Since DeCodeMe (Reykjavík, Iceland), 23andMe (CA, USA), Navigenics (CA, USA) and Pathway Genomics (CA, USA) opened their doors beginning in 2007, direct-to-consumer (DTC) personal genomic services have sparked fears, among academics and public officials alike, that the new field of personal genomics (PG) endangers both the integrity of science and the rights of citizens [1–3]. Worry about DTC genomics first took the national stage in 2006 when the US Federal Trade Commission published a report warning consumers that some ‘at-home’ genetic tests lacked scientific validity [10].

In that same year, Gordon H Smith, chairman of the Congressional Committee that oversaw the 2006 hearing on home DNA testing, called DTC genetic testing ‘modern-day snake oil’ [4]. While many attempt to distinguish PG from the mostly ‘nutrigenomics’ companies reviewed in 2006, PG companies have raised similar concerns. Just this year, Representative Bob Latta dubbed genomic DTC companies, including PG companies, the ‘snake oil salesmen’ of the ‘high-tech community.’ Representative Latta joined other members of the Committee on Energy and Commerce’s Subcommittee on Oversight and Investigations in his assessment that DTC personal genetic testing posed serious threats to consumers [102].

Although such critiques may be prudent, I suggest that framing PG as a dangerous corruption of science and a violation of democratic rights misses most of what is interesting and important about this rapidly emerging new terrain of science and governance. Rather than threatening the integrity of scientific or democratic practice, through yoking the locus of agency in liberal democracies – the ‘person’ – to the locus of agency in the life sciences – genomes – PG has created a potent zone for biosocial formation [5]. It is a zone where much is at stake – both for how we conduct scientific explorations of life and for how we order and govern democratic polities in an age increasingly mediated by the genome sciences.

In order to bring into focus these more fundamental issues, and frame what I hope will be more productive discussions and debates about this new field, in this article I focus my attention on efforts to construct the person and the ‘genomics’ of PG. While debates about personal genomics focus largely on relatively narrow issues of fraud and deception, this emerging new scientific and political terrain poses more fundamental questions about how the study of biological life, as well as the organization of democratic life, should proceed in genomic times.

KEYWORDS: biocuration n biosociality n citizen science n direct-to-consumer genomics n genome interpretation n personal genomics

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At stake in the debate about personal genomics is what kind of person can be trusted to interpret genomes. Deciding this hinges not just on determining if consumers can interpret genomic information, but on deciding which biological and medical experts (if any) can perform these interpretive acts. Understanding why personal genomics has generated such tension and attention requires bringing these struggles, over who can interpret ‘the code of life’, into focus. While debates about personal genomics focus largely on relatively narrow issues of fraud and deception, this emerging new scientific and political terrain poses more fundamental questions about how the study of biological life, as well as the organization of democratic life, should proceed in genomic times.
trace the emergence of the figure of the person in genomics. I argue that this figure was not forced into the space of genomics by suspect commercial forces; rather, genome scientists played a central role in constructing this person as they sought the research subjects needed to transform genomics from a set of novel scientific tools to a proper human science. Persons were not brought in because genomics had become a solid body of knowledge with a product ready to sell to them, but because to become a human science, genomics needed access to human beings and their genomes.

These could not be any old human beings, but rather ones that could be secured as liberated and free, and not subject to scientific exploitation. The figure of the person crafted in liberal democratic theory – a rational person capable of self-governance through access to knowledge – appeared to offer up such free persons. However, just as genomics offered no solid already established body of knowledge ready to be consumed, political theory offered no already formed concept of what constituted a free person capable of using genomic knowledge. Instead, in order to form, PG initiatives needed to at once constitute genomics and persons that could work together.

It should not be a surprise that this proved a complicated task. To be successful, it required negotiating answers to questions not just about whether consumers were capable of making sense of genomic information, but questions that preceded PG about what kinds of persons had the skills to interpret genomic information, and about the kind of thing genomics might be. The paper presents the possible persons – early adopter, impersonal objective scientist and connoisseur – and genomics – new web application, objective human science and curated consumer product – that have to date been put forward by PG practitioners. My goal is to demonstrate that the tensions currently surrounding PG will not be resolved if regulators and scientists continue to presume that there are already established persons and genomics to which PG should be made to conform, for this is exactly what is at stake. PG has hit a nerve not just because it raises questions about fraud and corporate responsibility, but because it raises more fundamental questions about the nature of biological and medical expertise and the constitution of free persons in an age where we expect genomics to be meaningful, both in democratic polities and the life sciences.

From primitives to populations to persons: constituting genomics as a proper human science

Despite being described as the ‘holy grail’ of biology, many biologists understood that on its own, the sequence of the human genome would mean very little [6]. The Human Genome Project (HGP), while enormously valuable for constructing novel technologies needed to read genomes, did not establish genomics as a human science. For this to happen, more than just the few human beings whose DNA the HGP sequenced would have had to have participated. For meaningful and fundamental new knowledge capable of creating a new human science to arise, scientists needed to study the genomes of many more human beings.

However, it was much less clear how to study diverse human genomes. By most accounts, the controversies sparked by the first prominent effort to archive and study human genomes, what became known as the Human Genome Diversity Project (HGDP), threatened all subsequent efforts. Organizers of the HGDP proposed to study human genetic diversity through first ‘picking’ and then sampling ‘isolated’ indigenous populations. This proposal sparked harsh criticisms from some critical biological anthropologists and many indigenous rights organizations who charged organizers with treating indigenous peoples as mere objects of study rather than as human beings with rights (namely, the rights to be informed about the research and to consent to participate in it). For many, this constituted biological racism and colonialism [7,8].

The resulting high-profile controversies surrounding the HGDP caused many researchers to worry that the whole area of human genetic diversity research might be sunk before it had even started [7]. When the US National Institute of Human Genome Research (NHGRI; MD, USA) did brave the waters as the HGP came to a close, they placed much attention on distinguishing their efforts from those of the HGDP. First, they changed their object of study. Instead of ‘diversity’, The NIH would study the less overtly political ‘variation’. Indeed, they named their effort the International Haplotype Map Project (the HapMap Project), a name with little meaning for anyone other than genome scientists. Second, they placed great emphasis on the ethical, legal and social issues (ELSI) raised by their research, explaining that research subjects would be able to participate in research design and regulation [9]. As a hallmark of this commitment, they promised that all genetic
information would remain anonymous, and that communities would participate in the review and design of the project. Far from the indigenous populations whom many believed HGDP organizers had treated as mere objects of research, NHGRI sought to produce subjects who actively participated in the creation of the HapMap [10].

Although this approach successfully steered the initiative clear of any broad public controversies – indeed, some who had been critical of the HGDP lauded its participatory approach – it did not calm discord in the human genomics community. Many genome scientists criticized the HapMap leadership’s approach to ethics, accusing it of a kind of political correctness that not only impeded human genomic research, but created new ethical and political dilemmas [10]. By insisting that subjects’ identities and individual phenotypes not be linked to their genotypic information, some feared that NHGRI would both prevent critical research and impede HapMap subjects’ ability to directly benefit from the research. Indeed, several human genomics researchers began to worry that if the NHGRI approach remained dominant, the field would soon hit a wall. Fundamental new biological knowledge about humans simply could not be created without wide-scale linkage of genotypic and phenotypic information [Author’s fieldnotes].

A 1960s liberal democratic empower communities approach could take genomics only so far. To progress, neoliberal reforms which promoted the powers of individual persons to consume genomic information would be needed.

Rise of the learned person
The idea that an ordinary person could understand and want to participate in genomics is not a new one. Indeed, when HGDP organizers found themselves accused of racism and biocolonialism and surrounded in controversy, some interpreted the situation as the unfortunate result of ‘American activism’ that had allowed ‘an utterly unrepresentative view’ (that of indigenous rights organizations and advocates) to dominate perceptions and opinions [Anonymous interviewee, Oxford, UK, Pers. Comm.]. Those who claimed to represent indigenous peoples may protest in North America, but as a prominent population geneticist in Europe explained to me in 1999, “to anyone who knew anything about population genetics it was clear that you were going to have to study variation between individuals” if you were going to make sense of ‘the basic information’ provided by the HGP. Not only did population geneticists understand this, so did all ‘educated’ people. For example, he explained, the educated and ‘sophisticated’ people of the Orkneys (UK), the Shetland Islands (UK) and other peoples of Western and Eastern Europe embraced genomic research. Indeed, Latvia, Estonia, Iceland, Sweden, UK and at least 14 other countries would all go on to establish population-based biobanks and genomic research [11,12]. The key, this population geneticist argued, is education: “You have much less of a problem with informed consent if you are dealing on the whole with an educated population” [Anonymous interviewee, Oxford, UK, Pers. Comm.].

This interview happened a decade ago. Since then, learnedness has become a central criterion for becoming a subject of genomic research. The preferred subject of human genomic variation research is no longer the ‘vulnerable’ isolated indigenous population but the ‘empowered’ person. This person is a rational individual, capable of self-governance and imbued with rights – centrally, the right to consume. Although the contexts of genomic research have changed considerably since the HGDP debates of the early 1990s, this new empowered and individuated genomic subject emerged in response to issues similar to those salient in the 1990s: the need to recruit subjects to study human genetic variability, and the need to avoid the perceptions of research subject exploitation. The need to avoid accusations of research subject exploitation was of particular concern to many of the key actors who launched PG, many of whom either directly or indirectly participated in the HGDP debates and understood that accusations of exploitation could prove disastrous. Thus, they understood that it would be crucial to work against any perception that PG companies might take advantage of subjects. One way to achieve this was through enrolling subjects who appeared to possess the needed expertise – in short, those who were educated.

Indeed, these are exactly the subjects that PG initiatives targeted. As one PG policy analyst explained to me, from the start their company sought people already knowledgeable about and interested in genetics as it ensured that their customers knew about and freely consented to their service:

“It [participation in the company’s service] has always been by people who are interested. We kind of expect that they will learn enough about genetics to be interested. And so the people who are interested in buying this service are probably going to be from a demographic that’s better equipped to deal
with it... The price was originally quite expensive, so that's already targeting a certain demographic that is more likely to be educated." [Anonymous interviewee, Mountain View, CA, USA, Pers. Comm.]

Although early critics of PG noted that the high price of the PG services would have a negative exclusionary effect, this policy analyst explained how such prices also proved productive for the companies. From the company’s perspective, high prices produced a higher class of consumer who would possess greater knowledge.

Who are the learned?
Not only did companies seek the learned, they attempted to expand the pool of people who could count as a part of this class. To do this, they turned the exploitation charge upside down. Drawing upon arguments that sound remarkably like ones science and society scholars might make as they critique the deficit model of science–society relations that blames ‘the public’ for inadequately understanding science, some prominent PG leaders began to argue that the ‘the lay public’ could understand genetics [13,14]. As Linda Avey, one of the original founders of 23andMe, explained in her inaugural post to her personal blogsite, The Life and Times of Lilly Mendel:

“I still strongly believe in the main reason why my co-founder, Anne Wojcicki, and I started 23andMe in the first place – to take genetics out of the protective realm of the scientific community and make it accessible to the lay public …very vocal scientists seem to be quite threatened by this notion of democratizing DNA. They characterize it as ‘trivializing’, which simply doesn’t make sense. I just don’t agree that providing people with their genetic data, which would be virtually impossible for them to derive on their own, demeans or trivializes it. Rather, I think the research community has taken the notion of ‘human subject protection’ way too far, to the point of unchecked paternalism (for more on this, check out Anne’s post here [103]). And I do think the lay public is capable of understanding that what is currently known about their DNA is mostly a work-in-progress”[104].

Instead of viewing genetic information as something that can harm subjects, Avey argues that it can benefit subjects. Indeed, so much so that far from protecting subjects from their genomic information, we should do what we can to give them access to it. Genomics should no longer be the exclusive preserve of genome scientists. By breaking down the barriers between scientists and lay people, 23andMe argues that PG does not place subjects at risk; rather, it extends their democratic rights through making it possible for them to consume a new valuable commodity: their genomic information. Thus, it announced on its home page after dropping its price from US$999 to $399 in the fall of 2008: ‘23andMe Democratizes Personal Genetics’. Similarly, Misha Angrist, member of the ELSI Advisory Board of the non-profit Personal Genome Project, describes PG as part of the ‘citizen science’ movement [15]. While PG initiatives target a specific demographic that they can trust to be educated – for example, members of the digirati – they also increasingly argue that access to genomic information is the right of all.

However, there is, of course, a potentially large discrepancy between these two kinds of persons: the in-the-know digerati on the one hand, and all people on the other. Indeed, it is this discrepancy that highlights questions about the nature of persons that PG poses: is genomics a domain that only members of the scientific and technological elite (e.g., the digirati) can understand and participate in? Or can it be democratized and extended to all persons (e.g., the lay public)? If it can be democratized, who counts as a person in the democratic polities that PG envisions?

Although PG poses these fundamental questions, these are not questions that the organizers of PG initiatives or their regulators have acknowledged to date. Instead, the answer is presumed. PG companies commonly answer: yes, the person of PG can be anyone. Regulators commonly answer: no, not just anyone understands the scientific and technical complexities one must grasp in order to participate in genomics; only experts can do this. Thus, fundamental issues about the construction of scientific expertise and democratic personhood raised by PG have been largely overlooked.

To bring these questions into view, in the rest of the article, I trace the dilemmas that resulted from PG companies and policy-makers alike, attempting to make sense of the emerging field by assuming that the contours of both the genome sciences and liberal democratic persons already existed, and that they could be used as benchmarks for PG. I argue that in many important ways these benchmarks did not exist, and that instead at stake in PG are fundamental questions about the place of genomics in contemporary sciences and democracies.

Co-constituting democratic persons & epistemic value: PG ‘chicken & egg’ problem
At some level, PG initiatives understood from the outset that they faced a ‘chicken and egg’ problem; that is, they presumed the existence
of a valuable science of genomics and consumers eager for this value when it was exactly these things they sought to build. At least in the case of 23andMe, many joined the effort precisely because they saw the limits of the existing modes of doing genomic research: US government approaches to ethics restricted the number of people who could or would volunteer for research at the very same time that finding genotype–phenotype associations required access to growing numbers of research subjects. A private approach to recruitment unfettered by government restrictions promised to address this problem. However, for this approach to work and for companies to produce consumers who were also research subjects, they first needed a valuable product. Yet the very existence of the companies marked the failure of existing models of genomic research to yield many commonly agreed upon valuable findings. As one genome scientist who had spent time developing products at a leading PG company explained:

“Well, that’s kind of a chicken or egg problem. You need to have this industry developed, become mature enough so we can figure out what the right answer is. And the way to do that is to allow the early adopters to be able to play and fund it effectively.” [Anonymous Interviewee, Santa Cruz, CA, USA, Pers. Comm.]

Companies faced a dilemma, in order to gain the participation of the persons (and associated genomes) needed to transform the power of genomic tools into scientific knowledge they needed to be able to argue that their services drew upon already established scientific knowledge. But that knowledge did not exist yet. To produce it, they needed persons willing to give up their DNA and money. Who might those persons be? As this genome scientist suggests, the early adopter.

Can the person of PG be an early adopter?

Treating the person of PG as an early adopter initially made a good deal of sense, particularly for a company like 23andMe that represented itself as extending the participatory tools of Web 2.0 into the field of genomics. For many years, this model had been a norm in web technology companies that regularly beta-tested their technologies on so-called ‘early adopters’. The approach assumed a mutual benefit – consumers gained the distinction of being the first to try the latest new gadget and playing a role in shaping it while companies received user feedback that allowed them to create technologies of greater value to consumers and companies alike.

Soon, however, questions arose about whether genomics fit this model. In 2008, the first reports, articles and blogs began to appear noting disagreements among PG companies’ test results [2,16,105,106]. Unlike an early adopter model in which the product is allowed to evolve over time incorporating the values of users, these reports operated from the assumption that the PG tests should provide consistent correct answers, and should not evolve or vary. Instead of a new technology in which the incorporation of user values had become commonplace in many sectors, they held PG to the norms associated with science in which one was guided by invariant truth, not changeable values.

In short, the reports troubled the early model of PG which envisioned genomics as another new web-based application, and persons as early adopters. They also began to make clear that to succeed, PG initiatives could not assume, but would have to craft, genomics and persons that could work together. Given the status of the authors of these early critical reports, leading technology bloggers and genome scientists (such as Craig Venter), the kind of genomics that companies next sought to produce was the kind the reports seemed to demand: an objective impersonal science.

An impersonal personal genomics?

As the historian of science Lorraine Daston argues, most assume that objectivity “is and has been a monolithic and immutable concept, at least since the 17th Century” [17]. On this view, achieving objectivity depends on restraining emotions and personal views, on making judgments that are not guided by one’s particular position in the world. In short, objectivity is impersonal and aperspectival. It is this aperspectival and impersonal approach to objectivity that guided the summer 2008 effort to create objective industry standards in the nascent field of PG. Coordinated by the Personal Medicine Coalition (PMC), this effort included the three main PG companies in existence at the time: 23andMe, Navigenics and deCODEme. Company representatives reported that all three made an honest concerted effort: regulators did not push the companies to work together, and when they did come together they shared algorithms and methods. Their goal was to put aside personal interests in order to create standard ‘objective’ approaches to genomic
Curated genomics: a thing of value

Limits of impersonality

A different picture of this effort to create industry standards emerged in interviews with the participants. While everyone I spoke with confirmed that all three companies shared a similar algorithm, they also all agreed that such an algorithm would lead to different results depending on the data inputted into it, and, importantly, many unresolved questions remained about which data that should be: Which SNPs (or variants in the genome) should one pick to analyze? Which papers about these SNPs could be used to determine the odds ratios (e.g., the odds that a person will develop breast cancer)? While the creation of DNA chips and the exponential drop in the cost of sequencing over the last 5–10 years made massive amounts of SNP data available, these developments also generated new questions about how to select SNPs for analysis: How does one sort through the hundreds of billions of sequenced nucleotides to make trusted claims about the links between genetic variants and meaningful traits? How can one decide what data should be cast aside as junk and what data should be trusted? Which ‘reads’ of genomes are clean enough to find SNPs, and which are filled with errors? Which associations are real, and which are not?

In short, crafting objective standards proved much more difficult than anyone first thought. Far from the ‘corporate corruption of objective science’ story often associated with PG, the summer 2008 PMC effort to create industry standards demonstrated that PG confronted a much more difficult problem: crafting a form of objectivity that works for human genomics. Would standards be required? If so, must they cover all aspects of human genomic research? Or might the field need different approaches guided by different epistemic and social values, or even different tastes? In the wake of the limited success of the PMC effort, PG initiatives began to take on and answer these questions. Their central problem: How to sort through the enormous amount of genomic data to come up with reliable, valuable information for consumers. Their answer? Curate.

Curated genomics: a thing of value for a person with taste?

While on one level the explosion of sequence data created a data analysis bottleneck, it also presented companies with an opportunity. Specifically, it created a space for value in the many senses of that term. There is too much data for humans to sort through, so choices must be made about what data to sort and how. These choices embody values, and they create data that is of value. As any bioinformatician will tell you, reads of genomes are a dime a dozen. They are cheap and plentiful, too plentiful. With sequencing costs dropping exponentially, more and more people are getting in the game, producing sequences at greater and greater speeds. However, there has not been a similar decrease in the cost of making sense of the resulting genomic data. As one molecular biologist and PG practitioner explained:

Interviewee: “People are wondering ‘Why isn’t the price [of PG services] going down if the price of genotyping itself is going down even faster than Moore’s law for computer processing? Why isn’t the price of PG going down that fast?’ The answer is that the data generation is getting cheaper and cheaper, but the interpretation has not followed suit yet.”

Author: “It is not going down at the rate predicted by Moore’s law?”

Interviewee: “No it is going up with time probably [laughter]” [Anonymous Interviewee, Silicon Valley, CA, USA, Pers. Comm.].

What became clear in my interviews with PG practitioners is that the work put into finding interesting and meaningful SNPs is the valuable work of the companies. Thus, creating common approaches and resources only goes so far. As this PG practitioner went on to explain:

“We had done this joint article with [two other PG companies] and there was some suggestion of let’s have some central registry where basically you...
Recognizing these limits, the companies largely dropped the effort to create standards. Instead, they began to acknowledge, and even highlight, the subjective dimensions of evaluating and certifying studies.

Some members of the Congressional Oversight Committee noted this phenomenon with some amount of puzzlement in their voices. Why, they seem to implicitly ask, would the companies highlight their different standards? [108].

However, for companies, there was a simple answer to this. Different strategies for evaluating data reflected their companies’ interests, values and intended users. Navigenics, for example, seeks users interested in improving their health, and produces a product that provides consumers with information about genetic variants that they deem important shapers of health (what they describe as ‘utility and relevance’), and health conditions about which something can be done (what they call ‘actionability’). By contrast, 23andMe seeks to attract the digirati and those interested in participating in this new cutting edge science; thus, it wants to be recognized as the most accurate and transparent. These different goals and values shape which SNPs the companies choose to analyze, as well as how they do the analysis.

The terms PG companies have chosen to describe this work is curation. The term is apt: curation, as opposed to objectivity, implies taste and values. It also solves the problem of difference. As a scientist employed by a PG company to be a biocurator explained to me:

“Well I think most people have read some of the papers talking about the differences between the companies. What they take away is the companies give you different results and they don’t always understand why and a lot of it is we have different criteria for what we put up on the website. I don’t think that necessarily the general public or even scientists reading these have followed up and realize that it is different curatorial practices” [Anonymous Interviewee, Silicon Valley, CA, USA, Pers. Comm.].

Unlike objectivity, which carries along with it the expectation of a single true answer, curations are supposed to differ – indeed, that difference is the value. This collection of paintings, of records, of genetic variants has been carefully selected by a ‘discerning eye’, not the ‘God’s eye’ of aperspectival objectivity [18,19]. This is what distinguishes them from all the rest, and why I should value them. Unlike objectivity, curation allows for difference; like objectivity, it is considered a thing of great value.

Perhaps using the term curation should be read as a sign of PG companies’ entanglement in the latest marketing fads. As the New York Times recently reported, everything from craft fairs to food stalls to sneakers now are curated [18]. It has ‘become a fashionable code word’ designed to set one’s products above the rest. This may very well be the case. Yet curation, which has historically been a part of the life sciences in the form of specimen collection, might also be closer to the truth [20]. It may better describe what the scientists employed by PG companies do: sort through vast amounts of data with a discerning eye that is informed by specific tastes and values. If truth in advertising is what so many want from this new industry, perhaps in this instance, as uncomfortable as it might be, this is what they are delivering.

But who can and will consume PG companies curated genomic information? To succeed, curation needs to deliver both epistemic and consumer value. To what kind of consumer can this value be offered?

Genomic connoisseurship?

Following Navigenics bash in lower Manhattan in April of 2008, in September of 2008 23andMe threw a ‘spit party’ in the Chelsea District of New York City (NY, USA) as part of New York Fashion Week. The likes of the Murdochs and Trumps, as well as an assortment of high-end fashion designers attended. In between drinking cocktails they spat into tubes in order that 23andMe might analyze their DNA. As reported in the New York Times Sunday Style section: “Co-founded by Anne Wojcicki, the wife of a founder of Google, the company, which has token financial backing from Harvey Weinstein and Wendi Murdoch, hopes to make spitting into a test tube as stylish as ordering a ginger martini” [21]. Genomic information curated and ready for those with fashion and style? A person of material wealth who also seeks symbolic distinction? Certainly this is what 23andMe hopes for, and seeks...
to create. As a scientist close to the industry explained: “At the original price especially it was certainly an item of conspicuous consumption... It’s like, ‘Oh! I’ve had my genome scanned’” [Anonymous Interviewee, Silicon Valley, CA, USA, Pers. Comm.]. Indeed, some of the early members of the spitterati received 23andMe ‘I spat’ beanies [22,108].

If the genomics of PG is a curated genomics, then its person is once again a person in the know. However, rather than a person with scientific knowledge, this person knows about fashion and taste. Rather than the educated, this is the connoisseur.

Genomic connoisseurship for the masses?

In the last part of 2008 and 2009, it appeared that curated genomics and connoisseurs might come together to form PG. In late 2008, TIME announced that 23andMe won its invention of the year award [23]. Co-founders Anne Wojcicki and Linda Avey appeared on Oprah, the Today Show and in the pages of *Fortune and Forbes* magazine [109]. 23andMe quickly became the chic new thing – that which marked one as a person with distinction [23].

However, the possibility for the creation of such a person seriously diminished in May 2010 when Pathways Genomics announced its partnership with Walgreens and its plans to offer genetic testing to consumers from the shelves of their local pharmacy [110]. This move took PG out of the realm of connoisseurship and into the realm of mainstream consumer culture. It also awakened a slumbering US FDA. As FDA administrator Jeff Shuren explained at the Congressional hearings, while the FDA had already been considering the issues raised by DTC genetic testing, the Pathway Genomics effort to sell in Walgreens was a clear sign that something needed to be done [111].

The specter of the masses had been raised. While 23andMe might be able to argue that they were democratizing genomics through reaching the spitterati, as soon as PG reached Walgreens, it sparked fears about what the uneducated and irrational masses might do once they accessed genetic information off the shelf at their local pharmacy. Would they, as Congressman Phil Gingrey suggested, jump off a building [102]? Or as FDA’s Shuren argued, might they ‘make a decision that adversely affects their health, such as stopping or changing the dose of a medication or continuing an unhealthy lifestyle, without the intervention of a learned intermediary?’ [111].

At play in these discussions were centuries old debates about the threat of the mass to the proper functioning of a democracy, and the role of education and expertise in countering this threat. Dating back as far as the French and American Revolutions, political philosophers such as Rousseau and de Toqueville worried about mob or mass rule: If the people should be, as John Locke argued, the locus of government, how can we ensure that they do not just pursue their individual interests instead of the common good [24]? Rousseau and many other liberal political philosophers argued that education would raise people up from their base interests to the level of rational discourse needed for the people to rule [25]. Shuren clearly feared that without the help of education and educated experts (what he called ‘learned intermediaries’), the ill-informed masses who shop at Walgreens might act to harm themselves. These individual harms could become collective harms as they created new problems for an already overburdened American healthcare system.

Some, such as Shuren, suggest that a possible answer to this problem is to require a ‘learned intermediary’ to shepherd individuals through the genetic testing process. Yet, as Shuren’s own oral testimony indicated, the answer is not so simple:

> “Some companies now are making claims about high-risk medical indications like cancer, about the likelihood of responding to a specific drug. In many cases, the link between the genetic results and the risk of developing a disease or drug response has not been well-established. Even the experts don’t know what the results mean” [102].

It is important to note that Shuren does not say here that experts do not know how to interpret the results of all genetic tests. For example, most would argue that ‘experts’ know how to interpret BRCA1 results. However, even in these more clear cut cases there is debate about who counts as an ‘expert’. For example, James Evans, in his recent editorial in the *New England Journal of Medicine*, questions whether primary care providers know how to interpret most single-gene tests, such as BRCA1 [26].

Concerns that even experts do not know how to interpret genomic information arose frequently in the Congressional hearings. Harkening back to the early criticisms made by prominent tech bloggers and genome scientists, members of Congress who sat on the DTC genetic services oversight hearing noted that different companies often provided customers with different results. Given this, members of Congress asked
the assembled experts and company representa-
tives how consumers could possibly interpret the
data. Congressman Christensen, herself a doctor, asked if doctors can interpret the results, and thus serve as an aide to consumers. The author of the Government Accountability Office report, Gregory Kutz, answered: “The genetic experts we spoke to said that most doctors would not be able to interpret”. Christensen responds: “So who can interpret it?” Later in the hearing, Jim Evans, medical geneticist and editor-in-chief of Genetics in Medicine, answered: “No one knows how to interpret these data. And that is quite clear” [102].

As these excerpts illustrate, what began as a question about the nature of democratic persons, and the role of expertise in their formation, quickly became a question about the nature of scientific persons, and the very existence of expertise. This, I argue, is why PG has generated such tension and attention. While the rise of the consumer in the field of genomics has raised questions about the ability of consumers to interpret genomic information, it has also brought to visibility a much less easily resolved set of questions about who can count as an expert in the field of genomics: a doctor? A computer scientist? A bioinformatician? A medical geneticist? A genome scientist? Anyone at all? While these questions about the nature of epistemic authority in the realm of genomics have been much less visible in the debates over PG, in many ways they account for why the PG debates are unlikely to be resolved anytime soon.

While it is beyond the scope of the paper to explore these questions in detail, I end by considering some of the major fault lines, and demonstrate that what is at stake and what demands greater attention from policy and professional bodies is nothing short of deciding who has the power to interpret and understand human biology in an age where governments and scientists alike feel pressure to make good on their investments in the genome sciences.

**Would you trust a computer to interpret the Bible?**

A major factor that accounts for the rise of genomics is the automation and scaling-up of genetic research made possible by computers. As Christine Hine writes in her recent book, *Systematics as Cyberscience*:

“Genome work is highly data intensive: sequencing of DNA produces masses of data, which can be meaningfully handled only by automatic analysis, searching and pattern matching. Procedures for submitting data to public databases are well established and integrated with journals in the field. If one is looking for a model of cyberscience, then genomics would seem an obvious place to start” [29].

Biology, by contrast, is seen as the ‘least technologically sophisticated and most craft based of the sciences’ [29]. Indeed, Hine cites a study published in 2000 which reported that biologists had less experience with email than other fields of science, and sent and received the least amount of email messages [29].

In the postgenomic era, however, no strict separation between biology and genomics can be drawn. To make genomic data meaningful (i.e., to create new fundamental biological insights), the computer engineers and bioinformaticians who were so central in completing the HGP must now work with biologists. Conversely, it is increasingly difficult to be a practicing biologist and not engage with genomic data. Not surprisingly, in this moment there are significant discussions and struggles over the place of informatics in analyzing genomic data.

Of course, no one is arguing that computers should play no role in analyzing data. The use of computers to generate genomic data almost
ensures that they must be involved; the amount of data to be analyzed is just too large to do otherwise. As one bioinformatician recently explained to me: "You get to a point with so much data that you need computers to reduce to something understandable" [Anonymous Interviewee, Santa Cruz, CA, USA, Pers. Comm.]. The struggle is over how much interpretive work the computer should do.

One trend is to completely automate the discovery process. Many of the new Silicon Valley (CA, USA) start-ups in genomics are developing tools designed to allow researchers to automate more and more of their data discovery process. As one Vice President of Products explained in a recent presentation to potential consumers at the University of California, Santa Cruz (CA, USA), his company’s service allows you to ‘blast your data against every dataset ever created’ [Author’s fieldnotes]. In using the term ‘blast’, the speaker playfully referred to a central genomic tool: the Basic Local Alignment Search Tool, or BLAST. His company would greatly expand the powers of BLAST by allowing you to use it on ‘every dataset ever created’. Doing so would enable you to learn something that you might never have hypothesized, for example – that tibolone might exacerbate breast cancer. Indeed, using this approach, hypotheses are no longer needed. As the company website advertises, ‘6 Billion Data Correlations, One Click’ [112]. With that kind of power, there is no need any longer to guess what portion of the genome might be relevant. You can just ‘BLAST’ it all!

While this might be the trend in Silicon Valley, a different approach to computers still holds at the benches of many practicing life scientists. Bioinformatics is still a relatively new field, and most practicing biologists received no training in it. Thus, as many of the computer scientists and bioinformaticians with whom I work observed, computer-mediated analysis is still a challenge for many.

Many also do not trust the computer to do much interpretive work. Sequencing is one thing; interpreting that sequence is a different order of work. Indeed, some liken this work to translating the Bible [Author’s fieldnotes]. And would you trust a computer to interpret the word of God?

Which humans?

Yet, to say that a computer cannot be trusted to do the fundamental and critical work of interpretation is not to settle the matter. Among those who think humans, and not computers, should do the bulk of the interpretive work, the question still remains: Which humans? Do you need biochemists? Medical geneticists? Physicians? Computer scientists? This is an open question, and an area of contestation with a long history [6]. As one computer scientist succinctly explained to me, part of what is at stake is authority and power. You could, in theory, teach a computer to do much of the annotation work that genome scientists currently preside over at a much faster pace. However, to date, most of the work of annotating genomes has not been automated. To some computer scientists, this appears to be an enormous waste of human energies. However, if we bring into view questions about the construction of professional expertise and authority, the resistance looks less irrational.

To teach the computer to do what life scientists once did is to deskill physicians, medical geneticists and others whose professional life is devoted to interpreting biological life. To put it more succinctly, using computers in this way threatens the authority of biologists, medical geneticists and physicians to interpret the biological world, and to create medical and biological knowledge. Indeed, it may force some of these current experts into obsolescence unless they can retrain and learn the trade of bioinformatics.

Given the enormous symbolic and economic power genomes now are afforded, perhaps it is easy to see why resistance might emerge, and why changes in practices for interpreting genomes might evoke such passion and debate. The struggle is over which persons will have the power to play key roles in interpreting the ‘book of life’.

Conclusion & future perspective

It is the kind of person who can be trusted to know and interpret genomes, viewed by many as the book of life, that is at stake in PG. While to date the discussion of this issue has focused almost entirely on whether this person can include private consumers and members of what many PG practitioners call ‘the general public’, I argue that the issue is much broader. As its heart it is a struggle over what kind of knowledge and skill sets are needed to interpret genomes, and what kind of persons possess these forms of expertise. In an age where genomes are increasingly understood to contain the book of life, it is these persons who will become important as today’s biological and
medical experts. In other words, the kind of person at stake in PG is as much the life scientist and the physician as it is the consumer. To understand why PG has generated so much attention, and to begin to effectively address this tension, we must bring into sharper focus this much less visible struggle over what and who can interpret the genomes.

Personal genomics opens the doors of the genome sciences, inviting in new interpreters right at the moment when significant questions about how and why we interpret genomes are gaining greater significance. Although most of the limelight in the last several years has been on lowering the cost of sequencing, making genomes meaningful requires not just cheap genomes, but interpreted genomes. While the cost of genomes might be declining at a rate faster than Moore predicted, interpretation defies plotting. It is much harder to place a value on interpretation, precisely because it is the very constitution of value that is at stake. Who will create this value? What will be this value? These are the questions at stake in PG, and these questions, rather than questions about fraud or corruption of science by corporate interests, require greater attention. How do we transform the exponentially growing mounds of genomic data into fundamental new insights about biological life? What kinds of standards and expert bodies are needed to guide this process? What role, if any, should consumers and citizens play in this process? These are not questions that will or should be solved by the FDA. Rather than continuing to act as if there are clear answers and standards that some bad actors corrupt and regulatory agency enforce, it is time to take stock of these more fundamental questions that PG provoked and made evident.

Personal genomics raises too many consequential questions about the nature of genomics and the future of the life sciences for the story of this new field to be one of swinging the doors of genomics open to new entrants. For the same reason, the doors will not shut just because someone cries snake oil. In conjuring up the powerful figure of the person, the locus of agency in liberal democracies, and joining it to the figure of the genome, the life

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**Executive summary**

**The terrain of personal genomics**

* Personal genomics presents a new terrain of science and governance that hit a nerve not just because it raises questions about fraud and corporate responsibility, but because it poses more fundamental questions about how to combine persons and scientific knowledge together in an age where we expect genomics to be meaningful, both in democratic polities and the life sciences.

**From primitives to populations to persons: constituting genomics as a proper human science**

* The figure of ‘the person’ was not forced into the space of genomics by suspect commercial forces; rather, genome scientists played a central role in constructing this ‘person’ as they sought the informed, freely consenting research subjects needed to transform genomics from a set of novel scientific tools to a proper human science. The figure of the ‘person’ crafted in liberal democratic theory, a rational person capable of self-governance through access to knowledge, appeared to offer up such subjects. However, just as genomics offered no solid already established body of knowledge ready to be consumed, the law offered no already formed concept of what constituted a free person capable of deciding to participate in genomic research.

**Co-constituting democratic persons & epistemic value: personal genomics’ ‘chicken & egg’ problem**

* To form, personal genomics initiatives needed genomics and persons that could work together. To date, three different pairings have been tried: genomics figured as new web applications that might be taken up by early adopters; as objective human science practiced by scientific experts; and as a new curated consumer product used by connoisseurs. All three pairings raise unresolved epistemic and political issues that are at the heart of the tensions provoked by personal genomics.

**Who will interpret the genome?**

* While the rise of the consumer in the field of genomics has raised questions about how competent consumers are to interpret genomic information, it also raises less easily resolved questions about who can count as an expert in the field of genomics (i.e., a physician? A computer scientist? A medical geneticist? A geneticist?). What began as questions about the nature of democratic persons and the role of expertise in their formation proved so intractable because they linked to these fundamental epistemic questions about construction of expertise about genomes.

**Conclusion & future perspective**

* What is at stake in personal genomics is the kind of person who can be trusted to know and interpret genomes, viewed by many as the ‘book of life’. While to date the discussion of this issue has focused almost entirely on whether this person can include private consumers, at its heart it is also a struggle over what kind of methods and knowledge, and thus, what kind of scientist, is needed to interpret genomes. In other words, the kind of ‘person’ at stake in personal genomics is ‘the life scientist’ as much as it is ‘the consumer’. Given this, the issues raised are unlikely to be resolved by a regulatory agency such as the US FDA. Instead, we require broader efforts that address underlying questions about who has the authority to shape the future of medicine and the life sciences in an age of genomics.
science’s celebrated locus of agency, PG has created a potent zone, a crucible of contemporary biosocial worlds.

Acknowledgements
The author would like to thank Misha Angrist, Rebecca Herzig, Kate O'Riordan and three anonymous reviewers for their very helpful feedback on this article. The author is also grateful to Alberto Camбросio for the invitation to present an early version of this essay at the May 2010 Gordon Cain Conference, ‘Personalizing Medicine Here and Now: Empirical Studies of Post-Genomic Medicine’, and for the helpful comments of the other conference participants. The author would also like to thank Whitney Boesel for her excellent research assistance and for her help formatting this paper for publication.

Financial & competing interests disclosure
The author has no relevant affiliations or financial involvement with any organization or entity with a financial interest in or financial conflict with the subject matter or materials discussed in this manuscript. This includes employment, consultancies, honoraria, stock ownership or options, expert testimony, grants or patents received or pending, or royalties. No writing assistance was utilized in the production of this manuscript.

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- of interest
- of considerable interest

15. Classic collection of essays that address questions about the role of citizens in the production of scientific knowledge.

Websites

The ‘persons’ & ‘genomics’ of personal genomics


103 The Spittoon: Research participants have a right to their own genetic data http://j.mp/RHIrX


