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A Case for Genetic Testing: Issues, Controversy, and Unintended Consequences

Introduction

The United States has a dubious history in regard to racial injustice towards people deemed inferior or “different.” From the unfortunate times of slavery to the Civil Rights Movement to the present time, discrimination has been an issue that only a minority of individuals has faced. However, the notion of discrimination has a new connotation ever since 2003 when the Human Genome Project was completed. The goal of the HGP was to sequence the entire human genome in order to have a genetic map that could unlock the basis for genetic diseases and disorders, to develop faster, more efficient methods for DNA sequencing and sequence analysis, and transfer this information to the private sector.ⁱ One unintended consequence of the HGP is that it actually challenged previously held doctrines of genetic differences among the races since genetic variation between individuals was found to be only 0.1%. Thus, people have over 99% of the same genetic configuration, with about 20,000-25,000 genes in human DNA. This information proves critical in correctly diagnosing and treating patients with previously misunderstood conditions and ailments.

Some practical uses of the HGP are the development of genetic tests to screen for genes implicated in the progression of genetic diseases and disorders. Genetic disorders are caused by mutations, such as the erroneous addition, deletion or substitution of base pairs in DNA sequences. These mutations can occur spontaneously or be inherited from parents in the form of chromosomes, which contain genes made up of DNA that guides cells to create proteins that are necessary for normal function.ⁱⁱ As a result of these mutations, the body may synthesize defective proteins or not create them all together, thus leading to medical conditions. The problem of using genetic testing lies in employers and insurance companies using these genetic tests to discriminate against

people with less desirable genes that might incur a significant cost if hired or given an insurance plan. Other ethical issues arise with the use of genetic screening, such as privacy of genetic information, and the implementation of laws to regulate and keep pace with the improvement of medical technology.

In this paper, the use of genetics will be examined as both a harmful method in the construction of social and political views that promoted racial inequalities, and a beneficial medical innovation in the fight against disease. By accounting for historical transgressions in the development of new technologies and legislation to prevent further medical calamities, genetic testing can allay the potential fears and ethical concerns of the public while aiding in the treatment and prevention of genetic conditions. Thus, the HGP and other medical research advances have the unique opportunity to get rid of these biological misconceptions, thereby curing the social and political ills of injustice and exploitation which appeared to be based on faulty biology.ⁱⁱⁱ Ultimately, knowledge eventually overcomes prejudice, but the delay may be long unless active steps are taken to implement the improvements in knowledge, (Race Science and Society) and that is why furthering the use of genetic testing and research is pertinent and necessary.

The historical significance of genetics

Before the concept of the HGP was even conceived, medical researchers used race as a legitimate biological construct in order to justify unethical medical experimentation and promote “differences” as the result of perceived genetic inferiority. Below are just a few examples of these medical calamities:

“A notoriously syphilis-soaked race”

The 1932 Tuskegee Experiment is perhaps the most notorious experimental abuse of medical research towards African-Americans and undoubtedly transformed African-American perceptions of the health-care system. The U.S. Public Health Service designed this experiment with the ultimate goal of studying the progression of syphilis in black men. The PHS promised free medical treatment to six hundred sharecroppers in Macon County, Alabama, but lied to the men about this treatment and waited until the subject’s death in order to perform an autopsy. The erroneous but widely held belief at

that time was that syphilis did the worst damage to the neurological systems and brains of whites, while wreaking its worst havoc on the cardiovascular system of blacks, sparing their primitive and “underdeveloped” brains.^{iv} This belief was rooted in the specific racial dimorphism of syphilis that conferred differing outcomes to this disease based on genetic makeup. Thus, the Tuskegee Experiment is an important marker of the use of genetic differences to justify medical research and reinforce the social and political injustices at that time. It was not until concrete medical advances like the HGP that people began to change their notions of genetic inferiority, effectively proving the benefit of science in helping counteract unethical and baseless racial distinctions.

“A black disease”

In the late 1960s, employers and the federal government started instituting genetic screening for Sickle Cell Anemia in order to protect employees by avoiding their placement in work environments that could trigger illness such as a sickle cell crisis (Medical Apartheid). The primary targets of this testing were African-Americans because they demonstrated an increase risk of SCA since it struck one out of every four hundred African-American newborns at that time (Medical Apartheid). However, with this genetic testing came an abuse of power due to widespread discrimination towards African-Americans. For example, the U.S. Air Force Academy issued a directive barring admission of all black sickle cell *carriers* as a result of four black men dying in basic training in 1968 and 1969. An autopsy confirmed that the men’s red blood cells were sickle-shaped, but there was disagreement as to whether the sickling was the cause of death or a result of it. Sickle-cell disease is recessive, meaning that a person needs two copies of the gene to develop the disease. However, medical testing did not account for the recessive properties of SCA and thus any individual who possessed the gene was assumed to be genuinely ill with SCA, even if they were only a sickle-cell carrier. This blatant genetic discrimination prompted the passage of the National Sickle Cell Anemia Control Act in 1972, which sought to end this unfair treatment of African-Americans. With improvements in genetic testing, the public became aware that genes implicated in disease offer a *potential risk* of developing a condition, not a definite sign that a person will actually acquire the disease.

These examples serve to illustrate the power that scientific knowledge has in overcoming ignorance and injustice. The researchers at that time believed that differences in skin color meant dramatic differences in genetic makeup, but what they did not know was that most genetic variation is present between people of the same cultural background, not among races. However, due to these past historical relationships with medical research, the general public still fears genetic discrimination with the abuse of medical information.

Faces of discrimination

Jacob

Jacob, a boy who carries a gene for a disorder called Long QT Syndrome (LTQS), was denied coverage under his father's health insurance policy because of his "pre-existing condition." LTQS is a rare and little-known genetic disorder that sometimes triggers sudden cardiac death. Those who carry the gene may be healthy until they suffer an attack without warning, but carriers can control their risk of cardiac arrest with preventive beta-blocker therapy. Jacob's father wanted Jacob to be insured, but even after their state enacted a law prohibiting genetic discrimination, Jacob's insurance company still refused to cover him. After fighting the insurance company for a year and a half, Jacob's family finally won and got Jacob the health insurance he needed.^v

Many people have similar stories to Jacob, and as a result public fear of the misuse of information has increased according to a 2004 study by the Genetics and Public Policy Center at Johns Hopkins University which found that 92 percent of Americans oppose allowing employers access to their genetic information and 80 percent oppose allowing health insurers access (Faces of Genetic Discrimination). The public's attitude toward genetic testing is not improving as a result of the stance of health insurers, who, according to The American Council of Life Insurance, advocate the use of information about genetic predispositions to assign risk categories in medical underwriting and possibly refuse to insure those individuals who carry those genes.^{vi} Employers have a keen interest in the health of their employees due to economic pressures of reducing labor costs to increase profits and productivity, and thus want to use genetic testing as a way to screen employees in order to reduce absenteeism, and decrease life and health insurance costs.

What employers and insurers do not comprehend is the negative consequences of these actions that can negate the promises of genomic research because of fears of

discrimination. People will be less willing to advance scientific discovery by not participating in research trials or getting genetic tests to assess their risk of familial diseases. This consequence will undercut the profits of employers and insurers because the public will not be able to seek preventive treatment for his or her ailments, waiting until the condition is symptomatic and thus drawing on insurance to cover the incurred costs of not knowing their personal health risk for pre-existing conditions. Hence, it should be in the best interests of employers and insurers to promote genetic testing without actively seeking the knowledge of individual employee risks because treating a chronically ill employee is more financially damaging than providing treatment for preventive care. Simply, it makes good business sense.

Less Reason to Fear

To allay public fears in response to the growing use of genetic testing and research, the Genetic Information Nondiscrimination Act of 2007 (GINA) was:

...created to protect individuals from discrimination in health insurance and employment on the basis of genetic information. Establishing these protections will allay concerns about the potential for discrimination and encourage individuals to participate in genetic research and to take advantage of genetic testing and new therapies. The legislation will provide substantive protections to those individuals who may suffer from actual genetic discrimination now and in the future. These steps are essential to fulfilling the promise of the human genome project and improving the health and longevity of the American people.^{vii}

With the completion of the HGP, the Ethical, Legal, and Social Implications Committee (ELSI) was created to develop guidelines to assist federal and state agencies with preventing genetic discrimination from health insurers.^{viii} The ELSI recommended that health insurers be prohibited from using genetic information or an individual's request for genetic services to deny or limit health insurance coverage, establish differential rates or have access to an individual's genetic information without that individual's written authorization (Genetic Discrimination in Health Insurance).

With scientific advancement comes great responsibility in protecting freedoms not fathomed before. Thus, it is important to have legislation keep pace with a rapidly changing scientific world in which new innovations are possible, leaving room for increased misuse of this technology. Hence, legislation is pertinent to address public concern and account for the historical significance of genetic research in order to learn

from past medical calamities in order to build trust and faith with the public so that they are willing to actively participate and take full advantage of these discoveries. The benefits of genetic testing and research should outweigh the potentially negative ethical, social, and political aspects of this technology, but only if due consideration is given to establishing parameters that allow for responsible use of this technology and the information gained from these advances.

The Benefits of Genetic Testing: BRCA1 and BRCA2

Genetic tests are used to determine individual risk for developing a medical condition because if a person has a gene mutation, his or her chances of developing a disease are increased. Thus, knowing the status of your genetic makeup could prove useful as a pre-emptive measure to prevent a certain condition from manifesting itself. For example, mutations in certain genes make women more susceptible to developing breast and other types of cancer. These genes, BRCA1 and BRCA2, confer a risk of cancer three to seven times higher than a person who has normal, unaltered genes.^{ix} Hence, knowing the genetic risk of having these genes can allow women to seek preventive care and explore other medical treatments that can help delay or thwart the development of breast cancer. Some therapeutic options include:

- **Surveillance:** Carefully monitoring symptoms of cancer to catch the early stages of the disease. This can take the form of mammography's or a clinical breast exam.
- **Prophylactic Surgery:** Women can remove much of the at-risk tissue, such as breast tissue, fallopian tubes and ovaries, in order to reduce the risk of developing cancer.
- **Risk Avoidance:** This method focuses on behaviors that decrease the risk of cancer such as exercising and decreased alcohol and cigarette consumption.
- **Chemoprevention:** This strategy focuses on the use of natural or synthetic substances to reduce the risk of developing cancer, or reducing the chance that cancer will come back. (taken from Genetic Testing for BRCA1 and BRCA2: It's your choice)

As illustrated, genetic testing allows people to make an informed decision about their futures by affording them the knowledge of their genetic predispositions. This knowledge has wider implications, such as privacy issues, but with the GINA of 2007, people should feel secure with participating in genetic research and thus benefit from technologies truly aimed at helping alleviate impediments to good health. In addition, genetic tests can confirm a diagnosis so that a primary physician can develop an effective treatment plan. But most importantly, genetic tests can include testing of, i) prenatal fetuses as a means to determine whether a fetus is at increased risk of genetic disorders or physical deformities; ii) newborns for genetic diseases, and iii) potential parents to determine whether they carry genes for a disease that could be passed on to future children (Mayo Clinic).

Limitations of genetic testing

Previously it was not feasible to identify people at risk for genetic conditions before they became ill with a disorder. Now, researchers can conduct genetic tests to ascertain the genes associated with certain diseases, but it is important to realize that even the best medical advances have limitations which detract slightly from their effectiveness.

Sometimes genetic tests are unsuccessful at detecting mutations for certain diseases. This is due to the fact that some diseases have multiple genetic alterations that are capable of causing symptomatic conditions, and thus a genetic test may not be able to identify all the mutations because the causes of some genetic diseases and disorders are still unknown. In addition, a positive test result does not guarantee that a person will actually develop that disease because tests can only tell a person a statistical probability of developing a disease. One main reason for this is because some diseases need several mutations to occur in order to cause any clinical problems. Hence, having one gene alteration does not automatically mean that a patient will develop a disease.

Furthermore, mutations in genes can be spontaneous and the results of environmental factors like smoking or exposure to chemical substances. Thus, a negative result from a genetic test does not equate to a decreased risk of developing disease. Another limitation of genetic testing is that a positive result does not provide information about the severity of disease. For example, cystic fibrosis can have drastic symptoms that

affect an individual's quality of life such as chronic lung infections, or the disease can have manageable symptoms that can be alleviated by medication, diet, or exercise. Finally, genetic testing might be a privilege of the wealthy since these tests can cost up to a few thousand dollars, effectively preventing people of lower socio-economic status (SES) and the uninsured from accessing this technology. With improved efficacy of genetic testing, some of these limitations may be overcome. Thus, it is imperative to recognize that no medical innovation will be perfect, but this fact should not dictate the potential benefits and uses of these technologies unless the treatment is considerably risky and unreliable.

Genetic Testing and Implications for Doctors

Several studies have indicated that racial and ethnic minorities tend to receive a lower quality of healthcare than non-minorities, even when access-related factors, such as patient's insurance status and income, are controlled.^x This may be the result of some researchers speculating that biologically based racial differences in clinical presentation or response to treatment may justify racial differences in the type and intensity of care provided (Unequal Treatment).

Genetic testing presents yet another method that doctors must familiarize themselves with in order to better serve patients. It is important that they understand the wider implications of genetic testing and how this prevention strategy is viewed by the patient, who may not want to learn his or her associated risk of certain diseases due to fear of discrimination. Thus, physicians need to provide genetic counselors to assist patients in accurately interpreting their test and work with clinicians in developing an appropriate treatment plan that serves to decrease the likelihood of the patient falling victim to genetic disorders as a result of these mutated genes. Taking into account the limitations of genetic testing, clinicians need to disclose the risks associated with alternative prevention strategies. But most importantly, physicians and the healthcare system need to make these tests more widely available and affordable in order to decrease health disparities because genetic tests can be expensive, and serving minority communities who have an increased risk of being underinsured could make this treatment

option class specific because a person's SES would dictate whether they would receive the test and afford the ensuing treatment if alterations in their genetic makeup is found.

Biologically, a race is a result of the process by which a population becomes adapted to its environment. The particular array of traits which come to be the most frequent, and hence to characterize the group, are probably those which now or at some past time proved to be successful in a particular environment (Race Science and Society). Race is simply not a fixed or static category but a dynamic one. Thus, physicians need to pay less attention to the physical manifestations of these genetic differences and look at the underlying basis of disease in order to eliminate the risk of allowing stereotypes to adversely alter the effectiveness of the patient-clinician encounter and the inequalities that result. Genetic testing provides clinicians the opportunity to truly ascertain the minimal role perceived genetic "differences" play in the outcome of care and thus inadvertently disprove any previously held notion of biological inferiority as attributed to race. Improving scientific knowledge about the way genetics shapes and influences our health can decrease clinician's reliance on stereotypes while enhancing clinical outcomes by allowing genes to solely dictate the effectiveness or type of treatment given. This is because having genetic tests will give the physician a genetic road map of his or her patient and thus tailor medication dosage and alternative therapies to personally fulfill the patients needs and maximize effectiveness.

Conclusion

Time will only tell whether we truly have learned from our past history in order to face a growing problem that will eventually affect every person because sooner or later, discrimination will be generalized as scientific advances are better able to identify specific genes of chronic diseases, which a minority of people will have for any given genetic condition. This may have the unintended consequence of making discrimination salient to all people, and thus has the potential to change their views on judging people off superficial qualities and allowing these judgments to hinder the development of personal relationships.

Nevertheless, many genetic traits are mediated by genes which lie completely outside of the individual's control. Thus, it would be unjust to penalize or judge people

with certain genes that were given to them at conception because this would further exacerbate the problem of healthcare inequalities and discrimination. Genetic research and testing has a unique chance to positively impact patient's lives by providing them with information about genes that predispose them to disease, and eventually, finding a cure in those genes. The only way to achieve this is to remain dedicated in funding research that advances our knowledge of the underlying mechanisms that cause genetic diseases and disorders, and accounting for the ethical and social concerns with legislation that minimizes or eliminates the potential abuses of power. Simply, genetic testing is worth the investment.

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