ARE OUR GENES UP TO THE TEST?

STEFAN VITOROVIC

Genomics, Bioinformatics, and Medicine
Dr. Doug Brutlag
May 19, 2005

Introduction

The sequencing of some three billion chemical base pairs comprising the entire human genetic code by the Human Genome Project is complete. Now, the question arises: "What exactly do the 3,000,000,000 A's, T's, G's, and C's mean?" The manifold applications of this enormous wealth of information are some of the central topics concerning researchers in fields ranging from anthropology to behavioral genetics to pharmacogenomics. A number of the pre-conceived notions in modern society have been challenged by the findings: humans and chimpanzees are found to be 99% genetically identical; humans have *only* 20-25,000 encoding genes; the genetic variation between any two random individuals is *only* about 0.1%. Genetic sequencing and analysis have changed the way scientists think about what it means to be human and what makes people different from as well as surprisingly similar to each other.

The recent breakthroughs in genomic research have enabled scientists to identify the genetic basis for human diseases, ushering in a new era of individualized prevention strategies and "genetically-tailored" medicine. Genetic testing can already determine an individual's predisposition for several cancers, heart disease, diabetes, and Huntington's disease; one day, a simple screening procedure could give people a summary of their genotypes in a matter of minutes (Washington Post). These advances hold much promise for improving human health; however, genetic research comes with manifold non-medical consequences. A plethora of social, ethical, and legal questions arise, including the privacy of a person's genetic information, the legality of employers and insurers screening workers and policy holders for predispositions to diseases, and the necessity of laws and government regulations to prevent genetic discrimination.

If employers and insurance agencies could have access to the results of someone's genetic screening, would the companies actually be able to use a person's genetic heritage against them? Prominent scientists and governmental agencies, including the NIH and the Department of Health and Human Services, have recognized the dangers that come with knowing an individual's genetic information. Genetic sequencing procedures could be used unfairly by employers to screen individuals for particular traits, including intelligence or genetic diseases, resulting in the denial of employment or benefits. Insurance companies could administer genetic tests to people before they are approved for coverage. If the results show that someone has a predisposition for skin cancer, for example, the company could charge them higher rates or deny them coverage even if they are in good health and show no sign of developing cancer. Additionally, the allele frequencies of certain genetic disorders are different between specific ethnic or racial groups, which could result in discrimination towards these groups by employers and health care providers (Genetic Information and the Workplace). The genetic information, with its legal, social, and ethical consequences, seems to be even more complex than scientists could have ever imagined.

Genetic discrimination is unfair to workers and their families. It is unjustified – among other reasons, because it involves little more than medical speculation. A genetic predisposition toward cancer or heart disease does not mean the condition will develop. To deny employment or insurance to a healthy person based only on a predisposition violates our country's belief in equal treatment and individual merit.

- ***President George W. Bush***

Real People – Real Discrimination

Timothy

Timothy was genetically screened to find out is he had an increased susceptibility to any of the known genetic diseases. He learned that he had a single mutation for Gaucher's disease, which made him a carrier. Since the disease is recessive, having the single mutated allele meant that he would never develop Gaucher's disease *himself, but that he might pass on the mutation to his children. When Timothy revealed this information on a job application, he was denied the position solely on the basis of his genetic mutation, which had no influence on how qualified he was for the job.*

Mary

Mary is fifty-three years of age. When applying for a position at an insurance company, she revealed that she had the genetic allele for hemochromatosis; however, she did not have any of the symptoms of the disease and was in perfect health at the time of the applicant interview. During the second interview for the job, she was told that the company wanted to hire her but that it would not provide insurance coverage because of her genetic screening results. Mary agreed to these conditions of employment. In the final interview, the interviewer told her that the company thought she was well-qualified for the job, but they were unable to hire her because she revealed the genetic disease.

Although the majority of people are aware of discrimination based on age, gender, ethnicity, or sexual preference, many people do not realize the prevalence of genetic discrimination today. There are numerous well-documented cases of health insurers and employers using a person's genetic information against them. A survey of U.S. firms in 2001 by the American Management Association found that some employers are gaining access to the private genetic information of employees for a number of reasons: one percent of all genetic testing towards screening for sickle cell anemia, 14 percent towards screening for workplace susceptibility, and 20 percent towards collecting family medical histories of employees (Hudstead). The survey did not include questions about how the companies used the genetic information; however, the AMA did acknowledge the information gained by the employers was enough to discriminate in various employment decisions, such as hiring, firing, and promoting. The increased access to employee's genetic information and the possibility of requiring genetic screenings at the workplace can only serve to increase the public's fear of genetic testing and the misuse of their private medical information.

Genetic discrimination is not something to be regarded as a "threat of the future." Already, there are thousands of health insurers and employers who conduct genetic tests to determine whether an individual should be hired or if they are eligible for health insurance coverage. A 1996 survey of 917 individuals at risk of developing a genetic condition and parents of children with specific genetic diseases identified more than 200 instances of genetic discrimination; in other words, almost 1 out of every 4 people with a "high risk" of a genetic disease felt that they were treated unfairly just because of their genetic heritage. These cases involved discrimination by health insurance providers, employers, and other organizations that use genetic data. A survey of genetic counselors, primary care physicians, and patients in 1998 found 550 people who had been denied either insurance coverage or employment because of a genetic predisposition for a particular disease (Genetic Information and the Workplace).

One of the most well-known and widespread examples of genetic discrimination occurred in the early 1970s to identify African Americans who carried a gene mutation for sickle cell anemia. The employers used genetic screenings to target African-American employees, since they were known to be at an increased risk for the disease; many of these genetic tests were conducted without the consent of the individuals. Applicants who carried the mutation for the disease were denied jobs in the company even if they were asymptomatic and might not ever develop the disease. Congress passed the National Sickle Cell Anemia Control Act in 1972 to prevent this kind of genetic discrimination in the workplace. Nevertheless, this kind of blatant employer discrimination towards individuals susceptible to disease has left a very negative image of genetic screening and even the entire HGP (Genetic Information and the Workplace).

Past Policy Initiatives

With a new gene being discovered every few days, the wealth of genetic sequencing data has become staggering. In a recent position paper on genetic discrimination, the Council for Responsible Genetics (2001) stated that "genetic information is being generated much more quickly than our legal and social service systems can respond." According to the Council, current national and state measures to prevent genetic discrimination and to protect people's genetic privacy are "inadequate." The National Human Genome Research Institute (NHGRI) has also recognized the potential for misuse of genetic information. Thirteen years before the human genome was even sequenced, the head of the HGP and co-discoverer of DNA, James Watson, decided that at least 3 percent of the entire HGP budget should be devoted to ethics of genomics. To deal with such issues, the Ethical, Legal, and Social Implications (ELSI) Working Group was formed in 1990 as a central part of the HGP. The program supports ethics and policy research ventures, workshops, and conferences to address the pressing questions that come with genetic information. The Policy and Program Analysis Branch studies and tracks both national and state policies that have an impact on the NHGRI, enabling them to develop legislative proposals.

In 1995, the ELSI Working Group and National Action Plan on Breast Cancer (NAPBC) cosponsored workshops on genetic discrimination in health insurance and the workplace. The findings and recommendations of the workshops, which were published in *Science* (Hudson, 391), have served as the basic foundation for policy-making in the Executive branch and for laws enacted in state and national legislatures. The recommendations focused on two forms of genetic discrimination (employment and insurance) and are summarized below:

Employment Recommendations

- Employment organizations should be prohibited from using genetic information to affect the hiring of an individual or to affect the terms, conditions, privileges, benefits or termination of employment unless the employment organization can prove this information is job related and consistent with business necessity.

- Employment organizations should be prohibited from requesting or requiring collection or disclosure of genetic information prior to a conditional offer of employment, and under all other circumstances, employment organizations should be prohibited from requesting or requiring collection or disclosure of genetic information unless the employment organization can prove this information is job related and consistent with business necessity, or otherwise mandated by law.

- Written and informed consent should be required for each request, collection or disclosure.

- Employment organizations should be restricted from access to genetic information contained in medical records released by individuals as a condition of employment, in claims filed for reimbursement of health care costs, and other sources.

- Employment organizations should be prohibited from releasing genetic information without prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure will be made.

- Violators of these provisions should be subject to strong enforcement mechanisms, including a private right of action.

Insurance Recommendations

- Insurance providers should be prohibited from using genetic information, or an individual's request for any genetic services, to deny or limit any coverage or establish eligibility, continuation, enrollment, or contribution requirements.

- Insurance providers should be prohibited from establishing differential rates or premium payments based on genetic information, or an individual's request for genetic services.

- Insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information. Insurance providers and other holders of genetic information should be prohibited from releasing genetic information without prior written authorization of the individual. Written authorization should be required for each disclosure and include to whom the disclosure would be made.

These recommendations have been endorsed by many different organizations, including the American Psychiatric Association, the American Medical Association, the American Cancer Society, and the American Workrights Institute. The American Cancer Society, for example, is in strong favor of genetic research, but it fears that the potential to misuse personal genetic information is very “dramatic and chilling.” Along with the ELSI Working Group’s recommendations, they advise Congress to provide monetary support and authorization for studies on the ethical, legal, and social issues related to genetic testing. Additionally, the American Cancer Society feels that insurers should be required to meet the patient care costs for individuals participating in approved clinical trials on genetic research (ACS Genetic Testing). Having insurance companies compensate people for participating in genetic testing studies will encourage more people to participate in such studies and will speed the discovery of new methods and applications of the research (Faces of Genetic Discrimination).

The European Union has also created a task force that seeks to address the multitudinous issues surrounding genetic information, especially its use in the workplace and in health insurance policies. The European Society for Human Genetics (ESHG) was created to address such issues from a multinational perspective. In their 2001 report, *Genetic*

Information and Testing in Insurance and Employment, the ESHG states that the national legislatures in the EU are making progress in enacting laws that prevent the misuse of genetic information. They stress the importance of setting specific definitions for genetic testing, genetic information, and genetic discrimination so as to avoid any confusion. The ESHG also feels the need for more medical, epidemiological, and psychosocial research to better understand exactly what the genetic information reveals. Additionally, the group feels that genetic information that has no bearing on one's *current* ability to perform a job should not be used in an employment decisions. They suggest that third-party, independent testing agency provide genetic monitoring only for professions that expose people to harmful substances (e.g. chemicals, radiation), and, even then, the information should be accessible only by the employee. Such recommendations, along with the broad guidelines of the ELSI Working Group and American Cancer Society, will help persuade legislators to ensure the privacy of people's genetic code by passing specific regulations and, consequently, help regain the general public's trust of genetic testing.

Existing Legal Protections

There is no federal legislation that specifically and comprehensively deals with the misuse of genetic information in the employment and insurance. Incidentally, the only laws that can be applied to prevent such infringements are those that deal with other forms of discrimination. Because of the lack of definitive federal laws against workplace and insurance discrimination, individual states have enacted their own laws to deal with this problem; however, current state laws differ significantly in the coverage and protections afforded by the legislation as well as the means of enforcement. Many organizations feel that there is a pressing need for specific federal laws that ensure genetic information will be used to improve the health of citizens and not to discriminate against them.

The 1996 Health Insurance and Portability and Accountability Act (HIPAA) is the only federal law that addresses genetic discrimination. It prevents insurance companies from disclosing someone's private genetic information or from changing one's entire policy as a result of genetic information. The act, however, does not deal with the issue of health insurers requiring genetic screening or the use of genetic information in the workplace in general. The main source of prevention against genetic discrimination in the workplace is the 1990 Americans with Disabilities Act (ADA), which is enforced by the Equal Employment Opportunities Office (EEOC). Although the act does not specifically address genetic discrimination by employers, it does protect against disability-related discrimination. Under the act, people with diagnosed genetic diseases would have the same protection as individuals with other disabilities; nevertheless, carriers of genetic mutations for disease or individuals with asymptomatic genetic diseases are not protected under the clear disability-based discrimination prohibition of the law.

In response to the plethora of committee and government agency recommendations, the Genetic Information Non-Discrimination Act of 2003 was passed after years of negotiations by the unanimous vote of 95-0 in the Senate on October 14, 2003. The act explicitly "prohibits genetic discrimination in health insurance and employment." Health insurers are prohibited from denying coverage or charging higher premiums "based solely a genetic predisposition for developing a disease in the future" and employers are barred from using genetic information as a factor when making hiring, firing, placement, or promotion decisions (Statement of Administration Policy). It will be the first national legislation that focuses solely preventing genetic discrimination, particularly by employers and health insurance companies. This act, if passed by the House of Representatives, will set a precedent not only in America but for the entire world; it will show that the U.S. government is committed to protecting individuals' genetic information, allowing society to reap the benefits of genetic testing without having to live in fear.

Future Policy

John

John is an eighteen-year-old with a high probability of developing Huntington's disease because of one of his parents carries the genetic allele. He wanted to enlist with the US Marines in order to serve in the Persian Gulf War; however, he thought that the knowledge of his risk for Huntington's would disqualify him for military service even though it was unlikely that he would any of the disease symptoms during his time of duty. As a result, he answered "no" on a military questionnaire inquiring about his hereditary disorders, and he did not list Huntington's disease as part of his family medical history. John had to serve in constant fear of having his genetic condition revealed to the Marines and the other soldiers. He feared the possibility of being discriminated against by his fellow soldiers or, even worse, being discharged from military service.

We are on the verge of a true revolution in medicine. But there is a chance it will not happen as we hope. It will not be a failure of the science. There is increasing evidence people ***fear*** *their genetic information will be used to deny them health insurance or a job. This* ***fear*** *is keeping them from seeking medical help. If people believe that a new system of individualized medicine will lead to denial of health insurance or other benefits, they will not take advantage of what the new system could offer. The revolution at hand may not be realized because people are* ***afraid*** *to take part in it.*

- Dr. J. Craig Venter, President and Chief Scientific Officer
Celera Genomics

Although the potential benefits of genetic research are relatively obvious to the scientific community, the general public is not nearly as supportive of the "genomic revolution." The potential threat of genetic discrimination by insurers and employers is one of the main reasons that some people fear genetic information even though they would like to reap the medical benefits. A Time Magazine and CNN poll in June, 2000 shows the "conflict" of interests that come with sequencing the human genome: "46 percent of those surveyed responded that they thought the Human Genome Project, when completed, would likely be generally harmful, but 61 percent said that if they could gain information about their genetic code or DNA, they would want to know what diseases they were predisposed to get" (Potential Benefit and Risks). Numerous studies suggest that this fear of discrimination can keep individuals at high risk from getting a genetic sequence analysis and participating in clinical trials that could save their lives. A 2003 study of 470 people at Johns Hopkins Hospital had nearly half of the participants rate their fear of genetic discrimination as high. Those individuals were also less likely to get a genetic test or discuss genetic information with their physician. A 2000 survey of 296 cancer geneticists showed a similar concern even within members of the medical community. Sixty-eight percent of the geneticists said that they would not bill their insurance company for the cost of testing procedures that showed them to have a predisposition for a particular disease because of a fear of genetic discrimination (Faces of Genetic Discrimination). Evidence even suggests that this fear of discrimination is increasing (Appendix). Since the majority of genomic research in publicly funded through agencies such as the NIH, it is vital that the public be made aware of the benefits of genetic sequencing and, even more importantly, the efforts being made by numerous committees, agencies, and the legislature to prevent the misuse of such information so that people do not fear genetic testing.

To deal with the public's misinformation about genetics and their fear of genetic screening, a national public education campaign should be launched. The effort should be coordinated by the NIH, which will focus on informing citizens of all ages, areas, and socioeconomic standings of the benefits of genetic testing and the government's protection of their genetic privacy rights. A large portion of the campaign should be centered on early education at the middle and high school level. When students learn about the HGP or genetic testing in their classes, they should be made aware of the medical benefits of such testing, such as its potential to save millions of lives. To reduce their chances of fearing genetic discrimination, they should know that the government has erected adequate safeguards. To inform adults and senior citizens, the NIH should sponsor community lectures about genetic screening. Additionally, the government should require individual employers and insurance companies to make certain that their employees and policy-holders understand their genetic information will have no bearing on any of the companies' decisions. This will make them feel safe in their workplace and with their health service provider.

The second component of this education campaign is a nation-wide, publicly-funded advocacy initiative. A coordinated effort by the National Advocacy Council and the NIH should focus on removing the misconceptions and fears of genetic screening. The campaign should target numerous sources of media, including television, radio, information pamphlets, and posters, seeking to quiet the myths about genetic information and inform the populace of legislative efforts to protect them from genetic discrimination. Along with the public education campaign, the national advocacy efforts will help remove the stigma from genetic testing and regain the public's confidence in genetic research.

Along with national public education about genetic research, there should be an international organization that discusses the ethical, legal, social, and economic issues associated with genetic information, especially as it applies to the workplace and health insurance. The World Health Organization should hold an annual international conference that invites all the nations concerned about the implications of genetic research. People from various professions, including lawyers, physicians, ethicists, politicians, insurance representatives, and other citizens should be encouraged to discuss their concerns and present their policy recommendations. Since the issue of genetic discrimination has become truly international in scope, with numerous countries already assembling national DNA databases, the issues should also be addressed in an international forum. This exchange of ideas will reveal what policies have been successful in preventing genetic discrimination by employers and insurance providers; it could influence other nations who have not yet considered the consequences of genetic research into enacting legislation that prevents problems before they arise.

The framework of the policy should be centered on the policy recommendations for genetic discrimination and employment given by the ELSI Working Group. Genetic information should not be used by health insurers to deny or limit coverage or even to set premium rates. Additionally, the health care providers should be prohibited from requesting the collection of genetic material or from releasing genetic information without expressed written consent of the individual. The recommendations for employment discrimination should also be followed. The use of genetic information should be prohibited from any employment decisions, such as hiring and job benefits, and the companies should be not be able to request the collection of any genetic information. Like insurance providers, employers should not have any access to genetic information in medical records and must obtain written consent before releasing any such information. Any violation of the provisions should be strictly punishable by the law.

Some would argue that such strict legislation could actually be detrimental to the people that it is seeking to protect. They fear that preventing an employer from conducting genetic screening and monitoring in jobs that expose workers to potentially harmful substances, such as radiation or strong chemicals, could keep them employees from noticing any genetic damage or mutations that result from the workplace conditions. While federal policy should take such specific cases into account, they should still prohibit the employee from disclosing their genetic information with the company. Instead, the employee can choose to be genetically monitored by an independent testing organization, which would reveal the results only to the individual and not their employer. Thus, worker safety does not need to be compromised in order to prevent workplace discrimination.

Research in the 21st Century has opened the door to a host of new biological and medical technologies that have the potential for improving quality of life. Advances in the field of biotechnology, in particular, have enabled researchers to better understand human diseases, allowing them to focus on strategies for disease diagnosis, treatment, and even prevention. Biotechnology cannot realize this potential unless proper precautions are taken to prevent the exploitation of genetic information. The widespread ignorance and misconceptions about genetic screening serve as a hindrance to the advances in medicine and only further fuel the fear of discrimination. It is imperative that the general public acquire a better understanding of the benefits of genetic screening and of the legislation to prevent genetic discrimination. There is a need not only for specific national legislation to prohibit the misuse of genetic information, but also for international efforts that address the issues and ensure biological advances do not compromise the well-being of America and the rest of humanity.

[Genetic discrimination] can slow the pace of scientific discovery that will yield crucial medical advances... Without protections in place, individuals who do participate will represent a self-selected group that could skew research results, producing a negative impact on all of us who look to genetics to help find better ways of

diagnosing, treating and preventing disease…The longer this problem remains unresolved, the greater the damage that will be done to U.S. science and medicine.

- James D. Watson, President
Cold Spring Harbor Laboratory, and
Dr. Francis S. Collins, Director
National Human Genome Research Institute
National Institutes of Health

Works Cited

Bush, George. *Presidential Radio Address to the Nation.* 23 June, 2001. A*vailable at* http://www.whitehouse.gov/news/releases/2001/06/20010623.html.

Collins, Francis & James Watson. "Genetic Discrimination: Time to Act." Science. 745. (2003). A*vailable at* http://www.geneticalliance.org/PDF/Colins_editorial.pdf.

Venter, Craig. *The Potential for Discrimination in Health Insurance Based on Predictive Genetic Tests: Hearing before the Subcomm. on Commerce, Trade and Consumer Protection of the House Comm. on Energy and Commerce*, July 11, 2001. *Available at* http://energycommerce.house.gov/107/Hearings/07112001hearings322/Venter520.htm.

Faces of Genetic Information. How Genetic Discrimination Affects Real People. The Coalition for Genetic Fairness. (2003). *Available at:* http://www.nationalpartnership.org/portals/p3/library/GeneticDiscrimination/FacesofGeneticDiscrimination.pdf

Heading Off Genetic Bias. Washington Post. 26 April 2004. *Available at* http://infoweb.newsbank.com/iw-search/we/InfoWeb?p_action=doc&p_docid=100D8EAA11F2A28E&p_docnum=13&p_nbid=F5BS59YNMTEwMDQwNzY4OS4zODYwNDI6MTo2OjEyOC4xMg

Hustead, Joanne. *Privacy Concerns Raised by the Collection and Use of Genetic Information by Employers and Insurers: Hearing before the Subcomm. on the Constitution of the House Comm. on the Judiciary. 2001 American Management Association Survey.* 12 September 2002.

Miller, Paul. *The Controversy over Genetic Discrimination.* (2000). A*vailable at* http://www.state.mn.us/ebranch/dhr/acc_disc_gen.html.

National Human Genome Research Institute. Ethical, Legal and Social Implications Program Reports. http://www.genome.gov/12010621

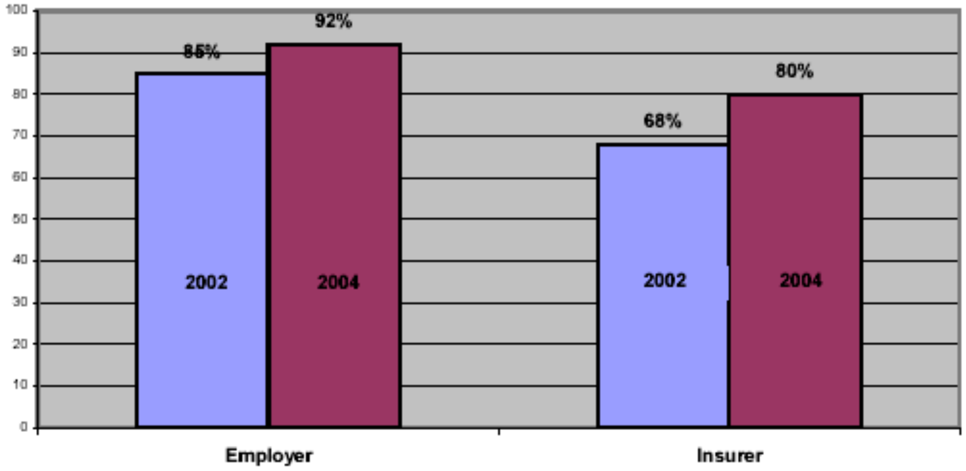
National Institute of General Medical Studies. *Report of the First Community Consultation on the Responsible Collection and Use of Samples for Genetic Research.* (2000). *Available at* http://www.nigms.nih.gov/news/reports/community_consultation.html#populations

Potential Benefits and Risks. Report of the First Community Consultation on the Responsible Collection and Use of Samples for Genetic Research. 26 September 2000. *Available at* http://www.nigms.nih.gov/news/reports/ community_consultation.html#populations

Rothenberg, Karen, et al. "Genetic Information and the Workplace: Legislative Approaches and Policy Challenges." Science. 275. (1997): 1755-57.

Statement of Administration Policy. Executive Office of the President. American Non-Descrimination Act of 2003.

Appendix



Americans' Growing Concerns about Access to Genetic Information

Percentage of Americans Who Say:
Employers / Insurers Should Not Have Access to My Genetic Information

Source: JHU Genetics & Public Policy Center Survey "Public Awareness and Attitude about Genetic Technologies" (2002, 2004)

The figure shows that there is a recent rise in Americans who feel that insurers and employers should not have access to their genetic information. In the two-year period between 2002 and 2004, there was a 7% rise in people who did not want employers to have information about their genes and a 12% rise in those who did not want insurers to have access to their genes.