

Users' Attitudes, Perception, and Concerns in the Era of Whole Genome Sequencing

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Abstract

Advances in Whole Genome Sequencing (WGS) will soon allow most individuals in developed countries to have access to their sequenced genome, potentially yielding revolutionary medical and societal implications. However, WGS also comes with the risk of amplifying a number of ethical and privacy concerns. In this talk, we begin to assess the perception of genetic tests, the attitude toward different WGS programs, as well as the perception of privacy and ethical issues with WGS. We report on a series of semi-structured interviews, involving 16 participants, and analyze the results both quantitatively and qualitatively. Our preliminary study yields some interesting findings and highlights the need for more ethnographic studies in the field.

1 Introduction

In the past half a century, DNA sequencing—the process of determining the precise order of nucleotides within a DNA molecule—represented one of the most active and fast-paced research frontiers.

The first complete human genome was sequenced in 2007 as part of the 13-year Human Genome Project. The last five years have then witnessed a race toward faster, cheaper, and more accurate Whole Genome Sequencing (WGS) technologies. Costs have quickly plunged from Human Genome Project's \$1B to \$250K in 2008 and to less than \$5K in 2010. In 2012, it was reported that the \$1,000 mark would soon be crossed.

The landscape of companies and technologies in WGS is fast-evolving, nonetheless, it is not far-fetched to predict that, in 5-10 years, most individuals in developed countries will have access to their sequenced genome. The emergence of affordable WGS technologies represents, undoubtedly, an exceptional breakthrough, due to the related medical and societal implications. Experts predict that advances in WGS will unlock the full potential of *personalized medicine* [2]—the practice of tailoring pre-symptomatic examinations, diagnosis,

and treatment to patients' genetic features. A number of companies (e.g., 23andMe.com and Knome) already provide customers with detailed reports on predisposition to diseases and conditions. A few drugs (e.g., for cancer, HIV, or thrombosis treatment) are paired with genetic tests needed to assess either the correct dosage or their expected effectiveness.

The availability of patient's wholly sequenced genome enables clinicians, doctors, and testing facilities to run a number of complex genetic tests in a matter of seconds, using specialized computational algorithms (as opposed to more expensive and slower laboratory tests). On the other hand, however, WGS also comes at the risk of amplifying important security, privacy, and ethical concerns, thoroughly overviewed in [1], that stem from the unparalleled sensitivity of DNA data.

The emergence of personal genomic tests as well as the (likely imminent) availability of affordable Whole Genome Sequencing (WGS) motivate the need for better understanding the associated perception and concerns of involved users. Computational genomic tests that involve end-users should be usable by, and meaningful to, regular non-tech-savvy individuals. This translates into non-trivial questions, such as: how much understanding should be expected from a user running a test? Do privacy perceptions and concerns experienced by patients correspond to what the scientific community would expect? How can we identify effective mechanisms to communicate potential privacy risks associated with genomic information and its disclosure?

A few studies have analyzed individuals' response to learning the results of some genetic tests and potential discrimination concerns associated with genomics. However, to the best of our knowledge, no study, thus far, has studied focused on the implications of WGS.

In this talk, we start to address this gap by presenting the results of a series of (IRB-approved) semi-structured interviews, involving 16 participants, aiming to assess the perception of genetic tests, the attitude toward different WGS programs, as well as the perception of pri-

Exp. A	Assessing the perception of today’s genetic tests
Exp. B	Comparing the attitude toward different WGS programs
Exp. C	Assessing the perception of potential privacy/ethical issues with WGS
Exp. D	Comparing the response to medical, genomic, and personal information loss

Table 1: Overview of study experiments and goals.

vacy and ethical issues with WGS. Results are analyzed both quantitatively and qualitatively, yielding several interesting findings related to the issues of control, trust, and discrimination, and highlighting the need for more ethnographic studies in this field.

Talk Outline: We provide background information about genomics and related privacy/ethical threats. After presenting our methodology, we analyze study results and provide a few recommendations, based on our findings, for user-centered designs of security- and privacy-enhancing tools for storing and testing genomic data.

2 Interview Methodology

Our study consisted of a series of semi-structured interviews with 16 participants (8 female, 8 male). It was reviewed and approved by PARC’s Institutional Review Board. Participants were recruited using social networks and internal mailing lists of a university and a company, announcing a study on the “knowledge and perception of DNA testing.”

Participants ranged in ages from 18 to 74, with 43% of them being between 25 and 34 years old. All were college educated, with 75% of them possessing a post-graduate degree. We deliberately decided to recruit users with higher education and more participants in the 25-34 age range as they constitute the representative population for personal genomic tests. We also collected information about their personal yearly income, and assessed their Westin’s Privacy Index. (The Westin’s Privacy Index [3] classifies users among privacy fundamentalists, pragmatists, and unconcerned, according to their responses to a three-question survey.)

Interviews lasted approximately 30 minutes (on average) and consisted of three parts: (1) first, we asked participants to provide 1-2 examples of genetic tests and describe their familiarity with genetics; (2) we interviewed participants while guiding them through a set of slides depicting a few hypothetical scenarios (this constituted the core of our study); (3) to collect demographic information and assess their Westin Privacy Index [3], participants were asked to fill out a short survey.

Our interviews aimed to conduct the four experiments presented in Table 1.

3 Summary of the Results

1. Perception of genetic tests depends on the related perceived medical benefit. Participants were mostly inclined to genetic tests that can help fight diseases, and less to tests that can help prevent them. Also, they were neutral w.r.t. discovering their ancestry.
2. On the account of trust, participants preferred that doctors administered medical genetic tests, rather than specialized personal genomics companies.
3. Participants raised the issue of control and preferred to retain and own their sequenced genome, and minimize their fear of potential discrimination.
4. Labor and healthcare discrimination were the top concerns among interviewees, with an increase unfairness feeling associated with the former, and a lack of confidence in the protection granted by the law. Fear of discrimination was predominant, compared to privacy threats, which were not fully understood by participants.
5. Participants were more frightened by having their financial identity and/or personal data stolen (with a bias toward the former for high-income, privacy fundamentalist users) than medical and genomic information. However, the perception of genomic information loss varied significantly among different participants and suggested that participants were more scared of their insurance provider or their employer using genomic information against them than of a hacker having access to it.

Conclusion. We conclude our talk by presenting a few recommendations, based on our findings, for user-centered designs of security- and privacy-enhancing tools for storing and testing genomic data. In the process, we encourage discussion and feedback from domain experts in the audience.

References

- [1] E. De Cristofaro. Whole Genome Sequencing: Innovation Dream or Privacy Nightmare? arXiv preprint 1210.4820, 2012.
- [2] G. Ginsburg and H. Willard. Genomic and personalized medicine: foundations and applications. *Translational Research*, 154(6), 2009.
- [3] P. Kumaraguru and L. F. Cranor. Privacy indexes: A survey of Westin’s studies. <http://reports-archive.adm.cs.cmu.edu/anon/isri2005/CMU-ISRI-05-138.pdf>, 2005.