

Privacy Concerns in Personal Genomics

STEP paper submission

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Executive Summary:

The President, as expected, recently signed the Genetic Information Nondiscrimination Act (GINA) into law. This bill will hopefully shield much of the public from genetic discrimination. The bill, in addition to other legislative and regulatory actions across the country, however, will fail to effectively protect the privacy of consumers of the personal genomics industry which has only just recently emerged. Succinctly, personal genomics refers to the growing number of services -- all purportedly educational and research-driven to escape FDA scrutiny -- that promises to provide customers with a glimpse of their genomic sequence, and thus their medical future. This paper compares personal genomics to the Web 2.0 phenomenon, noting in particular the propensity of users to unashamedly share their (and often their friend's) personal data -- effectively contributing to the erosion of online privacy. Personal genomics, with its focus on providing the lay public with complex genomic data, that is to all intents and purposes more revealing than their medical records, and encouraging them to share it with their friends, raises many of the same non-trivial privacy issues. With the concern that over-regulation of the industry, fueled predominantly by privacy concerns, will chill the important collection of genomic data, this paper proposes a number of suggestions that could easily be implemented by the nascent personal genomic

industry to reduce these privacy concerns, yet retain the flexibility necessary to nurture the underlying science and technology.

Introduction

The sequencing of the entire human genome was a triumphant coda to the innumerable successes and discoveries of twentieth century science. And like many of those publicly funded discoveries, genomics has been hastily transformed into a consumer technology: personal genomics. Personal genomics --in contrast to the relatively established single-gene testing industry-- fundamentally refers to the large-scale sequencing, deciphering and exploration of individual genomes. Personal genomics companies provide services that run the gamut from the cataloging the hundreds of thousands of discrete DNA sequence variations (approximately 0.2% of your genome) that potentially correlate with a genetic condition, to providing you with your entire 6 billion base-pair genetic sequence.

The Technology

The underlying science and technology is the result of a confluence of a number of biotechnological successes. Nobel Prize winning DNA sequencing technologies gave way to the genome project, a decade long government sponsored endeavor through the nineties and into the new millennium that gave us a representative sample of the entire human DNA sequence. However, contrary to conventional wisdom and media hype, this information was not very informative on its own. To add value to the raw sequence data, scientists have been analyzing and annotating the genome in an effort to catalogue and determine the function of not only the 20 to 25 thousand genes coded for by our DNA, but small sequence variations (in both coding and non-coding regions of DNA) between individuals, including single nucleotide polymorphisms (SNPs). These polymorphisms, although themselves often benign, are informative as they will frequently correlate with genetic diseases, health conditions, or physical manifestations such as hair color, eye color or height.

Rising computational power with concomitant plunging costs in digital storage and computational speed, coupled with dramatic expansions in sequencing abilities and breakthrough high throughput experimental techniques have helped to turn these successes in DNA sequencing and genetic variation analyses into a viable, commercializable consumer technology. Given the current rate of scientific advancement in genomics, costs for personal genomic screenings will continue to fall precipitously making the technology accessible to an ever widening audience.

The growth of web-sites devoted to educating and informing the concerned patient exposed an undercurrent of consumer anxiety that is only just being tapped by the personal genomics industry which promises to add to the information that you can bring to your doctor on the next visit. Add to this the potential for determining actionable health information and early detection of diseases from a complex code implanted within us at conception, and you have the next 'killer app'. Analogous to the continued marketing success of full body CT scans that are commercially offered to empowered medical consumers in their quest for self knowledge and

wellness, personal genomics provides a similarly tantalizing technology to that market demographic, without any appreciable health risks.

Government Efforts to Regulate

Efforts by New York and California have tried to constrain direct to consumer genomic testing technologies. California recently sent cease and desist letters to 13 personal genomics companies reminding them that state law requires the authorization of a medical doctor and the cooperation of a licensed diagnostic laboratory for each consumer test. And, two bills have been introduced in Congress calling for greater monitoring of these personal genomics companies and attempting to harmonize state and federal regulation of the industry.

In addition, the President recently signed the Genetic Information Nondiscrimination Act (GINA) into law effectively prohibiting insurance providers and employers from discriminating on the basis of genetic testing. With Congressional assurances of privacy and immunity from discrimination, consumers are even more likely to quickly jump into the personal genomics market.

Unfortunately, personal genomics creates new and unforeseen privacy issues that will not be remedied by these state and Federal actions.

Personal Data Sharing

In an effort to distinguish themselves within the current small field of personal genomics firms, companies have portrayed themselves as either primarily health or recreationally minded. Some claim to provide more user-friendly and medically actionable statistics for their customers or limit customer access to only vetted gene-disease relationships. This data can then be passed on to a physician to determine what if anything the patient should do. Other companies give their users access to the entire set of raw genomic data, and provide a more recreational genomics atmosphere, providing tools to share and compare data with other users.

And, in addition to ostensible medical uses, personal genomics also provides social applicability in the form of ancestry exploration or researching prospective dates, and potential entertainment value through the eventual comparing and cataloguing of the polymorphic genes of successful athletes, intellectuals and celebrities.

Privacy Concerns

In contrast to medical records that are traded almost exclusively among authorized doctors, personal genomics will allow equally if not more revealing information to be viewed, traded, and potentially even data-mined, in the online bazaar. Similar to the devastating erosion of online privacy where effectively indelible web pages disclose personal information, confidential emails are rapidly and widely circulated, and surfers unwittingly drop revealing digital bread crumbs, personal genomics undercuts privacy to a new degree.

And, like many users on social networking sites, consumers may not realize how much of their privacy is compromised. But, unlike many of the web 2.0 neophytes who casually and cavalierly post their entire lives online, personal genomics will not only have privacy repercussions for the consumer, but also for any of his relatives; an individual's genome reveals half of the genome of his parents and children and a substantial fraction of his sibling's. Just like posting a picture on MySpace or Flickr can reveal a lot about you and those in the frame with you, when someone shares his genotype, by choice or otherwise, he is exposing substantial private information about himself and his close relatives.

Although we do not currently understand even a subset of the genetic influences on our lives, eventually we will; but by that time it will be too late to retract the genomic data that many of us imprudently uploaded.

Naively one might assume that Federal legislation like GINA, designed to harmonize what was until now a patchwork of state and local laws regarding genetic discrimination, would protect consumers from these privacy concerns. But while insurance companies and employers are prohibited by GINA to ask for genetic information, they are allowed to access freely available information, the type that is produced by personal genomics companies and shared by their consumers, and will likely be collected and indexed by enterprising marketing firms. Further, health insurers and employers are only a small subset of people that can discriminate based on genetics. Life and disability insurance providers, for example, are not included in the current legislation. GINA only limits discrimination, but one can imagine that personal genetic information can be used for a host of other purposes, from unauthorized scientific research to selective dating or just general voyeurism.

Who Should Regulate the Industry?

At this juncture there are two possibilities, independent self regulation by the industry or overburdening government regulation. This paper suggests the latter.

Notwithstanding the possible repercussions to consumers and their relatives in terms of job loss, inability to obtain insurance, or general social stigma that will most likely occur despite the best intentions of Congress to fight genetic discrimination, placing high barriers to acquisition of genomic data through government regulation of the personal genomic industry may chill the use of personal genomics and the concomitant important collection of data for vital research purposes. There is a point at which the complexities of compliance with government regulations effectively serve as a ban on the technology. For risk averse biotech companies and wary consumers this threshold is easily met.

And, despite the aforementioned privacy concerns, individuals should be free to share their own genomes, and notwithstanding paternalistic efforts to control the disclosure of genetic information, the government probably does not have a strong enough privacy interest to constrain consumers' free speech. But, without substantial oversight, personal genomic companies might be unable to effectively deal with the varied ethical and moral concerns that might arise, and consumers will belatedly realize the devastating privacy implications for themselves and their families. It is therefore imperative that the personal genomics industry

proactively and independently incorporate the tools necessary to protect the privacy of their consumers.

Suggested Courses of Action

The generic answer to these concerns routinely involves the usage of boilerplate informed consent forms, the ethicist's acknowledgement of the individual's absolute personal autonomy. Typically though, this consent is limited to the acknowledging individual and bounded by the conditions outlined therein, but personal genomics asks the individual to effectively forego complete anonymity and privacy, to extend the reach of the consent beyond themselves to include their family and community members, and to expand it to incorporate information and experimentation not as of yet even imagined. Thus, the advent of personal genomics raises issues that could make the current application of informed consent meaningless.

Nonetheless, there are numerous other efforts that could be easily incorporated by the industry to help limit the aforementioned privacy concerns. Personal genomics companies may look to other industries for useful methods of protecting their consumers. For instance, like cigarette packaging, the results of your personal genomics screening could come with a simple yet effective warning about the potential privacy repercussions associated with sharing this very personal information. Personal genomics companies might also consider a requirement for an extensive live consultation with a genetic counselor, and a second tier of informed consent prior to allowing consumers to share the results might also be warranted.

In contrast to the current incomprehensible standard informed consent forms typically used within the medical community, personal genomics could require that all consumers complete a short online course outlining the potentials and pitfalls of the technology. Similar to the web technologies employed by online educational institutions and driving schools, the ability to share your data would be contingent on your successful completion of the online course including various quizzes along the way to test your comprehension of the material.

Personal genomics companies should also invest heavily in education campaigns informing the public as to the medical usefulness of the data, and the possible repercussions for the consumer and his family should the data be casually and widely shared. Given the heavy influence of Hollywood and other media on the general public's scientific education, powerful metaphors that tie into blockbuster movies such as GATTACA will be very effective in illustrating the potential harms and repercussions of carelessly sharing this data.

A comprehensive, lay-friendly, and scientifically reliable database providing actionable and relevant genetic information would also help to clarify the need for privacy in personal genomics and make the impact of sharing data more real to the consumer.

Additionally, corporations could choose to not offer personal genomics services to minors or others who might be unable to comprehend or predict the repercussions of their new-found genetic knowledge. Similarly, companies could voluntarily choose to not divulge the most egregious of the discovered information without further assurances from the customer that they understand the impact of such knowledge on their immediate family.

Conclusions

There are no simple solutions. As technology pushes forward ,substantial privacy issues will continue to rise. And, perhaps, just as the internet changed our perceptions of personal space and privacy, personal genomics will require society to reevaluate our current standards of medical confidentiality and privacy.