What are the stakes? Genetic nondiscrimination legislation and personal genomics

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'The emergence of commercial whole-genome testing has stirred many fears and concerns inside and outside of the scientific community.'

The US Genetic Information Nondiscrimination Act (GINA) was signed into law on 21 May 2008 after more than a decade of debate [1,101]. In light of the absence of a national health insurance scheme in the USA, many celebrated it as a much-needed legal remedy against health insurers and employers discriminating against applicants and clients based on genetic information [2]. Some commentators argued that GINA is especially valuable in times of aggressive expansions of direct-to-consumer (DTC) genetic testing companies selling their services over the internet [3]. These DTC genetic testing services cover a wide range of topics from nutrigenetics and genetic ancestry testing to testing for genetic predispositions for hair-loss. Since November 2007, most North American and European residents can also access information on their own genetic predispositions to a wide range of phenotypes directly over the internet. For between US\$985 and US\$2500 and a small amount of saliva, three companies examine between 600,000 and 1.1 million SNPs spread across their customers' genomes to inform them about their genetic 'lifetime risks' of affliction with a variety of diseases and conditions ranging from diabetes to Alzheimer's disease. While Navigenics (CA, USA) [102] focuses on strictly medical conditions, 23andMe (CA, USA) [103] and deCODEme (Reykjavik, Iceland) [104] also include 'recreational phenotypes', such as ear-wax consistency and eye color, in their basic set of phenotypes.

The emergence of whole-genome testing has stirred many fears and concerns inside and outside of the scientific community [4–6]. Besides calling into question the accuracy of its scientific underpinnings and clinical utility [7,8,105], it was feared that the information divulged to customers could cause needless anxieties or

even be the cause for groundless relief (for example, upon receiving information, which suggests that her lifetime risk for lung cancer is below average, a person could happily carry on smoking), that it could put an additional burden on doctors who are not equipped to interpret whole-genome test results [9,106], or that it would render naive consumers vulnerable to genetic privacy breaches from the side of nosy insurance companies. In addition, representatives of the personal genomics business have called for regulation of the newly emerging market to avoid potential consumers staying away from their services out of fear that 'charlatans' might misuse their data [8,10]. For 23andMe, which has financial and marital links with Google and therefore also with the electronic health record feature 'Google Health' [107], GINA is also to be welcomed for a different reason: Grant Wood of Intermountain Healthcare (UT, USA) was quoted on 23andMe's weblog that GINA makes it "easier for genetic data to be integrated with electronic health records" [8,108].

In this light, the collective sigh of relief that was heard when GINA passed Congress seems understandable. However, is GINA truly the answer to people's prayers?

Does GINA go far enough – or too far?

Let us now take a look at what GINA does and does not do. GINA prevents insurers and employers from discriminating against individuals based on genetic information ('genetic information' includes not only genetic tests but also information about family histories regarding particular diseases), and it stipulates that genetic test results must remain private [11]. Some have argued that the latter requirement exerts undue pressure on employers: it is now their responsibility to ensure that they do not, even unintentionally, divulge genetic information to any third party, or else they might face considerable fines [8].

While some consider the protection provided by GINA as unnecessarily tight (as it prevents insurance companies from doing what they have been doing for a long time: considering

family histories of diseases), others instead draw attention to what GINA does not do. First, GINA does not prohibit discrimination against people applying for life insurance or long-term care and disability insurance based on genetic tests. It also does not prevent healthcare providers from recommending tests to their patients (although they must not require that patients take them). GINA does not apply to members of the military. Finally, it does not prevent discrimination against those whose genetic disease has already been diagnosed (in contrast to those whose genetic tests show a susceptibility to a disease, which are protected by GINA). In other words, GINA protects those who may be or will be affected rather than those who are actually affected and the signs of the conditions are obtainable from other means than genetic tests or family histories. However, as many argue, this is only fair – because why should those who suffer from genetic diseases enjoy better protection against discrimination than those who suffer from nongenetic diseases and disabilities? [2]. In addition, if GINA had protected from potential discrimination all those who suffer from a ‘genetic disease’, would that not start a ‘war of definition’ over what counts as a genetic disease? Apart from some rare exceptions, it is hardly possible to say with certainty that any particular disease or disability is not at all ‘genetic’ [12–15,109].

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Before we look at the implications of GINA for personal genomics, let us first examine its effects for specific high-risk mutation tests for late-onset diseases such as breast cancer. Within the GINA regime, a woman clinically diagnosed with breast cancer could be turned down or ‘punished’ with higher insurance premiums, or discriminated against in the process of seeking employment, while the same could not legitimately be done based on a genetic test or a family history of breast cancer. In other words, while insurers and employers are not allowed to use test- and family history-based genetic information, GINA does not prevent them from using any genetic information at all – because

genetic information can also be obtained through other means than genetic tests or family histories. As Godard *et al.* point out, “insurers can use genetic information available in medical files; the registered information in medical files is usually more accurate and complete than what is known by the insurance applicants”. As a brief comparative side note, in Europe, regulatory frameworks regarding genetic nondiscrimination vary – in the UK, for example, a moratorium is in place between the government and the Association of British Insurers on the latter using results of DNA tests in insurance underwritings up to a certain value [12,110]. The moratorium, which is a voluntary agreement, stands until 2014. One should note, however, that this applies to life and private health insurance only, owing to the existence of the National Health Service. This is probably also the reason that the UK has not issued its own Genetic Nondiscrimination Act.

GINA & personal genomics

For users of the aforementioned commercial personal genomics tests based on whole-genome scans, GINA clearly protects them – if they are under US jurisdiction, of course – from insurers or employers discriminating against them based on the results of whole-genome tests (note that individual US states may have stricter safeguards that remain unaffected by GINA). However, presently, it remains very doubtful that employers or insurers would be interested in such information at all, given the typically very small effects of tested SNPs on the expression of certain phenotypes. However, GINA might relieve customers of personal genomics services of worries about the future for job or insurance applications: employers and insurance companies cannot demand them to disclose whether whole-genome scans have shown that they are ‘above average risk’ for anything.

‘Regarding whole-genome scans, it is very doubtful that at present employers or insurers would be interested in such results at all, given the typically very small effects of tested SNPs on phenotypes.’

But, again, what sounds great to some causes worries for others. Some are concerned that an unintended side effect of the enactment of GINA will be that people will feel ‘too safe’ – so safe that many more will be happy to purchase whole-genome tests [8]. For those who worry

about genetic privacy, the schemes employed by personal genomics companies that store data online [111] might bear additional hazards that GINA is not equipped to address. The main risk is data access by third parties which customers might not be aware of. For example, most personal genomics companies put the information which explains that data might be passed on to law enforcement authorities in the small print of their 'terms of service'. In addition, bloggers have already praised the website of deCODEme, for example, as "one hell of an interesting server to hack into" [112].

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Nevertheless, all in all, the enactment of GINA is a very good development. It moves issues of privacy protection in the Web 2.0 world more into the center of public debate. Many potential unintended consequences of DTC genetic testing, as well as the practice of web-exhibitionism (I have stopped counting how many people posted their whole-genome data online), can probably be avoided simply by increasing public awareness and debate. In addition, as Altman *et al.* pointed out, protection from genetic discrimination will also help to ease the fears of

medical researchers concerning sharing datasets of their study participants, thereby removing a 'key obstacle for data sharing' in the medical research context [16]. Perhaps most importantly, as more and more people will realize that the predictive value of genetic susceptibility tests alone is very limited in most cases, we are likely to experience a decrease of interest in commercial services that offer DNA analysis alone. Personal genomics might remain with us as a fun tool for those who have money to spend, but unless it is being connected with other lifestyle and phenotypic data, individual whole-genome data will not rock the world. The insurance business, of course, knows this already.

Acknowledgements

I thank David Gurwitz, Shiri Shkedi, Hendrik Wagenaar and my colleagues from the 'Genes Without Borders' project (funded by the Austrian Federal Ministry of Science and Research: www.gen-au.at) for helpful discussions and comments on the manuscript.

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 the 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.

Bibliography

- Hudson KL, Rothenberg KH, Andrews LB, Kahn MJ, Collins FS: Genetic discrimination and health insurance: an urgent need for reform. *Science* 270, 391–393 (1995).
- Hudson KL, Holohan MK, Collins FS: Keeping pace with the times – the Genetic Information Nondiscrimination Act of 2008. *N. Engl. J. Med.* 358(25), 2661–2663 (2008).
- Howard HC, Borry P: Direct-to-consumer genetic testing: more questions than benefits? *Personalized Med.* 5(4), 317–320 (2008).
- Ready or not. *Nature* 452, 666 (2008).
- van Ommen GB, Cornel MC: Recreational genomics? Dreams and fears on genetic susceptibility screening. *Eur. J. Hum. Genet.* 16(4), 403–404 (2008).
- Kaiser J: It's all about me. *Science* 318, 1843 (2007).
- Janssens AC, Gwinn M, Bradley LA, Oostra BA, van Duijn CM, Khoury MJ: A critical appraisal of the scientific basis of commercial genomic profiles used to assess health risks and personalize health interventions. *Am. J. Hum. Genet.* 83(3), 539–599 (2008).
- Allison M: Industry welcomes Genetic Information Nondiscrimination Act. *Nat. Biotech.* 26(6), 596–597 (2008).
- Hunter DJ, Khoury MJ, Drazen JM: Letting the genome out of the bottle – will we get our wish? *N. Engl. J. Med.* 358, 105–107 (2008).
- Brownlee S, Lenzer J: Direct to consumer genetic testing: knowing me, knowing you. *BMJ* 336, 858–869 (2008).
- Korobkin R, Rajkumar R: The Genetic Information Nondiscrimination Act – a half-step toward risk sharing. *N. Engl. J. Med.* 359(4), 335–337 (2008).
- Godard B, Raeburn S, Pembrey M, Bobrow M, Farndon P, Aymé S: Genetic information and testing in insurance employment: technical, social and ethical issues. *Eur. J. Hum. Genet.* 11(Suppl. 2), S123–S142 (2003).
- Wilson D: Acquisition and disclosure of genetic information under alternative policy regimes: an economic analysis. *Health Econ. Policy Law* 1, 263–276 (2006).
- European Society of Human Genetics: Genetic information and testing in insurance and employment: technical, social and ethical issues. *Eur. J. Hum. Genet.* 11(Suppl. 2), S11–S12 (2003).
- Zimmerman R: What is genetic information? *Genetics Law Monit.* 1(5), 9–13 (2001).
- Altman RB, Benowitz N, Gurwitz D *et al.*: Genetic nondiscrimination legislation: a critical prerequisite for pharmacogenomics data sharing. *Pharmacogenomics* 8(5), 519 (2007).

Websites

101. The Genetic Information Nondiscrimination Act at the National Human Genome Research Institute
www.genome.gov/24519851
102. Navigenics
www.navigenics.com
103. 23andMe
www.23andMe.com
104. deCODEme
www.decode.me
105. Boseley S: DNA test website raises accuracy fears. The Guardian online (22 January 2008)
www.guardian.co.uk/science/2008/jan/22/genetics.health
106. Henderson M: Handle with care: genetic tests are risky, and I've got the proof. The Times online (1 March 2008)
www.timesonline.co.uk/tol/news/uk/science/article3463550.ece
107. Google Health website
www.google.com/health/html/tour/
108. Andro Hsu at 23andMe's weblog "The Spittoon", 'Capital Hill, Health IT, and a movie' (11 July 2008)
<http://spittoon.23andme.com/2008/07/11/genetics-day-on-the-hill/>
109. The Wellcome Debate at The Wellcome Collection
www.wellcomecollection.org/exhibitionsandevents/pastexhibitionsandevents/The-Wellcome-Debate/index.htm
110. Department of Health, 'Concordat and moratorium on genetics and insurance' (2005)
www.dh.gov.uk/assetRoot/04/10/60/50/04106050.pdf
111. '23 and You': Linda Avey and Anne Wojcicki talk with Michael Specter on genetics. From 'Stories from the Near Future', the 2008 New Yorker Conference. Conference Video
www.newyorker.com/online/video/conference/2008/23andyou
112. Weblog 'Decode Your Genome'
http://blogto.com/tech/2008/01/decode_your_genome/