

Editor's Note

In recent years the policy of the American Journal of Medical Genetics regarding Conference Reports or Symposia summaries has been highly selective. We chose to make this Symposium summary avail-

able to our readers because of the timeliness of the topic.

John C. Carey
Editor

Conference Report

Preempting Genetic Discrimination and Assaults on Privacy: Report of a Symposium

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At a symposium in June, 2002, biomedical researchers, clinicians, legal experts, policy-makers, and representatives of the insurance industry and the advocacy community gathered to address issues of genetic privacy and discrimination; and to identify research, legal, and policy gaps needing to be filled. They concluded that over the next decade, as more genetic information becomes available and the public becomes more aware of individual risks, concerns about privacy and discrimination will become increasingly important. Documented cases of genetic discrimination are rare and largely anecdotal, yet individuals with genetic conditions harbor significant fears about discrimination. Current laws enacted to protect individuals from workplace and insurance discrimination offer some measure of protection, but leave many unfilled gaps. Moreover, the use of genetic information in potentially discriminatory ways is not limited to employment and insurability. Existing laws do little to protect people seeking life, disability, or long-term care insurance. And the courts have used genetic informa-

tion in a wide variety of cases including paternity, criminal, and tort (personal injury) cases. Genetic information that might jeopardize an individual's right to privacy may also be obtained in the course of research studies, including through the collection of DNA and tissue samples. The insurance industry, State and Federal agencies, and the advocacy community are all making efforts to address some of these gaps through legislation and education of clinicians, the public, and policy makers.

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INTRODUCTION

With the announcement in February, 2001, that a first draft of the entire human genome sequence had been deciphered, scientists and clinicians were giddy at the possibilities for newer and better therapies as well as a deeper understanding of disease mechanisms. Even then, however, cautionary questions were being asked about the potential for misuse of the vast amount of information generated. At a symposium in June, 2002¹, biomedical researchers, clinicians, legal experts, policy-makers, and representatives of the insurance industry and the advocacy community gathered to address issues

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of genetic privacy and discrimination; and to identify research, legal, and policy gaps needing to be filled. They concluded that while at this time, concerns about genetic privacy and discrimination have not yet become a burning issue for the public at large, the landscape will change over the next decade as more genetic information becomes available and the public becomes more aware of individual risks. As the technology continues to speed forward, education of clinicians, the public, and policy makers will need to accelerate as well.

“MOONSHOT OF BIOLOGY”

The path from discovery of a gene to a clinical application can be long and sometimes arduous; and as a result, only a handful of genetic tests are currently available clinically. But the Human Genome Project (HGP) has resulted in a rash of gene discoveries, and clinics are bracing for a host of new tests to become available. The HGP has been likened to other big science, government-funded projects, such as that which sent a man to the moon and the Manhattan Project, because of the wide ranging technological implications and the associated ethical, legal, and social issues that must be addressed. Edward R.B. McCabe (Los Angeles), outlined how the HGP is changing the paradigms for both clinical medicine and research. In the clinical arena, genetic testing of asymptomatic individuals is already resulting in a shift from treatment of symptoms to predictive and preventive care; and from defining patients as those who are sick to those who are at risk. Meanwhile, the emerging science of genomics requires a reconsideration of our current understanding of the basic science at its foundation. It is not wrong, said McCabe, “It’s just so incomplete that it ends up not being terribly informative.”

When the HGP began, the scientists involved felt a keen obligation to address ethical, legal, and social implications at the same time that the research was being conducted, said Kathy Hudson (Baltimore, MD). As a result, the National Human Genome Research Institute (NHGRI) established the Ethical, Legal and Social Implications (ELSI) Program, and earmarked 5% of all NHGRI funds for this Program. In the past decade ELSI has funded research on a range of issues, including privacy and the fair use of genetic information. NHGRI also has a policy office that seeks to harvest fruits of ELSI research and translate these into policy recommendations, including those having to do with genetic discrimination in health insurance and employment, and with privacy of genetic information in the research setting, according to Hudson.

PERCEPTION OR REALITY?

While documented cases of genetic discrimination are rare, individuals with genetic illnesses nonetheless harbor significant fears of discrimination. And these fears, according to Kimberly Quaid (Indianapolis, IN) and Barbara Bernhardt (Baltimore, MD) affect individual’s decisions to seek medical care and participate in research. Quaid discussed her experience working with families affected with Huntington disease (HD), a de-

vastating, progressive, and ultimately fatal neurologic disease which is inherited in an autosomal dominant fashion and which has nearly 100% penetrance and no effective treatment. What this means is that the child of a person with HD has a 50% risk of inheriting the mutant gene that causes the disease; and those who do inherit the mutation are almost certain to later develop and die from the disease. Bernhardt discussed work with breast cancer patients who are considering genetic testing for the *BRCA1* and *BRCA2* breast cancer susceptibility genes. Unlike HD, breast cancer is a relatively common and genetically complex disease with incomplete penetrance, for which treatments are available.

Since the genetic mutation that causes HD was identified in 1993 and a test became available that would inform individuals whether they had inherited the gene, many clinicians have been surprised that only about 5% of at-risk individuals chose to be tested. According to Quaid, fears of genetic discrimination in the areas of employment and health insurance keep people from getting tested; and if they do seek testing they often do so anonymously. Similar concerns have been expressed by breast cancer patients. Bernhardt has documented the fact that patients harbor significant concerns that they, or their family members, will encounter discrimination from health insurers.

Evidence for discrimination is largely anecdotal, and is not limited to employment and insurance. In one highly publicized case [Verlinsky et al., 2002], a woman who carried a gene variant that made her highly susceptible to developing early Alzheimer disease, sought preimplantation screening of her embryos to ensure that she would not pass the gene on to her offspring. In an editorial accompanying the report of this procedure, two ethicists questioned the ethics of the procedure, when the woman was unlikely to “be able to care for or even recognize her child in a few years” [Towner and Loewy, 2002].

Quaid, equating the ethicists’ concerns to discrimination, noted that the woman was using the technology to ensure that she would not pass the disease on to her offspring. “Unlike the eugenics movement of the 20th century, genetic discrimination against individuals in the 21st century will be fueled not by inference but by actual knowledge of genotype,” she said. “The discrimination of the future [will be] an insidious one of policies and guidelines for fertility clinics, reimbursement decisions of insurance companies limiting access to technology, and employment decisions.”

HOLES IN THE SAFETY NET

Given that the risk of genetic discrimination is at least theoretically high, several speakers discussed the extent to which federal laws have succeeded in protecting privacy and curbing discrimination in the workplace and from health insurers, as well as in research settings and in the courts. Four federal laws were discussed: the American with Disabilities Act (ADA), the Health Insurance Portability and Accountability Act (HIPAA), the Occupational Safety and Health Act (OSHA), and

the Family and Medical Leave Act (FMLA). The conclusion was that while these laws offer some measure of protection, they leave many unfilled gaps.

Problems arise because genetic and other medical information may be collected by insurance companies and employees for legitimate reasons, yet may be used in unintended ways. Employers may collect genetic information about employees and their children, for example, through medical exams required for employment, claims submitted to employee-sponsored health plans, and employee requests for workers' compensation, sick leave, family or medical leave, and disability accommodations. The ADA, OSHA, and other laws that allow or require employers to collect this information were enacted primarily to protect employees from toxic substances in the workplace and from discrimination on the basis of disability. Protecting privacy was not a preeminent goal; and in fact, according to Joanne L. Husted (Washington, DC), privacy protection has often been overlooked. The laws generally regulate the use of information to matters for which the information was gathered, but do little to regulate access and disclosure.

HIPAA, for example, prohibits excluding an individual from a group health plan because of a past or present medical condition, and specifically states that a genetic predisposition is not a preexisting condition. Further, it restricts group health plans from disclosing information to employers, and requires consent for disclosure to others except in cases where there are legitimate public health concerns or for law enforcement or government oversight. Yet this law leaves many gaps. First, it applies only to group health plans and does nothing to protect the millions of people who are covered only by individual health insurance policies or those who are uninsured. Moreover, HIPAA regulates the group health plan but does not regulate employers, who may still require employees to take genetic tests and disclose information. While employers are prohibited from using that information in employment decisions, the enforcement mechanism in HIPAA is weak and, to date, largely untested.

Husted cited several cases in which medical/genetic information collected by employers was used in a discriminatory way against workers. Terri Sargent learned that she had Alpha-1-antitrypsin deficiency, a genetic disease from which her 37-year-old brother was dying. Fortunately for Terri, a preventive replacement therapy is available, and she began treatment at a cost of \$3,800 per month, which was covered by employer-provided health insurance. One month after the treatment began, despite the fact that she was healthy and had received excellent performance reviews over more than 3 years of employment, she was abruptly fired, losing not only her job but her health insurance as well. She took her case to the Equal Employment Opportunity Commission (EEOC), claiming that she was wrongfully fired and a victim of genetic discrimination in violation of the ADA. In November, 2000, the EEOC ruled in her favor [Goldman, 2001].

In another recent case, Burlington Northern Santa Fe Railway allegedly began to surreptitiously test employees for a rare genetic mutation possibly associated with

carpal tunnel syndrome. In a federal lawsuit, employees and their union contended that the secret testing violated the ADA and several state laws. The company apparently was searching for employees predisposed to carpal tunnel syndrome, presumably in order to avoid workers' compensation claims [EEOC, 2002]. The case was settled out of court, leaving the thorny legal issues unresolved, said Husted.

USE OF GENETIC INFORMATION IN THE COURTS

Genetic tests have also been ordered by courts for a wide range of cases outside of employment and insurability, including paternity, criminal, and tort (personal injury) cases. Diane Hoffman (Baltimore, MD) described numerous cases in which both defendant and plaintiffs have used genetic information to try to prove or disprove causation, in cases such as malpractice or toxic exposure. These cases are problematic, said Hoffman, because the courts lack clear guidelines about how to evaluate genetic information. For example, a genetic predisposition to a condition does not necessarily rule out the contribution of a toxic exposure to an injury. Genetic tests might also be used by a defendant to reduce damages by showing a plaintiff's shortened life expectancy.

In one child custody case, at a father's request, the court ordered a woman with a family history of HD to undergo genetic testing for the mutation, presumably to prove her unfitness as a mother. The woman fled the jurisdiction rather than submit to this test [Rothstein, 1994]. A family history of HD or early onset Alzheimer disease has also reportedly been used by adoption agencies to deny prospective parents' applications.

These requests for genetic information are permitted under Rule 35 of the Federal Rules of Civil Procedure, which allows court ordered mental or physical examinations when relevant to the allegations made in the lawsuit. Genetic testing can be included as part of a court-ordered examination. This intrusive form of discovery, said Hoffman, is used most often by defendants to disprove a plaintiff's claim of physical or emotional injury. Without some protections, she maintained that the law has the potential to cause litigants emotional or psychological harm, especially if the individual learns through the testing that he or she has a genetic disorder for which there is no cure or treatment. Yet, few guidelines exist for judges ruling in these cases.

UNEXPLORED TERRAIN

Beyond the courts, Hoffman noted several other areas in which lack of privacy protection might allow genetic test results to be used in unintended ways. Existing laws do little to protect people seeking life, disability, or long-term care insurance. Hoffman described one case in which a couple was denied admission into a continuing care retirement community because one of them tested positive for the ApoE4 allele, which is associated with a predisposition to Alzheimer disease [NHGRI, 1997]. Genetic testing conceivably could also be used by private schools making decisions regarding admissions, track-

ing, accommodations, and participation in athletic programs. Currently, there are no federal laws that prevent such discrimination in these settings; state laws are rare, and protection is spotty.

PRIVACY RISKS IN RESEARCH

Genetic information that might jeopardize an individual's right to privacy may also be obtained in the course of research studies. Federal regulations for the protection of human subjects were adopted in 1981, and since 1991, Institutional Review Boards (IRBs) have been charged with ensuring that researchers follow basic principles including informed consent. However, as described by Mary Kay Pelias (New Orleans, LA), emerging technologies in molecular genetics and bioinformatics have generated new issues related to human subjects, their families, and the use and disclosure of genetic information.

For example, genetics research may yield sensitive information about not only the individual directly involved in a research study, but also about family members [Botkin, 2001]. Whether these family members should also be considered "human subjects," and thus covered by human subjects protections and informed consent guidelines is now under review by the National Human Research Protections Advisory Committee's Working Group on Genetics.

Protection of privacy has also emerged as a serious concern in the collection of tissue and DNA specimens in clinical research. DNA and tissue may reveal information that individuals may not want to know or may not want others to know, said Ellen Wright Clayton (Nashville, TN). This places genetics researchers in a quandary: how to gain useful clinical information from the sample without jeopardizing the individual's right to privacy. Clayton reported a case that illustrates the problem. An inmate who volunteered to participate in genetics research was subsequently linked through his DNA to a crime other than the one for which he had been incarcerated.

Directly identified data holds the greatest risk of breach of confidentiality, either because of unintended access to the information by "hackers" or because of recontact of the subject by researchers. Stripping identifiers from the data or encrypting the data could protect individual privacy, but with a potential loss of relevant information. Clayton described a clinic in Marshfield, Wisconsin, that has established a DNA repository and associated data warehouse with numerous protections, including encryption of clinical data; a server that is not connected to the internet and thus not at risk of hacking; and a system of data control in which investigators must submit their queries to one of only a few "data miners" who have access to the full data set. In other words, the warehouse stores a great deal of information, but with very limited access to protect confidentiality.

CAN PUBLIC POLICY MEND THE HOLES?

On day two of the symposium, speakers addressed public policy issues from divergent viewpoints. Nancy

Wexler (New York, NY) questioned why our society has no-fault auto insurance and no-fault divorces, but health insurance and employment that are fault-based. She cited several cases of individuals pushed to extremes including murder and suicide in efforts to protect their ill family member's dignity and avoid inadequate care. The most heinous repercussion of the lack of adequate care and insurance coverage is on a person's self esteem, she said. She also noted that the Department of Defense considers a hereditary or genetic disease to be a pre-existing condition. No matter how long an individual has served in the armed forces, medical benefits can be stripped away after diagnosis of genetic disease, said Wexler.

Following Wexler, insurance executive John W. Rowe (Hartford, CT) used the symposium to announce a proposal to establish insurance industry-wide "practice guidelines" regarding access to genetic testing and interpretation of results. Rowe's company, Aetna, proposed that health plans cover the cost of genetic testing for individuals at risk of developing a genetic-based disease only when the results of the test may affect treatment. Testing of uninsured family members would also be covered when the test results might affect treatment of the insured. The insurance company would also cover genetic counseling and support physician education. Rowe noted that decisions on what will actually be covered are usually made by the employer (via decisions in the types and amounts of coverage to be provided as part of the policy), not the insurance company. In terms of use, Rowe proposed that insurers not base eligibility decisions on genetic testing or require testing as a pre-condition to obtaining health insurance.

Loopholes in HIPAA were addressed by Representative Louise Slaughter (Democrat, NY). In 1995, she introduced the first piece of legislation to ban genetic discrimination. That bill, HR602, would cover all health insurance programs and prohibit enrollment decisions and premium increases based on results of genetic tests. Slaughter said that in spite of the many supporters of the proposed legislation there had been no hearings on this bill in 6 years, primarily because of opposition from the health insurance industry.

Ann M. Carroll (New York, NY) described the public policy landscape for preclinical genetic testing in New York. Failure of Congress to address issues of genetic privacy and discrimination, she said, has forced states to take the lead. Over half of the states have enacted confidentiality protections that limit the disclosure of genetic information. A majority also prohibit discriminatory uses of genetic information by employers and health insurers. Only a few states, including New York, require pre-test informed consent.

In highlighting what is not covered by New York State law, Carroll identified issues that still need to be addressed not only in New York, but across the country. Protection is only mandated for predictive uses of genetic testing for asymptomatic individuals. Left unprotected are symptomatic individuals, or those who seek testing for reasons other than to determine future risks, such as pharmacogenetic testing performed to guide medical treatment by determining whether an individual has

inherited a gene variation that affects the probability of having a favorable or adverse response to a particular treatment. Confidentiality protections should be extended to cover all forms of genetic information, said Carroll.

The symposium identified many concerns, real and perceived, as well as gaps in our knowledge about genetic privacy and the risks of genetic discrimination. Yet, there are few research tools or systematically collected databases to address these gaps. Government and the private sector may need to make a more concerted investment of resources to develop our knowledge base and educate, as the public becomes more engaged and aware of the issues surrounding genetic privacy and discrimination.

SPEAKERS CITED

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