Biological Citizen Publics: Personal Genetics as a Site of Scientific Literacy and Action

Stacey Kuznetsov[1], Aniket Kittur[1], Eric Paulos[2]

Human-Computer Interaction Institute[1]
Carnegie Mellon University
Pittsburgh, PA, USA
{stace, nkittur}@cs.cmu.edu

Electrical Engineering and Computer Sciences[2]
University of California
Berkeley, CA, USA
paulos@berkeley.edu

ABSTRACT

Low-cost genetic sequencing, coupled with novel social media platforms and visualization techniques, present a new frontier for scientific participation, whereby people can learn, share, and act on data embedded within their own bodies. Our study of 23andMe, a popular genetic testing service, reveals how users make sense of and contextualize their genetic results, critique and evaluate the underlying research, and reflect on the broader implications of genetic testing. We frame user groups as citizen science *publics*—groups that coalesce around scientific issues and work towards resolving shared concerns. Our findings show that personal genetics serves as a site for public engagement with science, whereby communities of biological citizens creatively interpret, debate, and act on professional research. We conclude with design trajectories at the intersection of genetics and creativity support tools: platforms for aggregating hybrid knowledge; tools for creative reflection on professional science; and strategies for supporting collaborations across communities.

Author Keywords

Genetics, scientific literacy, biological citizenship, publics

ACM Classification Keywords

H.5.m. Information interfaces and presentation (e.g., HCI): Miscellaneous.

INTRODUCTION

Since the completion of the Human Genome Project [19], an international research effort that mapped the human DNA in its entirety in 2003, genetic research and its underlying technologies have advanced in radically new and unexpected ways. The cost of genetic sequencing has decreased exponentially over the past decade. Affordable genetic testing services and intuitive visualizations of the results are increasingly turning personal DNA into an object an inquiry. This creates a new frontier for scientific participation, in which people can make sense of, share, and act on information embedded within their own bodies. Little is known about this space within creativity literature: how do people interact with and make sense of the underlying scientific information, and how does the understanding of personal genetics influence their sense of self and their daily lives?

We present a study of 23andMe [1], a low-cost ($99), online service and community for personal genetic testing. While the DNA testing itself is done in professional laboratories, 23andMe serves as a site for sense making "from below": forum and community features enable participants to share experiences, narratives, and intuitions about their results. Similar to other citizen science efforts [e.g. 5, 13, 28], 23andMe relies on lower-cost sensing (genetic sequencing) and increased computational power (for processing genetic information), as well as new social media tools to support the emerging communities of participants. However, public participation in personal genetics also presents a shift from people 'as sensors'—*i.e.*, gathering information about external environments—to communities who collect, make sense of, and act on information embedded within their own bodies. Thus, participation in genetics is often motivated by and brings about a host of new concerns, from discovering personal and intimate information about oneself to understanding patterns in human migration and evolution. These issues reflect opportunities and challenges arising from the convergence of biology and computation [27].

Research contributions

We frame 23andMe participant communities as citizen science *publics*—groups that coalesce around scientific issues and work towards resolving shared concerns [7].

Figure 1. 23andMe spit kit.

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Drawing on concepts of biological citizenship [24] and biosociality [21], we first reflect on how services such as 23andMe serve as sites for creative sense-making around biological traits and concerns. We then detail our analysis of 23andMe forum threads and a qualitative study of six individuals who joined and used the service over the course of 3 months. Our findings reveal why participants joined 23andMe; how they contextualized the data within their lives and environments; how they critiqued and evaluated the underlying research; and their reflections on the broader implications of genetic testing. We conclude with a discussion of *biocitizen publics* and suggest three opportunity areas for Creativity and Cognition: 1) tools that creatively visualize genetic information along with self-reports and human experiences; 2) technologies that support creative reflection on scientific research; and 3) feedback systems whereby members of the general public can contribute to and influence professional research.

BIOLOGICAL CITIZENSHIP AND SCIENCE PUBLICS

The concept of biological citizenship was first discussed in the context of people's claims to welfare for biological damages (i.e., Chernobyl Nuclear Power Plant workers who demanded compensation for the 1986 disaster) [21]. Of course, biology has, in some ways, always been intertwined with ideas of citizenship: from the pragmatic association with where one is born, to the more contentious questions of national identity being shaped by race and family lines. Globalization has arguably blurred geographic boundaries by increasing connectivity between cultures, economies, and community practices [24]. Advancements in genetics contribute to this trend by offering new information regarding personal, family, and cultural backgrounds. These developments bring the concept of citizenship as a purely national concept into question [24] and give rise to new types of agency that can be exercised by biological citizens.

Genetic information can serve as both an individuating and a collectivizing force [22]. On one hand, it reveals unique features of individuals (*e.g.*, distinct ancestral backgrounds or unusual biological traits). At the same time, people are collectively making sense of and sometimes act on shared genes or genetic conditions [22]. This *biosociality*—the forming of communities around biological characteristics—leads to new types of activism [20, 24]. For instance, groups formed around genetic conditions (e.g., Huntington's disease[1]) influence professional science, both by contributing their own tissue samples and medical data to research, as well as by shaping the research itself through advocacy, funding, and public awareness campaigns.

Citizen science publics and interaction design

Building on existing trends in quantified-self [e.g., 14], interaction design will soon be addressing personal sensing beyond step counts, blood pressure, *etc.* and considering personal DNA. Our study is among the first to examine personal and collective interpretations of this data, and can be seen as parallel to prior research in eHealth [e.g., 13, 17]. Unlike 23andMe participants, however, eHealth users are relatively sure of their diagnosis, and their discussions focus on treatments, preventative action, and social support [13]. Genetic data, on the other hand, has a higher degree of uncertainty—genes are viewed as potentials for traits rather than definite indicators. Moreover, while eHealth sites tend to focus on diseases [ibid], 23andMe also offers data about physical and mental abilities and ancestry. Users are therefore interested in understanding themselves and fulfilling their potential through their genes.

The resulting communities can be viewed as citizen science publics: participants collectively interpret, critique, and make impact on professional research. Other citizen science efforts [5, 13, 28, and others] rely on low-cost sensing to support publics around ecological concerns. In the context of personal genetics, concerns are deeply intimate and idiosyncratic, often made more unique by people's specific environments, lifestyles, and physical bodies. Genetics publics thus draw on heterogeneous methods and materials, including sensing technologies, social media tools, and the human bodies themselves to construct, communicate, and pluralize scientific knowledge [24].

Prior studies show a limited public understanding of science, often due to mixed messages from television shows and science fiction movies, as well as pre-existing mental models of genetic tests, diseases, and kinship [3, 18, and others]. While scientific literacy has been a major focus within HCI [*e.g.*, 23, 25, 26, 28], research has not explored scientific literacy in the context of genetic publics. Tools that enable collective sense-making around genetic concerns are reminiscent of politically-oriented approaches to link people through their actions [11, 15]. For instance, DiSalvo proposed *tracing* to expose hybrid "networks of materials, actions, concepts and values that shape and frame an issue over time" as a design strategy for supporting publics [8]. We contribute to this research by focusing on knowledge production and agency within a personal genetics community.

ABOUT 23ANDME

Founded by Linda Avey and Anne Wojcicki in 2006, 23andMe is a biotech startup aimed at providing low-cost genetic testing. The service offers "a comprehensive genetic scan of a subset of the SNPs (single nucleotide polymorphisms, or DNA variations) in your genome which correspond to the SNP data being studied by the research community" [1]. 23andMe works as follows: user can order a 'spit kit' online, which arrives a week later; 2) the kit is used to collect and preserve the participant's saliva sample, and is mailed back to 23andMe; 3) after a 4-6 week processing period, the results are viewed and shared online.

[1] E.g., Huntington's Disease Society of America; hdsa.org.

At the time of research, the service offered genetic health results—from one's ability to taste bitter flavors, to hereditary illnesses such as Parkinson's disease—as well as genetic ancestry. Shortly after our study, the FDA ordered 23andMe to stop offering health results to new users, and this order is currently under negotiation with the service. The health results profile over 240 conditions, ranging from multiple sclerosis, to Alzheimer's disease, cystic fibrosis, or sarcoma, as well as traits such as alcohol flush reaction, hair curl type, lactose intolerance, smoking behavior, photic sneeze reflex, and drug response—sensitivity to coumadin, phenytoin, warfarin and others. Ancestry results include maternal and paternal line haplogroups (genetic populations that share a common ancestor), overall composition broken down by geographic region, and percentage of Neanderthal DNA. The site also provides social networking tools: relative finder, which connects users based on shared DNA; forums, whereby users can discuss topics such as health, ancestry, specific haplogroups, Alzheimers disease, or general questions about the 23andMe service.

A platform for citizen-driven genetic research

23andMe links its results with corresponding academic publications, enabling users to learn how the findings were produced and 23andMe's confidence in its data. Some 23andMe results are improved through surveys and questionnaires on the site. These cover ancestral and health history, and personal traits such as computing one's empathy quotient, determining if one's personality is planned or spontaneous, or smoking behavior. The site also provides surveys that lead to discoveries—helping scientists identify genetic variants that are associated with traits such as dimpled chin, freckling, or earlobe type. In addition, 23andMe invites community members to propose their own research projects. Members can submit research proposals, which are evaluated by 23andMe committees of professional scientists [1]. Upon approval, members can design studies, recruit respondents, and analyze the data through 23andMe. Ongoing projects aim to identify SNP's that might be associated with specific traits, including Parkinston's disease, sarcoma, and Alzheimer's.

RESEARCH METHODS

Our research includes several strands of investigation. We began by reviewing and coding public 23andMe forum posts. Our research covers both the initial posts and the corresponding responses within 150 threads from Labs, Measures of Intelligence, Health, Relative Finder, and Hereditary forum topics. We identified 238 themes, which were affinity diagrammed into topical categories. These high-level groupings served as focal points for our in-depth qualitative study of 23andMe users. The study followed six individuals as they joined 23andMe and interacted with the service over the course of 3 months. Participants completed initial semi-structured interviews about their motivations for joining 23andMe, prior knowledge of genetic testing, as well as their personal health, family history, personality and intelligence. After the initial interview, participants signed up for the 23andMe service with private accounts (that are not accessible to the researchers) and completed the spit kits on their own. Participants attended follow-up interviews when their data became available online, probing their reactions to and understanding of the results, whether or not their expectations were met, and how the information might impact their lives in the future.

Data from the first two interviews, along with the themes derived from the forums, was synthesized into two co-design activities for our third set of interviews. These final interviews were conducted about a month after participants' data was first made available on 23andMe, probing how the service affected their lives over the past month, and asking them to complete the co-design activities. In a concept generation phase of this interview, participants were prompted to envision future genetic services, what types of organisms might be tested, and for what factors/traits. Photographs (cards) presented a range of settings (e.g., home, park, restaurant), organisms (plants, pets, other humans), and types of sensing (scanning, swabbing, drawing fluid). After this brainstorming, participants were asked to talk through several scenarios where they might use one of the sensing methods to test a particular organism in a specific context. Participants were asked to speculate on issues such as comfort level, data sharing, privacy, and implications of these types of testing. Participants were compensated $10 per hour for their time during the interviews, and reimbursed for the 23andMe service. Data from the interviews was transcribed and coded to themes.

Ethics and privacy

Due to the sensitive nature of genetic testing, our research engaged with a range of ethics and privacy issues. On one hand, we introduced users to 23andMe given the risk of the service revealing information that could drastically impact individuals' understanding of themselves and their families. The effects of personal and community interactions with genetic data is highly debated: direct-to-consumer genetic tests have been shown to motivate healthy behavior as well as cause health anxiety [e.g. 12], and this is also reflected by the site's negotiations with the FDA. Moreover, there is also a possibility of the service itself having a breach in privacy and/or using participants' information towards undesirable research. We mitigated these issues by being transparent: our recruitment and consent materials stated that there were potential risks of privacy loss as well as the genetic results being surprising and/or upsetting. We emphasize that all work in personal genetics must consider and engage with such possible unwanted consequences.

About the participants

Participants were recruited with flyers posted at local bulletin boards, coffee shops, gyms, and restaurants, and pre-screened to ensure a range of ages, backgrounds, and family situations, as well as a gender balance. Our study

included 6 participants (ages 24-64, 3 male): five completed all interviews, and one completed only the first two due to a delay caused by a 23andMe DNA processing error. Participants' occupations included a massage therapist, an engineer, a federal contractor, a musician, a project assistant, and a retired music teacher. None of the participants had a genetics or related background, and only P1 had used genetic testing before the study to find out her ancestry. We continue by detailing our findings across four themes: i) motivations for joining 23andMe; ii) contextualizing 23andMe data; iii) validating 23andMe results; and iv) the broader implications of genetic testing.

MOTIVATIONS FOR JOINING 23ANDME

Participants and forum contributors cited health, ancestry, identity, and community as key motivations for joining 23andMe. What often set these apart from motivations of other citizen science communities is the highly personal and intimate nature of the information being sought after.

Health

All participants described themselves as health conscious, and linked health with a combination of environmental, lifestyle, and genetic factors. Three of the participants were interested in 23andMe primarily for health reasons. P1, for instance, wanted to learn if breast cancer, which ran in her family, was caused by genetics:

> *I'd love to see the health side of my background. Both my grandmothers had breast cancer. My maternal grandmother died from it my paternal grandmother had them removed and she survived. But none of my aunts have had it. So I wonder if it was genetic or if it was something environmental.* (P1)

Similarly, P2 wanted to learn about drug responses, and whether they were linked with his ethnicity. P1, P2 and P6 were interested in '*actionable information'* to reduce the risks of developing genetic diseases. It's important to note, however, that two of the participants, P4 and P5, were more skeptical of the role their genes play in their health.

> *I just think that we have so much more control over our health than geneticists and most people lead you to believe… I just wouldn't be too concerned about anything that indicated like oh you have an elevated risk for this cancer or that or this because I just feel like I know that the way I live my life has way more to do with it than just some genes.* (P4)

Above, P4 believes that her lifestyle influences her health over genetics. P4 and P5 both stated that they would not be concerned about their health risks on 23andMe.

Ancestry

All six participants were interested in their ancestry, and had their family histories passed down to them by word of mouth, birth and marriage certificates, or comprehensive written family trees and genealogies. To varying degrees, all participants described mysteries or disagreements about their pasts, and were hoping to lear more through 23andMe:

> *I'd like to know whether what I've been told by relatives you know how accurate it is 'cause I know they traced the family tree of my mother's mother's mother's family but the rest of it you kinda go by family tales.* (P5)

The unknowns included inconsistencies in documents such as birth certificates, as well as questions about specific family members' backgrounds. Participants also wanted to rectify disagreements about the ethnic and geographic composition of their ancestors, such as, for instance "*rumors about Chinese ancestors*" (P2) or whether or not her paternal side, which has been believed to be pure English, has any "*Irish blood*" (P5). Moreover, participants were interested in early migrations ("*information about where my ancestors migrated from*", P2; "*what different migrations of people out of Africa you're most closely related to and that really interests me*", P4). These motivations were also reflected by the Ancestry and Paternal/Maternal Line forum postings (e.g., "*I'm adopted (the reason why I joined 23andMe) so I really don't know much about my family or relatives*."[2]).

Personal identity

While health and ancestry were cited as the primary motivations, participants also tended to link genetic information with ideas about personal identity.

> *I like exploring existence and just the mind and body and just curious. Just understanding more and more about myself* (P6)

> *I'm just interested in finding out about my genetic code and what part of that plays into who I actually am.* (P3)

In the excerpts above, participants express a desire to learn more about themselves through the use of 23andMe. These comments highlight the ways participants view their genes as playing a key role in who they are. To different extents, this idea was reflected by all participants, who discussed 23andMe as a resource to learn more about oneself.

Community and connectedness

Finally, several participants also highlighted the value of 23andMe as a community tool. For example, P3 was interested in the "*less clinical"* aspects of genetic testing:

> *You can like see different people in the community and see who you're related to and it seems less clinical I guess than if I were to just test for diseases and be like oh I'm a carrier for this.*

Similarly, P4 suggested that the service might be "*fostering a sense of community and interconnectedness within human beings*". Forum posts, especially in the Community category, reflected this idea as well:

> *Who would ever join this thread if they didn't want to find out something about people who are genetically similar to them, especially when they have rare, or rare-ish combinations? That's why I joined 23andMe...*[3]

[2] https://www.23andMe.com/you/community/thread/15124/

[3] https://www.23andMe.com/you/community/thread/12766/

Interestingly, individual genes were often used as "pivots" on which to find other users that had similar traits or conditions (e.g., "*Is There Anyone Else with 2Copies of the Gene for Intelligence?*"). However, these connections were not motivated by forming social bonds such as making friends, but rather for informational reasons (*e.g.*, to learn about the experiences, backgrounds, and health problems of those with similar traits, disease risks, or ancestry).

To summarize, participants and forum contributors cited a host of personal and intimate reasons for joining 23andMe including mitigating personal health risks, rectifying discrepancies in family histories, reflecting on personal identity, and connecting with other 23andMe users.

CONTEXTUALIZING AND LINKING GENETIC DATA

Participants expressed a range of initial reactions to their data, from feeling like she won a '*genetic lottery'* (P1) and describing the information as '*futuristic*' and '*cool'* (P3, P6), to being somewhat disappointed with a lack of specificity in the ancestry data (P2, P4). Over time, participants tended to link their genetic data with various aspects of their lives, as well as environmental factors, and cultural and historic knowledge. These links often served to determine causality—to explain why or how participants came to be who they are, as well as to make sense of their surrounding world. The links also served to alter lifestyle and behavior, as well as predict implications for future relationships, and generations. We detail several of these connections and their implications below.

Past experiences

The study participants, as well as the forum contributors, compared 23andMe results with personal experiences, inferring the genetic data to be correct when these matched. For example, P1's odds of post-operative illness were consistent with her prior experience with anesthesia and her non-verbal intelligence results on 23andMe matched earlier SAT test scores. Similarly, P2 linked his odds of developing keloids with past injuries, while P3 associated his genetic 'inability to taste bitter flavors' with his preference for bitter foods such as coffee or beer. Posts across the Health forums expressed similar connections (*e.g.*, "*My 23andMe health risks does state I have a high risk for asthma… I am sensitive to certain things like wood smoke, some flower fragrances and some perfumes*."[4]).

Background and family history

Similar to drawing on their personal experiences, 23andMe users also linked their genetic data with what they knew of their family histories and backgrounds, and in many cases, used these comparisons to validate the 23andMe results. It was not uncommon to observe participants cross-referencing their high-risk traits with specific family members who experienced those conditions (e.g., "*I know people in my family who've had a lot of these so it seems like to match up*", P3). For instance, P4 noted that intolerance of cumadin and eye degenration, which 23andMe showed her at risk for, run in her family; while P1 associated her Eastern European background, as shown on 23andMe me, her dad's side "*because there's Lithuanian and some other things over there*". These findings were consistent with our forum analysis, which showed other examples of traits being linked with family histories (e.g., "*I am a carrier (for hemochromatosis) and my Aunt died from the disease*."[5]; "*I've found synasthesia to be genetically linked on the maternal side of my family*.[6]).

Resolving unknowns about the past

In addition to associating 23andMe data with known family facts, participants also tried to use the service to resolve inconsistencies and unknowns. For instance, P1 speculated that her surprising Ashkenazi heritage, as shown by 23andMe, might explain a mysterious name change in the family. Likewise, forum posts included links between ancestry results and specific family members.

> *It may clear up the question of her race. I have found Jacob Cassell, which may confirm the Cherokee rumor in my family.*[7]

Other attempts to explain background questions were less successful, especially in cases when 23andMe results did not provide enough detail. P2, for instance, could not infer whether his background included Chinese ancestry, because his heritage was shown broadly as "South East Asian". Similarly, P5 could not determine if her paternal side contained Irish heritage based on the "European" category. Moreover, all female participants were disappointed with the fact that the service could not profile their paternal side.

Lifestyle and behavior changes

Five participants also linked 23andMe results with changes in day-to-day behaviors. For example, P2 who was shown to have a high chance of blood clots by 23andMe, planned to get an exercise ball and walk more, P3 noted that his increased risk of developing a heart condition, as suggested by 23andMe, "*reminds me that I should be healthy… eat healthy and it can be avoided*". P6 also reconsidered his diet and exercise based on his inherited traits:

> *Like the fact that I'm likely* [lactose] *intolerant—that made me interested in realizing maybe I should stay stay away from milk cause I've noticed if I drink a lot of milk I get a little stuffy. The muscle type, that I'm likely not a sprinter that made me think about how I should exercise.*

Forum threads also showed a host of similar examples, whereby results influenced participants' behaviors:

> *Since the 23 & Me results I am reducing my fat intake.*[8]

[4] https://www.23andMe.com/you/community/thread/8777/

[5] https://www.23andMe.com/you/community/thread/563/

[6] https://www.23andMe.com/you/community/thread/14056/

[7] https://www.23andMe.com/you/community/thread/18896/

My take-away from this is: stop eating meat. It has a high correlation with stomach cancer and if you are potentially at a higher risk it is in your best interest on so many levels to minimize risk.[9]

However, although the majority of participants and many forum posts linked genetic risks with lifestyle changes, P4 was less concerned about the role her genes play in her health. Throughout the interviews, P4 emphasized that environmental factors influence her disease risks more than her genes do, and she was therefore not planning to make any changes based on the 23andMe results.

Cultural and historical context

Finally, participants also contextualized their genetic information within their broader understandings of history, culture, religion, and evolution. For example, historical knowledge was used to speculate on and explain unexpected 23andMe results:

So it says 0.7% South Asian, which I can see that because you know just historically there's a lot of trade between south Asia and the Philippines there's a kingdom down there. (P2)

In the above excerpt, P2 notes that his South Asian heritage, as shown on 23andMe, could be explained by ancient trade routes. Similarly, P4 associates her surprising Balkan lineage with a broader view of fluidity across cultures:

It did show that I had some Balkan ancestry… it kinda goes to show how you know we think of there being some kind of stability with like ethnic groups of people but of course all kinds of people have been migrating all over for a really really long time… there's just a lot more fluidity.

Interestingly, some of the results were also associated with cultural stereotypes (*e.g.*, "*I don't have the alcohol flush reaction, which is usually I thought was mostly Asian people who have that*", P3; "*I'm an Asian that's bad at math*", P2, based on measures of intelligence results).

Evolution

Similar to placing genetic results in a historical or cultural context, participants and forum contributors also linked genetic information with their ideas about evolution. P5 speculated about how evolution might have played a role in creating the gene that prevents people from tasting cilantro, while forum posts hypothesized about evolutionary causes of certain genetic traits or mixing with Neanderthal DNA:

Is it something that millennia ago that people were in a certain area and it was lifesaving to them to—you do not touch the cilantro. (P5)

I have 3.1% Neanderthal genes, which puts me in the top 98th percentile of all humans. Since evolutionary biologists and geneticists believe the Neanderthal and modern human mixing occurred in southern Europe, that could explain it.[10]

These excerpts exemplify how 23andMe users linked genetic test results with potential evolutionary causes.

To summarize, this section highlighted how 23andMe results were contextualized within and linked to users' environments, lifestyles, family backgrounds, and broader cultural and historical knowledge.

MAKING SENSE OF PERCEIVED INACCURACIES

While contextualizing 23andMe data within aspects of their lives, participants and forum contributors found instances where they did not agree with the results—from traits such as eye color, photic sneeze reflex, or smoking behavior, to their ancestry such as haplogroup information that did not reflect their country of origin. Although most participants (5 out of 6) appreciated being able to see the studies 23andMe drew upon to present the data, they also tended to cross-check information with other genetic testing services, as well as sources such as Wikipedia, Mayoclinic, Webmd, and friends who they considered to be experts. Oftentimes, these inquiries led to users to question, debate, or refute scientific information. Many factors—from environmental influences, to study limitations and biases—were drawn upon to determine whether the genetic data was reliable. Below, we detail how participants made sense of and interpreted discrepancies between their perceptions of themselves and their external world, and the genetic data that reflected the invisible information within their bodies.

Nature vs. nurture

All of our participants, as well many of the forum posts we analyzed, discussed genetic testing as an indicator that has a degree of uncertainty. It was not uncommon to hear our participants refer to 23andMe traits and conditions as a "propensities", or "not definites". To varying degrees, all participants acknowledged 23andMe results as predispositions rather than guarantees (e.g., "*whether they're activated has to do with a lot of factors*" P4). Participants and forum posters emphasized the role that environment and lifestyle plays in gene expression:

This risk is not taking into account me, but only my genes. (P3)

*It's always going to be a complex interplay of nature and nurture; genetic factors or predispositions probably (at least IMO) going hand in hand with environmental / cultural factors, *individual* predispositions, etc.*[11]

These excepts show that, while in many cases, participants did not doubt the accuracy of the genetic tests per say, they attributed inaccuracies in their results to the influences of environmental and lifestyle factors.

[8] https://www.23andMe.com/you/community/thread/10116/

[9] https://www.23andMe.com/you/community/thread/9664/

[10] https://www.23andMe.com/you/community/thread/11378/

[11] https://www.23andMe.com/you/community/thread/15866/

Small datasets and preliminary research

In other cases, 23andMe users critiqued the results for being based on small (inconclusive) datasets. It was not uncommon to hear participants refer to 23andMe results as based on "*preliminary research"* (P3), or findings that are constantly changing based on new or incoming data (P1).

> *There's more studies more research going on so I guess within the framework of the limited knowledge that we have now and our understanding of things now as a snapshot I guess I trust this as much as you can* [trust] *what we know now.* (P2)

Above, P2 notes that 23andMe results are dependent on '*what we know now'*, and may change as new data comes in. P6 and P3 also pointed out that 23andMe tests for a small subset of genetic mutations. For instance, P3 commented that the service "*only tests for 3 of 100s of possible mutations you might have in the BRCA* [breast cancer] *gene*"; while P6 also critiqued the 23andMe service for not taking into account how different genes might interact with each other.

Limitations and biases of supporting studies

In some cases, participants and forum contributors also identified limitations and biases in the underlying research. It is important to note that even prior to joining 23andMe, all participants expressed a skepticism towards scientific publications—from questioning data that is "*constantly changing*" (P2), to suggesting that findings may be influenced by corporations, researchers' "*pre-conceived ideas*" (P4), or financial and political motivations (P5). Given participants' initial skepticism towards scientific research, it is not surprising that they also identified limitations in studies cited by 23andMe. Most commonly, they noted that that the related studies did not apply to their gender, ethnicity, or age group (e.g., "*this health risk assumes I'm European and of a different age"*, P3; "*maybe if I was that group it would be accurate*", P2). Furthermore, participants also pointed out that many of the sample sizes were too small (e.g., a study of 139 people), or had un-accounted variables (e.g. "*who are you studying will skew results*", P4).

In addition, several forum discussions expressed concerns over potential biases in the underlying research.

> *In many fields it is rare for a person to strive for the truth ahead of getting published, getting tenure, or other renown.*[12]

> *I think the test has a major flaw in that all the people are white… So would it not make sense that white people would do better on this test than Asians, Mexicans or African-Americans?*[13]

These excerpts illustrate potential research biases that were of concern to 23andMe users: ulterior political or financial motives of the underlying studies, and racial bias.

Inaccurate 23andMe survey responses

Finally, participants and forum contributors questioned the accuracy of some of the 23andMe results that were based on the site's surveys. For instance, P5 noted that she guessed her survey answers when she could not remember her family history, and was worried that others might be doing the same, thereby skewing the data. Moreover, P3 pointed out that there was no mechanism for changing one's survey responses if they were accidentally entered incorrectly. Several forum posts expressed similar concerns (e.g., "*I really have to question the effectiveness of some of their* [23andMe] *research questionnaires*"[14]).

To summarize, this section outlined several ways by which participants and forum contributors made sense of instances when their 23andMe results did not match with what they believed to be true about themselves. Among the discussed factors were the influence of environment and lifestyle over genetics, as well as lack of data, limitations of supporting studies, and inaccuracies in 23andMe survey responses.

BROADER IMPLICATIONS OF GENETIC TESTING

Finally, our forum analysis and discussions with participants revealed ways that 23andMe users reflected on the broader implications of genetics. Below, we detail users' speculations about potential positive and negative consequences, and new ways of seeing that might emerge as genetic testing becomes more widespread.

Potential positive consequences

All participants emphasized that genetic testing poses unprecedented opportunities for healthcare.

> *I think it just would be empowerment for people to be able to watch out for their own health. I think it would be on a societal basis … I would think people would take a little better care of themselves or at least would know what to watch out for.* (P1)

Above, P1 highlights how access to genetic testing might empower people to mitigate disease risks and/or take better care of personal health. To varying degrees, all participants also highlighted opportunities for improved preventative care and diagnostics, and drugs being designed to suit individuals based on their genes. Participants also pointed out that services such as 23andMe could '*advance scientific knowledge*' for researchers and the general public (P1, P2), or serve as an *'educational tool'* to show how '*humanity is evolving'* (P4).

More broadly, several participants also commented on the implications of large communities forming around shared ideas rooted in genetics.

> *It brings people together with all this medical information already tied to them… so it's a good method of inquiry for a group because this group already exists and they have this huge pool of data.* (P3)

[12] https://www.23andMe.com/you/community/thread/538/

[13] https://www.23andMe.com/you/community/thread/13697

[14] https://www.23andMe.com/you/community/thread/8139/

I guess just like one thing with the internet is it does like bring together large groups of people instantaneously pretty much so you knows it's good that there's always at least the availability at least to start like a massive movement almost at the drop of a hat where you can rally people around an idea. (P4)

These excerpts illustrate how participants viewed 23andMe as resource for bringing people together to learn new information or to work towards changing the status quo.

Potential negative consequences

Alongside these envisioned positive outcomes, participants also discussed a range of privacy and ethics concerns associated with genetic information being aggregated by companies such as 23andMe and available online. These ranged from questions of data ownership and discrimination by employers or insurance companies, to more extreme visions of dystopian futures where people might be disempowered or separated into cast systems based on genetics. Interestingly, all participants also agreed that the potential benefits of genetic testing outweighed the possible negative consequences. Despite their privacy concerns, for instance, all participants were not too worried about a breach of security to the 23andMe site, which was noted in the 23andMe terms of use, and likened this possibility to someone stealing their credit card information.

New ways of seeing

Finally, participants also reflected on future genetic testing technologies as not necessarily a means to a scientific end (*i.e.*, diagnosing a disease), but also as a new way of seeing or understanding the world. For instance, when asked to envision the implications of rapidly sequencing any genetic material, P4 discussed the value of *seeing* or knowing things more intimately:

It's not because you're tying to find something out its the act of knowing like you know something more intimately because you've seen a different side of it. (P4)

Here, P4 reflects on widespread genetic testing as an opportunity to observe living and organic materials differently. To varying degrees, other participants expressed similar ideas, noting that tools for rapid genetic sequencing might help identify surrounding organisms or learn more about the world ("*it would be easier to figure out what things were made of*", P4; "*it might be really neat for findings things*", P1).

To summarize, this section highlighted participants' perspectives on the bigger implications of genetic testing, which ranged from positive consequences for healthcare and bringing people together, to questions about ethics and privacy, as well as new ways to see the world differently.

BIOCITIZEN PUBLICS

Thus far, we detailed our study of 23andMe, including users' motivations, practices, challenges and reflections on the broader implications of genetic testing. Our findings are, in many ways, aligned with Rose et al.'s analysis of biological citizenship [24], particularly by showing how widely accessible genetic data contributes to the blurring of citizenship as a purely national concept grounded in geographic boundaries. Indeed, learning about ancestry was a key motivation for joining 23andMe, and this information resulted in feelings of 'connectedness' to other community members. Most directly, these trans-national connections were made evident through 23andMe's relative finder, which revealed genetic kinship ties across the world; as well as forum features, whereby users interact with others who are, as one member put it, 'genetically similar'. More broadly, the service showed trends in evolution and human migrations, which in the words of one participant suggested 'fluidity' rather than 'stability' between ethnic groups.

With ideas about biological citizenship thus rooted in global inter-connectedness, users of 23andMe coalesce around scientific findings not as passive consumers of data but as active, trans-national participants who interpret, contest, and/or validate their results. New practices, centered around contextualizing and making sense of genetic data are giving rise to sub-communities or *publics*. Similar to other citizen science groups arising out of shared concerns (e.g., local air quality), 23andMe publics are predicated on pressing questions of personal identity, personal health, or family history. Also, like the traditional citizen science efforts to gather local and professional knowledge, 23andMe users share and reflect on personal experiences, lifestyle choices, environmental factors, and cultural beliefs along with scientific (genetic) data. These heterogeneous information sources are aggregated across the 23andMe platform, whereby users draw on the site's research and social tools to create hybrid assemblies of personal narrative, pluralistic discourse, and academic research.

Finally, when these assemblies of hybrid knowledge reveal discrepancies between genetic test results and what participants know about themselves and their world, users collectively contest the underlying data. The emerging dialogues critique the biases, methodology, and scope of professional research: from identifying unfair funding influences, to speculating about the importance of environmental factors that may have been overlooked by studies, or pointing out limitations in participant pools. With this framing of 23andMe users as active science communities, there are many opportunities for HCI to support and sustain the resulting biocitizen publics. Not unlike HCI's involvement with other citizen science groups, future design trajectories might include: platforms for aggregating different types of knowledge; tools for contesting and legitimizing scientific research; and enabling agency within and across genetics communities.

Interactive systems for visualizing hybrid information

Our findings suggest that genetic test results were rarely, if ever, considered in isolation. Instead, participants entangled 23andMe data with personal experiences, family narratives, lifestyle changes, and cultural/historic information. C&C

can support this creative sense-making of hybrid information through new visualizations and sharing tools.

One opportunity lies in treating genes as *informational pivots* that creatively aggregate information about environments, lifestyles, and backgrounds across users. For example, future interactive systems could use graph visualizations: genes can be presented as nodes with which users associate personal experiences, family histories, or cultural and historic knowledge. In addition, systems can link personal narratives and experiences with genetic test results. While the 23andMe service currently only supports text-based input across forums, future systems can enable rich multi-modal metadata to be attributed to specific genes. For instance, users may want to share visual or audio experiences (photos, videos) of living with certain genes.

Considering personal genetics as a first-class organizing principle throughout online services also has the potential to change the way we organize, seek, and share information. With connectedness being a key value for 23andMe users, this approach could more intuitively reveal links between biology, people, and environments. Interfaces with genes as pivots could support DiSalvo's concept of *tracing* [8] to enable fluid navigation between scientific data and other factors such as local history, morals, and personal relationships. Building on 'politics of scale', such platforms can also enable people to become connected not only through their actions [11] but also through their genes.

Tools for creative reflection on scientific research

Aggregating diverse forms of knowledge along with genetic data led participants to critique the underlying research. Our findings show that participants actively problematized or validated 23andMe results. Here, C&C is presented with opportunities to support creative and critical reflection on scientific research. Most directly, sharing mechanisms could enable people to discuss and evaluate the underlying work. For example, future personal genetic systems could enable users to comment on and rate study size, data quality, biases, claims, and other aspects of the research that is drawn upon to present the genetic results. In addition, systems could also more deeply engage people with the scientific method, enabling members of the general public to effectively formulate hypotheses, explore the underlying data, and validate the results. These approaches could embrace *agonistic pluralism* to create productive conflict and people to contest the status quo [9].

Of course, tools for contesting professional research raise questions about the scientific literacy of participants. Earlier research has commented on the limitations of more traditional tools to codify and transfer scientific knowledge [*e.g.*, 16] and services such as 23andMe present new opportunities for disseminating information to people with varying degrees of expertise. The service already supports scientific literacy by communicating information in a variety of ways, from short layman summaries or star confidence ratings, to extensive excerpts from academic publications. Citizen science systems in other domains (e.g., environmental monitoring) could adopt similar or new visual techniques to make scientific data more transparent and legible. For instance, systems focused on factors such as air quality or phenology could more transparently present aspects of the contributing research, such as sample size, duration of studies, reproducibility, or funding sources.

In parallel, creativity support tools can more deeply engage members of the general public in discourse around bioethics, healthcare, and public participation in science. For instance, work in tangible interaction can overtly reveal recent trends in biotechnology research by incorporating genetic information and organic materials into tangible artifacts. New interactive experiences might highlight different biological aspects of the living world. Enabling people to see more intimate information within their bodies and the living systems around them (i.e., new ways of seeing) might bring about new forms of reflection, discussion, and action within and across groups.

Supporting new forms of activism

Finally, as 23andMe users made sense of their results, they inevitably commented on the broader implications of genetic testing. From the potential improvements to public healthcare and ways to bring large groups of people together, to the possibilities of seeing the world in new ways, or concerns about ethics and privacy, 23andMe users engaged with the larger issues around genetics. For creativity systems, this presents opportunities to support new collaborations and activism across communities. With critique of genetic research being a prevalent practice throughout 23andMe, interactive technologies can enable groups to more directly impact professional science work. For example, new tools might allow 23andMe users to create and contribute to advocacy initiatives around genetics research that is relevant to their lives. This could take on the forms of public awareness campaigns to nudge science agendas, tools to encourage more people to participate in science studies, or platforms for raising money to fund new research projects more directly. New systems can also serve to democratize science by interfacing genetic research with related healthcare and policy debates and decisions.

CONCLUSION

As genetic testing continues to become more accessible, communities of participants will grapple with increasingly complex scientific information. This creates opportunities to re-envision how people engage with the intimate data embedded in their own bodies and the living systems around them, and support the emerging citizen science *publics* as they debate and act on genetic research. As a first step, our work examined the practices of 23andMe users, focusing on how participants contextualize their genetic results, critique and evaluate the underlying research, and

reflect on the broader implications of genetic testing. Our findings revealed new research areas for creativity and cognition: platforms for aggregating and visualizing hybrid knowledge; tools that enable creative reflection on scientific research; and systems for supporting collective action within and across genetic communities. These directions can enable broader scientific participation and support citizen science publics at the intersection of genetics and interaction design.

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