

COMMENTARY

Misdirected precaution

Personal-genome tests are blurring the boundary between experts and lay people. **Barbara Prainsack, Jenny Reardon** and a team of international collaborators urge regulators to rethink outdated models of regulation.

Since the introduction of direct-to-consumer, whole-genome testing in 2007, a handful of new companies have simultaneously fascinated and exasperated observers. 23andMe in Mountain View, California, deCODE Genetics of Reykjavik, Iceland, and Navigenics in Redwood Shores, California, are some of the companies offering consumers disease-risk information based on a genome-wide analysis. To do this, the companies look at up to a million of the single-point genetic variations known as single nucleotide polymorphisms (SNPs).

Despite the wonder of having one's genetic information probed and the allure of celebrity spit parties — publicity events that had the rich and famous providing DNA samples for analysis — these companies' business model raised hackles and fears in the research and public-health communities. Many said that clinical utility was unclear, doctors would be unable to interpret the information, customers would be unnecessarily frightened or erroneously relieved about disease risk, and privacy would be endangered in unprecedented ways¹⁻³.

The companies have successfully navigated legal challenges from California health authorities about lab certification and licensing, but commentators have called for regulatory oversight, or even tight regulation, of personal-genomics services^{4,5}. We believe that anticipatory governance is premature without a better understanding of how SNP-based whole-genome information is used by, and what it means to, a wide range of users. At present, the only anecdotal evidence available is from wealthy (and presumably also healthy) early participants. An understanding of what a broader range of users hope to learn from this type of whole-genome information, and whether it would lead to actual life and behaviour changes, would help in assessing whether personal-genomics services are likely to be adopted in large numbers. This could happen in their current form as stand-alone services, or whole-genome data could enter clinical practice as part of patients' electronic health records, together with family histories and lifestyle information.

Personal-genomics services should not be allowed to circumvent governance for the reasons they propose. Companies argue that individuals should have the right to decide whether to take genetic tests and participate in



genetic research, and that state protection is paternalistic and patronizing, impeding individual rights to consume and participate in the production of genomic knowledge. Although we welcome a shift from genetic protectionism to a situation in which individuals become experts on, and active governors of, their genomes, society should not succumb to fantasies about 'empowered' individuals making free, informed choices in an unregulated genomic marketplace.

Protectionism and empowerment are simply different sides of the same governance coin. Both imagine that good governance derives from decisions that are uninfluenced by political and economic forces. But we do not live in a world where such imaginative fictions of freedom are helpful — the close relationships between modes of producing knowledge and producing economic value are too obvious.

Converging roles

If anything, personal genomics has rendered this relationship even closer. 23andMe, for example, encourages customers to upload health, physical and lifestyle information, and to participate in genetic research. For the first time, we see research participants paying to be enrolled. Here, the notions of donor, customer, patient and activist are merging. Increasing efforts of public and non-governmental agencies and private companies — such as the National Human Genome Research Institute of the US National Institutes of Health based in Bethesda, Maryland, the National Geographic Society in Washington DC and 23andMe — to include 'people' in the design and regulation of research promise a 'democratization' of scientific practice. But democratization should not be assumed to be an *a priori* good; it can produce unexpected costs for researchers and research subjects alike⁶.

The emphasis on individual empowerment often disguises the fact that personal genomics is pushing the individualization of responsibility for health one step further. The quantity of information that individuals need to consider when making choices about their health is on the rise⁷. Apart from increasing the burden of individual responsibility (and the blame for poor health), it is questionable how free and

independent individual choice in this context is: although personal-genomics companies proffer education, those who sell products are seldom the best educators of their potential customers.

Regulation will be effective only if it is informed by the results of a systematic examination of these issues. We recommend that public authorities make it a priority to fund empirical research exploring what individuals expect from personal genomics, and in what way genetic-susceptibility information is likely to affect practices and lifestyle choices. The Coriell Personalized Medicine Collaborative of the non-profit Coriell Institute for Medical Research based in Camden, New Jersey, and a public-private partnership between Scripps Translational Science Institute in San Diego, California, with Navigenics and Microsoft have led by example as they explore systematically how users are making sense of personal genetic tests. Depending on the outcomes of studies such as these, governments should decide to what extent existing regulations of DNA testing should be extended to personal-genomics services, and in what contexts new legislation is necessary.

The best solution is unlikely to be the simple extension of existing regulation of laboratory tests, and of genetic testing for medical purposes⁵. This is because existing regulatory regimes of traditional medical genetic testing are based on assumptions that are no longer tenable in the post-genomics era. For example, the California Department of Public Health, when sending cease-and-desist letters to several personal-genomics companies, assumed that a medical test is a distinct entity governed by a clearly discernible set of experts: doctors

and public-health authorities. This no longer holds true. Genomics blurs the boundaries that make such clear distinctions possible. A genome scan reveals information that is medical, gene-

logical and recreational. And those who scan and interpret the data are not distinct bodies of experts, but instead, novel configurations of geneticists, customers, ethicists, bioinformatics experts and new media executives.

Moreover, some commentators argue that the principle of medical genetic testing in a clinical context doesn't apply, because many doctors know less about genomics than personal-genomics customers themselves⁸. The

"Regulatory frameworks from the genetics age are ill-suited for personal genomics."



industry tends to agree with this criticism, but perhaps for a different reason: the more that physicians need to be involved and trained, the slower the growth of the industry.

In this world of converging roles, both protectionist regulation and notions of consumer empowerment will fail, because they rely on clear boundaries that no longer hold. These are boundaries between experts and lay people; between academic knowledge and economic power; and between patients and donors. Effective responses to this situation require clarification of the novel issues created by the convergence of information about health, consumer and lifestyle choices, and genealogy; novel relations between geneticists, patients, consumers and corporate executives; and the continued intensification of collaboration, on both the research and the patient/consumer sides.

The spell is breaking

Efforts to regulate personal genomics using strategies from the genetics era miss two crucial points: the business is still very much 'in the making', embedded in dense relations between data, services, economic models and research endeavours; and it is also likely that genetic discrimination will cease to be the main concern. These two points are connected. Personal-genomics customers are already going through a process of disenchantment: it is increasingly clear how little power SNP-based readouts of a person's 'genotype' offer for predicting future ailments in an individual. Reported frustrations of 'early adopters' with the kind of information

they've received show that the fascination may be fading. Similarly, we predict that insurance companies will find little to gain from SNP data alone. SNP data are meaningful when embedded in lifestyle data, medical records and family disease histories, and this is exactly where the field will develop. Google Health (<https://www.google.com/health>), a free electronic health-record feature launched earlier this year, already encourages users to store medical records and family histories on the Internet. Given that Google and 23andMe are technologically and financially linked, a possible way of making use of personal-genomics test results could be to link them with other data in one's electronic health records.

The questionable predictive medical value of SNP-based testing, and enthusiastic rhetoric about empowering individuals, should not lead to the conclusion that the field should remain unregulated. However, regulatory frameworks from the genetics age are ill-suited to the task, and premature regulation could have unintended negative effects. Research needs to address the question of how people will use such data. Current arguments for regulation of this nascent industry place a premium on genetic information as a determinant of future health or illness. This is misguided, partly because the arguments rest on a distinction — that might be obliterated — between genetic data and other types of data. We should enter these waters with our eyes open, but not be afraid to get wet. ■

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