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Redefining Genetic Discrimination:
The Ethical Future of Applied Genetics in Medicine

The Oxford English Dictionary offers two distinct definitions of "discrimination." First, discrimination is "[t]he unjust or prejudicial treatment of different categories of people or things" (OED 2014). Under this definition, a future of genetic discrimination within the medical field and larger scientific community sounds threatening, harkening back to the birth of eugenics and the horrific genetic cleansing agendas and "scientific experiments" of the Nazis in the Second World War. However, genetic discrimination may not be inherently evil or negative in nature. The OED secondly defines discrimination as "[r]ecognition and understanding of the difference between one thing and another" (OED 2014) Rothstein and Anderlik drew from both of these definitions, which they respectively called the "civil rights" and "actuarial" definitions, to form a hybrid meaning of their own: discrimination as "drawing a distinction among individuals or groups plus an element of either irrationality or social unacceptability or both" (Rothstein 2001). In this paper, I will explore the future of ethics of applied genetics in medicine in order to answer the following question: What could a future of genetic discrimination look like? The answer will largely depend on which definition of discrimination the medical and scientific communities decide to uphold and how the law protects privacy and other rights regarding people's genetic information.

There are two major applications of genetic information in medicine: preventive health and therapeutics. Diagnostics, too, represents a grey area that falls somewhere in between these two medical instances of applied genetics. It seems that most of the debate about use and abuse of genetic information centers around its preventive health applications, in terms of equal treatment with regards to insurance costs and employment opportunities. In contrast, for example, few would argue against the use of genetic testing to design individualized gene-based therapies to cancers or immunological diseases; one of the only issues raised in opposition to this approach is sometimes the high costs of designing such treatments and the disparities in who might be able to afford them (Kalish 2002).

According to the philosopher John Rawls, society and the legal system ought to strive for "fair equality of opportunity;" in other words, we should design social institutions so that any inequalities work to the benefit of the least advantaged members of society (Rawls 1971). Under such a justice system, any person ought to have an equal chance of employment in an applications process or enrolment with a health insurance company, regardless of what disease predispositions their genes may encode. Following Rawls' logic, genetic screening should not be allowed in employment or enrolment processes to determine ability to work or premium cost.

Health care insurers, however, would disagree with the Rawlsian approach to what is and is not fair. Insurance works by pooling risk, and therefore insurance companies employ risk classification by such characteristics as age, individual and family health history, health status, occupation, and even specific biological and behavioral markers of health such as serum cholesterol or alcohol and tobacco use (Peters). When our health care system is structured and approached as a capitalistic venture or a gambling game of chance and weighed risks rather than a basic human right, of course people are right to be worried about whether or not their genetic predispositions for certain diseases might mean that they are subjected to higher insurance premiums. Privacy protections for genetic information, therefore, are of the utmost importance to patients considering whether or not to get tested for various genetic conditions, especially if such conditions may run in their family or could be passed along to the next generation. Concerns about privacy, therefore, join the long list of critical issues, like the impact of a Huntington's disease or breast cancer likelihood test result on their mental health, that individuals must consider before opting into genetic testing (Willing 2001).

In an interview with Diane Horn on KCMU 90.3 FM-Radio in 1998, engineering technology and public policy expert Phil Bereano said, "The dangers of genetic testing depend not upon science, but upon law and public policy" (Horn 1998). While I would fundamentally disagree with his wording – science is, itself, highly politicized and cannot be ignored in this equation – I do understand his underlying message that there are legal protections required to ensure that genetic information is not abused to endanger the rights of any person. What, then, are the existing laws and public policies in the United States that are in place to protect individuals against these "dangers of genetic testing"?

At the time of Bereano's interview, state laws varied with regards to the degree of protections against genetic discrimination that were offered to state residents. As a result of this variability, some unfortunate cases of rights violations did occur because of inadequate protections. For example, African Americans in the US Air Force used to be screened for sickle cell trait (the presence of a single gene, not the pair of genes that actually causes sickle cell disease) in order to determine eligibility for Air Force pilot training programs (Horn 1998). The Air Force claimed to be worried that changing oxygen levels in flight could trigger the disease onset even in those who were merely carriers, which is simply not a medically viable concern (Horn 1998). Other cases of genetic discrimination in the face of insufficient legal protection include numerous examples of employers secretly testing their employees for certain genetic predispositions without asking consent or even alerting them about doing so. Specifically, black individuals were tested for sickle cell disease, while Latinos were tested for hypertension, presumably due to relative rates of such conditions in these racial and ethnic groups (Horn 1998). This example parallels gender discrimination, since these same employers were also found to be testing women to see whether they were pregnant as well (Horn 1998). On that note, some have argued that separating out different kinds of discrimination such as specifically *genetic* discrimination rather than general health status-based discrimination, citing that not only is it difficult to define "genetic" and infeasible to detach genetic information from other health information, but distinct treatment increases the stigma attached to genetic conditions in the first place, lending "legitimacy to genetic reductionism and determinism" (Rothstein 2001). Despite such arguments, public policy and research have continued to focus on genetic discrimination, and by 1996, the Council for Responsible Genetics had identified over 200 cases (Brant-Rauf 2004).

Not until the Genetic Information Nondiscrimination Act (GINA) of 2008 did the federal government intervene, establishing a minimum standard of protection that must be met in all states; its terms do not weaken any stronger protections provided by specific states' laws (NIH 2014). Under GINA, unfair treatment on the basis of differences in one's DNA that may affect their health is considered illegal, effectively preventing employers or health insurers from discriminating against any individual on genetic grounds (NIH 2014). However, GINA does not cover life insurance, disability insurance, or long-term case insurance, nor do its protections apply to persons in the military, veterans, or Native Americans receiving care from the Veterans Health Administration or Indian Health Service (GHR 2014). Additionally, employers with fewer than 15 employees are not required to abide by GINA either (GHR 2014). Clearly, some weaknesses and loopholes remain in the current policy that may be financially harmful and, indirectly, lead to unequal health coverage for certain individuals.

Over the years, studies have been conducted to assess public opinion on genetic testing and genetics in general. Of course, as research Celeste Condit pointed out in her own research on this issue, “[b]ecause the public is a collective and, therefore, heterogenous and abstract concept, its thoughts are difficult to identify” (Condit 2001). Between 1960 to 1995, portrayal of genetics in US mass-media magazine articles was overwhelmingly “mostly positive,” with only about 10 percent portrayals being “mostly negative” and another 20 percent or so presenting balanced opinions (Condit 2001). It should be noted that these mostly positive portrayals of genetics significantly preceded GINA. Condit’s conclusions were that by 2001, the American public was not “acting in hasty ignorance in response to the rapid development of genetic technologies” but, rather, displayed a “cautious optimism” and tentative support for medical applications of genetics like DNA testing to diagnose or anticipate inheritable diseases (Condit 2001). After the Act, by 2011, research revealed that Americans tended to support scientific research on genetics, as well as legal protections against genetic discrimination for United States citizens (Aimeling 2013). One study revealed that out of a nationally representative sample of over two thousand American adults, a majority believed the federal government should increase spending on genetic research. Another salient majority opinion was that medical professionals ought to be involved in explaining the results of genetic tests rather than other genetic testing options such as direct-to-consumer companies (Aimeling 2013). Another study examining deterrence from participating in genetic research found that 60 percent of participants would provide their DNA to a biobank if given the opportunity, even though 90 percent expressed privacy concerns and 37 percent worried the study data generated could be used against them in some capacity (Kaufman 2009).

As for specific applications of genetics in medicine, a study by Bates et al in 2005 found significant public disapproval of “designer babies,” or the selection of preferred traits in one’s offspring (Bates 2005). The same study noted a strong approval for manipulation of genetics via medical interventions in order to correct for disease, however (Bates 2005). Awareness of the potential for social stigmatization was the most common reason participants gave for the distinction in their opinion between these two applications of genetics in medicine (Bates 2005). Interestingly, some minority groups such as African Americans within this study recognized that looking at race seriously through genetic research could bring advantages, despite substantial concerns about racism in medicine and science in general; inclusion of African Americans in research protocols would, participants noted, result in better medical treatments for their community (Bates 2005). The results of this study confirmed previous findings from past research publications and suggest an overall positive public perspective on medically applied genetics, with a healthy dose of wariness regarding the possibility of negative genetic discrimination.

In conclusion, it seems a shame that the mainstream media and the scientific and medical communities have already loaded the term "genetic discrimination" with negative and dangerous connotations. Personalized medical treatments (like genetic tests to determine drug sensitivity, gene therapies, and stem cell therapies) have the potential to benefit cancer patients, immunodeficient patients, and many others, and all *because* they are genetically discriminatory in nature – or in other words, because they were designed by differentiating between individual treatment options based on the patient's particular genotype (Kalish 2002). Negative instances of genetic discrimination can also certainly occur, but they are mostly confined to genetic testing related to preventative medicine and health, rather than therapeutics and treatment of disease. With careful design and enforcement of legal protections, the negative aspects of genetic discrimination threatening their use in preventive health care could be re-spun in a more positive light; certainly, the risk reduction and cost-beneficial realities surrounding genetic testing and counseling are promising enough to bear further examination into how to protect patients' privacy and equal rights while also promoting patients' and parents' future children's health. Meanwhile, ongoing research continues to identify more and more ways to use genetic information to affect positive change in the therapeutic approaches to disease. It may be time to reflect on the construct of genetic discrimination in medicine and to redefine this term to embrace the great potential of personalized treatment based on genetically perceived distinctions.

Resources Cited

Aimeling, Rene, and Shana Kushner Gadarian. "Public opinion on policy issues in genetics and genomics." *Genetics in Medicine* (2014) 16, 491-494. 7 November 2013. <http://www.nature.com/gim/journal/v16/n6/abs/gim2013175a.html>.

Bates, B. R. et al. "Warranted concerns, warranted outlooks: a focus group study of public understandings of genetic research." *Social Science & Medicine* 60 (2005) 331-344. <http://www1.chapman.edu/~cosgr100/bevan/publications/9research.pdf>.

Brant-Rauf, P. W., and S. I. Brant-Rauf. "Genetic testing in the workplace: Ethical, legal, and social implications." *Annual Review of Public Health* 25: 139-153. 2004.

Condit, Celeste. "What is 'public opinion' about genetics?" *Genetics: Nature Reviews | Perspectives.* Volume 2, 811-815. October 2001. <http://www.nature.com/nrg/journal/v2/n10/pdf/nrg1001-811a.pdf>.

GHR (author unknown). "What is genetic discrimination?" Genetics Home Reference: Your Guide to Understanding Genetic Conditions. June 2, 2014. <http://ghr.nlm.nih.gov/handbook/testing/discrimination>.

Horn, Diane. "Genetic Discrimination: A Primer." Interivew with Phil Bereano on KCMU 90.3 FM-Radio (Seattle, Washington, USA) January 19 1998. Council for Responsible Genetics. <http://www.councilforresponsiblegenetics.org/ViewPage.aspx?pageId=98>.

Kalish, Douglas I. "Discrimination is Good." TCS: Tech. August 6, 2002. <http://www.techcentralstation.com/1051/techwrapper.jsp?PID=1051-250&CID=1051-080602A>.

Kaufman, David J., et al. "Public Opinion about the Importance of Privacy in Biobank Research." *American Journal of Human Genetics* Volume 85, Issue 5, Pages 643-654. 13 November 2009. <http://www.sciencedirect.com/science/article/pii/S0002929709004571>.

NIH (author unknown). "Genetic Information Nondiscrimination Act of 2008." National Human Genome Research Institute, National Institutes of Health. January 27, 2014. <http://www.genome.gov/10002328>.

OED (author unknown). "Discrimination." Oxford Dictionaries. Oxford University Press. 2014. <http://www.oxforddictionaries.com/us/definition/american_english/discrimination>.

Peters, Ted. "Genetic Discrimination." Counterbalance: New Views on Complex Issues. <http://www.counterbalance.org/genetics/discrim-frame.html>.

Rawls, John. A Theory of Justice. Belknap Press of Harvard University Press, 1971, 1999. Print.

Rothstein, Mark A., and Mary R. Anderlik. "What is genetic discrimination, and when and how can it be prevented?" *Genetics in Medicine*, September/October 2001, Vol. 3, No. 5, pp. 354-358.

Willing, Richard. "Gene tests bring agonizing choices." *USA Today*. April 23, 2001. <http://biochem118.stanford.edu/Papers/Ethical%20Issues/Gene%20tests.pdf>.