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Congress of the United States
House of Representatives
Washington, DC 20515

January 18, 1996

BAN DISCRIMINATION BASED ON GENETIC INFORMATION CO-SPONSOR THE STEARNS-KENNEDY BILL (H.R. 2690)

Dear Colleague:

As knowledge about the genetic basis of common disorders grows, so does the potential for discrimination. Please join us in co-sponsoring H.R. 2690, the Genetic Privacy and Nondiscrimination Act.

The discovery so far of over 150 mutant genes make it possible for doctors to peer into their patients' medical future. Today, at least 50 genetic test for hereditary disease are available; by the turn of the century, DNA test are almost certain to be a standard part of medical exams. From a single sample of a patient's blood, doctors will be able to spot genetic mutations that signal the approach not only of rare hereditary diseases, but also the common killers, including breast cancer, heart disease, and diabetes.

As the attached article indicates (over), scientists are continuing to discover genes responsible for different diseases. **Yet while the possibility of identifying disease characteristics through an individual's genetic makeup provides hope to many sufferers, the potential for discrimination is great.**

The Genetic Privacy Act is intended to prevent such discrimination. It provides the following safeguards: (1) prohibits the disclosure of genetic information by anyone without the specific written authorization of the individual; (2) prohibits employers from seeking to obtain or use genetic information of an employee or prospective employee in order to discriminate against that person; and (3) prohibits health insurers from using genetic information to reject, deny, limit, cancel, refuse to renew, increase rates, or otherwise affect health insurance.

To cosponsor H.R. 2690, please contact Suzy Glucksman with Rep. Joe Kennedy at X55111 or Vernoica Crowe with Rep. Stearns at X55744.

Sincerely,

Joseph P. Kennedy II

Cliff Stearns

104TH CONGRESS
1ST SESSION

H. R. 2690

To establish limitation with respect to the disclosure and use of genetic information, and for other purposes.

IN THE HOUSE OF REPRESENTATIVES

NOVEMBER 29, 1995

Mr. STEARNS introduced the following bill; which was referred to the Committee on Commerce, and in addition to the Committees on Government Reform and Oversight and Economic and Educational Opportunities, for a period to be subsequently determined by the Speaker, in each case for consideration of such provisions as fall within the jurisdiction of the committee concerned

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A BILL

To establish limitation with respect to the disclosure and use of genetic information, and for other purposes.

Be it enacted by the Senate and House of Representatives of the United States of America in Congress assembled,

SECTION 1. SHORT TITLE.

This Act may be cited as the "Genetic Privacy and Nondiscrimination Act of 1995".

SEC. 2. FINDINGS AND PURPOSES.

(a) FINDINGS.—Congress finds the following:

(1) The DNA molecule contains information about an individual's probable medical future.

(2) Genetic information is uniquely private and personal information that should not be disclosed without the authorization of the individual.

(3) The improper disclosure of genetic information can lead to significant harm to the individual, including stigmatization and discrimination in areas such as employment, education, health care and insurance.

(4) An analysis of an individual's DNA provides information not only about an individual, but also about the individual's parents, siblings and children.

(5) Current legal protections for genetic information, tissue samples and DNA samples are inadequate to protect genetic privacy, and require further attention.

(6) Laws for the collection, storage and use of identifiable DNA samples and private genetic information obtained from those samples are needed both to protect individual privacy and to permit legitimate genetic research.

(b) PURPOSES.—It is the purpose of this Act to—

(1) define the rights of individuals whose genetic information is disclosed;

(2) define the circumstances under which an individual's genetic information may be disclosed; and

(3) protect against discrimination by an insurer or employer based upon an individual's genetic information.

SEC. 3. DEFINITIONS.

As used in this Act:

(1) DNA.—The term "DNA" means deoxyribonucleic acid.

(2) DNA SAMPLE.—The term "DNA sample" means any human biological specimen from which DNA can be extracted, or the DNA extracted from such specimen.

(3) EMPLOYER.—The term "employer" has the same meaning given such term in section 3(d) of the Fair Labor Standards Act of 1938 (29 U.S.C. 203(d)).

(4) GENETIC INFORMATION.—The term "genetic information" means the information about genes, gene products or inherited characteristics that may derive from an individual or a family member.

(5) GENETIC TEST.—The term "genetic test" means a test for determining the presence or absence of genetic characteristics in an individual, including tests of nucleic acids such as DNA, RNA

and mitochondrial DNA, chromosomes or proteins in order to diagnose a genetic characteristic.

(6) INSURER.—The term "insurer" means an insurance company, health care service contractor, fraternal benefit organization, insurance agent, third party administrator, insurance support organization or other person subject to regulation under State insurance laws. Such term includes self-funded health plans and health plans regulated under the Employee Retirement Income Security Act of 1974 (29 U.S.C. 1001 et seq.).

(7) SECRETARY.—The term "Secretary" means the Secretary of Health and Human Services.

SEC. 4. REQUIREMENTS FOR DISCLOSURE OF GENETIC INFORMATION.

(a) PROHIBITION.—

(1) IN GENERAL.—Except as provided in paragraph (2), regardless of the manner in which genetic information was received, or of the source of such information, including information received from an individual, an entity may not disclose or be compelled (by subpoena or any other means) to disclose genetic information about an individual unless such disclosure is specifically authorized by the individual involved or the legal representative of the individual

through a written authorization which includes a description of the information being disclosed, the name of the individual or entity to whom the disclosure is being made, and the purpose of the disclosure.

(2) EXCEPTIONS.—Notwithstanding paragraph (1), genetic information concerning an individual may be disclosed if such disclosure—

(A) is authorized under Federal or State criminal laws relating to the identification of individuals, or as is necessary for the purpose of a criminal or death investigation, a criminal or juvenile proceeding, an inquest, or a child fatality review by a multidisciplinary child abuse team;

(B) is required under the specific order of a Federal or State court;

(C) is authorized under Federal or State law for the purpose of establishing paternity;

(D) is for the purpose of furnishing genetic information relating to a decedent to the blood relatives of the decedent for the purpose of medical diagnosis; or

(E) is for the purpose of identifying bodies.

(b) APPLICATION OF SECTION.—The prohibitions of this section shall apply to any redisclosure by any entity after another entity has disclosed the genetic information.

SEC. 5. PROHIBITION ON CERTAIN EMPLOYMENT PRACTICES.

(a) DISCRIMINATION AS TO RIGHTS OR BENEFITS.—No employer may seek to obtain, obtain, or use the genetic information of an employee or a prospective employee, or require a genetic test of an employee or prospective employee, to distinguish between or discriminate against or restrict any right or benefit otherwise due or available to the employee or prospective employee.

(b) ENFORCEMENT.—The powers, remedies, and procedures set forth in sections 705 through 709 of the Civil Rights Act of 1964 shall be the powers, remedies, and procedures this section provides to any person alleging a violation of this section.

SEC. 6. REQUIREMENTS RELATING TO INSURERS.

(a) GENERAL PROHIBITION.—An insurer offering health insurance may not use genetic information to reject, deny, limit, cancel, refuse to renew, increase the rates of, or otherwise affect health insurance.

(b) PROHIBITION ON INDUCEMENT.—With respect to a genetic test conducted in accordance with subsection (c),

an insurer may not use such a genetic test as an inducement for the purchase of insurance.

(c) PERMISSIBILITY OF TESTS.—If an insurer requests that an applicant for insurance (other than an applicant for health insurance) take a genetic test in connection with an application for insurance, the use of the results of such test shall be disclosed to the applicant and the insurer shall obtain the specific written authorization of the applicant for such disclosure.

(d) APPLICATION.—This section shall apply only to insurance policies issued on or after the date of enactment of this Act, and to the renewal of policies issued before, on, or after such date of enactment.

SEC. 7. FURTHER RECOMMENDATION BY THE NATIONAL BIOETHICS ADVISORY COMMISSION.

Not later than August 31, 1996, the National Bioethics Advisory Commission shall prepare and submit to the appropriate committees of Congress a report containing recommendations on—

(1) the development and implementation of standards to provide increased protection for the collection, storage, and use of identifiable DNA samples and genetic information obtained from those samples; and

(2) the development and implementation of appropriate standards for the acquisition and retention of genetic information in all settings, including appropriate exceptions.

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Second Breast Cancer Gene Identified

Associated Press

NEW YORK, Dec. 20—An international team of scientists has identified a second major breast cancer gene that makes some women highly likely to get the disease.

In families with a strong history of breast cancer, women who inherit a defective version of the gene run about an 80 percent lifetime risk of the disease.

The gene is called BRCA2, and it follows the identification of BRCA1 last year. Only about 5 percent to 10 percent of all cases of breast cancer are thought to come from inheriting a faulty gene.

Together, the two recently identified genes account for perhaps 90 percent of these familial breast cancers, with BRCA2 responsible for maybe 40 percent.

Last year, scientists announced that they had narrowed the search for BRCA2 to a small area of chromosome 13. The new study says researchers have captured a chunk of the gene itself.

The work is reported in the Dec. 21 issue of the journal Nature by Michael R. Stratton of the Institute of Cancer Research in Surrey, England, P. Andrew Futreal of the Duke University Medical Center in Durham, N.C., and 36 others.

In another development today, Myriad Genetics Inc. of Salt Lake City announced that its scientists have recovered the entire BRCA2 gene. The company said it plans to use it in combination with BRCA1 to develop a test for predisposition to hereditary breast cancer.

The cause of the vast majority of cases of breast cancer is not known. Scientists hope studying inherited breast cancer genes will someday pay off with new treatments and ways to prevent the disease.

More immediately, the finding will let members of the relatively few families affected by BRCA2 find out if they carry the gene, which also raises the risk of breast cancer in men. But the researchers said it is too soon to start testing the general population for the presence of a defective BRCA2 gene.

Like BRCA1, the newfound gene seems to suppress cancer when it is working normally. But when BRCA2 is defective, this brake is lost.

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Genetic Bank to Store DNA Data for Families
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DENTON, Texas (AP) The University of North Texas and a private company have announced the formation of a ''genetic banking service'' to operate like a safety deposit vault for DNA.

The private, for-profit venture will store DNA specimens for 25 years for people who pay $175 each for the service, the Houston Chronicle reported today.

The point is to ensure that future generations will be able to examine a person's genetic code after the person has died, mainly to help their doctors fight diseases they may have inherited.

The new gene bank will avoid ethical tangles by gathering and storing data only, and not providing that data to anyone but members of a donor's family, an official said.

Officials of the new GeneLink data bank say there may be no better legacy to leave one's offspring than the genetic data they may be able to use to protect their own health.

Genetic markers already are being identified tentatively for physical disorders such as cancer and heart disease and even for other disorders long believed to be entirely behavioral, such as alcoholism and attention deficit disorder.

In years to come, there may be great advantages in being able to go back and scan the DNA of a relative who is no longer alive.

A person who wants to store DNA data will gently scrape the inside of his or her mouth with special swabs and mail them to GeneLink. GeneLink and UNT will store the swabs in refrigerated safes for 25 years, or longer if a new fee is paid.

Genetic discrimination

It should come as no surprise that the proliferation of personal genetic information—a foreseeable result of government and biotechnology industry investment in techniques that have produced the Human Genome Project—is resulting in genetic discrimination.

The Human Genome Project, a 15-year federal program jointly sponsored by the National Institutes of Health and the Department of Energy, has the goal of finding and chemically analyzing all human genes. Its budget is estimated to be $3-5 billion over the entire funding period. It is already stimulating a massive expansion of available personal hereditary information.

Genetic discrimination—the differential treatment of people arising from genetic information—may be an inevitable result of the study of human genetics, which thrives on correlating DNA variations with measurable human differences.

When genetic approaches are applied to complex human traits (such as sexuality, criminality, or common ailments), basic insights may be garnered. But descriptions of human characteristics relying solely on genetic data will always be too simplified and reduced. More generally, it is self-evident that we are more than the sum of our genes and should never be slaves of our DNA.

Nonetheless, it is naive to believe that adverse genetic discrimination will not occur when these powerful but limited genetic methods are applied to complex phenomena in a society that has flirted with eugenics (human improvement through genetic manipulation); discriminated against groups on sexual preference, racial, gender, and other grounds; and allowed important businesses to claim that "equity" demands some individuals be denied benefits that are, in fact, basic entitlements (like healthcare, financial safety, or fair access to governmental employment).

My colleagues and I have been studying genetic discrimination since the mid-1980s. (Our study is a collaborative effort among members of the Boston-based Genetic Screening Study Group and is currently supported, in part, by a grant from the Department of Energy.) Using survey instruments and a case-based analytic method, we have reviewed hundreds of self-reported incidents. In addition, we have interviewed individuals "at risk" for suffering genetic discrimination who claimed not to have experienced this problem.

The results of these investigations not only confirm the existence of genetic discrimination currently, but suggest that a range of institutions are engaging in discriminatory practices. We've documented a variety of personal behaviors directed at coping with genetic discrimination's effect. Individual and family avoidance of genetic testing in order to escape discrimination is an ironic outcome in the world of the Human Genome Project, since those who are shunning personal hereditary data are also paying for it with their tax dollars.

Our study of genetic discrimination has been primarily focused on proving its occurrence and describing the incidents. But research on this topic has also highlighted the struggle our nation faces over healthcare, social fairness, the provision of equal rights, the protection of privacy, and the right of independent non-coerced choice in personal matters.

Discrimination and access

It has been argued that genetic information is not unique nor does it differ from other types of clinically relevant data. For instance, it has been compared to blood pressure measurements in its predictiveness and impact. Certainly, any information found in the medical record can become part of processes that may lead to loss of entitlements. But genetic information does have special characteristics:

1. It is informative and relevant from conception to death. DNA instructions do not seem to change substantially over a lifetime, though environmental modifications may alter their expression significantly.

2. The results of genetic testing can reveal other personal traits (race, ethnicity, gender) and mark members of a testee's family as "at risk."

3. Genetic information can be associated with personal and family stigmas as well as a special social status resulting from our society's involvement with eugenic practices.

Though an elevated blood cholesterol and a DNA test to detect the presence of a gene that can produce high blood cholesterol may be similarly predictive and produce identical responses from perusers of the "private" medical record, the implications of the gene test are currently considerably greater.

If illnesses and predispositions are defined by genetic information rather than clinical symptoms and signs, the class of sufferers termed the "asymptomatic ill" will grow. These people's illness only resides in their DNA sequences; it may never manifest itself as a functional disease. Yet individuals from this group are increasingly excluded from healthcare under present social conditions. The Human Genome Project and its products may so graphically highlight the inequities of our healthcare financing and delivery system that change will be fostered.

Unfortunately, however, healthcare reform may not occur. Coverages for genetic testing, counseling, and treatments could remain limited and inadequate in the future.

Or future healthcare planners might use genetic information to manage (limit) benefits as insurers do now. The result will be continued genetic discrimination in healthcare despite reforms intended to deliver universal access to care.

We may find that we are all genetically inclined to some potentially disabling condition.

"Fair" discrimination

As incidents of contemporary genetic discrimination have been described, commentators have argued that though painful, such outcomes are morally and practically defensible: There is "good" discrimination and "bad."

An extreme position has been articulated: Society should in the near future organize all its benefits, goods, and services using genetic information. Under this scenario, healthcare would be allocated by tracking disease genes, education would be offered to people demonstrating genetic intelligence, surveillance of those with hereditary behavioral disorders would increase, and manual labor might be reserved for those with genetically strong backs.

A more moderate version of this argument suggests that the "public" should have the right to protect itself against adversities involving the genetically predisposed. This position was dramatized by a recent account of a family of air-traffic controllers who had inherited the gene for the neurodegenerative and psychiatric disorder, Huntington's chorea (also called Woody Guthrie's disease). This family's genetic counselors faced pressures to inform federal officials and employers, as well as care for these needy people.

Should the privacy of genetic information be subject to violations for the public good? Consider the many genes now amenable to testing that predispose the bearers to a substantial risk for subsequent blindness. Would we deny access to driver's licenses, gun permits, certain jobs, and other rights to those who had inherited blindness-related genes—irrespective of their visual acuity?

The public may have the right to assess the functional ability of people when these individuals apply for special permits, but there should not be a program instituted by the state to measure and assess individual genetic propensities.

Similarly, illnesses should continue to be defined, and their existence determined, by clinical criteria. The concept of "genetic diseases" obfuscates the fact that illnesses affect humans while genes are only risk factors in a complex physiological cascade that can lead to disease under permissive conditions. Though genetic testing may facilitate the identification of states (traits) that can be ameliorated by special treatments or preventive approaches, these traits are not illnesses. Unfair outcomes from lumping predisposition and illness together must be anticipated.

Protecting the "disabled"

The Americans with Disabilities Act of 1990 is an important advance in civil rights designed to improve the lives of individuals with special needs or disabilities. Its interpretation and enforcement have yet to be fully determined. This law may extend protection, particularly in public and business settings, to people who are are not functionally affected but are perceived to be "disabled." If interpreted broadly by the executive branch and enforced effectively, it could help bring an end to some forms of genetic discrimination now perpetrated by employers and insurers.

But is it appropriate to consider those with unexpressed genetic conditions as disabled? With continuing investment in the Human Genome Project, we may find that we are all genetically inclined to some potentially disabling condition. The universality of a propensity to disease or disability might lead to both the acceptance of the "disabled" into the political and social mainstream and the end of genetic discrimination.

But it probably won't. It is misleading to lump the "disabled" and "genetically labeled" into one group. Today, genetic information is an intrinsic part of all human existence; disability (defined significantly by responses to it) is not. While not slowing the pace of disability rights reform, genetic information should be protected separately. It ought to be recognized like other human characteristics, such as gender and race, that are not an acceptable basis for exclusion or discrimination in our society.

The right "not to know"

The choice to undergo genetic testing for

Insurance companies have denied payment to healthy individuals whom they perceived as "genetically ill."

any reason necessitates that the option to forego such an analysis be available. This is a basic principle of choice and consent. Incidents of genetic discrimination illustrate that the right to be ignorant of one's heredity, *to not know* genetic information, may be eroding.

Consider the case of a pregnant woman at high genetic risk for producing an ill child. The affected family is told by their HMO that the prenatal costs and child's medical care will not be covered if prenatal diagnostic information is ignored. Irrespective of overt pressures to abort genetically "affected" fetuses, the cost of healthcare can be used as a powerful coercive force on prospective parents and may eliminate the choice to not undergo genetic testing.

In addition, insurance companies have denied payment of contracted benefits to healthy individuals whom they perceived as "genetically ill"—as having a hereditary "pre-existing" condition. The military has refused to pay disability compensation to service members who develop symptoms of a genetic condition while enlisted, claiming the illness existed in these healthy people before recruitment.

If an individual must know all his or her genetic predispositions in order to enter into a valid contract with an agency, then to "not know" is to give up healthcare financing, driving, a mortgage, disability protection, other insurances, and forms of employment. Is there really a choice in the decision to undergo genetic testing when so many benefits are contingent on the existence and knowledge of genetic information?

No simple solutions

Since the problems associated with genetic discrimination are complex and arise from both historical and socioeconomic facts, simple solutions are unlikely. Research and education will certainly play an important role in any reform process. Legislation may also be needed.

Two years ago, the California State Legislature passed a bill that, had it not been vetoed by Governor Pete Wilson, would have amended the Unruh Civil Rights Act to include "genetic characteristics" as a protected entity in the state. Under the terms of this bill, the use of genetic information would have been curtailed in insurance and employment practices. The veto came after opposition and lobbying by an association of state employers. This occurred even though the Americans with Disabilities Act will have a far greater impact on this sector than the bill that was killed.

Consideration is now being given to reintroducing this legislation—a worthy endeavor of the representatives of the people of California. The vetoed legislation has stimulated similar efforts in other states; Wisconsin, for instance, banned DNA testing or even inquiries about it by health insurers.

Passing laws in the absence of public understanding and consensus can cause problems and be an ineffective form of social change. But in the case of genetic discrimination, much as with gender and race, changes in society may necessitate that public policy and the law be used to protect the vulnerable while a process of education is concurrently undertaken. The existence of discrimination arising from genetic information may force the public to clearly restate the historic commitment to equality in the face of difference. ●

PAUL R. BILLINGS, MD, PhD *is an internist and human geneticist. He is chief of the Section of General Internal Medicine (Palo Alto VA Medical Center) in the Department of Medicine at Stanford University's School of Medicine.* *This work was supported in part by Grant #DE-FG02-92ERG1395 from the U.S. Department of Energy.* *Billings can be reached at (415)852-3492.*

Genetic Discrimination and Health Insurance: An Urgent Need for Reform

Kathy L. Hudson, Karen H. Rothenberg, Lori B. Andrews, Mary Jo Ellis Kahn, Francis S. Collins

The accelerated pace of gene discovery and molecular medicine portend a future in which information about a plethora of disease genes can be readily obtained. As at-risk populations are identified, research can be done to determine effective prevention and treatment strategies that will lower the personal, social, and perhaps the financial costs of disease in the future. We all carry genes that predispose to common illnesses. In many circumstances knowing this information can be beneficial, as it allows individualized strategies to be designed to reduce the risk of illness. But, as knowledge about the genetic basis of common disorders grows, so does the potential for discrimination in health insurance coverage for an ever increasing number of Americans.

The use of genetic information to exclude high-risk people from health care by denying coverage or charging prohibitive rates will limit or nullify the anticipated benefits of genetic research. In addition to the real and potentially devastating consequences of being denied health insurance, the fear of discrimination has other undesirable effects. People may be unwilling to participate in research and to share information about their genetic status with their health care providers or family members because of concern about misuse of this information. As genetic research progresses, and preventive and treatment strategies are developed, it will be increasingly important that discrimination and the fear of discrimination not be a roadblock to reaping the benefits. To address these issues, the National Institutes of Health–Department of Energy (NIH-DOE) Working Group on Ethical, Legal, and Social Implications (ELSI) of the Human Genome Project and the National Action Plan on Breast Cancer have jointly developed a series of recommendations for state and federal policymakers which are presented below.

K. L. Hudson is assistant director for Policy Coordination, National Center for Human Genome Research, National Institutes of Health (NIH). K. H. Rothenberg is Marjorie Cook Professor of Law and director of the Law and Health Care Program, University of Maryland School of Law, and member of the National Action Plan on Breast Cancer (NAPBC). L. B. Andrews is chair of the NIH-Department of Energy Working Group on Ethical, Legal, and Social Implications and professor at Chicago-Kent College of Law. M. J. Ellis Kahn represents the Virginia Breast Cancer Foundation and the National Breast Cancer Coalition and is co-chair of the Hereditary Susceptibility Working Group, NAPBC. F. S. Collins is director of the National Center for Human Genome Research, NIH, and co-chair of the Hereditary Susceptibility Working Group, NAPBC.

In the past, genetic information has been used by insurers to discriminate against people. In the early 1970s, some insurance companies denied coverage and charged higher rates to African Americans who were carriers of the gene for sickle cell anemia (*1*). Contemporary studies have documented cases of genetic discrimination against people who are healthy themselves but who have a gene that predisposes them or their children to a later illness such as Huntington's disease (*2*). In a recent survey of people with a known genetic condition in the family, 22% indicated that they had been refused health insurance coverage because of their genetic status, whether they were sick or not (*3*).

As a case example, Paul (not his real name) is a healthy, active 4-year-old, but he has been twice denied health insurance. Paul's mother died in her sleep of sudden cardiac arrest when Paul was only 5 months old. Paul's maternal uncle also died of sudden cardiac arrest when he was in his twenties. After these sudden and unexpected deaths, Paul's family began a hunt to discover the cause. Their search finally led to a research geneticist who was able to determine that several family members, including Paul and his mother, carried an alteration in a gene on chromosome 7. This gene is one of several genes that causes the long QT syndrome, so-called because of the distinctive diagnostic pattern on an electrocardiogram.

Several years ago, Paul's father, Bob, lost his job and with it the group policy that provided health insurance coverage for Paul and him. Paul's father has repeatedly applied for a family health insurance policy with a major insurance company. The company agreed to cover Bob but refused to issue a family policy that would cover Paul because he has inherited the altered gene for the long QT syndrome from his mother.

The story of Jackie and Emma further illustrates the social, ethical, and legal dilemmas presented by the revelation of genetic information. Sisters Jackie and Emma, along with many other members of their family, have been tested as part of a research protocol for alterations in the gene, *BRCA1*, that confers hereditary susceptibility to breast and ovarian cancer. Both were offered an opportunity to learn the results of their genetic tests and both accepted. They each learned they carry an altered form of the gene, putting them at increased risk for breast and ovarian cancer.

After finding out the results of her genetic test, Emma had a mammogram that showed a very small lesion in her breast. A subsequent biopsy revealed carcinoma, and Emma decided to proceed with a bilateral mastectomy because of the substantial risk of cancer arising in the opposite breast. Her lymph nodes were negative for cancer, so her prognosis for cure is very good.

Emma's sister Jackie also tested positive for the same alteration in the *BRCA1* gene, though no cancer was detected. Although the benefit of prophylactic mastectomy in reducing the risk for breast cancer is not yet known, she decided to have a bilateral prophylactic mastectomy. Emma and Jackie feel strongly that they have benefited from knowing this genetic information but are fearful that it will be used against them and their family by insurers and employers. They both keep their genetic status secret and are so fearful of losing their health insurance that they used assumed names when sharing their story at a recent workshop on genetic discrimination (*4*).

Emma and Jackie's story is not unique. An estimated 1 in 500 women carry a mutation in the *BRCA1* gene that may confer as much as an 85% chance of breast cancer and a 50% chance of ovarian cancer (*5*). Although substantial uncertainty exists about the relative value of the available options (surgery compared with intensive surveillance) for a woman with a *BRCA1* mutation, it is likely that ultimately this information will be medically useful.

Health Insurance in the United States

Because of high costs, insurance is essentially required to have access to health care in the United States. Over 40 million people in the United States are uninsured (*6*). Group insurance, individual insurance, self-insurance, and publicly financed insurance (for example, Medicare and Medicaid) are the principal forms of health insurance in the United States for the ~240 million Americans with coverage. Most people get their health insurance through their employer. Many employers provide health insurance coverage through self-funded plans in which the employer, either directly or through a third party, provides health insurance coverage. For individuals and small groups, insurance providers use medical history as well as individual risk factors, such as smoking, to determine whether to provide coverage and under what terms. This is

known as underwriting. Insurers argue that underwriting is essential in a voluntary market to prevent "adverse selection," in which individuals elect not to purchase insurance until they are already ill or anticipate a future need for health care. Insurers fear that individuals will remain uninsured until, for example, they receive a genetic test result indicating a predisposition to some disease such as breast or colon cancer.

In the absence of the ability to detect hereditary susceptibility to disease, the costs of medical treatment have been absorbed under the current health insurance system of shared risk and shared costs. Today, our understanding of the relation between a misspelling in a gene and future health is still incomplete, thus limiting the ability of insurers to incorporate genetic risks into actuarial calculations on a large scale. As genetic research enhances the ability to predict individuals' future risk of diseases, many Americans may become uninsurable on the basis of genetic information.

State and Federal Initiatives

A recent survey has shown that a number of states have enacted laws to protect individuals from being denied health insurance on the basis of genetic information (Fig. 1) (*7*). The first laws addressing genetic discrimination were quite limited in scope and focused exclusively on discrimination against people with a single genetic trait such as sickle cell trait (*8*). Since the Human Genome Project was launched in 1990, eight states have enacted some form of protection against genetic discrimination in health insurance. The recently enacted state laws are not limited to a specific genetic trait but apply potentially to an unlimited number of genetic conditions. These state laws prohibit insurers from denying coverage on the basis of genetic test results, and prohibit the use of this information to establish premiums, charge differential rates, or limit benefits. A few of these states, including Oregon and California, integrate protection against discrimination in insurance practices with privacy protections that prohibit insurers from requesting genetic information and from disclosing genetic information without authorization.

Two factors limit the protection against discrimination afforded by current state laws. First, the federal Employee Retirement Income Security Act exempts self-funded plans from state insurance laws. Nationwide, over one-third of the nonelderly insured population obtains health insurance coverage through a self-funded plan. Second, nearly all of the state laws focus narrowly on genetic tests, rather than more broadly on genetic information generated by family history, physical examination, or the medical record (*7*). Limiting the scope of protection to results of genetic tests means that insurers are only prohibited from using the results of a chemical test of DNA, or in some cases, the protein product of a gene. But insurers can use other phenotypic indicators, patterns of inheritance of genetic characteristic, or even requests for genetic testing as the basis of discrimination. Meaningful protection against genetic discrimination requires that insurers be prohibited from using all information about genes, gene products, or inherited characteristics to deny or limit health insurance coverage.

No federal laws are currently in place to prohibit genetic discrimination in health insurance (*9*). The Clinton Administration's proposal to reform the health care system and provide health insurance for all Americans did prohibit limiting access or coverage on the basis of "existing medical conditions or genetic predisposition to medical conditions" (*10*). Congressional efforts to reform the health care system in 1995 have been much more modest and are targeted at guaranteeing access, portability, and renewability of coverage and at leveling the playing field in the insurance market so that the same rules apply to insured and self-funded plans. Recent federal health insurance reform proposals attempt to guarantee the availability of health care by prohibiting insurers from denying coverage on the basis of health status, medical condition, claims experience, or medical history of a participant. Most of the proposals permit exclusions for pre-existing conditions, but these are time limited.

It is not clear if the current health insurance reform proposals would prohibit insurers from denying coverage on the basis of genetic information. Genetic information is distinct from other types of medical information because it provides information about an individual's predisposition to future disease. In addition, genetic information can provide clues to the future health risks for an individual's family members. If enacted, current health reform proposals would prohibit denying insurance to those currently suffering from disease or with a past history of disease. But these proposals may not protect people like Paul, who are healthy but have a genetic predisposition to disease, from being refused insurance coverage. Current proposals also may fail to protect couples who, although healthy themselves, carry the gene for a rec ve disorder such as cystic fibrosis that might affect their children or future children.

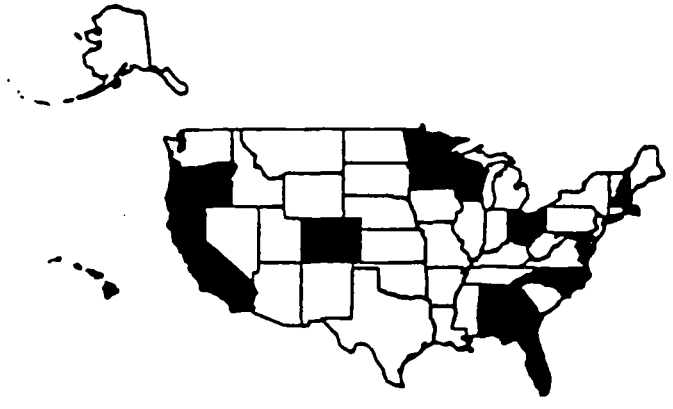

Fig. 1. State laws on the use of genetic information in health insurance (*7*). States shown in purple were the first states to enact legislation addressing genetic issues in insurance. Florida and Alabama laws prohibit insurers from denying coverage on the basis of the sickle cell trait. North Carolina prohibits insurers from denying coverage because the applicant has the hemoglobin C or sickle cell trait. Maryland prohibits discrimination in rates based on any genetic trait unless there is actuarial justification. States shown in green (California, Oregon, Colorado, Minnesota, Wisconsin, Ohio, Georgia, and New Hampshire) prohibit insurers, to varying degrees, from requiring or requesting genetic tests or their results, from denying coverage on the basis of genetic tests, and from using tests to determine rates and benefits. California, Colorado, Oregon, and Wisconsin laws include provisions to protect the privacy of genetic information. States shown in orange (Massachusetts and Hawaii) have related bills pending.

Recommendations

Planners of the Human Genome Project recognized from the beginning that maximizing the medical benefits of genome research would require a social environment in which health care consumers were protected from discrimination and stigmatization based on their genetic make-up. Genome programs at both the DOE and the National Center for Human Genome Research, a component of NIH, have each set aside a portion of their research budget to anticipate, analyze, and address the ELSI of new advances in human genetics. The original planners also created the NIH-DOE ELSI Working Group, which has a broad and diverse membership including genome scientists; medical geneticists; experts in law, ethics, and philosophy; and consumers, to explore and propose options for the development of sound professional and public

policies related to human genome research and its applications. The ELSI Working Group has long been involved in discussions about the fair use of genetic information. In a 1993 report, "Genetic Information and Health Insurance" (*11*), the ELSI Working Group recommended a return to the risk-spreading goal of insurance. The Working Group suggested that individuals be given access to health care insurance irrespective of information, including genetic information about their past, current, or future health status. Because denial of insurance coverage for a costly disease such as breast cancer may prove to be a death sentence for many women, the National Action Plan on Breast Cancer (NAPBC), a public-private partnership designed to eradicate breast cancer as a threat to the lives of American women, has identified genetic discrimination in health insurance as a high priority (*12*).

Building on their shared concerns, the NAPBC (*13*) and the ELSI Working Group (*14*) recently cosponsored a workshop on genetic discrimination and health insurance (*4*). Scientists, representatives from the insurance industry, and members of the ELSI Working Group and the NAPBC participated in the 1-day session. On the basis of the information presented at the workshop, the ELSI Working Group and the NAPBC developed the following recommendations and definitions for state and federal policymakers to protect against genetic discrimination.

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

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

3) Insurance providers should be prohibited from requesting or requiring collection or disclosure of genetic information.

4) 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.

The definitions are as follows. Genetic information is information about genes, gene products, or inherited characteristics that may derive from the individual or a family member. Insurance provider means an insurance company, employer, or any other entity providing a plan of health insurance or health benefits including group and individual health plans whether fully insured or self-funded.

These recommendations have been endorsed by the National Advisory Council for Human Genome Research (NACHGR) (*15*). The NACHGR stresses the positive value of genetic information for improving the medical care of individual patients and the need to ensure the freedom of patients and their health care providers to use genetic information for patient care. The NACHGR views the elimination of the use of genetic information to discriminate against individuals in their access to health insurance as a critical step toward these goals.

The ability to obtain sensitive genetic information about individuals, families, and even populations raises profound and troubling questions about who will have access to this information and how it will be used. The recommendations presented here for state and federal policy-makers are intended to help ensure that our current social, economic, and health care policies keep pace with both the opportunities and challenges that the new genetics present for understanding the causes of disease and developing new treatment and preventive strategies.

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2. P. R. Billings *et al.*, *Am. J. Hum. Genet.* **50**, 476 (1992).
3. E. V. Lapham (Georgetown University) and J. O. Weiss, *The Alliance of Genetic Support Groups*, Human Genome Model Project, preliminary results of a survey of persons with a genetic disorder in the family.
4. "Genetic discrimination and health insurance: A case study on breast cancer," Bethesda, MD, 11 July 1995, workshop sponsored by the National Action Plan on Breast Cancer (NAPBC) and the NIH-DOE Working Group on the Ethical, Legal, and Social Implications of Human Genome Research.
5. D. F. Easton *et al.*, *Am. J. Hum. Genet.* **52**, 678 (1993); D. Ford *et al.*, *Lancet* **343**, 692 (1994).
6. *Employee Benefit Research Institute Special Report SR-28*, issue brief number 158, February 1995.
7. K. H. Rothenberg, *J. Law Med. Ethics*, in press.
8. North Carolina, NC ST: 58-65-70 (1975), Florida, FL ST: 626.9707 (1978), Alabama, AL ST: 27-5-13 (1982). In 1987, Maryland passed a law, Art. 48A, 223(b)(4), prohibiting health insurers from discrimination in rates based on genetic traits unless there was "actuarial justification."
9. In March 1995, the U.S. Equal Employment Opportunity Commission (EEOC) released official guidance on the definition of the term "disability." The EEOC's guidance clarifies that protection under the Americans with Disabilities Act (ADA) extends to individuals who are discriminated against in employment decisions solely on the basis of genetic information about an individual. For example, an employer who makes an adverse employment decision on the basis of an individual's genetic predisposition to disease, whether because of concerns about insurance costs, productivity, or attendance, is in violation of the ADA because that employer is regarding the individual as disabled. Issuance of the EEOC's guidance is precedent setting; it is the first broad federal protection against the unfair use of genetic information.
10. *Health Security Act*, Section 1516, S. 1757/HR 3600.
11. "Genetic information and health insurance: Report of the task force on genetic information and insurance" (NIH-DOE Working Group on the Ethical, Legal, and Social Implication of Human Genome Research, 10 May 1993).
12. The NAPBC has as its mission to reduce the morbidity and mortality from breast cancer and to prevent the disease. Specific goals include the following: (i) to promote a national effort to establish and address priority issues related to breast cancer etiology, early detection, treatment, and prevention; (ii) to promote and foster communication, collaboration, and cooperation among diverse pubic and private partners; and (iii) to develop strategies, actions, and policies to improve breast cancer awareness, services, and research.
13. NAPBC steering committee: Susan J. Blumenthal (co-chair), Zora Kramer Brown, Doris Browne, Anna K. Chacko, Francis S. Collins, Nancy W. Connell, Kay Dickersin, Artyne Draper, Nancy Evans, Harmon Eyre, Leslie Ford, Janyce N. Hedetniami, Mary Jo Ellis Kahn, Amy S. Langer, Susan M. Love, Alan Rabson, Jane Reese-Coulbourne, Irene M. Rich, Barbara K. Rimer, Susan Sieber, Edward Sondik, and Frances M. Visco (co-chair). NAPBC hereditary susceptibility working group: Kathleen A. Calzone, Francis S. Collins (co-chair), Sherman Elias, Linda Finney, Judy E. Garber, Ruthann M. Giusti, Jay R. Harris, Joseph K. Hurd Jr., Mary Jo Ellis Kahn (co-chair), Mary-Claire King, Caryn Lerman, Mary Jane Massie, Paul G. McDonough, Patricia D. Murphy, Philip D. Noguchi, Barbara K. Rimer, Karen H. Rothenberg, Karen K. Steinberg, and Jill Stopler.
14. ELSI working group: Betsy Anderson, Lori Andrews (chair), James Bowman (dissenting), David Cox, Troy Duster, (vice chair), Rebecca Eisenberg, Beth Fine, Neil Holtzman, Philip Kilcher, Joseph McInerney, Jeffrey Murray, Dorothy Nelkin, Rayna Rapp, Marsha Seldon, and Nancy Wexler.
15. NCHGR council members: Anita Allen, Lennette J. Benjamin; David Botstein, R. Daniel Camerini-Otero (dissents with recommendation 3), Ellen W. Clayton, Troy Duster, Leroy E. Hood, David E. Housman, Richard M. Myers, Rodney Rothstein, Diane C. Smith, Lloyd M. Smith, M. Anne Spence, Shirley M. Tilghman, and David Valle.

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