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BIOC 158

Final Project

07/07/2015

# Genetic Privacy: General Law and Application to Clinical Studies

"Genetic privacy" has emerged as a term to encompass an individual's right to protection from non-voluntary disclosure of genetic information [6]. Before genomics emerged as a widespread discipline, the concept of medical privacy could be maintained by ensuring that only the people who were involved in the medical treatment of the individual would be allowed access to his or her medical information. This is not necessarily feasible with many aspects of genomics. For example, large-scale genomic research, or compilations of large-scale databases containing genomic information that can be traceable to an individual inevitably allow breaches in traditional medical confidentiality. Because of this, we must consider letting go of the traditional concept of medical confidentiality, in order to keep up with the ever-changing aspects of genomic medicine. This paper will provide a general overview of current laws in place that concern "genetic privacy", as well as address the issue of clinical studies and "genetic privacy" by proposing a new open-consent model approach for clinical study protocols, that was developed within the Personal Genome Project conducted by the Harvard Medical School. This new approach will demonstrate the feasibility of the co-development of ethics and genomics in specific study protocol, and focus on the quality of consent to participation in studies using correlated genotype and phenotype data.

## Federal Genetic Privacy Law

"Genetic privacy" law emerged in 1974 when the Federal Privacy Act was passed. This law governs the collection, maintenance, use, and dissemination of personally identifiable information about individuals that is maintained in systems of records by federal agencies [7]. It prohibits the disclosure of information from a system of records without the written consent of the subject individual, unless the disclosure is pursuant to one of twelve statutory exceptions [7]. It also provides individuals with a way to seek access to and amend their records, and imposes agency record-keeping requirements [7]. Although this was a good beginning to regulating access to individual information, it only applies to federal agencies, which limits the impact of the law.

In 1996, the Health Insurance Portability and Accountability Act was passed, otherwise known as HIPAA. Within this act is the “Privacy Rule” with an effective compliance date of April 14, 2003 [7]. It regulates the usage and disclosure of “Protected Health Information (PHI)” held by “covered entities” including health insurers, medical service providers, employer sponsored health plans, etc. It was extended to independent contractors and entities that fit within the term “business associates.” In 2003, genetic information was ruled to fit under “Protected Health Information” [2]. This was a large step forward for "genetic privacy" in terms of covering more of the entities that had access to individual health information. However, HIPAA does not fully protect against genetic discrimination, nor does it apply to individual health insurers except if covered by the portability provision. More specifically, it does not prohibit the use of genetic information as a basis for charging a group more for health insurance; it does not limit the collection of genetic information by insurers; it does not prohibit insurers

from requiring an individual to take a genetic test; it does not limit the disclosure of genetic information by insurers [2]. Therefore, it does not prevent the very genetic discrimination that "genetic privacy" is supposed to prevent.

Beyond HIPPA is the Genetic Information Non-Discrimination Act of 2008, or GINA. This act prevents discrimination against employees or applicants because of genetic information [4]. More specifically, it prohibits the use of genetic information in making employment decisions, restricts employers and other entities covered by Title II of the act from requesting, requiring or purchasing genetic information, and strictly limits the disclosure of genetic information [4]. Therefore, GINA tends to cover some of the inadequacies of HIPPA. However, there are some caveats that need to be addressed. For example, GINA does not cover life, disability, or long-term care insurance, and does not protect individuals with pre-existing conditions, employers with less than fifteen employees, the U.S. military, the Indian Health Service, the Veterans Health Administration, or the Federal Employees Health Benefits Program [4].

Another health care policy that serves to remedy some of the weaknesses of GINA is the Patient Protection and Affordable Care Act, more commonly known as ACA. It protects those with pre-existing conditions from discrimination, including anything that is present in the results of a genetic test [1]. In addition, it requires insurance companies to publicly justify any unreasonable rate hikes, which serves to further prevents genetic discrimination [1]. Lastly, it prevents arbitrary withdrawals of insurance coverage, and guarantees your right to appeal any withdrawals [1].

## State Genetic Privacy Law

Although there have been various federal laws enacted, state laws still vary across the country. There are different categories for the levels of genomic privacy protections including: required personal access to genetic information, required consent to perform/require genetic tests, obtain/access genetic information, retain genetic information, disclose genetic information, defining genetic information and DNA samples as personal property, and specific penalties for "genetic privacy" violations [5]. As of yet, none of the fifty states have laws that require all of these levels of "genetic privacy" [5]. Alaska is the closest state to having a comprehensive "genetic privacy" state law plan, but it does not require personal access to genetic information [5]. It is also the only state that defines DNA samples as personal property [nscl]. There is such a wide range of laws depending on the state, so more federal laws are necessary to standardize "genetic privacy" across the country.

## Personal Genome Project Model

As demonstrated, there is clearly a need to adapt the current medical confidentiality and privacy structures in place to the emerging field of genomics. Though the laws currently cover much of the issues surrounding "genetic privacy", a new model has been proposed by the Personal Genome Project at Harvard that is meant to focus on the quality of consent to participation in studies using correlated genotype and phenotype data [6]. This concept of "genetic privacy" emerged over the past few decades as a consequence of developments in genetics and information technology, much of which included the “invisible” part of heredity at the molecular level, human

genes. Before, we only had access to information such as family health history, pedigree information, and physical features. Currently, rapid advances in genetic sequencing allow for fast, affordable, and ubiquitous whole-genome sequencing. These rapid advances allow us to accumulate massive quantities of data that can be used to establish informatics links among millions of human genome sequences and extensive phenotype analyses. These data can then be used to generate and test hypotheses concerning human health and the human genome. The caveat is that these data can lead to identification of the individuals whose DNA sequences they contain.

The first idea that needs to be parsed out, is the difference between access to genetic information, and the misuse of that information. The right to “"genetic privacy"” is difficult to justify on the theory that secrecy of the genotypes has intrinsic value. After all, 99.9% of the human genome is the same across the population [2]. Therefore, the key in figuring out how to conduct ethical studies with individual participants’ genomes, is to recognize that the issue at hand is not that the genomes should never be disclosed or used, but that the genomic data should not be misused, and that any possible situation that may arise for that individual’s genomic data should be disclosed before valid consent can be obtained.

Secondly, this new model rests on the belief that "genetic privacy" is just one instance of privacy, and does not adhere to the idea of genetic exceptionalism, which is the belief that genetic information is special and must therefore be treated differently from other type of medical information [3]. The fundamental basis for this model is the idea of “open consent.” This is proposed as a way to ensure that the study design and consent protocol that abandons some parts of traditional confidentiality, also preserves

trust. The moral goal is to “obtain valid consent by effectuating veracity as a precondition for valid consent and effectuating voluntariness through strict eligibility criteria, as a precondition for substantial informed consent” [6]. In essence, veracity is the primary priority, and autonomy is secondary to that. Although in medicine, physicians may withhold information based on “therapeutic privilege,” there is no such concept in research. When obtaining informed consent from participants in research, disclosing information that is incomplete or distorted could undermine overall trust in research and in science. Therefore, truthfulness and veracity are the primary priorities within the “open-consent” model.

This “open-consent” model implies that research participants should be informed of, and accept certain pieces of information. They accept that their data could be included in an open-access public database, and that there are no guarantees regarding anonymity, privacy, and confidentiality [6]. They also accept that participation involves a certain risk of harm to themselves and their relatives, and that they may not benefit in any tangible way [6]. They must also accept that compliance with monitoring of their well-being through quarterly questionnaires is required, and that they can withdraw from the study at any time [6]. Lastly, they must accept that though they may withdraw, complete removal of data that have been available in the public domain may not be possible [6]. Once these criteria have been met, the individual has given valid, informed consent, and ethical research may be conducted using genomes.

## Conclusion

We know that current genetic advancements have challenged the traditional normative framework for medical information confidentiality as well as for biomedical research. It is clear that the previous interpretations of privacy and confidentiality are no longer as comprehensive as before, and thus new standards are necessary in order to maintain ethical rigor when engaging with a patient's genetic information. As discussed, there have been federal laws enacted in order to address "genetic privacy" issues in a more progressive manner. However, there has not been a nationwide standardization of what "genetic privacy" consists of, and what should be regulated by law. "Genetic privacy" still very much varies by state.

Additionally, there have not been any specific provisions made for genetic research in which genetic samples are used for different contexts and purposes than those for which they have been collected. This raises serious questions about the validity of the consent that is being obtained by the researchers from the participants. The burden of proof should lie with the researchers and not with the participants. The Personal Genome Project headed by Harvard Medical School has proposed a new "open-consent" model to deal with the changing concept of consent and "genetic privacy". It opts for openness in its scientific design and for veracity as the leading principle in obtaining consent.

With each passing year, standards and definitions for "genetic privacy" are continually updated and improved upon. The "open-consent" model is just one way to address gaps in the ""genetic privacy"" policies and guidelines necessary for medicine

and genomics. Further interventions and models are required in order to eventually create a truly comprehensive model for “"genetic privacy"”.

Works Cited

1. "About the Law." *About the Law*. N.p., n.d. Web. 08 June 2015. <http://www.hhs.gov/healthcare/rights/>.
2. "Federal Policy Recommendations Including HIPAA." *Federal Policy Recommendations Including HIPAA*. N.p., n.d. Web. 08 June 2015. <http://www.genome.gov/11510216>.
3. "Genetic Exceptionalism." *Wikipedia*. Wikimedia Foundation, n.d. Web. 08 June 2015. <http://en.wikipedia.org/wiki/Genetic_exceptionalism>.
4. "Genetic Information Discrimination." *Genetic Discrimination*. N.p., n.d. Web. 08 June 2015. <http://www.eeoc.gov/laws/types/genetic.cfm>.
5. "Genetic Privacy Laws." *National Conference of State Legislatures*. N.p., Mar. 2008. Web. <http://www.ncsl.org/research/health/genetic-privacy-laws.aspx>.
6. Lunshof, Jeanite E., Ruth Chadwick, Daniel B. Vorhaus, and George M. Church. "From Genetic Privacy to Open Consent." *Nature* 9 (2008): 406-11. Print.
7. "Privacy Act of 1974." *Wikipedia*. Wikimedia Foundation, n.d. Web. 08 June 2015. <http://en.wikipedia.org/wiki/Privacy_Act_of_1974>.