond to and understand results when making health decisions. They found that in evaluating online genetic technologies, early users are both skeptical and optimistic. In line with other studies, most of the early users “did little or nothing to alter their health-related behaviors after having received their individual genomic risk assessment” (p. 285). Based on these findings, McGowan et al. assert that early users are not interacting with online genetic tests as medical devices, and that “early users of personal genome scanning can be characterized as lay experts on the value of these technologies” (p. 285). However, the conclusion by McGowan et al. that users do not see online genetics as a medical technology must be contextualized and qualified by its temporal limitations, since it is based on early users and research done in an earlier context, before recent substantial company efforts at legitimizing online genetics as scientific. McGowan et al. (2010) justify their study of early users by assuming that “they represent the users most able to understand the science and contribute to the implications of using scanning services” (p. 267). They list several reasons why they believe studying early users to be pertinent. For one, they claim that early users “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” (McGowan et al., 2010, p. 267). This may be problematic because, particularly in 2010, it was far too early to say that there was an ideal consumer that companies had in mind. Consider that more recently, in 2016, 23andMe briefly experimented with offering next generation sequencing to consumers, and then promptly abandoned these efforts (Pressman, 2016), revealing that online genetics companies are still not yet clear on how they want people to use their technologies. Another reason McGowan et al. give for studying early users is that “they are likely the most interested and invested consumers, most prone to find ingenious and creative ways of using this information;” and “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” (2010, p. 267). This assumption begs for testing with further research; It is not necessarily the case that early users are more knowledgeable, interested, invested, or creative in how they do this technical practice than later users.

As I discuss further in chapter 3, my study recognizes that “users matter” at all phases of technological development, rather than only at the early phases (Oudshoorn & Pinch, 2003). It is important to study current everyday users of online genetics, especially within the broader context of recent moves by industry to legitimize themselves as suppliers of scientifically valid products, efforts which have accelerated since 2017. In the years since McGowan et al. (2010) and the other preceding qualitative studies on users were published, online genetics has less of a niche activity; industry has linked themselves to scientific articles, gained regulatory authorization to market a growing number of health reports, entered into research partnerships, and had their data cited in academic journals. Current users matter, and empirical research on

how and why people currently engage in this evolving practice of online genetic testing is needed.

2.4 Research questions

This review has highlighted a lingering gap in the social science on online genetic testing around user experience and it justifies an investigation of current users. Reviewing the existing research on users, I have demonstrated that it mostly consists of quantitative methods, largely from medical and health science disciplines. It has often fixated on users' reactions to genetic information, with mixed findings. It has illuminated certain trends but has not gained deeper insight into the subjective experiences and perceptions of users, why they do online genetics, and the processes by which they interpret their results. To fill these gaps, my research will consist of a qualitative study design.

The review has also presented findings from qualitative studies related to online genetics users. Findings from users of clinical/offline genetics indicate a need to further explore how people interpret health information from genetic tests, as it is evident that it is not a straightforward process. Much of the existing qualitative empirical work on online genetics users has based its conclusions about users on analyses of company products, platforms, marketing, and stakeholders; rather than collecting data directly from users. Some of it has also looked at user perspectives indirectly through social media posts. A few qualitative studies have interviewed online genetics users for health more specifically, but they were all done with early users of online genetics. As noted in the beginning of this chapter, online genetics for health has changed considerably since 2017, with companies constructing online genetics as more medically and scientifically legitimate. Hence, it is important to investigate how current users perceive this technical practice, why they use it, and how they interpret the health information it

provides. Accordingly, the two research questions guiding this study are: (1) Why do people do online genetics for health? (2) How do users engage with and interpret health information from online genetics?

Chapter 3: Theoretical and Methodological Framework

In this chapter, I discuss the theoretical and methodological approaches of this study. To start, I construct my theoretical frame by drawing on concepts and theoretical insights from several authors. My overall framework consists of a “users matter” approach, which urges empirical consideration of how and why people do online genetics in practice. Within this broader framework, my approach recognizes the symbolic value which has been culturally attributed to genetic information and techniques. After developing my theoretical frame, I discuss the methodology of this study, which includes a qualitative research design, data collection through interviews, and an abductive data analysis approach.

3.1 Theoretical approach

3.1.1 Users matter

In their book *How Users Matter: The Co-construction of Users and Technologies*, Oudshoorn and Pinch (2003) outline their “users matter” approach to studying users, which entails paying greater analytical attention to the role users play in interpreting technologies. Oudshoorn and Pinch (2003) incorporate theoretical insights from a diverse body of scholarship in developing their framework. They start with the Social Construction of Technology (SCOT) approach, which was the first to challenge the deterministic view of technology, bringing attention to how users contribute to the shape that technologies take within society, among particular social groups. This approach was pioneered by Pinch and Bijker (1987), who demonstrated the agency of users by referring to the case of the bicycle: the bicycle developed as it did because, early in its development, other versions of the bicycle were deemed “unsafe” or culturally inappropriate by certain social groups like women (see also MacKenzie & Wajcman, 1999). The SCOT approach has mostly focused on the early stages of a technology’s

development, suggesting that users, as social groups, contribute to an artifact's "interpretive flexibility," or the way that a technology can hold different meanings and uses by different social groups, including different groups of users. SCOT theory suggests that as a technology develops, it goes through a variety of social processes called "closure mechanisms," which lead to the "stabilizing" of its meaning, where a dominant use is established for the technology (Bijker and Pinch, 1987). While the development of the SCOT approach was a key milestone in technology studies, it has some limitations: it has potentially overdetermined the role of people and social forces, neglecting non-human actors and features of the technologies themselves, and it has primarily focused on the early stages of technologies, not considering how users may shape technologies at later stages (Oudshoorn & Pinch, 2003, p. 4). Paraphrasing Mackay and Gillespie (1992), Oudshoorn and Pinch emphasize that SCOT "closed down the problem of users too early, and it did not show how users could actively modify stable technologies" (2003, p. 4).

Oudshoorn and Pinch (2003) also highlight another, newer set of approaches to users and technologies within STS which have brought attention to users: semiotic approaches. This theory includes the concept of "configuring" the user developed by Woolgar (1991), who suggests that designers of technologies design them with particular users and intended uses in mind, which they inscribe into the technology. He considers technology as text and suggests that users play a role as "readers" of the text, implying a process of interpretive flexibility. Semiotic approaches also include that of Akrich and Latour (1992), who established Actor Network Theory (ANT) and developed its notion of "script". The ANT approach considers how technical objects themselves "participate in building heterogeneous networks that bring together actants of all types and sizes, whether humans or nonhumans" (cited in Oudshoorn & Pinch, 2003, p. 10). Thus, ANT attributes agency to technical objects. According to ANT, technologies are

developed by designers with “scripts” embedded in them, which prescribe a dominant use for a technology. Users shape technologies either by adhering to intended scripts or resisting them (Akrich & Latour, 1992). Oudshoorn and Pinch (2003) note that, just as with the SCOT approach, this second STS approach to users also takes the technologies as the point of departure; users are limited to solely responding to the technologies, by either following, rejecting, or otherwise responding to scripts (p. 11).

To mitigate the overemphasis of STS approaches on technologies and designers, which tends to prioritize analysis of technologies and designers at the expense of users in the technological ecosystem, Oudshoorn and Pinch (2003) bridge STS approaches with work from feminist and cultural/media studies scholars to develop a more comprehensive framework which considers both sides of the user-technology relationship. These scholars encourage more research on users. Oudshoorn and Pinch explain that feminist scholars’ focus on users has been rooted in concerns about the invisibility of women in the analyses of technological development, as well as how technologies affect women in unique ways (see Pursell, 2001). Feminist historians first turned attention to users “to go beyond histories of men inventing and mastering technology” (Oudshoorn & Pinch, 2003, p. 4). Cowan (1987) has played a pivotal role in turning attention to users in feminist approaches to technology. Oudshoorn and Pinch (2003) highlight Cowan’s concept of “the consumption junction,” which argues for a focus on the consumer and “on the network relations in which the consumer is embedded” (p. 5). Cowan (1987) explains that rather than look at the network the user is embedded in from the “outside in”:

I focus on the consumption junction, the place and the time at which the consumer makes choices between competing technologies, and try to ascertain how the network may have looked when viewed from the inside out, which elements stood out as being more important, more determinative of choices, than the others, and which paths seemed wise to pursue and which too dangerous to contemplate (p. 263).

Thus, feminist approaches like Cowan (1987) urge sociologists of technology “not only to place the consumer in the center of the network (at the consumption junction) but also to view the network from the consumer's point of view” (p. 262). By focusing on users, work by feminist scholars also conceptualizes users as a diverse group (Star, 1991; Casper & Clarke, 1998; Saetnan et al., 2000). Oudshoorn and Pinch draw on Lie and Sorensen (1996) to suggest that rather than essentializing users, “the distribution of power among the multiple actors involved in socio-technical networks should be approached as an empirical question” (Oudshoorn & Pinch, 2003, p. 7).

Finally, Oudshoorn and Pinch (2003) incorporate the work of scholars working in cultural and media studies in their users matter approach. This set of approaches, in contrast to the preceding ones, has always explicitly given users of technologies central focus, along with the cultural contexts in which they engage with technologies (Silverstone & Hirsch, 1992; Lie & Sorensen, 1996; Sorensen et al., 2000). In the “domestication” thesis of cultural studies, analysis of users and technologies looks at how people “domesticate” technologies by putting them into practice, and how the cultural context which the users and technologies are embedded in structures this process (Lie & Sorensen, 1996). As Oudshoorn and Pinch (2003) explain, domestication is understood as a dual process in which both technologies and users may change: “The use of technological objects may change the form and the practical and symbolic functions of artifacts, and it may enable or constrain performances of identities and negotiations of status and social positions” (p. 14). Clarifying how they differ from semiotic theories, Oudshoorn and Pinch (2003) explain that domestication analyses “conceptualize the user as a part of a much broader set of relations than user-machine interactions, including social, cultural, and economic aspects” (p. 15).

Bringing together concepts from these four bodies of scholarship, Oudshoorn and Pinch (2003) summarize their users matter framework as an interest in “whatever users do with technology,” acknowledging that while a technology may have a dominant use, “there is no one essential use that can be deduced from the artifact itself” (p. 1). While the framework Oudshoorn and Pinch present emphasizes the importance of turning attention to users, it is important to reiterate that this approach is not intended to ignore the factors which restrict user agency, or the effects of technologies on users but rather is meant to simultaneously account for users and technologies in relationship. As such, Oudshoorn and Pinch’s theory of user-technology relations is embedded within wider moves in STS scholarship toward looking at the simultaneous “co-production” of science and society (see Jasanoff, 2004). Oudshoorn and Pinch propose studying technologies “in their ‘context of use’—the society and the web of other artifacts within which technologies are always embedded. In short, we look at how technologies are actually used in practice” (Oudshoorn and Pinch, 2003, pp. 1-2). In this study, I apply elements of this framework, namely a users matter approach, considering how current users of online genetics matter. By applying a users matter approach, I recognize that while online genetics companies have made recent moves toward legitimizing their services as medically and scientifically authoritative (see chapter 2), the question of how users interpret and use online genetics must be investigated empirically.

A few scholars have already argued for scholarly attention to users who participate in genetic testing. Hedgecoe and Martin (2008) have offered an important critique of STS scholarship on genetic technologies and their relationships to users. They cast light on the deterministic assumptions in much of the STS literature focusing on users of genetic testing, which has often theorized that the technology is “transformational” for users’ identities.

Hedgecoe and Martin (2008) contend that rather than developing a contextualized analysis of technological development, the transformational lens may lead analysts to simply reinforce the views of the popular accounts, notably among the technology's advocates, who may have over-emphasized the power of emerging genetic techniques. The transformational approach associates new genetic techniques with novelty and "revolutionary" changes in the technical and social realms. This transformational lens consists of both critiques as well as more positive analyses. The critical lens, which consists mostly of the geneticization literature, may present a fear-based oppositional stance to genetics, with "this sense of power being shared with the advocates of the technology" (Hedgecoe & Martin, 2008, p. 820). Abbey Lippman (1991) coined the term "geneticization," to refer to the notion that genetics reduces individuals to their genes. She argued genetics could lead to individualizing health problems, attributing a person's health to their genes, and omitting the social and environmental aspects of health (Lippman, 1991; 1994). Theoretical analyses which follow this approach argue that giving users genetic health information may lead to greater health anxiety or lead to individuals seeing their identities as completely based on their genetics (Nelkin & Lindee, 2004; Lee, 2013; Pender, 2012).

Similarly, a more positive lens, such as that found in the work of Rabinow (1996), Rose and Novas (2005), and Rose (2008), also attributes a sense of power to genetics. This literature has focused on the potential progressive effects of genetic knowledge for user identity. It has elaborated on Rabinow (1996), whose initial notion of "biosociality" suggested that genetic information could transform peoples' understanding of themselves and result in new communities built around new identities connected by shared genetics. The implication is that this knowledge could have positive effects for some users, such as those who develop a collective identity as patients of a particular medical disorder (Rabinow, 1996; Rose & Novas,

2005). Rose (2008) has built on this line of thinking, predicting that direct-to-consumer genetics would lead to a “transformation” of user identities which incorporate genomic knowledge as one source of information about one’s self. Hedgecoe and Martin (2008) note that these theorists, who claim that genetic knowledge may change user identities in novel ways and may potentially complicate traditional power dynamics in some cases, have also assumed that genetic technologies are transformative (p. 820). Hedgecoe and Martin assert that there has been limited evidence that genetic technologies have had such powerful effects, at either the individual or social levels (see also Akrich, 1992). In calling for caution and more empirically-based analyses of users, Hedgecoe and Martin (2008) bring attention to the insight developed by Akrich (1992) that the early stage in a technology’s development is when expectations are most deterministic.

Fishman and McGowan (2014) also advocate an approach to studying genetics and its users which avoids applying a transformational lens. They point out that company marketing has attempted to convey the “transformative possibilities” of online genetics for users, mainly positive transformations related to user empowerment and self-knowledge (p. 29). Indeed, a current visit to 23andMe’s page about its health reports states: “These reports provide you with more insights so you can be the best possible advocate – for you” (23andMe, 2019b). On the other hand, as Fishman and McGowan point out, critics in the medical and bioethical literature have expressed concern that users will not be equipped with the genetic literacy needed to understand their results and that they may react in inappropriate ways to the information (Mason, 2017; Finkel, 2019; Badalato et al., 2017). Like Hedgecoe and Martin (2008), Fishman and McGowan (2014) also problematize the two strands of literature which have developed among social scientists studying genetics users. First, Fishman and McGowan note how the geneticization group has argued that genetic knowledge leads to genetic essentialism among

individuals and within society and maintains historical systems of eugenics (Fishman & McGowan, 2014, pp. 30-31). The second group of theorists has built on the “biosociality” lens to: “celebrate the inherent potential in ‘knowing more’ about one’s self, characterising such self-knowledge as perhaps ultimately liberating and opening up new opportunities to affect self-change and sociality” (p. 32). While some critics, such as Arribas-Ayllon (2016), have criticized geneticization more heavily than the biosociality argument, Fishman and McGowan (2014) rightly critique both lenses, contending that the meanings users give to online genetics are not yet clear. They argue that studying the user perspective is critical, rather than relying on company marketing or critical yet non-grounded theories: “Beyond the broad brushstrokes of grand theories are the analyses that can only be achieved with empirical data to draw out the complex interrelationships between identities, technologies, self-knowledge, and the future” (Fishman & McGowan, 2014, p. 38).

A few scholars have operationalized the users matter approach or elements of this approach to studying online genetics users, either explicitly or implicitly (such as by avoiding a transformational lens). However, they have all done so in studies of early users, when online genetics was marketed as a recreational product rather than a health one, leaving a gap in research on how users matter in the current context (see chapter 2). For example, Harris et al. (2016) begin their chapter on users with reference to Oudshoorn and Pinch (2003), recognizing the importance of users. Their approach to online genetics users considers “how individuals engage in scripted and creative ways with online genetic testing” (Harris et al., 2016, p. 34). By applying the users matter frame, Harris et al. recognize that many users of online genetics may not identify with medical conditions and problematize theoretical claims which have assumed

genetic knowledge transforms user identities or develops new collective identities. The users matter approach allows Harris et al. (2016) to examine how users:

Worked both on and off ‘script’ – their genetic results were created within a 23andMe risk assessment framework, yet they contributed their own interpretations following wayfaring lines, accepting and refuting findings according to their own understandings of illness, often using the results to explain what they think they already knew (p. 46).

In their analysis, Harris et al. deem 23andMe genetic tests “ambiguous products situated in a grey area between recreation or entertainment, and health” (p. 70). Elsewhere they elaborate: “The industry has played with a number of ambiguities, including whether the products they sell are medical or recreational” (p. 8). Consequently, when analyzing why the 23andMe users they studied displayed a recreational attitude toward online genetics for health, Harris et al. suggest it is partly attributed to the company’s efforts to place themselves in that “grey area” (p. 35). Importantly, however, online genetics companies currently brand their services as more medical and scientific than recreational.

In their own study of early users, McGowan, Fishman, and Lambrix (2010) also apply a more user-focused theoretical approach, focusing on user perspectives to “recognize the co-constitution of users and technoscience in the social life of the emergent technology of DTC personal genome scanning” (p. 267). They use the concept of “interpretive flexibility,” from SCOT theory, to analyze online genetics users: “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” (McGowan et al., 2010, p. 266). They borrow concepts from Woolgar (1991) and Akrich (1992) to examine the agency of technology, acknowledging that: “technologies are developed with specific types of consumers in mind, and that the ideal consumer is inscribed into the process of technology development” and “‘built-in’ technology design inevitably has impact

on and constrains the ways in which consumers make use of it” (McGowan et al., 2010, p. 266). However, they also take concepts from Cowan (1987), Pinch and Bijker (1987), Bijker (1992) and Lie and Sorenson (1996), among others, to put analytical focus on users as well, recognizing that: “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” (McGowan et al., 2010, p. 266). However, they focused on how early users matter. Concluding their study, McGowan et al. assert that their findings demonstrate: “the largely informational rather than medical value that early users attribute to personal genome scanning technology in its current state” (2010, p. 262). Much has changed since that “current state.” Like Harris et al., their study was done before significant efforts to construct online genetics as scientifically legitimate by the industry.

Other studies have utilized some elements of the users matter approach, such as by rejecting a transformational lens and recognizing that how users react to genetic results may not necessarily be transformative or even straightforward. In her study, Nelson (2016) contends that while many have assumed users of genetic ancestry testing would “unconditionally accept” the information given to them by genetics, how people engage with genetic information is more complex. In Nelson’s view, “narrative and contextual framing” is crucial to users’ reception of genetic information: “Genetic markers in and of themselves have no meaning or value [...] we learn to read the significance of DNA in science labs and at genealogical gatherings” (2016, pp. 161-162). Nelson (2016) also suggests that the reason users may not find answers through genetic testing technologies is because users often have non-scientific aspirations for genetics: “Some of the questions posed to genetic science may be fundamentally irresolvable through DNA analysis [...] the issues, controversies, and questions we pose to science about race and the

unsettled past can never find resolution in the science itself” (pp. 163-164). Thus, her rejection of a transformational lens is linked to her understanding of genetics as social and symbolic.

Similarly, O’Riordan (2011; 2013) also rejects deterministic and transformational views of genetic technologies and puts analytical focus on how users engage with online genetics, implicitly incorporating elements of a users matter approach. She does this by acknowledging that genomic information must be interpreted by users; it does not itself tell a story: “Genome sequences or scans have to be narrated through annotation, commentary, interpretation, and explanation, in relation to the life of the protagonist. [...] they make up the elements that produce the story, but they are not storytellers” (O’Riordan, 2011, p. 129). Following these authors, my analysis will not assume that the way users engage with and interpret information from online genetics is necessarily straightforward or predictable. In applying the users matter approach, I recognize that users may have room for interpretation when they engage with their genetic results, although perhaps within limits (both technical and cultural). I am interested in how online genetics technologies are used in practice, from the perspectives of users.

3.1.2 Genetics as symbolic and social

In line with the users matter approach is an understanding that genetics may have “social power” and symbolic value. I follow Nelson (2016) by being open to the possibility that users have hopes or “aspirations” which exceed the technical capacities of the technologies involved, and that genetic testing may not necessarily or exclusively be about seeking scientific knowledge. Nelson (2016) postulates that people often have “intangible aspirations for DNA” (p. 5). Thus, she suggests, DNA has a “social power” which goes beyond its actual scientific properties and the limitations of genetic techniques. She further comments that individuals who pursue genetics may often do so for reasons that are, as Alvin Weinberg puts it, “trans-scientific”

(cited in Nelson, 2016, p. 164). In acknowledging this, she aligns herself with some of the viewpoints expressed by Nelkin and Lindee (2004), whom she quotes as having said that genetics “can play a role in many different stories” (cited in Nelson, 2016, p. 7). Nelson (2016) is primarily focused on the racial politics goals people may associate with genetics. With her concept of “reconciliation projects,” her approach to studying genetics users goes: “beyond the intimate, personal quests that we have now come to most closely associate with genetic ancestry testing. [...] How individual genetic data is transformed into collective cultural politics is the centrepiece of this book” (pp. 21-22). My analysis of users is open to the idea that online genetics, which is often interchangeably called “personal genetics” (Quitterer, 2014), may not always be solely about the personal. In addition to political goals, Nelson (2016) also makes note of individual trans-scientific aspirations people hold for DNA and genetic testing. She recognizes that for online genetics participants, “Identity and self-making are primary ambitions,” and that

Questions and desires, not pure ‘science’ alone, set the terms for how a personal reconciliation project—the pursuit of African ancestry—is carried out. Consumers come to DNA testing with genealogical aspirations: with particular questions to be answered; with mysteries to solve; with autobiographical narratives they want to complete (Nelson, 2016, p. 77).

However, for Nelson, these personal goals are presented as secondary to political goals for users, and rightly so, as she focuses her inquiry on a broader social group of users—black Americans—and their efforts at using genetics to effect socio-political change at a collective level. However, my approach to studying users draws on other scholars as well in order to make room for the possibility that not all users pursue genetics for resolving wider political issues; perhaps they also do so for personal reasons (which may nonetheless also be trans-scientific).

Nelkin and Lindee (2004) illuminate how genetic information has come to symbolize self-knowledge. They have developed the concept of the “DNA mystique” to understand the cultural and symbolic value of genetics:

Clearly, the gene of popular culture is not a biological entity. Though it refers to a biological construct and derives its cultural power from science, its symbolic meaning is independent of biological definitions. The gene is, rather, a symbol, a metaphor, a convenient way to define personhood, identity, and relationships in socially meaningful ways. The gene is used, of course, to explain health and disease. But it is also a way to talk about guilt and responsibility, power and privilege, intellectual or emotional status (Nelkin & Lindee, 2004, p. 16).

According to Nelkin and Lindee (2004), at the same time that a sense of mystique has been cultivated around the gene, the study of the gene through genetic science is popularly presented as “untainted by religious, political, or philosophical commitments” (p. xvii). In articulating their DNA mystique concept, Nelkin and Lindee focus on how people seek genetic knowledge and techniques for self-knowledge. They explain the promise of genetic determinism, that “if scientists can decipher and decode the text [of genes] [...] they will be able to reconstruct the essence of human beings, unlocking the key to human ailments and even to human nature—providing ultimate answers to the injunction ‘know thyself’” (Nelkin and Lindee, 2004, pp. 6-7). Furthermore, Nelkin and Lindee (2004) suggest that “the similarity between the powers of DNA and those of the Christian soul” is not just metaphorical: DNA has “taken on the social and cultural functions of the soul” (p. 42). In recognition of the ways in which a sense of mystique and symbolic power has been socially attributed to DNA, my study of users of online genetic testing is open to the possibility that they may not necessarily be motivated by science or exclusively by science; perhaps they may perceive DNA as a symbol for something else, such as self-knowledge.

A few other scholars illuminate how biological and health information may symbolize identity and selfhood. For instance, Quitterer (2014) contends that while “at first glance”

genomic information seems to be just about an individual's biology, it is different from other biological information like blood pressure, because genomic information has often been linked to our overall "blueprint"—which is concerned with not only our disease susceptibility, but also our phenotype (p. 47). As a result, it is possible that some people imagine their genome is linked to their overall identity, rather than solely their biological traits. Quitterer (2014) calls this the "genomic principle of identity (GPI)," explaining how "genomic information constitutes a principle of identity which comprises the biological, the autobiographical, and the social self" (p. 47). Moreover, Quitterer (2014) suggests that the "GPI" has replaced "traditional identity-providing concepts like the soul or the substantial form" (Quitterer, 2014, p. 47). This concept of the GPI implies that it is possible that users who pursue health information may not always be looking only for scientific knowledge or facts about their biology or health, but maybe also for material to construct their autobiographical and social selves.

To make more room for the possibility that health information from online genetics may be used to construct identity, I draw on Lupton (2016), who has focused on users of emerging digital health technologies which fall under the "quantified self" or "self-tracking" movement. Lupton explains that in discourses of self-tracking: "detailed quantifiable data have become valorised above other forms of information about one's life, health and wellbeing" (2016, p. 64). In her approach to studying digital health technologies, health data is conceived as one additional type of data about the self. Lupton (2016) presents several principles of quantified self-tracking discourses:

Data (and particularly quantified or quantifiable data) are an avenue to self-knowledge; [...] quantifiable data are more neutral, reliable, intellectual and objective than qualitative data, which are intuitive, emotional and subjective; self-tracked data can provide greater insights than the information that a person receives from their senses, revealing previously hidden patterns or correlations; self-tracked data can be motivational phenomena, inspiring action, by entering

into a feedback loop; everything can be rendered as data; and data about individuals are emblematic of their true selves (p. 65).

Based on Lupton's insights, it is possible that doing online genetics for health may be a self-tracking practice for some users, in which users may "quantify" the self, through digitizing their biological materials (saliva and the genetic data contained within it), seeing the resulting digital data from online genetics as a form of self-knowledge.

Harris et al. (2016) also suggest that users who have done online genetics may engage with it as a form of identity development. In studying the uploaded YouTube stories of users of online health genetics, they conceptualize user narratives as "autobiology": "Derived from the term autobiography, this is a story that encompasses the biological; a study of one's life including the molecular, the cellular, the genetic, the physiological and/or other biological elements" (Harris et al., 2016, p. 36). They propose this concept as an alternative to "biosociality," acknowledging that the effects of online genetics on user identities may not necessarily be as strong as the biosociality thesis suggests (p. 35). Harris et al. (2016) present four traits of autobiologies. First, they are stories involving technologies which have "travelled outside" of the clinic (p. 36). Second: "the context of storytelling differs from the more classical sociological genre of the illness narrative, in that the storytellers are not framed principally as patients, as they are not unwell" (Harris et al., 2016, p. 36). Third, "unlike pathobiographies or illness narratives where illness becomes an important part of one's identity, autobiologies are fleeting engagements" (Harris et al., 2016, p. 36). Fourth: autobiologies are primarily about the individual or the self, in contrast to stories in patient activist groups, which may serve to mobilize patient activism (p. 36). Following Harris et al., I utilize the concept of "autobiology" to understand how users (who may not necessarily identify with or perform the patient role) interpret online genetics for health.

The theoretical claims linking online genetics to recreation and identity demand further investigation because in the current context in which users do online genetics, it is no longer marketed as merely “recreational” or “informative” to users, as it was when these previous authors applied user-centred approaches. In order to truly understand whether and how users matter, these theoretical claims must be tested in the present context, when online genetics for health has been constructed by its designers as a scientifically legitimate practice for gaining insight into one’s health.

3.2 Methodological approach

3.2.1 Qualitative research

This study is an empirical inquiry into the user perspective. I am interested in why users do online genetics as well as how they respond to its results. This entailed asking the users themselves. To access user perspectives, I chose a qualitative research approach, conducting interviews with users. Sociologists have established that qualitative research is favourable over quantitative when attempting to analyze new social phenomena that are not yet well understood (Weiss, 1994). While the popularity of online genetics is growing at a consistent rate, it is still an emergent phenomenon, with limited research on users in particular. As Harris et al. (2016) have also emphasized, online genetics is “live” and in a state of flux. Qualitative research, which remains grounded and flexible in changing terrain, is best suited to studying online genetics users. As discussed in the literature review, much of the research on the perspectives and experiences of users of online genetics for health so far has used quantitative approaches, which entail survey methods. While these have yielded some interesting trends on users at a general level, qualitative methods helped me develop a more contextualized analysis of why an individual user may do online genetics and the complex ways in which they may engage with the information they receive from it. In line with my research questions and theoretical frame, I want to understand the complex and sometimes contradictory experiences and perceptions of users. Interviews allow space for subjectivity, ambiguity, and contradictions (Silverman, 2013).

3.2.2 Data collection

The key recruitment criteria for selecting interviewees was that they had done an online genetic test which provides health information. In addition, interviewees were also recruited if they had taken an online genetic test from a site which did not necessarily provide users with

health information itself, but from which users could download their raw data and upload it to third-party interpretation sites for health information. With my focus on users of online genetics for health, I did not recruit people who had done online genetics solely for ancestry-related information. Due to the specificity of this recruitment criteria, I anticipated that finding local interviewees would be a challenge, so I focused recruitment efforts to online platforms. I made online recruitment posts, which I shared on two online platforms: Facebook (social media site) and Reddit (online discussion site). On Facebook, I posted in public groups related to online genetics. I also shared a recruitment post on my Facebook profile. To ensure potential interviewees were not too close to me and did not feel any pressure to participate, I only planned to recruit acquaintances (rather than friends) through my social network and also invited them to contact me themselves. On Reddit, I made recruitment posts on sub-forums related to online genetics. In all these recruitment posts, I described my study and invited people to contact me if they were interested in being interviewed. I specified I was looking for people who had engaged with health information through online genetics, were over the age of 18, and spoke English. I also explained the interview could be done either in person (if they were local) or online through video call (see Appendix A).

I interviewed ten people in total (see Appendix B for participant demographics). Interviews were conducted between February and April 2019. One interview was conducted in person in Ottawa, but the rest were done online using video call services (including Facebook Messenger, Skype, and Zoom). Interviews were between one to two hours. They were audio recorded. The interviews were semi-structured, guided in part by an interview guide (see Appendix C). While I had particular topics in mind for interviewees to discuss, the interview was guided more by what the participants were interested in discussing. In line with my empirical and

abductive approach, it was important for me to remain open to what participants wanted to discuss. I took on a listener role and allowed them to lead, asking follow-up questions in order to invite elaboration as necessary. Interview questions were designed to investigate users' experiences and perspectives of online genetics. I asked interviewees questions related to: why they use online genetics services; what online genetics websites they have used; how they used all the different websites; how they responded to health information; whether they shared their data for company research. I also asked about their views and opinions related to doctors, as well as online genetics companies. Additionally, I asked them how they view their own knowledge of genetics and health, and what they think is the connection between genetics and health.

3.2.3 Data analysis

In maintaining a strong empirical approach to studying online genetics users, my data analysis followed an “abductive” approach. Abductive data analysis is an approach which selects “provisional” theories to guide the development of research questions and initial themes of interest (Thornberg & Charmaz, 2014; Bowen, 2009; Lichtman, 2017). As Thornberg and Charmaz (2014) explain: “a researcher who uses abductive reasoning constantly moves back and forth between data and pre-existing as well as developing knowledge or theories, and makes comparisons and interpretations in the search for patterns and the best possible explanations” (p. 162). In this approach, researchers “take advantage of knowing and using the literature, not for forcing the research into preconceived categories but as multiple possible lenses” (Thornberg & Charmaz, 2014, p. 162). While I developed a theoretical approach, it was considered tentative, flexible, and open for revision based on what was found in the data. This approach tried to stay close to the data, being as open-minded as possible when coding and analyzing data, even while being informed by some theoretical assumptions going into the research. The aim was to be

informed by theory, but, especially in the initial coding of data, remain open and flexible, rather than disregard codes which were not discussed in previous literature. This methodological approach facilitated my goal of conducting an empirical study of users.

Applying this approach, I embarked on a detailed and lengthy process of coding interview data. Interviews were transcribed into Word files, and also coded using Word. Coding was done in steps. I began with “initial coding,” which Thornberg and Charmaz (2014) define as coding without external reference, trying to simply describe what is in the data. Through initial coding, I developed a list of around 100 codes, which I compiled in a spreadsheet file in Excel. I narrowed this list of initial codes by using Excel to highlight the most common codes which appeared in my data; codes were then eliminated if they rarely appeared across the dataset in comparison to other codes. I then grouped together the codes into broader codes, a step called “focused coding” by Thornberg and Charmaz (2014, p. 157). These codes were then integrated into broader themes. Through this coding process and abductive approach, I eventually developed a list of themes which were partly constructed from existing theory as well as my empirical data. I then analyzed these themes in relation to my research questions, which were also modified to better correspond to the generated themes.

Chapter 4: Findings

4.1 Motives

In this section, I present prominent findings from my interview data that relate to the overall theme of user motives for doing online genetics. These key findings can be summarized as: For the most part, my participants do online genetics out of curiosity, even if some also have more specific health questions about which they are curious; rather than health, participants do online genetics to learn more about themselves and their identities. Most of my participants believe in the power of the data from online genetics to tell them something about themselves.

4.1.1 Curiosity and identity

My participants pursue online genetics for health for the purpose of identity development, rather than health concerns per se. All of the participants I interviewed mentioned curiosity at different points throughout their interview and highlighted it as the key motive which initially drove them to participate in online genetic testing. For the most part, they demonstrated a recreational attitude toward the practice; That they see it as another source of identity development as further illustrated by how many of them frequently talked about ancestry interests as well. Andrew told me his reason for taking an online genetic test: “Probably 80% curiosity and interesting things, and maybe 20% awareness of health things [...] generally speaking it was more for the novelty and the interesting parts, than trying to make real diagnostic decisions off it.” Later in the interview, Andrew also said he does not have any specific questions he wants answers to through online genetics, aside from whether he has “interesting” health conditions. He links his interest in online genetics to his interest in biohacking, saying these practices relate to “the idea of knowing yourself better;” for him “genomics seemed like the natural next step” on that path of self-knowledge.

Likewise, when I asked Ben why he went on many different websites to search for information on health, he replied: “Curiosity. I don’t feel sick [...] I’m not going to change anything. [But] I still wanna know.” When I probed further on what motivates that curiosity, he replied: “Hmm, good question. Genetics is cool. [Laughs.] I wanted to learn all I could from DNA. The ancestry, what country I come from, but also health stuff.” Additionally, later in the interview he questioned the benefits of online genetics for health, calling it “fun” for learning about ancestry and interesting genetic traits, but “maybe not so [much] health benefits.” (He added that there could be a potential health benefit if someone has a serious concern like breast cancer risk.)

Evka’s curiosity was evident as she listed a long list of online genetics services she used to find information related to both ancestry and health. She said that being a genetics researcher herself, she likes exploring the health information from online genetics out of academic curiosity. She definitely does not have specific health questions she is particularly interested in because, in her view, “the genes would not give the answers.” She said for her, online genetics “is a hobby for the rest of my life: Just looking, trying. But ordinary people they just want to know some answers (or sometimes they don’t know what they’re looking for, actually).”

Similar to the others, Felix said he is “just curious about” online genetic testing and he has never expected to find any “surprise” results from it. He seeks both ancestry and health information from online genetics. He is interested in less serious questions about personal genetic traits, like why he has “ginger hair,” but he also said he is curious about some health issues that no one else in his immediate family has and any potential risk factors for health issues in the future. Ghassan purchased a health report from 23andMe after receiving an ancestry report and described his interest in health as an add-on, due to a general desire to want to know more

about “the unknown.” He indicated he does online genetics because he is “just curious.”

Ancestry came up throughout his interview, and he frequently expressed how interesting his ancestry results are to him.

Hernandez referred to curiosity throughout his interview and identified himself as “a curious person that likes to learn new things.” About his reason for testing, he said: “Well I didn’t know where I was coming from. I mean I’m not adopted, but I was still curious about it. About my ancient origins.” Later he elaborated: “I was into genealogy many years before. I spent many years around Europe, and I was curious about where I was from. I also wanted to contrast all the data from other companies. I was curious about if the calculators were working well or not.” Hernandez would often respond to my questions with reference to genealogy-related interests and results first, rather than health. However, he did say that a background question for him while doing online genetics is whether his DNA predisposes him to clinical depression, a condition with which he struggles.

Carly is one of the few users who has more specific health questions which have led her to try online genetics. However, her health questions are also intertwined with questions related to ancestry:

We have some mysteries in my family in terms of our ancestors. My father didn’t know his father, though he knew he existed. [...] And on my mother’s side we had some question marks as well because my mother’s mother died when she was just 34. My mom was a child (11 years old), and so she didn’t know her. And my mother and I both have some health issues that relate to some chronic migraines. We wondered if there’s some sort of medical basis for this. And now my son, now he’s 6 years old, he’s starting to demonstrate some of the symptoms. Severe headaches, which cause him to vomit, which is pretty unusual; but that’s the age at which it started for me. [...] So I had questions on both sides of my family, both for who we were related to and also for medical conditions, that we hoped to get some answers for.

As I will elaborate in section 4.2 (on Interpretation), Carly has not found answers to these questions through online genetics. She said that while she is interested in health, it takes “second

priority to the ancestry question.” She also noted that initially she did not have access to health reports because it was during the period when the FDA prevented 23andMe from providing that information. She got health information from Promethease and from 23andMe later, when it gained regulatory authorization to provide health reports. Throughout her interview, Carly talked at length about her ancestry interests and findings.

Notably, Dina’s interest in genetics was catalyzed by the experience of having an adopted son with a rare and fatal disease. She explained: “One of the ways I controlled my stress and all the complex feelings was just to immerse myself in the science a little bit.” She also uses online genetics to search for ancestry and health-related information pertaining to her other adopted children. While the experience of being an adoptive parent has been a catalyst, Dina said she has remained interested in online genetics “to connect with and understand all the pieces that made me, Me.” She said the “genealogy side” was what got her into online genetics at first, and she frequently talked about both genealogy and health. For her, genealogy is “mostly a hobby and sort of an academic pursuit.” Dina also mentioned she has “mysterious” autoimmune “things” like unexplained rashes, which she has been trying to learn more about through online genetics. In her view, people who have unexplained autoimmune conditions may be driven to online genetics for answers out of desperation. To sum, Dina said she is “of two minds” about using online genetics for health: “I find it interesting and a curiosity; but I don’t necessarily look to it for a solution.”

Israa originally purchased an AncestryDNA test to learn about her heritage, because she often feels “ethnically ambiguous.” She said: “I just wanted to know the individual parts of who I am.” Israa later decided to take an additional test with 23andMe for more information and because she thought it “would be a fun one to do.” When describing her conversations with her

brother-in-law about her health results from Promethease, she emphasized that they are “lighthearted” rather than serious:

He was like, ‘Hey Israa, look you have a gene for bipolar. That explains a lot.’ Things you can make fun of someone for. Like below average cup size: He was like ‘Is that why you’re considering plastic surgery?’ Like dude stop [laughs]. But it was super lighthearted and humorous. It wasn’t like ‘Oh my God,’ or ‘That’s why?!’ It was more like ‘Why did Israa waste money on this?’

However, Israa also has a more serious health question she is curious about: her breast cancer risk. Her mother had breast cancer, and Israa hopes online genetics may give her more insight into her own future risk for breast cancer. (As I will reveal in section 4.2, Interpretation, Israa emphasized she has not found any insights on this question through online genetics.) She told me: “I’m so fixated on my future and trying to figure out where I’m going to be in five years, ten years or twenty years. I like to plan ahead. I don’t like surprises at all.” She told me that if she finds out she has BRCA 2 (the more serious breast cancer mutation), she would take proactive measures to protect herself, such as getting a prophylactic mastectomy.

Jennifer identified herself to me as someone who is “interested in health in general,” and that is the primary reason she took an online genetic test. She likes learning about how the body works, and medicine and biology. In terms of learning more about herself, she is interested in whether she has any current or potential future health problems. One health question she has had in the background while testing is whether her and her partner have any variants which, combined together, could impact their newborn child (at the time, they were planning to have a baby soon). When doing online genetics, she also wonders if she can find information about the low energy levels and sleep disturbances she often experiences. However, Jennifer is also interested in “any cute little genetic variants” she may have. She said she wants her parents to get tested so she can “cross-reference” their DNA to determine: “whose hair do I have, whose eye colour do I have, or different things like that. I think that’s just really fun and interesting.” She

emphasized that, going into online genetics, she “didn’t take it super seriously” and it is “more just for fun.”

4.1.2 Online versus clinical health practices

Some participants provided reasons for using an online genetics test to learn more about their health or ancestry, rather than taking a genetic test with a doctor or through some other means. Some also noted differences between being an online genetics user versus being a patient getting tested with a doctor. Andrew believes he gets information from online genetics he would not get from a routine doctor’s visit. Throughout his interview, Andrew demonstrated some skepticism toward doctors; although, as if to reassure me, he would periodically reiterate his “utmost respect” for doctors. Most of his skepticism is directed toward general physicians, rather than specialists like genomic oncologists or geneticists. He said: “My GP [...] would not generally be in a position (with the exception of explicit guides for certain medications) to make a useful interpretation of my genome.” He also stressed that he likes being informed of his own health information. Every time he visits a doctor, he sends them a form letter to obtain all his paperwork; he has accumulated a “big medical archive” of all his medical records. Andrew also connects his interest in online genetics to a general sentiment of being anti-elitist or anti-academic and pro-open sharing:

I like being able to do research and to find my own answers. And I acknowledge academia’s incredibly valuable. [...] But any time I can contribute to outside the institution or institutions that make their data publicly available I’m 100% for that. I wanna help with that and give back to that. Like I’m building software right now to view VCF files, because I really hate just searching through VCF files to find a snip I care about, so I’m trying to write a better viewer for that. [...] Like that ability to build and be free to create because people have made resources available that help me with that and facilitate that is a great thing, so I wanna give back to that how I can. Sorry, I’m really passionate about open source and contributing like that!

Ghassan and Ben highlighted their belief that doctors usually only see an individual when they have something “symptomatic,” a condition with identified symptoms. Ghassan elaborated that seeing a doctor involves looking at the “known” (a condition with identifiable symptoms and possible treatments), whereas online genetics is concerned with the “unknown.” When Ben was telling me about how he likes to learn on his own about his genetics, at some point he interrupted himself to add: “Sometimes I also talk with my doctor, but he doesn’t like to talk about this. [...] He tells me it’s not important now. He’s probably right. Like, if I come and say ‘Oh I have the mutation for Alzheimer’s,’ he says ‘Okay, you come back when you’re 60 years old’ [laughs].”

When Carly explained why she took an online genetics test despite some concerns she has on data privacy issues, she said she does not see many alternatives. She elaborated on this view, saying that doctors are not “as interested or concerned with the ancestry portion of things, and even if we got results regarding medical things, that doesn’t answer these other questions or doesn’t close any distance for us between potential relatives.” Carly also spoke at length about her issues with doctors she has experienced in the past in relation to a variety of medical conditions and highlighted that her being a “petite woman” may have to do with some doctors being dismissive toward her. For instance, she shared that once after she was hospitalized for a “massive kidney stone,” a doctor pointed at her Sprite can during the visit and said: “See the thing you’re drinking, that’s it, right there is the problem. You gotta cut down on your calcium and all that.” She followed his advice but was hospitalized again. She finally went to a nephrologist (kidney specialist) who did testing and informed her that the initial doctor who advised her was “wrong” and “lazy” in not doing additional testing. Based on this experience, she said:

I felt like a lot of the doctors who had seen me in their office, I’m in and out in less than two minutes. They probably don’t even know my face or couldn’t pick

me out of a crowd, and they're—he was really negligent. But there are other doctors too who I think were maybe more thorough but still they were focused on that one issue and they weren't focused on the bigger picture, of this constellation of issues I was having.

Carly said doing the 23andMe test for health fits into her aim of being a more proactive participant in her health and in her interactions with doctors. She highlighted other similar actions she has taken in order to be more empowered as a patient, like ordering SmartJane kits (online tests related to vaginal health) and UBiome kits (online tests related to microbiota and gut health).

Israa said she feels more comfortable asking her family members who are doctors about her genetics than she might feel with a doctor. When I probed why, she explained that with family she is less worried about asking “stupid questions” or having to prepare in advance to know what she would ask the doctor in her limited time with them. Additionally, Israa told me: “I definitely like to take matters into my own hands because I don't want to trust anyone else with it.” Israa explained: “I feel like I have more control [doing it myself]. And I don't really feel like I need a doctor right now. Maybe like ten years ago I'd definitely go to a doctor for this. but these tests are so accessible right now that I don't feel I need it at this moment.”

4.1.3 Power of data

Most of my participants convey a reverence for the power of data. Of course, by virtue of the fact that they have participated in online genetics they demonstrate their belief in the data's ability to tell them something insightful about themselves. Interview excerpts illustrate this point. For instance, Andrew said: “Genomes are appealing to me because it's you and it's the fundamental underpinning of humanity, of life itself.” He stated that while the field of genomics is currently “young,” one day the science behind online genetics will be able to account for human health: “I think (barring epigenetics and things I think are absolutely important that there

are also answers in) speaking strictly about the answers we can find from genomics, I think we're gonna see massive health wins from the science as the science catches up to it."

Throughout the interview, Andrew referred to data as the "oxygen" of science. When discussing how online genetics results showed him a slightly elevated risk for heart disease, he talked about the difference between seeing that result "in the data" versus hearing about it through his family: "The genetic data was kind of helpful just to be a little more concrete—it's not just like things your family have said, it's like: here's the data. Here's the hard scientific data." Andrew also made a point of discussing how artificial intelligence and machine learning would benefit genomics. He talked at length about IBM Watson:

The main use of Watson has been in healthcare diagnostics. Because Watson can look at a cohort of like 20,000 people that all took the same medication and come up with records way more accurate and way faster in terms of side effects, efficacy than humans can. Not that I trust computers but not the people, but just that it's such an overwhelming amount of data, I would be far more ready to accept suggestions about treatment based upon my genomics from a digital system than a doctor who is not specialized in that. I would trust a genomic oncologist or a geneticist far more than I would trust an automated recommendation system. And I trust an automated recommendation system far more than I would trust my general physician.

Carly expressed her belief that there are some cases where someone's genetics are "just determined to make themselves known." She gave the illustration that genetics play a large role in determining a person's outcomes by talking about the autism that runs in her family.

Meanwhile, she said, other types of conditions may not be linked to genetics, such as weight issues: "Of my siblings and I, only two of us struggle with our weight and the others are fine. So that might be something that's more impacted by environment and other issues. It's hard to know. So I guess the short answer is it's variable; depends on the issue at hand."

Dina frequently criticized "genetic determinism" throughout her interview, especially when discussing practices like giving people nutritional advice based on their genetics (which

she thinks is “pseudoscience”). However, later when I probed her on how important she thinks a person’s genetics are to their development, she said: “To me, [it’s] 65% nature and 35% nurture, based on the research I’ve seen. It depends on the trait.” Dina also related that when she parents her children, she holds the belief that “genetic inheritance is a huge thing that they bring with them; they’re not a blank slate.” She talked a lot about the inheritance of temperament and mental health. For instance, she shared:

When my youngest son was younger, I belonged to a support group for parents of bipolar children. These families, the ones that were biological and not adoptive families, once their child was diagnosed, often one of the parents got diagnosed later. And then they began to say Oh we had Aunt Tilly who committed suicide, and we had Uncle Joe who drank himself to death, and you begin to see the mental health issues. [...] You don’t necessarily realize the patterns, because you know Aunt Tilly had an unhappy marriage or whatever, and Uncle Joe fell in with the wrong people in teenage years and ended up with a drinking crowd. And you don’t begin to link those things.

Dina also frequently emphasized the power of data. She said 23andMe is able to do “ground-breaking research” because they have huge datasets. She highlighted: “In the medical field if you get a study of 1000 people, that’s big—but the commercial genetics companies are sitting on data that is a vast treasure trove of possible medical information.” Dina also expressed hope that in the future, with “enough data and computing power” to find patterns in human health, the genetic science will be strong enough to completely explain human health. Most of the participants have the belief that sharing their data with online genetics companies allows them to contribute to research. For instance, when I was talking to Ben about data sharing among companies, he said: “They should do research. I’m happy to contribute my DNA for new research.”

Like Dina, Felix also expressed the belief that, with time, the science behind genetic testing will only get more accurate. Moreover, he believes the ratio of influence between genetics and environmental factors in determining a person’s outcomes is: “Maybe like 70% genetics, 30% environmental.” He later added: “Well there are some health conditions that you’re born

with that you can't change. you can't stop somebody from not having down syndrome. But you can stop somebody from developing a certain cancer if they take certain precautions, although it doesn't guarantee 100% success either."

Ghassan said that in terms of its value for health, "[genetic data] gives you a guide, but it's not like the outstanding authority either." Yet, Ghassan indicated that seeing his genetic results for ancestry changed some of his pre-existing perceptions of his ethnicity and identity, making him see himself as "less French Canadian" after doing the test. He also summed up his belief in the power of data with this quote:

If you have this company that's like I'll give you this insight about whatever, then you're gonna want to do that. And based on the results you get you may sort of make that more part of your lifestyle in terms of learning things. And that goes with everything, in a sense, right? Like people read horoscopes and then they take that. I don't know if you do, but people look at that. People believe a lot in how certain things, I don't know, astronomy or whichever, Chinese New Year. Religion is a big part too. People always like getting that outside source that complements something in their life, to develop more about their identity.

Hernandez' initial view of what genetics could tell a person about their health was: "Everything. They [are] still discovering every day new stuff and new things. They can tell us everything. It's just codified over there. I see it as a strong encryption that we're trying to decrypt. I mean everything [is] encoded over there." It was only after I probed whether he thought any other factors contributed to health that he altered what he said and added that genetics makes one "predisposed" to certain things. He then went on to give a detailed example of how environmental conditions can affect health: he said that gluten intolerance has spread across Spain (where he is from) now because of the switch to a more productive type of non-organic wheat and conventional farming rather than organic farming. He later said the genome "is like a song. I mean you can write a song, but the way of singing it can be different." Overall, however, Hernandez expressed the view that to get real insight into himself and his health he

needs more data, such as that offered through whole genome sequencing (at the time of our interview, he had just sent his genetic sample to one more online genetics company, Dante Labs, for this reason).

Israa expressed that the reason she does online genetics is because she thinks that genetics can tell her something that other sources cannot. She elaborated: “I eat really well and I exercise really well and I don’t consider myself to be a stressed out person. So I think I just wanted to know more about that last factor [genetics].” She also holds the view that the science behind online genetics will one day be robust enough to explain more of human health; for instance, if online genetics companies started whole genome sequencing (she was unaware that some companies do currently market such a service).

4.2 Interpretation

In this section, I present key findings from my interview data that fall under the theme of interpretation. These findings can be summed up as: My participants, most of whom identify themselves as more knowledgeable and scientific than other users, have not found meaningful or insightful results through doing online genetics for health. Most of my participants present themselves as engaging with online genetics in a critical way.

4.2.1 Results

Most of my participants do not feel that they have found meaningful results or gained insight into themselves (regarding health or ancestry) through participating in online genetics. As mentioned, Andrew does not have any specific health questions he seeks to answer, aside from being curious about whether he has “any interesting medical conditions.” He highlighted that “the answer was mostly no.” He went on to elaborate: “I didn’t find out anything I would consider lifechanging or upsetting. Like I didn’t find out I had Parkinson’s or Alzheimer’s or that kind of thing, which I know really doesn’t rule out the risk, but it does make it less of a certainty. Beyond that, there were some things that statistically speaking everyone’s got some of.” Similarly, while Ben was telling me his dislike of the way Promethease generates health reports for users, he interjected: “In any case, I never found anything, nothing.”

While Carly was telling me about the questions she is interested in when doing online genetic tests, she added “none of my tests ended up revealing anything.” Later on, while discussing her interest in learning more about possible genetic vulnerabilities to migraines and brain aneurysms, she said: “But the testing didn’t really give any information about that, because they only have so many conditions and things that they’ve mapped.” She also said that while she continues to keep up to date with new things she can test for in online genetics, she still has not

“found anything significant or anything that relates to this specific issue that we have questions about.” Even on the ancestry side, she said she has not located relatives from the branch of the family she has hoped to learn more about through online genetics. Additionally, Carly said she does not even bother with using third-party sites for health interpretation: she used Promethease once, did not find anything interesting, and never went back. In sum, she said that when she got her results from online genetics:

I was curious and I was excited to read through them, but then didn’t really find much. Which you know sometimes no news is good news, right? [Laughs.] There was nothing there to alarm me. But: there was nothing that was really insightful either—at least for the specific issues that I was most concerned about.

At one point, Dina said she has found some useful information from online genetics when it comes to a few questions she has had related to the genealogy and health of her adopted children. However, in the rest of the interview she presented herself as holding a critical stance toward online genetics and explained how because of this skepticism she has not find meaningful insights from engaging in the practice. For instance, when explaining that she does online genetics out of “curiosity,” she added: “And I didn’t see anything big that popped out of it [...] So it’s more at this point: I use my knowledge to help others not be stupid [laughs].” Her reference to helping others is about her activity on online discussion forums, where she still participants for the purpose of advising other users on how to interpret their online genetics results in a critical way. She said of her online genetics results: “The primary finding was that there’s nothing in my genetics in terms of today’s science that pops out.”

When Felix told me why he does online genetics, he said he is mostly curious but has a few health-specific questions in mind. He then said: “And when I run [my data] through Promethease, I got some more answers.” He called some of these answers “a little unexpected.”

However, he eventually said: “I didn’t see anything absolutely shocking like Oh my God, I can’t believe I discovered this.” Ghassan also indicated he has not found find anything “exciting” through online genetics. In fact, he said he was “expecting more” when purchasing health reports from 23andMe. When I probed him further on why, he said:

[For] my ancestry results, I’ve got a nice colourful picture: a little bit of this, a little bit of that, I’ve got a little bit of variation. I was a little surprised about certain things that I had. So when I upgraded to the health report I was like Oh, well maybe I’ll get even more surprises. Especially with the fact that the ancestry is nine reports and then there’s 85 things that you could be susceptible to [for health]. [...] I guess because there’s only like two predispositions that came up, I was just like I expected a little bit more in that case, because I was a little bit more surprised with the ancestry part. [...] I guess I thought I’d have more at stake or something.

Despite having more of a health-specific question (regarding her breast cancer risk) than other participants, Israa also expressed throughout the interview that she does not feel online genetics has given her any profound or meaningful insights into herself or her health. For instance, she said: “Really what Promethease was telling me was things that I already knew, such as a small increase of my risk for breast and ovarian cancer. And telling me that I already have dark hair and dark eyes. So there wasn’t anything ground breaking. There wasn’t anything super shocking.” She also said that although she did 23andMe to find out health information she might not have gotten from the AncestryDNA test, so far she has not discovered anything new about herself through it.

4.2.2 Not an exact science

All of my participants believe that the results of online genetics vary and the information they receive from it is open to interpretation. They often acknowledged how different testing companies, services, doctors, and sources have different ways of interpreting the same data. Additionally, some of the users discussed the doctor’s role in interpretation. Andrew emphasized

his view that interpretation of his genome could differ between different types of healthcare providers. As mentioned, he would trust the interpretation given by a specialist like a geneticist rather than a general physician. At one point he said this is because: “There’s no one right answer [...] we are just not at the point with the science where there’s one right decision based on your genome.” This is the same reason he said he does not make health decisions or diagnostics based on the results of online genetic testing.

Ben often criticized services like Promethease throughout the interview. At one point, when I was asking him about the difference between doing a genetic test with a doctor versus online, he said the raw data can be the same, but “the interpretation can be very different.” When I asked him how, he replied:

[Promethease] makes people scared and the doctor would be better; saying it’s wrong magnitude don’t worry, it’s not important for you. [...] There’s many posts online with people being super scared of stuff, and they wouldn’t have if they didn’t test. [...] If they do it with a doctor, the doctor will tell them not to worry. But I don’t think a doctor has time for that, for everyone.

Carly said that, “ironically,” because she has not found answers about her migraines that she had hoped to find through online genetics, “I finally decided that I needed to just go to a specialist.” Later, when I asked how her role as a patient with a doctor (doing a clinical genetic test) may compare to her role as an online genetics user (doing the test online herself), she said that while a doctor may give her more “explicit answers” to questions she has, she would likely also have a more “passive” role in the interaction. She believes that doctors usually use their discretion and tell patients what they think they “need to know.” At another point in the interview, Carly said people would benefit from following up with a doctor if they actually find a serious or significant result from an online genetic test. She stressed:

Then you need to take that information and you need to go to a specialist, a genetic specialist or doctor to get more help with that . [...] So yeah, I don’t think that the sites obviously—and they tell you as much, really, when you read all the

legalese [laughs] and all the information, look: ‘We are not medical doctors giving you advice, we’re just giving you this information, we can’t tell you what to do with it. You’ve gotta figure that part out for yourself.’

Dina also said someone should follow up with a geneticist if they actually find health information related to a serious condition, to confirm it. Dina herself has brought up her online genetics results to her immunologist and has also mentioned her macular degeneration result to her retina specialist. On another note, when Felix was telling me why he would rather have an online genetics service that gives him as much raw data as possible (versus one that offers more high-level, summary reports), he explained:

I would like to have the bigger picture so I can have more room to interpret it myself. Because it’s not an exact science. There are lot of studies that believe a certain gene can cause this, but then other studies will say the same gene can do the opposite thing. So if one website says this gene will definitely give you cancer and another study says it definitely won’t, I don’t want to be worried that I’ll get cancer when there’s only a 50/50 percent chance, I might actually not. I’d like to have the full picture.

Ghassan has only used 23andMe. When I asked how he feels about other types of online genetics services, he said that “someone like him,” who does not have a “natural sciences” background, would likely not benefit from having access to raw genetic data. He added there is no “protocol” for him to determine which interpretation of his genetic data is correct or not. He also said he would expect that the information from a doctor’s genetic test would be different in how it is interpreted or presented to him. Hernandez also presented the view that genetic data is open to interpretation. One of the reasons he uses many different online genetics services is “to contrast all the data from other companies.” This is the same reason he would rather get genetic results from a doctor and from online genetics, rather than choose only one of these options. He said the reason getting raw data is important to him rather than getting a more summarized interpretation of his genetics is because: “I don’t trust [testing companies]. They are not doctors

or geneticists. They're just people that know about computing or bioinformatics. They're not doctors."

Israa highlighted that some of her family members who are doctors are an invaluable resource for her to interpret health information from online genetics. She has asked her sister and brother-in-law questions about her results. She related:

They were able to understand what I was asking them right away and explain it to me. Because there was even one result where it was talking about some blood thinner metabolism, and I was like what the f--k is this?! They were like it's a blood thinner, and now you know not to take too much of that, I'm like okay. If I didn't have them there to ask more questions, I probably would have been more confused.

4.2.3 Expertise

Most of my participants constructed themselves in the interviews as advanced users of online genetics. They have a self-described high-level of knowledge in terms of their understanding of online genetics. Andrew defines his expertise as "far above average for an amateur and embarrassingly little for a professional." Likewise, Ben calls his own "slightly above average," noting that he studied biology. Carly said her expertise is limited because her academic background is in English and library science and she does not have a science degree. However, she said that since she is a research librarian at a university serving nursing and pre-med students, she feels she is "a little more knowledgeable than the general public, but by no means an expert." Dina told me she probably knows "more than the average person" about online genetics, adding: "I read a lot of heavy-duty studies when my son was sick." Evka definitely considers herself more of an expert user compared to the average user; in much of her interview she questioned whether the "lay user" should even be doing online genetics for health. Felix also said his expertise is "maybe slightly more than the average person, since I've done some research on it," but: "I'm not a professional or anything."

Hernandez demonstrated that he felt his expertise is above average, since, when talking about Promethease, he said: “Well, [it] was fine for beginners. When I started with bioinformatics or computing I didn’t understand a lot, so Promethease was cool and fine to understand a bit more.” Additionally, like Dina, he said he continues to participate in online genetics forums to advise other users on how to use online genetics and understand their results. Similarly, Jennifer has a PhD in Psychology, and therefore some statistical literacy, and considers herself better equipped than other users to understand the validity of the information from online genetics.

Ghassan was one of the two participants who stressed that he does not have an advanced level of knowledge related to online genetics and health: “I will say that I worked at a hospital, doing data, not anything aside from that. But, at the same time, I stopped science after grade 10.” However, the other participants also sometimes expressed doubts about their level of expertise. Israa, for example, expressed that she does not know as much as a biology major like her should know about genetics, in her view.

Despite the participants describing the limits of their expertise to me, most participants said they nonetheless prefer to engage with their raw, uninterpreted genetic data themselves and emphasized that they prefer being able to analyze their raw data to draw their own conclusions/interpretations for themselves. Andrew stressed his interest in seeing his raw genetic data strongly throughout his interview. He said: “With Dante Labs’ results I’m getting raw genomic data—I’ve been focusing on learning a lot more, learning about bioinformatics pipelining.” He also talked a lot about trying to get access to source files rather than reading health reports. At one point he stressed:

I absolutely 100% want all of my data. No questions asked. That is completely a top priority for me. In fact, I would say that is the top priority. I would be willing

to get my genes sequenced by a company that had lower read depths, or you know, more read errors, but get the raw data than a company that would guarantee me a perfect sequencing but wouldn't give me any of the data.

Dina said that she prefers seeing her raw genetic data and interpreting it on her own, clarifying: “For me, I would rather go solo, but I think I’m unusual that way. I think most people could use support.” Evka holds a similar view, saying that while health reports interpret raw data, her final goal is to do that for herself: “I was there basically for the data. [...] But I think ordinary people should get more the services which have the interpretation.” Felix said Promethease is his favourite online genetics site to use. He added: “Some of the other websites give you a basic overall, more general kind of idea. But this, the SNPedia stuff goes pretty in depth. And it’s updated on a regular basis.” He said that while Promethease may be “difficult to navigate for somebody who’s not familiar with it,” it is “detailed” and that is what he prefers. Hernandez emphasized the importance of him being able to access raw data, as well as data from his whole genome (he recently sent a genetic sample to Dante Labs at the time of the interview, because they promised to do whole genome sequencing). Israa said part of the reason she did the 23andMe test, after having already done one from AncestryDNA, is because she knew she could get her raw data from there and upload it to Promethease. For her, she feels that 23andMe and AncestryDNA are “user-friendly,” while Promethease has “a learning curve.” However, she prefers using third-party sites like Promethease and accessing her raw data file because: “when you download your raw data file all the genes are listed with the specific letter of the DNA; so if there’s one gene that I wanna know, I can open up the Word file and CTRL+F for it and find it and look at what my sequence is.”

Ben sometimes seemed torn over the value of having raw genetic data available to users in general, but in his own case, he prefers being able to access his raw data. He said: “It’s a very difficult question. But I think maybe generally it’s a good idea [for companies] to give all [raw

data] but be clear. As a user I'd like them to give all but also, if they give all, they should state some things are for sure, some things are maybe sure, some things are very unsure." At the same time, he thinks 23andMe is "limited" in terms of what it gives to users. Carly said she prefers 23andMe health reports versus services like Promethease, because: "It's easier to make sense of the results and it's easier to move through the information on the site." Ghassan also said he finds the health reports by 23andMe "pretty comprehensive," and does not see a need to get his raw data. He explained:

As a consumer I wouldn't find it very helpful. For me, I'm doing it so that way I get information about myself interpreted, versus just getting a bunch of sequence of whatever data. I'd be like Whoa what do I do with this? I personally wouldn't know. I didn't go to university for natural sciences, so there's only so much I could do with that. [...] I understand however that the trick is that you know if you do leave it to interpretation, then well, by virtue of interpretation, you may get possible distortions of some information, or you know the confidence of the findings may not be optimal. So you have to be careful who your provider is in terms of that interpretation as well. But I don't know. I'm pretty happy and okay.

4.2.4 Critical engagement

One of my most significant findings is that all of my participants, some more than others, demonstrate a critical engagement with online genetics, which they associate with a "scientific" approach. Andrew's critical attitude toward online genetics was evident throughout our time together. When discussing why he did the test, he emphasized: "With the exception of a couple extremely well-studied areas, the general idea of making health decisions purely off your genomic data is a little fool-hardy at this point." Additionally, overall, he conveyed that he is always critically engaging with the information provided to him through online genetics. He spends a lot of time doing research and trying to investigate and verify the health information presented to him by different online genetics services. One quote in particular illustrates his critical engagement with the information and the type of research he often mentioned doing:

[SNPedia] only takes you so far, but it is a helpful kind of hitchhiker's guide to the genome I think, at least for the remarkable variants that we've found. Again, not that that's diagnostic in any way, but at least it tells you what paper should I go to read on that, you know? It's a really helpful appendix. It's like a Wikipedia reference section on steroids. There's a nice little summary of the abstract, that's nice; and then [you figure out] is this worth my time to go and read? And there have been some that have been really scary. Like you have like 25 times increase risk of prostate cancer: like, oh sh*t, let's go look at that study. [And then it says]: 'Our cohort of nine individuals.' Like okay, no, we're disregarding that [laughs], that is worthless.

Likewise, throughout the interview, Ben made skeptical comments on what online genetics could give to users. Notably, however, in much of the interview he critiqued Promethease in particular, saying they are "overselling" or exaggerating the capabilities of the service within their promotional material. For instance, he said that when his friend received a report on being predisposed to a mental illness, he objected that online genetics cannot be used to analyze mental health issues. He also criticized other online genetics applications he used, saying they have the "same problem" as Promethease: "They want to say a lot about everything, but they don't acknowledge limitations very well." Specifically, he said that online genetics services "do wrong for diseases with many snips in them." He conveyed that online genetics may be more useful for those rare diseases which can be identified with a few key mutations (the examples he gave are serious conditions like breast cancer). Later in the interview he also stressed the importance of doing research and engaging in online discussion forums when gathering information online; he thinks reading and believing information from one source alone is "stupid." Ben is in a lot of online discussions forums related to online genetics. In his critique of Promethease, he highlighted a key frustration for him is that he feels many of the forums are often hosted or moderated by online genetics companies. He added that he wishes there were forums run by experts or those affiliated with academia to discuss online genetics.

In the interview, Carly also demonstrated a critical attitude toward online genetics, despite being one of the few participants who has more specific health and ancestry questions that have driven her to pursue this practice. She is critical both in terms of what online genetics can tell users, as well as regarding her experience of being a participant in research done by 23andMe. As for what online genetics can tell someone about their health, Carly believes it can show someone their “risks,” but she believes that people often misunderstand their results. She complained about seeing misguided interpretations of users on online discussion forums, with people asking “It shows I’m at risk for X: How scared should I be about X?” She emphasized her view that online genetics cannot answer such questions. Much of Carly’s criticism of online genetics was expressed in the context of discussing a weight loss study by 23andMe that she had tried to participate in. She gave a detailed account of that experience, which she called “a total mess”:

They rolled it out, they didn’t give us the information in time. They were sending survey updates of our progress when they hadn’t even sent us the diet instructions. They had separated us in three different groups. I didn’t even know which group I was in yet. When I tried to find the landing page for the program of the website, one didn’t exist. So I had to go to the news announcement page for this rollout and post comments in their comments section. And I saw like a dozen other people with the same questions. [...] I got so frustrated, I decided look: this is not how real science is done. [...] I said look I’m opting out because this—everybody’s not starting at the same time, not everybody has the same information. [Laughs.] How can you trust the legitimacy of the results, when there’s total lack of consistency between your three different groups and your controls? [...] That whole experience definitely made me feel less great about 23andMe having my information.

As mentioned, Dina often differentiates between herself as a more knowledgeable online genetics user and those who she perceives as less knowledgeable and more likely to engage uncritically with the information provided by online genetics. While telling me why she took the test, she paused to emphasize that as online genetics has gotten more popular, many of the user base is “not super scientific,” and “tend to think of genetic determinism.” She mentioned that she

has a background in statistics and model-building in empirical sociology, which helps her to better contextualize the results of online genetics, realize that “correlation is not causation,” and that the results often have to “be taken with a big grain of salt.” She often brought up naturopaths in her interview or those who make nutritional recommendations to users based on their genetics, criticizing them as “pseudoscience” and “genetic determinism.”

Evka expressed a very critical attitude toward online genetics during our interview, especially about its value for other users who she considers less informed than her. She acknowledged the limitations of the information for learning about one’s health. She said many users do not realize that “when they have a snip for something, there is 98% or something that they don’t know about the same gene, the same pathway. So they usually say I found this snip and it explains everything—because it does not, you don’t know the rest of it.” Evka also often expressed concern throughout her interview about “laypeople” misunderstanding the results of online genetics. For example:

Most of them think one snip can solve everything and [ask] questions like: ‘I found this snip about schizophrenia: does it explain why my dad was weird?’ And it’s like no [laughs]. And more often they say ‘I have no serious variants, which means I’m completely healthy and that’s just great.’ Which is also wrong. Well first of all I would say they have pretty big expectations from the health reports, at least. Because they expect answers, many of them expect answers because why else would they do it?

While Felix thought that genetic science as a broader field has yielded some important insights into the causes of disease, he said there is a lot of uncertainty in the science. He said: “Ten years down the road the science behind online genetics would be more reliable than it is now.” I probed him on what he thinks it would be like to be a user in ten years. He responded: “I might not have to do as much research because the research that would be out there would already be more solid. So I wouldn’t have to constantly check to see if there’s an update on this gene or this study or this result.”

Hernandez performed a critical attitude throughout his interview as well. As discussed, one of his motivations for using many different online genetics services is to compare the information from different “calculators” or algorithms used by the companies. He said that when he first looked at his genetic results: “I accepted all the information but I had also some doubts. Like for example, where Italian calculations came from, other calculators from other companies didn’t find it, things like that.” As mentioned, he does not fully rely on interpretations from any one source, as he said that he recognizes that the sources providing the information are not doctors.

Israa highlighted that Promethease (which she used for health information) is using what she considers to be a limited methodology to analyze her DNA, by not looking at all of her genomic data. She thinks that she is not “getting the full picture” from genetic testing because of this issue. When she was talking about her interest in possibly trying whole genome sequencing, she paused to note “but I know that it’s not perfect technology.” That is also the reason she has ordered different tests. She said when she interprets genetic information, she tries to use “a little bit of logic,” and note things like “if 40% of AncestryDNA users have a bad gene that only 5% of the human population has, then there’s obviously a huge error going on.” One quote that sums up her critical but open attitude well is:

You really have to know what you’re looking at when you’re doing it. You can’t be emotional or hysterical. You have to understand that you can’t change your DNA. And that it’s not a crystal ball—anything can happen. Just because it says you have a risk for something doesn’t mean it’s gonna happen. And it’s possible you could get a disease that your DNA didn’t call for. So it’s not a set in stone thing. But I think if you’re really interested, know what you’re looking at, and how to interpret the information, and having resources that will guide you when you don’t know what you’re looking at; I think it’s a positive experience.

Similar to Ben’s views on Promethease, Jennifer repeatedly expressed that she feels 23andMe does not do a good job of highlighting the limitations of their health reports. She said

that while the health reports themselves are straightforward and simple enough for a “layperson” to understand: “for somebody who knows nothing about genetics or statistics or anything like that, it’d be nice for them to know that this, while it’s still fun and interesting, it’s not 100% reliable.”

Chapter 5: Discussion

In this chapter, I discuss the implications of my findings for my research questions. This chapter is divided into two sections, one for each of my two research questions.

5.1 Motives

The first research question I address, related to user motives, is: Why do people do online genetics for health? My findings indicate that: All of my participants are driven by a curiosity into themselves, seeing health information as just one more source for gaining self-insight. In addition to this main motive, some also have specific health questions they wish to answer. Additionally, some of my participants are also interested in potential social benefits through online genetics, like challenging expert control of knowledge and contributing to novel medical research. However, these motives are not important for them, and they are skeptical about online genetics' ability to deliver on them.

5.1.1 Health-related questions

All of the users hope to learn something new about their health through online genetics, which they hope to then integrate into the broader, more general self-learning experience. In addition to this general aim, four of the users are more strongly motivated by specific health questions: Carly, Dina, Israa, and Jennifer. Importantly, even these users told me they are primarily motivated by curiosity. For two of my participants, Andrew and Carly, online genetics is just one more tool through which they try to be engaged in their health. Andrew also collects all of his medical records and is interested in biohacking. Carly engages in similar practices, like online SmartJane kits for vaginal health and uBiome kits for gut health.

These types of health-related motives are similar to those which other scholar have found in relation to the use of digital health technologies. Lupton (2013) explains that for users of

emergent digital health technologies, including online genetics, “patient consumerism” connects the ideals of patient engagement with digital medicine to promote the “digitally engaged patient” (p. 258). She argues that users believe that being an engaged and healthy patient requires gathering a variety of information about one’s biology, genetics, habits, and physiology (Lupton, 2013, p. 260). In my study, Carly similarly emphasized the importance of getting a “holistic” understanding of her health through doing online genetics. This finding also supports Ruckenstein’s (2017) point that some users of online genetics may wish to challenge the Foucauldian “clinical gaze” they feel when they see doctors for certain health problems. One of Ruckenstein’s participants highlighted that for some health issues, doctors do not see patients in a holistic light, which was part of this participant’s motive for using 23andMe (p. 1032).

My findings also indicate that missing family health history may motivate those users who have more specific health concerns when pursuing online genetics. Other researchers have found that people with limited access to family health history are motivated to pursue practices like online genetics (Lee, 2018; Crouch, 2015). For instance, Crouch (2015), who focused on clinical genetic counselling, found that some parents of adopted children may feel that they are lacking information important to their understanding of their children’s health, and “many viewed [whole genome sequencing] results as a way to fill in these gaps in knowledge” (p. 67). In my study, Dina first got into online genetics when her son was diagnosed with a fatal genetic disorder. Having adopted all three of her children, she decided to learn more about their family health history. Additionally, a few of my participants connect the personal health questions they have to issues that they have not found answers for in their family health history. For instance, Carly frequently talked about how not having known her father has made her more interested in using genetic knowledge to learn more about herself and her health. The rest of the six users did

not explicitly mention any health questions they are specifically interested in, aside from a general interest in learning more about their health. Now, let us turn to the motive that users are primarily focused on: self-insight.

5.1.2 “DNA mystique”: Self-insight

For the most part, my participants pursue online genetic testing with hope that it will tell them something interesting or insightful about themselves, with their health conditions or traits or their ancestry being just individual components of their identity. For instance, Andrew highlighted his goal with online genetics of “knowing yourself better.” Similarly, Dina explained that she does online genetics to discover “all the pieces that made me, Me.” Evidently for participants, health information from online genetics is just another source of information about the self. To further illustrate how taking online genetic health tests is primarily about self-knowledge and meaning-making for these users, consider that many of this study’s participants (eight out of ten) frequently brought up ancestry throughout the interviews, even though my focus was on health. Two of the ten users (Andrew and Jennifer) did not talk about ancestry as prominently as the others, although even they had taken an ancestry test before. As for the rest, they frequently talked about how interesting their ancestry results were. Most of the interviewees actually started their engagement with online genetics through genealogy or ancestry testing, where they subsequently sought health information. One quotation perfectly illustrates how my interviewees approach online genetics with the same intentions of identity development as used by those practicing religion or philosophy; Ghassan said:

If you have this company that’s like I’ll give you this insight about whatever, then you’re gonna want to do that. [...] And that goes with everything, in a sense, right? Like people read horoscopes and then they take that. [...] People believe a lot in how certain things, I don’t know, astronomy or whichever, Chinese New Year. Religion is a big part too. People always like getting that outside source that complements something in their life, to develop more about their identity.

This quotation indicates a perception among my participants that online genetics and the science behind it can reveal something insightful or fundamental about themselves. This supports Nelson's (2016) insight that users genetic technologies seem to have expectations or hopes which may go beyond the actual technical capacities of the technologies (p. 77). My participants seem to have the types of "metaphysical" or "trans-scientific" aspirations Nelson (2016) highlighted people holding for new genetic technologies (p. 164). Additionally, Nelkin and Lindee's (2004) concept of a "DNA mystique" has relevance to my research. The notion of "mystique" captures the sense of reverence that most of my participants hold for genetic data. Nelkin and Lindee argue that in this age of genetic essentialism, people have not necessarily stopped asking questions related to the meaning of one's life or one's selfhood, but they have just shifted away from the traditional sources to which people previously put such questions, like philosophy, religion, or the humanities, instead looking to genetics to answer these questions (p. xviii). My study adds to this literature because it reveals that my participants appear to derive meaning from figuring themselves not as genetically essentialist per se but as scientifically-minded and well informed by facts and current techniques; thus, they pose meaning-making or identity-building questions to practices they consider more scientific, such as online genetics.

The concept of the DNA mystique presents an analogy between faith in religion and science. To further understand the relevance of this analogy for online genetics, consider the following quote by Lone Frank (2011, p. 146), on what she thinks people are ultimately looking for in personal genetics:

Of course, health is important, but when it comes down to it, our diseases and infirmities are not really the most interesting thing there is about being human. [...] Where it really gets exciting is at the intersection between the shell of the physical being and the person we recognize as human. How do we get from genes to what we call, for lack of a more scientific term, the soul?

Frank is a science writer who has written an autobiographical account of her journey through personal genetics, titled *My Beautiful Genome*. Throughout the book, she emphasizes her belief that genetics will help people realize their purely biological underpinning and provide scientific answers to questions people would typically put to other sources, including spirituality. As a science writer, she has much in common with the majority of my interviewees. The quotation above echoes the one by Ghassan in my study: casting personal genetics as a search for self-insight and meaning.

5.1.3 Recreation

Through her ethnographic research on African-American users of direct-to-consumer genealogical tests Nelson (2016) concluded that “forms of genetic testing considered ‘recreational’—such as those used to infer an individual’s ancestral origins—can have as great an impact on society as testing in seemingly more weighty sectors like medicine” (p. 27). In her book *The Social Life of DNA*, Nelson problematizes a widespread view that hierarchizes importance in different forms of genetic testing where health and medical practices are at the top. My participants trouble this hierarchy in a different way; the users I interviewed, who have sought out online genetic health information, highlight their recreational motives. Participants discussed how they are largely drawn to this practice through curiosity and questioning its truth and personal and societal impact. For example, Ghassan said he does online genetics because of curiosity about the “unknown.” Likewise, Hernandez said he does it because he is curious and enjoys learning “new things.” My interviewees often emphasized that they like doing online genetics in their leisure time, with Dina and Evka referring to it as a “hobby.” Indeed, even the participants who conveyed more specific health motives also identified curiosity and recreation as their key motive. For example, Jennifer called it “fun and interesting,” and said she is

interested in “cute little genetic variants” she may have. Additionally, even though Israa tested to learn more about her breast cancer risk, she recalled that when she later had a conversation with her sister and brother-in-law (both doctors) about her Promethease results, it was “super lighthearted and humorous.”

The sense of recreation that my interviewees approach online genetics with is consistent with what Harris et al. (2016) have found in their research on online genetics users. They analyzed YouTube videos uploaded by people who had taken 23andMe genetic tests (which included health reports). The videos consisted of “unboxing” their test results for the audience. Harris et al. also analyzed Lone Frank’s account of her journey through personal genetics as a practice related to identity formation. Through analyzing these narratives, Harris et al. concluded that the users represented in them did not have health concerns and thus did not hold serious attitudes toward their results. Harris et al. speculated that this may be because the users do not associate with a particular patient identity and perhaps also because of the medium of YouTube, which may invite a more “playful” attitude and a “casual performance” of their narrative (p. 50). However, for my participants, the former point is relevant: they hold a playful and casual attitude toward online genetics because they are not patients. As many of them emphasized to me throughout their interviews, they are not “sick,” and they engage in this practice largely out of “curiosity”—for fun. Ultimately, the casual, recreational attitude toward online genetics may be more prominent than Harris et al. anticipated, and not just an attitude that is performed by users who have uploaded their accounts to YouTube. Harris et al. also suggested that users of 23andMe pursued the test for recreational reasons because 23andMe branded itself as a recreational product (at the time, before they decided to seek FDA authorization later on, they were trying to avoid regulatory scrutiny by marketing their health information as merely

recreational) (p. 35). My study suggests otherwise and contributes to existing literature by demonstrating that despite recent efforts by online genetics companies to construct themselves as medically and scientifically valid sources of health information, the current users in this study have interpreted online genetics in different ways, pursuing it for purposes of recreation and identity formation.

5.1.4 Social motives

Nelson (2016) found that the lens of genetic essentialism did not fully account for the motives of the users she studied, which included both personal motives related to identity but also the social motives she calls “reconciliation projects” (pp. 21-24). Thus, Nelson stressed that in terms of ancestry testing, genetic essentialism misses the point, because users are concerned about much more than themselves. Unlike Nelson, for my users it is evident that online genetics is very much about personal identity and meaning-making for themselves, as individuals. For the users I interviewed, individual motives are more important than social motives. Moreover, I have found that while some of my participants are also interested in social or political causes, these motives lack the sense of profundity or urgency that is emphasized in Nelson’s research.

The social motives of my participants are twofold: user empowerment and participation in scientific knowledge formation. Some participants hope to challenge expert or academic control of knowledge and information related to genetics and health, including experts like doctors as well as academics. For instance, in Andrew’s view, conventional holders of knowledge (such as the academy and doctors) appear to have a monopoly over genetic data and science. He hoped that sharing his data in online databases would allow him to contribute to “open source” institutions with “publicly available” data. Similarly, Israa and Carly’s comments regarding the potential for online genetics to empower them in interactions with doctors reveals a

perception among some of my participants that online genetics can facilitate user empowerment. Furthermore, Ben stated that academic articles are not necessarily a better source for users to seek genetic information. Hernandez told me that in his eyes, online genetics companies and universities are equally trustworthy (or rather, he remains suspicious of both types of institutions). Thus, notions of user empowerment and challenge of traditional expertise is the first type of social motive a few of my users are interested in. Similar social motives were found in Ruckenstein's (2017) study of online genetics users: some of her participants were content that their data was "no longer available only to the professional elite" (p. 1035).

Almost all of my participants are interested in contributing their data to novel research and science which addresses unanswered questions in human health or genealogy. For instance, Dina said:

I feel really good about contributing my data. In fact, to some extent, I feel that it's a way of achieving immortality of a kind. My record will be there forever. And whether it's for genealogy or for science, the record of my variants and all the [health questionnaire data]. I feel that it is a privilege to contribute to it to be honest.

This motive has some similarities to what Nelson (2016) found: her participants believed genetic testing could contribute to knowledge and education about the history of black people and their historical oppression in the United States. However, her participants were motivated by political causes rather than scientific; whereas, my participants are motivated by the hope that contributing their data to genetic databanks could fuel scientific research, either by the company or by third-parties that the companies share their data with. They hope this advances knowledge about the causes of disease and treatment. Likewise, Berrios et al. (2018) found that "altruism" partially motivated those who chose to enroll in genetics research done through genetic counselling clinics (p. 266).

It is probable that participant perceptions that they can contribute to social causes has been encouraged by company marketing, as it is in line with the messages that online genetics developers and executives have used to promote their consumer goods. Both online genetics testing services and sites which offer interpretation of genetic data have aligned themselves with socio-political goals. Hogarth (2017) has highlighted how, since its incorporation, 23andMe's founders have aligned themselves with Silicon Valley ideals, presenting themselves "as disruptive agents of change seeking to challenge the biomedical establishment" (p. 259). An analysis of the 23andMe platform by Saukko (2018) highlights that by having users fill out questionnaires, connecting their responses with genetic and phenotypic data, and delivering them back to users in reports called "insights," 23andMe cultivates a "participatory air" for users on its platform (p. 1320). Third-party genetic interpretation sites, where users upload their raw data for further health insights, also appeal to similar social-political causes. Interestingly, some of these sites also position themselves as countering the simplistic health information that users may receive on more popular sites like 23andMe, as Nelson and Fullerton (2018) found in their interviews with site developers. Additionally, McGowan et al. (2017) have highlighted that the developers of SNPedia (which connects users' genetic data from Promethease with academic studies) describe their platform as an attempt to bring science out of the "ivory tower," and promote "participant-driven genomic research" (p. 503).

However, the few participants who mentioned social motives did not dwell on them often, making it clear that what primarily motivates them is curiosity about themselves and personal insights that online genetics may offer. This casual, emotion-free attitude is in line with what Harris et al. (2016) anticipated: upon analyzing the type of participation represented on 23andMe's platform, they concluded that its participatory culture lacks the "emotional

investment of patients and affected families” one may typically see in “disease-specific patient-centred” research networks (p. 83). Furthermore, many of the participants expressed skepticism about the extent to which online genetics, at least in its current form, can contribute to these causes, emphasizing that the science is not yet advanced enough to identify clear causes or treatments for common diseases. Therefore, while social motives are present for some of these users, they are not as relevant or important as they are for the users in Nelson’s (2016) research.

Additionally, one participant, Carly, also shared her disappointment in her experience of participating in a weight loss research study by 23andMe. This is unsurprising, as other research has critiqued the social promises made by digital health platforms. For instance, Tempini and Del Savio (2018) have discussed the emerging phenomenon of “digital orphans” of “patient-powered research networks”: on certain digital health platforms, users have contributed data for the purposes of research but have yet to see fruitful results. In addition, Spector-Bagdady (2019) have argued that while a growing number of studies have been published with data from online genetics company databanks, there is a need for greater transparency, “to ensure the proper notification of contributors and to further understand the use of public research funds for private collaborations” (p. 1). This calls into question the open science ideals that some of the interviewees associate with online genetics. Moreover, they seem unaware of the commercial potential in the transfer of personal data. This was evidenced most recently in July 2018 with the deal between 23andMe and GlaxoSmithKline, where 23andMe gave “exclusive rights to its customer database for GlaxoSmithKline to use for drug discovery” (Finkel, 2019, p. 5).

5.2 Interpretation

In relation to interpretation, my second research question asks: How do users engage with and interpret their online genetics results? My findings indicate that: The users I interviewed see

themselves as maintaining a critical, “scientific” engagement with online genetics for health. Consequently, they do not feel that they have gained truth from online genetics. This suggests that, at least for this group of users, online genetics is not as “transformative” as some critics and proponents have assumed. These findings demonstrate how users matter.

5.2.1 “Scientific” attitude

Most of my interviewees conveyed to me that they hold a critical attitude toward online genetics which they view as part of a scientific approach. Most of them highlighted the limitations of online genetics, its tools and methods, throughout their interviews. Almost all of them emphasized that the science behind online genetics is not fully developed yet, and thus the results one receives from it should be considered inconclusive and up for review. Interestingly, many of them critiqued genetic determinism and mentioned their frustration at fellow users they have interacted with online who they believe take the results of the test too literally, misunderstanding how scientific knowledge is produced and its limitations. Moreover, most of my users, except for Ghassan, identified their level of scientific expertise as above-average (although all of them qualified it and said they were not quite experts). They often criticized online genetics companies for not making the limitations of their reports clear to users, conveying their belief that while more advanced or scientifically-literate users (which they identify themselves as) may understand the limitations of the information presented to them, the average lay user may misunderstand the accuracy and relevance of online genetic reports.

Most of my participants believe that online genetics results, rather than being purely factual, are open to interpretation and the science behind them is not exact. They often emphasized to me that different companies, interpretation sites, doctors, and other experts may all interpret the same results differently. For instance, Andrew and Ben told me there is “no right

answer” on what their genetics mean, and interpretation of data can be “very different” between sites and even between doctors. Ghassan, who identified himself as someone not educated in “natural sciences,” stressed that there is no “protocol” for interpretation as far as users like him are aware. Moreover, it seems that one of the motivations for my users to use a multitude of online genetics services is because of this recognition that the data is open to interpretation: as Hernandez highlighted, he uses multiple online genetics sites to compare the information and judge accuracy.

In connection with this point, some of the users I interviewed acknowledged and highlighted the role that a doctor may play in filtering and interpreting the information from genetic data for a patient. Aside from Andrew, my participants did not necessarily always associate the interpretive role of doctors as negative, even though many also wish to disrupt doctors as a gate-keeper of medical knowledge. Some of them highlighted the importance of doctors and other healthcare professionals in assisting users to better understand their online genetics results. Three of the participants discussed their results from online genetics with healthcare professionals. Interestingly, some said seeking a doctor for interpretation would be more important for users who are less scientifically-literate than they are, and less capable of understanding the limitations of the information presented. Thus, while my participants acknowledge the value of consulting doctors to contextualize genetic results (and a few of them did so for some of their genetic results), they seem to believe this is more important for other users, who they see as less knowledgeable than themselves. In constructing themselves as scientifically-literate or advanced users of online genetics, they expressed the belief that they do not need additional outside sources to understand their data.

For my participants, key to the critical, “scientific” identity that they construct for themselves is a perception that genetic data speaks for itself. As a consequence of this belief, their engagement with online genetics involves an attempt at stripping away all sources of potential interpretation which would give meaning to their data. Yet it is unsurprising, then, that participants are ultimately unable to finding broad meaning in their practice because, as Nelson (2016) highlights, “narrative and contextual framing” is necessary to make sense of DNA: “Genetic markers in and of themselves have no meaning or value; they are like letters on the page of a book for someone who has not been taught to read. But we learn to read the significance of DNA in science labs and at genealogical gatherings” (pp. 161-162).

Based on recent decisions and comments by CEO Anne Wojcicki, it appears that online genetics company 23andMe is well-aware of their role in providing meaning-making and interpretation to users. In 2016, the company decided to abandon efforts to explore next-generation sequencing, which would have changed the company’s service to give consumers more in-depth reports with a greater amount of data. When explaining the rationale for the seeming move backwards, Wojcicki emphasized that it was not due to financial issues, low demand, or regulator objection, but rather: “One of the things people are still figuring out with next-generation sequencing is ‘Exactly what does all that information mean?’” (Lee, 2016). Additionally, she has made comments that next-generation sequencing would make the results consumers receive too “complex,” likely requiring interpretation from a doctor, which she considers contrary to 23andMe’s aim of being a direct-to-consumer product (Pressman, 2016). These comments further illustrate that data from online genetics does not speak for itself; although most of the users I interviewed seem to believe so.

Other online genetics services which offer more complex health reports and allow users to interact with raw, uninterpreted genetic data, position themselves as more scientific in their attempt to present the complexity behind genetics, giving the user tools to interpret their genetic data on their own. As Nelson and Fullerton (2018) have explained, the intent behind these sites (like Promethease) is to give users more in-depth information, free of interpretation, and allow users to connect their genetic data directly to scientific articles. Therefore, it is no surprise that most of my participants have used these third-party interpretation websites and prefer them to simpler, user-friendly services like 23andMe which, while giving users more interpretation, also have more control over the information they present to users. The majority of my interviewees are interested in accessing their raw genetic data file and attempting to interpret it on their own through third-party interpretation websites. For most of them, this process usually consists of: (1) downloading their raw data file from testing sites like 23andMe or AncestryDNA, (2) uploading it to a third-party interpretation site like Promethease, and (3) receiving an annotated health report with links to relevant scientific journal articles on databases like SNPeDia, based on key variants found in their genetic data.

Even though most of my participants do not consider themselves to be experts, they do see themselves as more expert or scientifically astute than the average user and thus insist on seeing the raw data from online genetics and doing interpretation research themselves. This interest in analyzing raw data indicates a perception that the data speaks for itself; thus, if we have data in its naked form, it represents the ultimate truth, uncontaminated by interpretation or meaning from external sources (whether that source be a doctor or 23andMe). It seems these users are intent on making their engagement with online genetics as scientific as possible, with the belief that if they can stick to the data, they will get the personal insights hidden within it.

This reverence for data may also explain why my participants had limited critiques of data privacy issues.

Although most of my participants portray themselves as critical when interpreting their genetic results, they expressed the view that genetics—when done correctly—can tell people something fundamental about themselves. For instance, Dina and other users suggested that data-driven genetic science will eventually find all the answers to human health questions. Andrew also showcased his reverence for genetic data in the following quote, in which he explains why data is more reliable than family health history:

The genetic data was kind of helpful just to be a little more concrete—it's not just things your family have said, it's like: here's the data. Here's the hard scientific data. [...] I dunno I guess it's just more real to me. I don't disregard the other people, I believe them, I know they're telling the truth.

All in all, my interviewees conveyed that while there may be issues with online genetics, the ideal of doing science and gathering data to find out more about oneself is nevertheless a legitimate one; and for most, this is a key method for self-discovery. Also note that most of them maintain their hope that one day, the science will be good enough to explain human health and ancestry, but it just is not there yet, and we need more data. This persistent faith in data to one day give them the insights they are looking for explains why almost all of my participants said they continue to engage in online genetics, whether it be through online discussion forums, following new developments in genetics, tracking whether there are new relevant research findings, or searching for additional health reports and interpretation tools online.

While it may seem like a contradiction that my participants are critical of online genetics and yet hold hope for the potential of what it can tell them, these are two sides of the same coin: data itself tells a story, we just need to decode its language in full (and the user needs to learn to read it for themselves). My participants believe that to truly know themselves, they need pure

genetic data. Therefore, they embark on an effort of collecting that data, stripping away any attempts to skew it with meaning. This means that they critique online genetics companies for their incomplete or inaccurate interpretation of user data. However, they remain hopeful that if they can just access enough of their data, learn more about genetics and improve their expertise, and continue to resist external sources of interpretation, they can eventually get it to speak for itself. This contrasts with what Ruckenstein (2017) found, as most of the online genetics users she studied expressed frustration with raw data and said they did not feel they would be able to interpret it on their own, given limitations in their medical knowledge and skill with data (pp. 1033-1034).

5.2.2 No insightful results

It is possible that in my participants' attempt to stick with the raw data and make online genetics a purely "scientific" practice, they do not find the profound insights they seek. Notably, all my participants highlighted they have not found anything insightful about themselves or their health through online genetics. Throughout their interviews, most of them pointed this out to me. Even those users who highlighted specific health questions they were curious about similarly reported they have not found answers through online genetics. Ghassan even went as far as to say that he had been "expecting more" from online genetics when it came to health insights. Many of them conveyed that they had no response to their results, neither negative or positive. They made it clear they were not emotionally affected by the results either, such as when Carly told me that there was nothing "alarming" about her results, and Felix told me there was nothing "shocking" about his. Many of them said online genetics often confirms things they already know about themselves. Therefore, the response of the users I interviewed can mostly be described as neutral and disinterested, basically a non-response.

This lackluster response to the results of online genetic testing has been found by others who show that participants expect of genetic testing far more meaning than the tests per se can deliver. Nelson (2016) has argued, users often pose metaphysical or “trans-scientific” questions to DNA testing, which go beyond the actual technical limits of the test and the science behind it (p. 164). As I discussed at the start of this chapter, it is clear that the goals my participants have for online genetics go beyond the purely scientific, they are more like metaphysical curiosities related to meaning-making. However, the users in my study are really trying to make it scientific and, in their endeavour to do so, they do not find the meaningful insights they are looking for because they reduce the practice to an insular interpretation of raw data. The following quote by Dina makes the connection between a critical attitude and lack of personal insights clear: “The primary finding was that there’s nothing in my genetics in terms of today’s science that pops out.” To sum: as a result of their critical engagement with online genetics, the users I interviewed do not feel that they have gained much from it.

A critically engaged and underwhelmed response to online genetics is consistent with findings from several of the quantitative research studies on online genetics users. For instance, Egglestone et al. (2013) found only a minority of users made health changes in response to online genetics results; the vast majority did not. Likewise, Koeller et al. (2017) found that after receiving results, only 4% percent of users sought genetic counselling, and 38% said they would have sought it if it were available to them (p. 1275). In a study by van der Wouden et al. (2016), 63% of respondents said they would share their results with a healthcare provider; yet, after six months, only 35% had done so (p. 516). Additionally, Koeller et al. (2017) found that online genetics users who were more confident in their genetic or health literacy were less likely to respond to genetic test results. Survey research by Nielsen et al. (2014) found that many online

genetics users still value traditional experts as sources of knowledge for genetics and health: 47% of users said a research lab was the best source, 41% said a healthcare professional was the best source, and only 12% said online genetics was the best source (p. 100). Thus, my users' critical engagement and lack of response is consistent with some previous quantitative studies on online genetics users and health information. In terms of research on offline/clinical genetic testing, there are also some similar findings: in a study by Gordon et al. (2011), users were given genetic risk information for eight diseases from a medical research organization, and most were not reactive or "emotional" toward their results (p. 426).

These findings are also in agreement with the few qualitative studies on early users of online genetics which found that users questioned the validity of their online genetics results and were not responsive to them (McGowan et al., 2010; Ruckenstein, 2017; Harris et al., 2016). Harris et al. found that the early users they studied tended to "knit" the health results they got from 23andMe into their pre-existing beliefs about their health (p. 46). My study builds on this previous literature by indicating possible connections between users' motives and the ways in which they engage with and respond to online genetics: the users I interviewed found meaning in being scientifically-minded and self-proclaimed advanced users, which is linked to their insistence on interpreting raw data on their own. This then leads to a lack of insightful results, because the genetic data does not speak for itself when all external sources of interpretation are removed.

5.2.3 Not transformative

The lack of response to online genetics results amongst my participants reveals that, at least for the group of users I interviewed, this technical practice is not as "transformative" as many critics and proponents have suggested. Thus, my results support the users matter

framework applied in this study, which rejects the deterministic view of technologies causing social change. As Hedgecoe and Martin (2008) have highlighted, much of the genetic testing literature in social science has taken on a transformative tone in its analysis of how genetics may impact user identity. Some of this work is represented in the geneticization literature discussed earlier (Nelkin & Lindee, 2004; Lippman, 1991, 1994). The other subset of this work has emphasized the potential progressive effects of genetic knowledge for user identity. This work has elaborated on Rabinow (1996), whose notion of “biosociality” has suggested that genetic information could transform peoples’ understanding of themselves and result in new communities built around new identities connected by shared genetics. Rose (2008) has built on this line of thinking, arguing that direct-to-consumer genetics is leading to a “transformation” of identities which incorporate genomic knowledge as one source of information about one’s self.

Hedgecoe and Martin (2008) have rightly criticized this transformative view of genetics, whether critical or positive, as it has been developed without sufficient empirical support. They argue that rather than reflecting a more nuanced, empirical picture of technological developments and their effects on users, this transformational view may have led to simply reinforcing the promises of advocates regarding genetic testing technologies, who have an incentive to over-emphasize the transformative effects of genetics on users (see also Aradau and Blanke, 2015). Likewise, Fishman and McGowan (2014) have stressed: “utopic and dystopic claims about genomic information and identity have largely been made in the absence of examining the users themselves” (p. 30). Fishman and McGowan also found that both promotional material from companies and critiques by analysts have often over-emphasized the extent to which new genetic technologies are shaping users’ self-conceptualizations. These

authors, alongside Harris et al. (2016) and Saukko (2013), have encouraged more empirical work on online genetics users themselves. My study has been an effort to answer that call.

My study has problematized the transformative view of new genetic techniques and demonstrated how users matter since my participants, despite approaching online genetics with hope for personal insights, have not been responsive to their results. Whereas online genetics companies have constructed use of their technologies as positively transformative for one's self-knowledge, the users I interviewed conveyed a different view of this technical practice: they do not attribute transformative effects to online genetics on their identities. Moreover, while online genetics companies have made strong attempts in the past two years to present and market their health information as scientifically legitimate to consumers, the users I interviewed claim to resist some of the information online genetics provides. This resistance is situated within a broader effort among my participants to construct their identities as advanced, scientific users. These findings validate the users matter approach to studying users, which has argued that users play an active role in interpreting and attributing meaning to technical practices (Oudshoorn and Pinch, 2003).

Chapter 6: Conclusion

To reiterate, in terms of users' motives for doing online genetics for health, my findings revealed that these users pursue online genetics for health because of a curiosity about themselves, as they perceive health information from online genetics as just one additional source of self-knowledge. Additionally, some of the users I studied have more specific health questions in mind when they pursue online genetics. Social motives also play a minor role for some of them who believe that doing online genetics contributes to certain social goals like challenging expertise and sharing their data for research, although they maintain a skeptical attitude about the validity of the research done by online genetics companies. In terms of users' interpretation and engagement with online genetics, my findings indicate that the users in this study construct themselves as holders of a scientific attitude, which leads them to engage with online genetic health information in a critical manner. Participants insist on interpreting raw data from online genetics on their own, using tools from online genetics services (particularly third-party interpretation sites) to try to interpret their genetic data themselves. Indeed, being informed about current health innovations like direct to consumer genetic testing appears to be part of the identify formation for participants and it is through this that they read their results where they feel they have not learned anything new about themselves (including their health) through online genetics. This last finding also fits within the user centred theoretical approach to understanding technologies in society as in my study online genetics is not as "transformative" for users as many of its critics and proponents have claimed (see Hedgecoe & Martin, 2008; Fishman & McGowan, 2014).

My research has contributed to filling multiple gaps in the literature on online genetics users. This study adds to the literature on qualitative interview studies with users of online

genetics for health. Reviewing the existing research on users in chapter 2, I showed that it has largely relied on quantitative surveys, published within the medical and health science disciplines (Frieser et al., 2018; Koeller et al., 2017; Wang et al., 2018). This research has often fixated on peoples' reactions to genetic results, with mixed findings, yielding some interesting trends but without deeper insight into the perspectives of users, why they do online genetics and how they read and interpret their results. To address this gap, I sought insight into the complexities of users' experiences and perspectives through interviews. In chapter 2, I also noted some existing qualitative studies related to online genetics users. However, much of the existing qualitative empirical work on users has drawn its findings and conclusions about users on the basis of analyzing company products, platforms, marketing, and stakeholder statements; rather than collecting data directly from users (Nordgren & Juengst, 2009; Nelson & Fullerton, 2018; Hogarth, 2017). Some of the qualitative research has also studied user perspectives indirectly, such as through analysis of users' posts on YouTube (Harris et al., 2016). A few qualitative studies have actually done interviews with users, but these were all done with early users of online genetics for health (Ruckenstein, 2017; McGowan et al., 2010). As noted in chapter 2, online genetics has changed considerably since 2017, with the industry trying to legitimize their services as more medical and scientific than recreational. This warranted an empirical investigation into current users' perspectives, through interviews, to determine how users matter: how they perceive online genetics, why they do it, and how they interpret the health information it gives them.

I posed two research questions to guide such an investigation: (1) Why do people do online genetics for health? (2) How do users engage with and interpret health information from online genetics? To address these questions, I applied the users matter approach developed by

Oudshoorn and Pinch (2003), further contributing to the literature by leveraging a newer approach to studying users. Within this broader framework, my theoretical approach was also open to the idea of the symbolic value culturally attributed to genetic information and technologies (Nelson, 2016; O’Riordan, 2011; Nelkin & Lindee, 2004). In line with my theoretical approach, I used interviews and abductive data analysis to collect and analyze my data.

My study has validated the importance of a users matter approach, which goes beyond the transformational lenses of grand theories which have been applied to users of online genetics so far (see Hedgecoe & Martin, 2008). Previously, the users matter approach had only been tested in the early development of online genetics (Harris et al., 2016; McGowan et al., 2010), when companies were still marketing their health-related information as more recreational and informative than scientifically valid. The users matter framework applied in this study has demonstrated that users play a role in interpreting technical practices, and that user identity and wider social context plays a role in the impacts that innovations have in people’s lives (Oudshoorn & Pinch, 2003; Lie & Sorenson, 1996; Cowan, 1987; Silverstone et al., 1992). In my research I set out to study users and techniques in their context of use. Applying this approach has revealed that despite recent efforts by online genetics companies to construct themselves as medically and scientifically valid sources of health information; the users in this study have interpreted online genetics in different ways, largely pursuing it for purposes of recreation and identity formation and often questioning its medical and scientific legitimacy because they centrally identify as scientifically astute and skeptical in their engagements with emergent technologies. To underscore, like Nelson (2016), my study has demonstrated that the ways in which people do online genetics is also part of their identity construction; my findings suggest

that users construct their identity from online genetics not just through the genetic information it provides (which many are not responsive to), but also through their forms of engagement with it in a process of identity formation.

6.1 Limitations and Future Research

While this study has contributed to the gap in empirical qualitative research on perspectives of users of online genetics for health, it is important to note its limitations and link them to points for further research. While applying the users matter approach has led to important insights into how current users are doing online genetics in practice in this study, I could have established a fuller account of users if I had been able to apply a more comprehensive analysis of the user-technology relationship by collecting data from both users and the online genetics platforms that they use. Such a comprehensive project was not possible to pursue for this thesis, given limitations in time and resources. However, I plan to conduct future research which complements user perspectives with the perspectives of designers of online genetics and the technologies themselves, allowing for a fuller analysis of the cultural context and wider sociotechnical network in which users are embedded. In the future, I wish to address the question of co-construction of user and technology through an analysis of online genetics websites focusing on the role of these sites in the user-technology relationship, asking: Are users constructed as consumers, patients, lay experts, or other via these websites? This textual data from websites could complement interview data from users, facilitating an analysis of how online genetics and users co-construct each other.

The sample used in my study also entails some limitations. First, it would have been beneficial to conduct a greater number of interviews. Second, most of my participants were recruited through public social media forums on Reddit and Facebook. There are some

limitations to this recruitment method. It is possible that participants on these platforms represent a niche group of users, as they may be more digitally engaged than other users by being active on online discussion forums. Many of the users I met online were very eager to participate in research, and it could be that they were more eager to participate in this study than other types of users would be. These interviewees could represent the voices of highly engaged users of online genetics, who spend a lot of time on online spaces and actively seek opportunities to have their voices heard. In another light, such limitations can be seen as the parameters of my study: this study may be more representative of those users who identify themselves as more scientifically- and digitally-literate than other users. While this is possible, further research is needed to validate these users' claims that they are unique in comparison to other users of online genetics, who they claim are less scientifically literate. If I were to conduct a future study, with more time and resources for recruitment, I would endeavour to recruit people who are less active on online discussion groups, who may have different types of technical literacy and identities than the users in this study. Efforts to diversify recruitment would be in line with the users matter approach, which has noted that users are a diverse group (Star, 1991; Casper & Clarke, 1998; Saetnan et al., 2000). The users matter approach calls for more empirical research on users doing online genetics in practice. As evidenced in my study, users clearly matter for how digital innovations like direct to consumer genetic health testing are understood and engaged with by citizens and the impact these technologies have on people's self-conceptions and practices. A user-centred study also underscores how thinking about the unique social situation of the user matters in how the technologies come to matter. Future research should consider how different, particular social groups of users perceive online genetic health information, why and how they do online genetics, and with what consequences.

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Appendix A: Recruitment Post

Post title: Participants Needed for Research on Users of Direct-To-Consumer Genetic Tests for Health

Hi,

As part of my Master's in Sociology at the University of Ottawa, I am seeking volunteers for my research. I am seeking those who have taken an online genetic test for health, and are willing to participate in a semi-structured, one-hour interview about their testing experience. This is a chance to share your perspective and contribute to research on this new practice!

Online interviews will be conducted through a video call service. The date and time of the interview is up to you. Please note that participants will be selected on a first come/first served basis. Interviews will be conducted in English. You must be at least 18 years old to participate.

For more information, or to volunteer, please send me an email.

Thank you!

Appendix B: Participant Demographics

Pseudonym	Gender Identification	Age Group	Ethnic Identification	Country
Andrew	Male	30 - 40 years old	European	United States
Ben	Male	30 - 40 years old	Chinese	China
Carly	Female	30 - 40 years old	Latino	United States
Dina	Female	60 - 70 years old	European	United States
Evka	Female	30 - 40 years old	Czech	Czech Republic
Felix	Male	20 - 30 years old	European	United States
Ghassan	Male	20 - 30 years old	Lebanese-French	Canada
Hernandez	Male	30 - 40 years old	Spanish	Spain
Israa	Female	20 - 30 years old	Arab	United States
Jennifer	Female	30 - 40 years old	European	Canada

Appendix C: Interview Guide

Motivations

1. Can you tell me about the online genetic testing company you took the test with?
2. Which websites have you visited to learn about your genetics and health?
 - a. What do you do on these sites?
 - b. How do you feel about the different types of sites you've used?
3. Why did you take the test? Why did it interest you initially?
4. Why do you do online genetics for health?
5. How would you describe your knowledge of genetics and health?

Data and Sharing

6. What happens with your data when you take an online genetic test?
7. Did you upload your results anywhere, or share them publicly?
8. Have you talked about your results with others?
9. Some online genetics companies conduct research with customer data. Is that something you're interested in?
 - a. Why?
 - b. Why not?

Response to Results

10. How did you respond to your results?
 - a. Were there things you were more or less accepting of?
11. Did you learn something you did not already know?
12. Do you feel that the information from online genetics has answered things you had questions about?
13. Did you understand your online genetic test results?
14. Did you change any of your health practices (eating habits, lifestyle, medication, etc.)

because of online genetics?

Views of Clinical Health

15. Have you or would you have taken a genetic test with your doctor or some other healthcare provider?
 - a. Why or why not?
16. How do you feel about doctors?

Views of Genetics and Health

17. What do you think your genetics can tell you about your health?
18. Is there anything genetics cannot tell you about your health?

Now

19. Where are you now?
 - a. Are you still actively doing online genetics for health?
20. Going forward, are you planning to keep up to date with new developments in online genetics and health?
21. Would you say you found the answers you were looking for, or is it still in progress?

Closing

- Is there anything else you would like to share with me?
- Thank you so much for taking the time to do this interview.