Genetic Privacy in the United States

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As genetic testing becomes less expensive and more efficient, it will become more and more common to test and gather personal genetic information. These advances hold great potential for preventing, diagnosing, and treating conditions. However, these advances also come with great implications for genetic privacy. There are current laws in place such as GINA and HIPPA to protect private genetic and health information, but there are still gaps in protections. This paper will give a brief overview of the history and arguments surrounding genetic privacy laws. Then it will focus on three privacy topics: life insurance, newborn screenings, and direct to consumer genetic tests.

History of Genetic Privacy Laws:

Over time, the laws protecting individual's genetic information have improved. The first major law was the Federal Privacy Act of 1974, which states that federal agencies cannot release personal information without the written consent of the individual[1]. Exceptions include information for the Census Bureau and Bureau of Labor Statistics[1]. A downfall of this law is that provisions are only applicable to federal agencies, limiting the scope of this law's impact[2].

A second major law in the history of genetic privacy is HIPAA, or Health Insurance Portability and Accountability Act, of 1996. It protects individual health information, and in 2003, the Privacy Rule, under HIPAA, ruled that genetic information is considered health information[3]. HIPPA does not treat genetic information as exceptional. Through HIPAA, individuals in group insurances cannot be discriminated against, charged more for premiums, or dropped for pre-existing condition due to genetic information[3]. HIPAA, however, does not apply to individual insurers, it does not protect against genetic test requirements, it does not specify that insurers are the only ones that can access the information (employers could theoretically access it), and it does not prohibit increasing group fees because of a group member’s genetic information[2,3]. Moreover, if an individual has been on the group plan for less than 12 months, they can still be discriminated against due to their genetic information[3].

The third major law is GINA, or Genetic Information Non-Discrimination Act of 2008, which states that personal genetic information cannot be used by insurance companies or employers to discriminate against individuals. This includes firing, hiring, promoting, denying coverage, raising premiums, and requiring genetic testing[4]. However, GINA does not cover life, disability, or long term care insurance. It also does not protect individuals with diseases that have already manifested, such that insurers could use the information of a woman who has breast cancer and a BRCA1 mutation since she has already been diagnosed[4, 5]. Nor does GINA apply to employers with fewer than 15 employees or to the U.S. military, the TRICARE military health system, the Indian Health Service, the Veterans Health Administration, or the Federal Employees Health Benefits Program[5].

The ACA does close some of the gaps of GINA. For example, in preventing discrimination for individuals with any pre-existing condition (of which genetic test results are a part of), individuals with manifest diseases, or diseases already diagnosed, are protected[6]. However, parts of the ACA may conflict with GINA. GINA prohibits the use of family history in programs, but the ACA Wellness Programs would require such family history. The Wellness Program is meant to prevent illness in individuals and promote health and wellness. Part of this includes conducting a family history in order to address possible risk factors. To do this, employers would have to request family history, which GINA forbids[6]. There have not been any problems so far, but this raises issues on the potential conflict of public health and privacy.

Current Genetic Privacy State Laws:

Despite laws such as HIPAA and GINA, states vary in their protections. Only 19 states have specific penalties for genetic privacy violations, 1 state (Alaska) declares DNA samples as private property, 5 states (Alaska, Colorado, Florida, Georgia, Louisiana) define genetic information as private property, 27 states require consent to release genetic information, 8 states require consent to get genetic information, 7 require consent to obtain access to genetic information, 12 require consent to perform genetic tests, and 4 require personal access to genetic information. No state has protections in all 8 categories, and Alaska is the closest to having all except requiring personal access to genetic information[7]. With such variation, it could be important to have a federal law that can supersede any existing gaps in state coverage.

Arguments against Genetic Privacy Laws:

Opponents of genetic privacy laws point to the potential negative impact on research and family prevention.

After the Privacy Rule of HIPAA, Armstrong et al discovered that there were fewer research subjects, because participants now had to complete a written consent form via mail. This extra step was a barrier. Of the individuals that returned the consent form, they were mostly "older, were more likely to be married, and had lower mortality rates at 6 months [or were healthier]"[8]. Authors argue that in limiting the pool of participants, this can lead to selection bias, which may confound results on genetic research. Additionally, the costs for "complying with the HIPAA Privacy Rule were $8704.50 for the first year and $4558.50 annually thereafter"[8].

Aside from the potential impacts on research and research groups, genetic information could be used to save people's lives. For example, there are clear links in genetic mutations to certain conditions. Mutations in MLH1 and MSH2 are linked to colon cancer, and information on one’s mutation could promote preventative measures, such as colonoscopies[9]. Discovering that one has such mutations however has implications for the parents and other family members. Opponents of privacy laws reason that privacy will reduce prevention and health promotion in close relatives. Should policies mandate patients to inform family members about their genetic information, because they could be affected? What are the rights of the family members to or not to know their potential risk? There is not one single right answer, but these are all important questions to keep in mind.

Arguments for Genetic Privacy Laws:

The major argument for genetic privacy laws is protecting individuals (and possibly their families) from potential discrimination. Individuals may also not want to participate in research studies without some sort of privacy protections, leading to an even larger issue of selection bias.

There have been mixed results as to whether individuals will or will not participate in research studies if there are no privacy laws. For some participants, the benefits of potential advances in knowledge may outweigh the risks, while for others privacy may be a pressing issue. Researchers have found that in one group, individuals were more concerned about the impact of learning genetic information rather than a breach of privacy[10]. Nevertheless, in other research groups, “participants often express anxiety and concern about privacy aspects…fearing they will be ostracized [by their family members]….[or endure] discrimination by insurance companies”[11].

Individual fear is not unreasonable, for there have been documented cases of genetic discrimination. As highlighted in future sections of this paper, genetic information has been sold for profit or in exchange for goods. Burlington Northern Santa Fe Railroad tested employees for genetic conditions without the consent of their employees, with hopes of determine which employee would not develop carpal tunnel syndrome[12]. A boy with Fragile X syndrome was dropped from his insurance company, and a social worker lost her job after her employer found out her mother died from Huntington's[12]. Privacy laws help prevent such discrimination.

Proponents of genetic privacy laws also argue that genetic information releases information not just on the individual but of the individual's family[13]. This raises questions of privacy laws protecting not just one but many people that are important to consider.

Genetic Privacy and Life Insurance:

GINA does not protect genetic privacy with life insurance. This leaves individuals with genetic predispositions vulnerable to higher costs for life insurance, if they are even covered. Proponents for privacy laws in life insurance argue the difficulty in applying genetic information, while opponents point to how insurance functions.

Insurance is based on a model of pooling individuals and risk sharing. In these pools, there are individuals with higher and lower risks for using medical care. If there is about an even number of low and high risk individuals, the lower risk individuals will help buffer the cost of the higher risk individuals, making it possible to insure such a diverse pool. When a pool consists of solely higher risk individuals, not only do the lower risk individuals drop out of the pool, but the costs for both the consumer and insurance company rise. Prices become inaccessible for most, and different companies may even stop offering life insurance[14]. The entire insurance model breaks down. In knowing one's predisposition, individuals with the APOE4 allele associated with Alzheimer's, for example, may want to suddenly purchase life insurance, leading to a selection of high risk individuals. This is the worst fear of opponents of genetic privacy laws in life insurance. In keeping genetic information from insurers, there may also be an asymmetry of knowledge, with individuals knowing more about their health than insurance companies[14]. Insurance companies can thus not accurately assess individual risk.

On the other hand, proponents of the individual's right to retain genetic privacy when applying for life insurance call to the ambiguity of determining risk based on genetics. For genetic conditions like Huntington's, where a specific mutation leads to the condition, there is a clear link between the mutation and the outcome. Yet, most conditions are multifactorial. There are genetic pre-dispositions, but there is no guarantee that the individual will develop the condition. This has led to a proposition that only genetic conditions with high penetrance, like Huntington's, be actively taken into account. Another potential solution, inspired by England's current system, is to offer everyone a basic level of life insurance. If individuals want further features, then they would be required to take a genetic test, in which only highly penetrable diseases would be taken into consideration[15].

Currently, in the U.S., states vary widely in their approach. Vermont make it illegal to use genetic results, North Carolina make it illegal to make decisions based on certain conditions like sickle-cell, New York requires informed consent, and Wisconsin says that when individuals sign, insurance companies automatically take on any risk[15].

Policy Implications:

Since there is a lot of variation among states, having a federal stance could help clear up discrepancies in policies. Which state has the better policy is up for debate and will not be contested in this paper. The only policy recommending is that in creating a federal policy, it is important to consider both the perspective of the insurance company and of the individual consumers to make a policy that does not break down the insurance model but also protects individuals from unfair genetic discrimination. Other tactics in different countries, such as England, could serve as inspiration for future polices.

Genetic Privacy and DTC:

DTC, or direct to consumer, testing companies pose compelling privacy issues. Since they are not currently regulated by government entities, DTC companies create their own privacy protections, which vary company to company. Most do not protect consumers from third parties, and some have had histories of serious breaches in privacy.

DTC genetic testing is important to analyze because of the amount of people it can reach with influential genetic information. DTC companies can provide patients with genetic predispositions, other genetic information, such as ability to metabolize alcohol and caffeine, and ancestry information, without requiring consultation with medical professionals[16]. Thus, individuals without health insurance now have access to this potentially important information for prevention and treatment, as do lower income individuals as it becomes more affordable.

Yet, despite these potential benefits, consumers are not protected against surreptitious testing. Surreptitious testing involves taking the “take home” tests and sending off DNA that is not of the sender but of another individual[13, 17]. DNA samples left anywhere (such as a cup) could be theoretically sent to analyze without the consent of the individual. It is particularly alarming if used in the context of parents analyzing child DNA for purposes such as paternity. Currently, there are no federal regulations preventing against surreptitious testing, and laws greatly vary among states. The only state with explicit measures is Alaska. Alaska “prohibits any person from collecting a DNA sample, performing a DNA analysis, or disclosing the result of such analysis without the consent of the person tested, and the law defines DNA analysis as testing to determine the presence or absence of genetic characteristics in an individual”[18]. And only Alaska, Georgia, Colorado, and Florida declare that DNA is individual property, leaving consumers in other states vulnerable to surreptitious testing[18]. DTC tests are a perfect gateway to surreptitious testing because of its structure.

DTC companies are also not subject to any existing federal laws like HIPAA. They self-monitor, which has its downfalls. For example, 23andMe sent the wrong genetic information to 96 individuals[17]. There were no federal or state policies preventing this or holding 23andMe accountable to ensure it does not happen again. Also, it is unclear what will happen to the information if the company goes out of business[17]. There are no clear expectations, leaving uncertainties in how companies will handle private genetic information.

Continuing further, DTC companies can give genetic information to third parties, and it is not always clearly communicated to the consumer; it can be un-consensual. 23andMe has partnerships with the Swiss firm Mondobiotech and the Parkinson's Institute, and Navigenics is conducting studies with the Mayo Clinic and Scripps Institute[17]. These collaborations are meant to advance genetic research and knowledge, and most of the time consumers are asked for consent to release their genetic information. However, in the fine print, 23andMe states that "even by refusing to participate, we may still use your Genetic and/or Self-Reported Information for R&D purposes…which may include disclosure…to third-party non-profit and/or commercial research partners who will not publish that information in a peer reviewed scientific journal"[17]. Since these statements are deeply hidden in fine print, 23andMe does not make it explicit or accessible for consumers to easily understand and consent to these statements. Consent is important, because even if the findings are not published in a peer reviewed scientific journal, 23andMe could theoretically give any individual data received (even if one changes one's mind) to any third party, such as family members or employers, for any use.

Policy Implications:

Since DTC companies vary in what services they offer, it would be difficult to have one broad policy for all DTC tests. Yet, the serious gaps in surreptitious testing, third party access and privacy breaches must be addressed, especially since DTC companies are starting to become more popular. Alaska's existing policy against surreptitious testing could be used as a starting point for a more standardized state and/or federal policy making such testing illegal. Companies need to make information on third party access clearer on consent and information forms, and a policy must at the very least penalize companies for sharing the wrong information, even if it is accidental. Policies must act preventatively not reactively.

Genetic Privacy and Newborn Screenings:

In the U.S, as in other countries, newborn babies are tested for different genetic conditions. An important screening is for phenylketonuria, a deficiency in the enzyme phenylalanine hydroxylase. If a newborn baby is diagnosed with phenylketonuria, they must automatically be put on a strict diet to prevent against life threatening complications[19]. Newborn screenings are very important for the wellbeing of our newborns. However, as with other genetic tests, there are potential privacy concerns.

There is a potential lack of consent. Genetic Alliance discovered that about "62% of mothers were not given any information on the newborn screening, were not given enough information to make a proper decision, or did not remember being told about it either way"[20]. It is important to make sure that all mothers completely understand the implications of the screening and consent to it, just as with DTC and third party information.

Another issue is the uncertainty in what happens to the blood card after the initial screening. In Texas, journalists discovered that the state was trading the newborn blood cards to private companies (which were under no regulation or privacy laws) for laboratory material[20]. Some of the blood cards could have been used to conduct research, but some could have been used for other purposes. There is no way of knowing, and this is part of the problem. In not having clear expectations and limits, it leaves room for possibilities to abuse consumer information, such as selling it for profit.

Policy Implications:

The Council for Responsible Genetics recommends an opt-in model of consent for newborn screenings, rather than an opt-out model where babies are automatically tested[21]. By opting in, mothers will hopefully be more aware of the implications of newborn screenings. Another potential policy is to only keep the cards for a limited number of times so that the data cannot be misused in the future. This is similar to the “use it or lose it” proposal, where genetic information is only used for its intended purposes, and then it is discarded[22].

Conclusion:

Throughout history, there has been an increase in genetic privacy protections, but there are still gaps in life insurance, newborn screenings, and DTC testing, to name a few. There is not a single solution for the complexities of genetic privacy, but there are tangible actions that can be taken. First, there could be more explicit protections against surreptitious testing, as in Alaska. Second, there could be clearer rules as to what are the roles and power of DTC companies and newborn screenings. Along with this are clearer implications for what happens if entities do not following these rules. Third, informational documents could make what can and will be done with the genetic information clearer and more accessible to ensure complete consent. Moving forward, it will be important to consider the perspectives of insurance and private companies as well as the right of consumers to privacy and protection. It will also be important to consider if there is a need to standardize genetic policy protections to cover all Americans, across all states. As technological and scientific advances revolutionize the field of genetics, its impact will be felt from genetic testing to genetic privacy laws.

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