Virginia (1996)	S.J.R. 50	Continues the subcommittee established in 1995 to study the legal and policy ramifications of breast cancer susceptibility gene research, including the ethical and legal issues of health insurance coverage and reimbursement.
Virginia (1996)	S 335 (Ch. 704)	Prohibits health insurers from terminating, restricting or limiting coverage or sale, canceling or refusing to renew coverage, excluding from coverage, imposing a waiting period, requiring a rider or establishing differentials in premium rates based on any genetic information. Establishes that information obtained from genetic screening or testing shall be confidential and shall not be made public or used in any way, in whole or in part, to cancel, refuse to issue or renew, or limit benefits under any policy, contract or plan.
Wisconsin (1991)	WI ST: §631.89	Prohibits health insurers from conditioning the provision of insurance coverage or benefits on genetic testing. Prohibits use of genetic testing information in determining rates. Prohibits health insurers from requiring a genetic test or inquiring as to whether or not a genetic test has been performed.

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New Jersey (1996)	S 695 A 1411	Prohibits health insurers from discriminating in the issuance, withholding, extension, renewal, or establishment of rates, terms or conditions on the basis of genetic information. Prohibits any person from obtaining or retaining an individual's genetic information without first obtaining authorization from the individual. Prohibits the disclosure of genetic information in a manner that permits identification of the individual without the authorization of the individual.
New York (1996)	S 4293-D A 5796-C	Prohibits authorized insurers from requesting or requiring an individual proposed for insurance coverage to be the subject of a genetic test without receiving the written informed consent of such individual prior to such testing.
North Carolina (1975)	NC ST: §58-65-70	Prohibits health insurers from refusing to issue health insurance because an individual has sickle cell trait or hemoglobin C trait. Prohibits health insurers from charging higher premiums based on sickle cell trait or hemoglobin C trait.
Ohio (1993)	OH ST: §1742.42; §1742.43; §3901.49; §3901.491; §3901.50; §3901.501	Prohibits health insurers from canceling, refusing to issue, or renewing coverage or limiting benefits based on genetic screening or testing. Prohibits health insurers from requiring a genetic test or making an inquiry as to the results of genetic screening or testing. Provides for consideration of genetic testing if the results are favorable to the applicant and voluntarily submitted. Establishes the Task Force on Genetic Testing in Health Insurance.

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Oklahoma (1996)	HCR 1113	Creates the Task Force on Prevention of Genetic Discrimination. The duties of the Task Force include: reviewing House Bill 2478; reviewing legislation from other states and any model legislation related to genetic discrimination; and, making recommendations to the legislature by January 1, 1997 concerning proposed legislation which the Task Force deems necessary to prevent genetic discrimination particularly with regard to insurance and employment.
Oklahoma (1997)	HCR 1012	Creates the Task Force on Prevention of Genetic Discrimination. The duties of the Task Force include: - reviewing H.B. 2478 of the 2nd Session of the 45th Oklahoma Legislature which proposes the creation of a Genetic Nondiscrimination Act for the State of Oklahoma - reviewing legislation from other states and any model legislation related to genetic discrimination; and - making recommendations to the Legislature by January 1, 1998, concerning proposed legislation which the Task Force deems necessary to prevent genetic discrimination, particularly with regard to insurance and employment.
Oregon (1995)	OR ST: 659.036; 659.227	Prohibits health insurers from utilizing genetic information to reject, deny, limit, cancel, refuse to renew, increase the rates of, or affect the terms and conditions of health insurance policies. Provides for informed consent and privacy protection of genetic information.
Tennessee (1997)	H 413	Prohibits insurers from denying or canceling health insurance coverage on the basis of genetic information or on the basis that the individual or a family member of an individual has requested or received genetic services. Prohibits insurers from varying the premiums, terms or conditions for health insurance on the basis of genetic information or on the basis that the individual or a family member of an individual has requested or received genetic services. Prohibits insurers from requesting or requiring the disclosure of genetic information. Prohibits insurers from disclosing genetic information without prior written authorization.
Virginia (1995)	S.J.R. 372	Establishes a subcommittee to study the legal and policy ramifications of breast cancer susceptibility gene research, including the ethical and legal issues of health insurance coverage and reimbursement.

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Genetic Discrimination: Perspectives of Consumers

E. Virginia Lapham,* Chahira Kozma, Joan O. Weiss

In a study of the perceptions of 332 members of genetic support groups with one or more of 101 different genetic disorders in the family, it was found that as a result of a genetic disorder 25 percent of the respondents or affected family members believed they were refused life insurance, 22 percent believed they were refused health insurance, and 13 percent believed they were denied or let go from a job. Fear of genetic discrimination resulted in 9 percent of respondents or family members refusing to be tested for genetic conditions, 18 percent not revealing genetic information to insurers, and 17 percent not revealing information to employers. The level of perceived discrimination points to the need for more information to determine the extent and scope of the problem.

The rapid advances in human genetics, largely fueled by the Human Genome Project (HGP), have resulted in the expansion of the number and range of genetic tests (*1*). These tests are capable of providing carrier and presymptomatic information including risk of future disease, disability, and early death. In addition, these tests may reveal genetic information not only about the health of the individual, but also about his or her family members (*2*).

Concern about access to genetic information by health insurers has historical support (*3*, *4*). In the early 1970s, several insurance companies discriminated against individuals who were carriers of sickle cell anemia, even though they were quite healthy (*5*). The use of genetic information to deny life insurance to individuals leaves their dependents more vulnerable to economic consequences than is the case with the 70% of adults who are covered (*6*). The use of genetic screening to identify workers who may be particularly sensitive to noxious environments has been the principal focus of concern about workplace genetic testing even when done for benevolent reasons (*7*). Issues of genetic discrimination in employment and insurance have become more urgent as a result of the genome project (*8*).

Overall concerns about privacy and confidentiality have led the Ethical, Legal, and Social Issues (ELSI) Branch of the National Center for Human Genome Research to identify this issue as a top priority with the goal of proposing legislation specifically designed to protect people against genetic discrimination (*9*). Additionally, several working groups and scholars are focusing on this issue and have developed background papers and policy recommendations about the use of genetic information in health insurance as well as other areas such as life insurance and employment (*10*, *11*). Despite these concerns about potential genetic discrimination and documentation of individual cases, there is little information about the incidence and range of the problem (*12*).

This report provides information on the experiences of 332 individuals with one or more family members with a genetic disorder who are affiliated with genetic support groups. The study was part of the Human Genome Education Model (HuGEM) Project of the Georgetown University Child Development Center and the Alliance of Genetic Support Groups. It was the first phase of the HuGEM Project with the aim of getting input from 300 consumers in order to develop, implement, and evaluate a collaborative education model for consumers and health care providers.

Participants were recruited primarily through the national, regional, and local genetic support groups affiliated with the Alliance of Genetic Support Groups. Notices were put in two issues of the monthly *Alliance Alert* and letters were sent to the directors of 101 genetic support groups (representing an estimated 585,800 members). The notices contained information about the study and requested volunteers that were at least 18 years old and with one or more persons in the family with a genetic disorder who would be willing to participate in a 30-min telephone interview to provide opinions on the ethical, legal, and social issues of the HGP as well as priority topics for education. Volunteers were assured confidentiality of their responses. Random sampling was considered and ruled out because of time, cost, and the primarily educational focus of the project. Thus, the findings are applicable only to this group. Support group leaders were requested to distribute the letter requesting volunteers at meetings and in newsletters. Persons interested in participating were to complete a form at the bottom of the letter or call a 1-800 number for more information.

As a result of information provided by the support groups to the members or through the *Alliance Alert*, a group of 483 persons (from 73 different groups) contacted the Alliance of Genetic Support Groups about the study. They were sent information about the study and about the Human Genome Project. Of these, 336 (70%) returned consent forms (*13*). From this group, four persons decided not to participate after the interviews started, 306 persons completed telephone interviews, and 26 requested and completed the questionnaire by mail, for a total return of 332 respondents from 44 states and the District of Columbia (*14*).

Respondents were primarily female, highly educated, married, and Caucasian (*15*)—characteristics believed to be typical of genetic support groups (*16*). Age categories ranged from the twenties through seventies with a median age in the forties. A range of religious preferences was reported (*17*). There was an average of 2.1 affected family members per respondent with a range of 1 to 12 affected members reported.

The study questionnaire was composed primarily of questions with multiple choice responses. Telephone interviews were conducted by four social workers, a genetic counselor, and a consumer administrator (*18*) and lasted an average of 40 min with a range of 29 to 90 min. The content covered five areas: demographic information; knowledge of the Human Genome Project (61% had heard about the HGP before volunteering to participate in the study, 74% considered the HGP very important to their families, and 81% considered it very important to society); personal and family experience in areas related to genetic testing and research; opinions on a range of ethical, legal, and social issues; and priority topics for education. The education priorities were used to develop and implement educational forums in the mid-Atlantic and Pacific Northwest regions and will be described elsewhere.

Respondents were asked whether they or other family members had encountered problems with health insurance, life insurance, and employment (*19*). The term "genetic discrimination" was not used in the survey. It is used in this report to describe prejudicial actions as perceived by the respondents that resulted from insurers' or employers' knowledge of an individual's genetic condition, carrier status, or presumed carrier status, based on observation, family history, genetic testing, or other means of gathering genetic information (*20*).

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Respondents reported 101 different primary genetic disorders. The 18% of families with two or more disorders were asked to select one for purposes of the study. Of the primary disorders 68% were single-gene disorders, 10% were chromosome disorders, 11% were multifactorial disorders, 11% were major malformation syndromes, and less than 1% were mitochondrial and endocrine diseases.

Data analysis included frequency responses and comparison of responses to the questions on genetic discrimination by education, religious preference, and health of respondent and they showed no statistically significant differences (Pearson value of $P < 0.05$ was considered significant). Gender and ethnicity showed no significant differences when controlled for sample size.

Consumer experiences with health insurers were deemed important because the availability of affordable health insurance often determines who does and who does not have access to health care (*4*). For many people with genetic disorders, health insurance may mean the difference between life and death (*21*).

Although considerable genetic information may already be available to insurers in medical records, 40% of the respondents recalled being specifically asked about genetic diseases or disabilities on their applications for health insurance (Table 1). It cannot be assumed that the remaining 60% had not been asked questions about genetic diseases and disabilities. Many of them volunteered the information that they had never applied for health insurance. Some were able to maintain the coverage they had prior to diagnosis of a genetic disorder. Others had not applied because they assumed the genetic condition in the family would result in being turned down. Whether or not this information was then used to deny insurance to these people based on their genetic condition is not known.

Twenty-two percent of the respondents (Table 1) said that they or a family member were refused health insurance as a result of the genetic condition in the family. Since insurers do not need to provide reasons for turning down applications, it might be argued that respondents may have subjectively assumed that the denials were made because of the genetic condition. In this study, however, 83% of those who were refused health insurance had also been asked about genetic diseases or disabilities on their applications. Looked at in another way, nearly half (47%) of those who were asked about genetic diseases or disabilities on an application for health insurance were subsequently turned down. As health and life insurers are primarily regulated by states and most states are just beginning to address genetic issues in legislation (*22*), it is not known how many insurers actually ask genetic questions on applications.

The 31% of respondents with health insurance coverage who were denied reimbursement for some service or treatment indicated reasons such as the treatments were considered experimental, and services such as physical or occupational therapy were not considered a medical necessity. Time limits for submitting claims were also an issue, with insurers not paying claims that were more than a year old even when they had been submitted within the year and returned for more information. In several instances, payment was denied even though preapproval for a treatment or service had been given.

The large majority (83%) of respondents (Table 2) said they would not want their insurers to know if they were tested and found to be at high risk for a genetic disorder. The rate decreased to 78% when a similar question was asked that added the condition, "if the insurer pays for the tests." Some of the respondents noted that they would pay for genetic tests themselves or not be tested if they wanted to keep their genetic information confidential. The fear of genetic discrimination, as shown in Table 3, resulted in 9% of the respondents or a family member refusing to be tested for a genetic condition. This fear eliminates the opportunities of individuals to learn that they are not at increased risk for the genetic disorder in the family or to make life-style changes to reduce the risks or seriousness of the condition. It may also affect the number of people willing to participate in scientific research (*10*). Fear also prevented 18% of the respondents from revealing genetic information to an insurance company.

Approximately 70% of adults in the United States have some form of life insur-

Table 1. Questions and responses about experiences of consumers in areas of health insurance, life insurance, and employment. The total number of respondents is 332.

Question	Responses (%)		
	Yes	No	Don't know
As a result of the genetic condition in your family, have you or a member of your family been—			
Asked questions about genetic diseases or disabilities on an application for health insurance?	40	55	5
Refused health insurance?	22	76	2
Refused insurance coverage of some service or treatment?	31	67	2
Refused life insurance?	25	70	6
Asked questions about genetic diseases or disabilities on a job application?	15	83	2
Denied a job or let go from a job?	13	85	2

Table 2. Questions and responses to opinions about genetic information in insurance and employment.

Question	Responses (%)		
	Strongly agree or agree	Disagree or strongly disagree	Not sure or don't know
Genetic testing should be part of pre-employment physical exams.	4	94	2
Health insurers should be able to get genetic information if they pay for the tests.	16	78	6
	Yes	No	Not sure
If you were tested and found to be at high risk for a genetic disorder with serious complications, which of the following would you want to know the results of the test?			
a. Your employers?	6	87	7
b. Your insurance company?	11	83	6

ance (23). It is widely available, and only 3% of those who apply for coverage are declined. Of the 97% accepted, 5% are required to pay higher than standard premiums (24). This may be compared to the respondents in this study in which 25% (Table 1) of the respondents or affected family member have been refused life insurance (25).

Two questions were asked about the employment experiences of the study population. As noted in Table 1, 15% of the respondents said that they or affected family members had been asked questions about genetic diseases or disabilities on job applications. This increased to 20% of affected respondents ($P = 0.006$). It is not clear how often this information was used to subsequently deny the job to the applicants but the possibility exists and was of concern to respondents. In this study, 87% of respondents (Table 2) would not want their employers to know if they were tested and found to be at high risk for a genetic disorder with serious complications.

Thirteen percent of all respondents (Table 1) reported that they or another family member had been denied a job or let go from a job because of the genetic condition in the family. This was true for 21% of affected respondents and 4% of unaffected respondents ($P = 0.00001$). The percent was reduced to 9% ($P = 0.006$) for those with an affected child, even though a higher proportion of these respondents were in the workforce than the total population.

During the course of the analysis, a question was raised as to whether the perceived problems encountered in job application or denial or dismissal emanated from an employer's perception of a visible disability. To approach this question, analysis was done for the 77 unaffected respondents whose only affected family member was a child of less than 16 years of age. It was found that 7% of this population was asked about genetic diseases or disabilities on a job application and 3% were denied or let go from a job. These numbers should only be used as a starting point for future analyses.

For the affected respondents, some specific examples highlight the kinds of problems experienced. A man with a sex chromosome disorder reported that he had been denied a job following a pre-employment physical exam after the doctor wrote the name of the possible disorder on his medical report. The employer, in this case, knew it was illegal to use the diagnosis in the hiring decision and told the applicant that he would deny the conversation in the future if asked. A woman with a skeletal disorder reported that she was given termination notice the day after she informed her employer of a genetic diagnosis. The notice was withdrawn after she sought legal counsel. Examples provided by other respondents focused on effects of the genetic condition that could come under the protection of the 1995 interpretations of the Americans with Disabilities Act (26). The dilemma for persons with genetic disorders is that they must show not only that they have a genetic defect but also that they were regarded as "disabled" by an employer and discriminated against because of that perception. This raises concerns about the privacy and confidentiality of genetic information in the workplace.

A total of 17% have not revealed genetic information to their employers (Table 3) for fear of losing their jobs or insurance coverage. This increased to 25% of affected respondents ($P = 0.00001$). Overall, 43% of the respondents reported that they or members of their family have experienced genetic discrimination in one or more of the three areas. This included health insurance only (9%), life insurance only (11%), employment only (6%), and more than one category (17%).

Additional studies of persons with genetic disorders are indicated to confirm or deny the perceptions of the consumers in this study. It is possible that members of genetic support groups who have experienced genetic discrimination may have been more motivated to volunteer for this study. On the other hand, persons with these resources of higher education and membership in support groups traditionally have the skills and means to work with and influence social systems and may have experienced less discrimination than other groups. With adequate funding, a random sampling of respondents from support group or clinic populations could be selected with probability methods and objective as well as subjective information could be gathered.

Another goal would be to design more detailed questions to elicit information on genetic discrimination from respondents. Distinctions between the implications of overt genetic disease and conditions on each person and the effects on unaffected family members, or persons who are carriers or do not overtly express the consequences of the genetic condition will require further study. Consumers may be willing to participate if confidentiality is assured and trust is established. In this study, it was also found important for the interviewers to have clinical as well as technical skills in interviewing to facilitate the comfort level of discussing sensitive issues. This would also be recommended for future studies.

Although the goal of the HGP (and other genetic testing and research) is to help people, it could also cause harm if the level of perceived discrimination is in fact true. Neither the authors nor the respondents (as indicated in earlier responses) are suggesting that the HGP should not continue. On the contrary, there is strong support to continue research and to find ways to deal with genetic discrimination including federal or state legislation, guidelines, and standards among insurers, employers, researchers, and health professionals, and citizen advocacy to establish protections.

Table 3. Percentage of respondents withholding information or refusing to be tested for a genetic condition as result of fear.

Question	Responses (%)		
	Yes	No	Don't know
As a result of a genetic condition, have you or a member of your family—			
Refused to be tested for a genetic condition for fear of your insurance coverage being dropped.	9	89	2
Not revealed genetic information to an insurance company.	18	79	3
Not revealed genetic information to an employer.	17	81	2

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12. See, for example, P. R. Billings et al., Am. J. Hum. Genet. 50, 476 (1992).
13. As the interviews progressed and it was apparent that few members of ethnic or racial minorities were volunteering, additional telephone outreach was made to support groups to increase awareness of the project and opportunities for volunteering. This was only partially successful as seen in the demographic data.
14. There were no statistically significant differences in the responses on the telephone and written interviews so they are reported here together.
15. Of the 332 respondents, 80% were female, 57% have at least a bachelors degree, and 90% were Caucasian. Other characteristics were: 75% were married and living with their spouses, 76% have children, and 63% work outside the home. Family relationship, whether or not the respondent or family members were affected, age of diagnosis, and current age were also recorded.
16. Precise data on the demographics of genetic support groups are not available. Impressions are from staff of the Alliance of Genetic Support Groups based on their conversations and communications with the member organizations and attendance at national, regional, and local meetings.
17. Religious preferences were Roman Catholic, 26%; Protestant, 41%; Christian-other, 9%; Jewish, 11%; other, 2%; and none, 12%.
18. The interviewers were trained in interview techniques by the principal investigator and participated in pretesting the questionnaire.
19. The questions on possible genetic discrimination were taken from a questionnaire developed by Dr. Dorothy C. Wertz, The Shriver Center, Waltham, MA, entitled, Ethical Issues in Genetics, Part I, p. 33, No. 34, and used with permission of Dr. Wertz (letter of 16 December 1993).
20. This definition of genetic discrimination differs from the one used by Billings et al. (12) as they did not include actions against persons who were symptomatic or visibly affected by their genetic disorders. The design of our questionnaire does not permit analysis according to the definition of Billings et al. (1993). Because the questions on discrimination ask about all family members at once, the questions do not distinguish among: (i) the direct consequences of ongoing genetic disease or conditions, (ii) the effect of genetic disease on other family members, and (iii) the consequences of genetic information gained through testing.
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25. Many respondents said they had never applied for life insurance because they assumed they would be turned down.
26. Since 1990, the Americans with Disabilities Act (ADA) has provided protection for persons with disabilities in the workplace. The ADA prevents employers from openly denying employment or firing an individual solely on the basis of a "disability" if there are "reasonable accommodations" that can be made in the work setting to allow the person to perform his or her job. In April 1995, the ADA was interpreted by the U.S. Equal Employment Opportunity Commission to include healthy people who are carriers of genetic disorders. Implementation in general relies on employers and employees knowing and similarly interpreting the law as well as having good faith efforts to comply.
27. We wish to thank the HuGEM Advisory Committee: N. A. Holtzman, F. Neal-Smith, E. O. Nightingale, K. H. Rothenberg, and L. Walters, for assistance in planning and carrying out the study. We also wish to thank L. Palincsar, M. A. Wilson, S. Rennert, and M. Volner, for their assistance with the telephone interviews and M. J. Verdieck and M. O'Reilly for research assistance. Most of all, we wish to thank the consumers who provided the data. Funded by the National Institutes of Health at the National Center for Human Genome Research in the branch of Ethical, Legal and Social Issues, grant number RO1-HG00786-03. This support does not constitute an endorsement of the views expressed in this article.

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Kap104p: A Karyopherin Involved in the Nuclear Transport of Messenger RNA Binding Proteins

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A cytosolic yeast karyopherin, Kap104p, was isolated and shown to function in the nuclear import of a specific class of proteins. The protein bound directly to repeat-containing nucleoporins and to a cytosolic pool of two nuclear messenger RNA (mRNA) binding proteins, Nab2p and Nab4p. Depletion of Kap104p resulted in a rapid shift of Nab2p from the nucleus to the cytoplasm without affecting the localization of other nuclear proteins tested. This finding suggests that the major function of Kap104p lies in returning mRNA binding proteins to the nucleus after mRNA export.

Transport across the nuclear envelope occurs through nuclear pore complexes (NPCs) and is governed by the interaction of soluble transport proteins (karyopherins) with the transport substrate and the NPC (*1–12*). Most of our understanding of the mechanism of translocation comes from studying protein import in semipermeablized cells (*1*) of model karyophilic proteins that carry a nuclear localization signal (NLS) from either the SV40 large T antigen or nucleoplasmin (*2*). These classical NLSs are recognized by karyopherin α in a dimeric cytosolic complex with karyopherin β (*3–8*). The complex docks at the NPC through its interaction with nucleoporins that contain characteristic repeated peptide motifs (*6–11*). The small guanosine triphosphatase, Ran, and p10 are required for the subsequent translocation of the substrate (and karyopherin α) through the NPC (*11, 12*).

Distinct saturable and noncompeting pathways for the import of different karyophiles have been uncovered through the use of microinjection studies in oocytes (*13–15*). Similarly, saturable noncompeting pathways exist for the export of macromolecules from the nucleus (*14, 16, 17*). The signals that mediate many of these processes are different from classical NLSs (*14, 15, 17–19*) and thus may use recognition factors other than karyopherin α and karyopherin β for nuclear transport. Here we characterize the first such factor, which we term Kap104p and which is required for the import of at least two yeast nuclear mRNA binding proteins.

The *Saccharomyces cerevisiae* proteins Kap60p and Kap95p are homologs of mammalian karyopherin α and karyopherin β (*20*). Sequence comparisons of Kap95p with the complete yeast genome database uncovered three additional proteins that are structurally similar to Kap95p; two of these, which we term Kap123p and Kap104p, have not been previously characterized (*21*), and the third, Pse1p, was identified as a multicopy enhancer of protein secretion (*22*). The sequence alignment of Kap104p with Kap95p is shown (Fig. 1A). The proteins bear substantial similarity over their entire lengths, and secondary structural predictions suggest that Kap95p and Kap104p share the same overall domain structure of HEAT motifs (*23*).

Deletion of *KAP104* resulted in a severe growth defect and temperature sensitivity (*24*). Immunofluorescence microscopy (*25*) with antibodies specific for Kap104p (in wild-type cells) showed that Kap104p was mainly cytosolic and was apparently absent from the nucleus (Fig. 1B). However, in *nup120Δ* cells, which cluster their NPCs to a region of the nuclear envelope opposite the nucleolus (*26*), Kap104p colocalized with the nucleoporin Nsp1p (*27*) (Fig. 1C). The ability to detect coincident staining of the nucleoporins and Kap104p under these conditions likely was due to an interaction of Kap104p with NPCs.

Subcellular fractionation (*28*) was consistent with the distribution of Kap104p detected by immunofluorescence. Kap104p was present mainly in the cytosolic fraction,

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Flunk the Gene Test and Lose Your Insurance

THE GORRELLS
Jane (front) and her daughter Danielle know they share a gene associated with colon cancer

DNA probes may offer dazzling insights. But they have helped foster a pernicious new form of discrimination.

BY GEOFFREY COWLEY

JAMIE STEPHENSON HAS SEEN FIRSThand what modern genetic science can do for a family. When her son David was 2 years old, a pediatrician noticed developmental delays and suspected fragile X syndrome, a hereditary form of mental retardation. A lab test confirmed the diagnosis, and the Stephensons spent several years learning to live with it. When David was 6, he visited a neurologist, who scribbled "fragile X" on an insurance-company claim form. The company responded promptly—by canceling coverage for the entire family of six. There is no medical treatment for fragile X, and none of David's siblings had been diagnosed with the condition. "The company didn't care," Stephenson says. "They just saw a positive genetic test and said, 'You're out'."

From the dawn of the DNA era, critics have worried that genetic testing would create a "biological underclass"—a population of people whose genes brand them as poor risks for employment, insurance, even marriage. The future is arriving fast. Med-

KEN SCHLES

ical labs can now test human cells for hundreds of anomalous genes. Besides tracking rare conditions, such as Huntington's disease and cystic fibrosis, some firms now gauge people's susceptibility to more common scourges. By unmasking inherited mutations in p53 (main story) and other genes, the new tests can signal increased risk of everything from breast, colon and prostate tumors to lymphoma and leukemia. Many of the tests are still too costly for mass marketing, but that will change. And as the Stephensons' story suggests, the consequences won't all be benign. "This is bigger than race or sexual orientation," says Martha Volner, health-policy director for the Alliance of Genetic Support Groups. "Genetic discrimination is the civil-rights issue of the 21st century."

No one would argue that genetic tests are worthless. Used properly, they can give people unprecedented power over their lives. Prospective parents who discover they're silent carriers of the gene for a disease like Tay-Sachs, which causes death by the age of 3, can make better-informed decisions about whether and how to have kids. Some genetic maladies can be managed through medication and lifestyle changes once they're identified. And while knowing that you're at special risk for cancer may be an emotional burden, it can also alert you to the need for intensive monitoring. Jane Gorrell knows her family is prone to colon cancer. Her father developed hundreds of precancerous polyps back in the 1960s, and both she and her sister had the same experience during the '70s. Their condition, known as familial adenomatous polyposis, has since been linked to a mutation in the p53 gene—and Gorrell has learned that one of her two children inherited it. Though the child has suffered no symptoms, she gets frequent colon exams and is helping researchers test a drug that could help save lives.

The catch is that no one can guarantee the privacy of genetic information. Outside of large group plans, insurance companies often scour people's medical records before extending coverage. And though employers face some restrictions, virtually any company with a benefits program can get access to workers' health data. So can schools, adoption agencies and the military. Employees of Lawrence Berkeley Laboratory (LBL), a large research institution owned by the Department of Energy and operated by the University of California, recently discovered that the organization had for three decades been quietly testing new hires' blood and urine samples for evidence of various conditions. They claim blacks were screened for the sickle-cell trait, Latinos for syphilis and women for pregnancy. "I can't say the information was put to some incredibly harmful use, because we don't know what happened," says Vicki Laden, a San Francisco lawyer who has tried unsuccessfully to sue the lab for civil-rights violations. LBL recently stopped the testing.

How often is genetic information used against people? No one knows, but there are signs that discrimination is fairly common—even in the absence of sophisticated tests. In one recent study, researchers led by Georgetown University sociologist Virginia Lapham surveyed 332 families belonging to genetic-disease support groups; 22 percent of the respondents said they'd been refused health insurance, and 13 percent claimed their perceived risks had cost them jobs.

In another 1996 study, a team led by Lisa Geller of Harvard Medical School documented more than 200 instances in which healthy people experienced genetic discrimination. As in Lapham's study, many participants told of losing their health and life insurance. But that wasn't their only problem. One respondent was denied a job *selling* insurance after he disclosed that he had hemachromatosis, a hereditary iron-storage problem that can be treated for about $1,200 a year. A social worker who had excelled during her first year at a Wisconsin health-care company was quickly forced out after mentioning that her uncle had Huntington's, a degenerative brain disease that victims' offspring have a 50 percent chance of developing. Carol Isaacson Barash, the bioethics consultant who administered the Geller study, recalls another case in which a woman in the early stages of Huntington's disease was unable to place her child with a private adoption agency. A public agency took the child, eventually matching her with a couple who had previously been rejected themselves—because one partner carried the Huntington's gene. "It's a stark commentary on how society regards people at risk," says Barash.

Not surprisingly, people from high-risk families have come to fear tests almost as much as disease. Lori Andrews, a Chicago law professor and former head of a federal task force on the social implications of genetic research, notes that only 15 percent of people with a Huntington's afflicted parent choose to learn their own status. Discrimination isn't their only concern; virtually everyone testing positive for the Huntington's gene develops debilitating symptoms during middle age, and doctors can do nothing to help. But people who might benefit from genetic tests are almost as leery. When researchers at the Georgetown University Medical Center surveyed 279 people from families plagued by breast and ovarian cancer, only 43 percent wanted to be tested for hereditary mutations in BRCA1, a recently discovered gene that is often implicated in those diseases. Many said the prospect of discrimination scared them off. Kendra McCarthy, a 47-year-old administrator at the Virginia Department of Mental Health, saw her mother die of breast cancer at 34, her father of esophageal cancer at 41. When scientists developed tests that might have gauged her own susceptibility, she always declined to take one. McCarthy doesn't regret that decision. She has developed

"Genetic testing has the potential to revolutionize medicine. But revolutions can have casualties."

—FRANCIS COLLINS
Director of the Human Genome Project

THE STEPHENSONS
When an insurance company learned that David (left) had fragile X syndrome, it canceled the whole family's health coverage

Information, Please

One of the wonderful things about science is that there are no stupid questions. Nobel Prize winners ask the same ones you do. Who cares if they are working to cure deadly diseases while you're only trying to make sense of the science pages. A primer:

What is a gene?

The basic unit of heredity, a gene consists of an ordered sequence of chemicals called nucleotides and designated by the letters A, T, C or G. A gene is situated on one of the 23 pairs of human chromosomes. It carries instructions for the production of proteins. The sequence of nucleotides serves as a chemical code, determining which protein the gene makes. Genes take the form of a double helix.

Where are genes?

Every cell in the body contains exactly the same genes (roughly 100,000). We have two copies (identical or just similar) of every gene. The only exceptions are sperm and egg cells, which contain one for each chromosome pair rather than both, and red blood cells. What makes one cell different from another (liver from muscle, for instance) is not their genes—which are identical—but which of the genes are turned on.

What is a mutation?

Any change in the normal sequence of nucleotides in a gene. A mutation can be the deletion of a nucleotide or the change of one nucleotide to another.

What causes a mutation?

A mutation can be inherited; if the egg or sperm cell that produced the child contained a mutation, so will every one of the child's 30 trillion cells. A mutation can also be caused by radiation, by "mutagens" (chemicals, such as benzopyrene, found in cigarette smoke) or by some viruses.

What happens if a gene is mutated?

It no longer produces the protein it was designed to. Since a cell has two copies of each gene, often it can get by with only one healthy version, and a mutation has no medical consequences. But if the mutation occurs in an egg or sperm cell, it can be passed on to children. And if both copies are mutated, or if the cell needs both copies to function, then the cell is no longer able to carry out its job.

KENDRA MCCARTHY
Though cancer stalks her family, she shunned genetic testing. 'If prevention was possible, I would say it makes sense.'

breast cancer, but she still has her health coverage, and a life-insurance plan to protect her two sons.

Besides depriving people of potentially useful information, the fear of discrimination can hamper scientific progress. Barbara Weber, a geneticist with the University of Pennsylvania Cancer Center, often asks women who learn they have BRCA1 mutations to take part in confidential follow-up studies. She wants to know which strategies are most effective for preserving their health. "We have the tools to answer these questions," she says. "All they have to do is tell us how they're doing every six months." Yet three fourths of the women she approaches say no.

The federal government is now taking steps to make genetic information less threatening. The recently enacted Health Insurance Portability and Accountability Act (better known as Kennedy-Kassebaum) bars insurers from treating genetic mutations as "pre-existing conditions" unless they're causing illness. The insurance act also guarantees coverage to anyone leaving one group plan for another, whatever his pre-existing conditions. That will make switching jobs easier for many people, but ethicists say the act is only a first step. Because it covers only group plans and doesn't deal with disability insurance, it won't do much for folks like Theresa Morelli, an Ohio lawyer who applied for independent coverage several years ago. Morelli was 28 and in perfect health when she met with an insurance agent and paid her first premium. A month later, she got her check back, along with a letter saying her application had been denied because her father had Huntington's disease. Morelli's dad had in fact received that diagnosis—erroneously, it turned out—and her doctor had made a note of it in her chart.

The restrictions on employers are as porous as those on insurers. The 1990 Americans with Disabilities Act bars companies from discriminating against people with disabilities—and it defines that term broadly enough to include genetic mutations that have yet to cause symptoms. But the act does nothing to keep employers from gathering medical information. "As long as employers have access to genetic information," says Lewis Maltby of the ACLU, "they'll have an irresistible incentive to use it."

Lawmakers are now racing to strengthen the protections. At least 15 states have recently placed restrictions on insurers or employers, and Congress will consider several bills in the new session. The insurance industry argues that it should be free to charge people rates that reflect their risks, at least when dealing with individuals and small companies. That way, says Richard Coorsh of the Health Insurance Association of America, each applicant pays the fairest possible price. But most ethicists contend that where health coverage is concerned, people shouldn't be penalized for risks they can't modify.

Discrimination isn't the only potential downside to genetic testing. Some of the new susceptibility tests are only vaguely predictive of illness—and no one forces the companies that offer them to counsel patients about what the results actually mean. Even when the tests are sound and the results secure, the knowledge they create can dash hopes and divide families. Unfortunately, principled ignorance doesn't always make life easier.

With ANNE UNDERWOOD *in New York,* KAREN SPRINGEN *in Chicago and* MARY HAGER *in Washington*

05/16/97 FRI 12:40 FAX 301 402 0837 NCHGR/OD

6

Advent of Testing for Breast Cancer Genes Leads to Fears of Disclosure and Discrimination

C-1 NYT 2/4/97

By GINA KOLATA

DR. BARBARA WEBER is still reeling from the experience of a patient who told her insurance company that she had a mutated gene that could cause breast cancer.

The woman had been tested for the mutation at Dr. Weber's clinic at the University of Pennsylvania, and Dr. Weber told her the results. Since some studies indicate that women with the mutated gene have a 80 percent chance of developing breast cancer, the woman wanted both of her breasts removed right away. Before she had the operation, she submitted a claim to her insurance company, Dr. Weber said, not disclosing that she had had the genetic test but reporting a strong family history of breast cancer.

The company turned her down, Dr. Weber said, on the ground that it did not pay for preventive medicine. So, at the woman's request, Dr. Weber submitted the woman's genetic test results. At that point, Dr. Weber said, the company told the woman that it would not pay for the surgery because she had a preexisting condition — a genetic defect — when she took out her health policy.

"It was absolutely unbelievable," Dr. Weber said. She said that the woman, who did not return telephone calls requesting an interview, had the surgery anyway. Afterward, Dr. Weber said, when pathologists examined the woman's breast tissue, they found a cancerous tumor that had been missed by mammograms.

Gene Wilkinson for The New York Times

Dr. Barbara L. Weber says patients who get gene testing are refusing to enroll in studies for fear of discrimination.

It is cases like this and fears of similar treatment that are convincing some women and researchers that it might be too dangerous to put genetic testing results on medical charts and in clinical records, where privacy cannot be assured.

Women worry that insurers will raise their rates, or refuse to insure them, that employers will not hire them or promote them, and even that friends and family members might treat them differently if they knew that they were tainted with a deadly gene.

Fears of genetic testing have been expressed by some people for several years, starting with tests for rare genetic diseases like Huntington's. But the recent discovery of two genes that can cause breast and ovarian cancer when mutated has made the issue more pressing.

Not only is breast cancer the disease that women fear the most, but the mutations, in genes known as BRCA1 and BRCA2, are relatively common in Ashkenazi Jewish populations (of Eastern European descent), affecting as many as 1 in 50 women.

In addition, many women who are not Jewish but have a strong family history of breast or ovarian cancer also inherited mutated genes. In the last year, several commercial laboratories have begun offering tests for breast cancer gene mutations and major medical centers across the country have set up their own testing programs.

Some legal experts say the fears of discrimination may be exaggerated. Federal legislation that

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will go into effect on July 1 addresses part of the issue, focusing on people who are covered under group medical plans. The law states that if an individual was in such a plan for at least 12 months and had a genetic condition diagnosed in the past 6 months, a new insurer cannot use that genetic information to deny or limit coverage.

Alexander Morgan Capron, an ethicist and law professor at the University of Southern California, said insurers could charge people with genetic conditions more for insurance. But, Mr. Capron added, 29 states including, New Jersey, have recently passed laws preventing health maintenance organizations and health insurance companies from charging people more because they have a gene mutation. In addition, the Americans With Disabilities Act prevents employers from discriminating against people who have disease-causing genetic mutations. Employers who insure their employees are exempt from the state laws, Mr. Capron said.

Richard Coorsh, a spokesman for the Health Insurance Association of America, said that people's fears that insurance companies would discriminate against them might be overblown. Many people are insured through their employers as part of large groups, and they would be covered even if they did have a cancer-causing gene.

But Mr. Coorsh said that those who change jobs or lose their jobs and have to seek insurance as individuals "will probably end up having to pay a lot more money" for health insurance if they test positive for a cancer gene. There have been few systematic studies to assess how real the risk of genetic discrimination is, but one survey, published in October in the journal Science, showed that the fear, at least, is widespread.

The study, by Dr. E. Virginia Lapham of the Georgetown University School of Medicine and her colleagues, involved 332 people who belonged to support groups for families with a variety of genetic disorders. Of them, 25 percent believed they were denied life insurance because of their disorder, 22 percent thought they were denied health insurance, and 13 percent believed they were not hired for a job or that they lost a job because of the disorder.

Whether or not these suspicions are justified, they are having an effect. Some researchers at major medical centers are worried enough to work under a cloak of secrecy. They test for the breast-cancer genes is under the aegis of a research program, which allows doctors to keep the results secret, coded and encased in locked files.

Some researchers advise women who are tested not to tell even their private doctor of their test results if the doctor insists on putting all relevant information in a woman's medical record. Some doctors agree not to write down test results, relying instead on their memories.

At treatment centers, which, unlike research centers, are not permitted to hide test results, some women have used aliases to protect their privacy.

"If we ever needed proof that the system is broken, this is it," said Dr. Francis Collins, director of the National Center for Human Genome Research in Bethesda, Md. "The system forces people to take drastic steps to protect themselves. It is putting a terrible burden on patients."

Doctors, too, are placed in an untenable situation, Dr. Collins said. "You are forced sometimes to have interactions with insurance agents or with other physicians or with H.M.O.'s where you would have to pretend you don't have the information," Dr. Collins said. "It's a very strange dilemma — to choose between patient confidentiality and telling the truth."

It is, said Dr. Thomas Murray, the director of the Center for Biomedical Ethics at Case Western Reserve University in Cleveland, "a classic ethical quandary," and one with no obvious solution.

Moreover, some cancer researchers note, there is a legitimate reason why insurers and employers might want to avoid people with genes that can cause cancers: the insurers because of the extra cost of treating them and the employers because of the extra insurance cost and because of lost productivity.

"I'm not sure you can totally blame insurance companies or employers," said Dr. C. Kent Osborne, a cancer researcher at the University of Texas in San Antonio. "It's a bigger problem than that. It's something we have to deal with as a society."

In the meantime, doctors and patients are trying to act expediently.

Dr. Funmi Olopade, who directs the breast cancer genetic testing program at the University of Chicago, was interviewed just after an appointment with a 25-year-old woman, who came to learn her test results. The woman was found to have a genetic mutation that can cause breast and ovarian cancer. The university keeps these records secret.

But, Dr. Olopade said, the woman asked her if she could tell her private doctor of her test and what it disclosed.

Dr. Olopade suggested that the young woman start by asking her doctor if he felt he had to put everything he knew about a patient into the medical records. If so, Dr. Olopade said, the woman might not want to tell.

And with good reason, said Dr. Mark Siegler, who directs the ethics program at the University of Chicago. More than a decade ago, before the proliferation of electronic records, before the merging of health maintenance organizations that both pay for and care for patients, he investigated the complaint of a woman who had come to the hospital for elective gall bladder surgery and had said that too many people were looking at her medical records.

"I started to ask how many people had legitimate access to her chart," Dr. Siegler said. "I stopped counting at 75." He concluded, he said, "that confidentiality really didn't exist." That is even more true today, he added.

Some doctors, like Dr. John Glick, the director of the University of Pennsylvania's cancer center, say they will not put genetic testing information in a woman's medical records if she asks that it be kept out. Instead, Dr. Glick said, he simply remembers what the woman said about her test results.

Other doctors refuse to go along with such requests. One woman, who tested positive for the breast cancer gene at the University of Pennsylvania, said her private doctor told her it would be dishonest not to include the test result in her medical records. So, the woman said, "I asked that my medical records be closed and I changed doctors."

The woman said she was worried about health insurance. "The company my husband works for just got bought out, so things aren't as steady as they used to be," she said. "You can survive breast cancer, but you can't survive if you don't have insurance."

Dr. Henry Lynch, who directs the

Some doctors won't put results of genetic tests in a woman's records...

cancer center at Creighton University in Omaha, tests women for the breast cancer gene as part of a research program in which he can keep the results secret, and as part of a clinical program, where he cannot. But some women in his clinical program use aliases. Others pay for tests out of their own pockets, hoping to keep the information out of the hands of their insurance companies.

Dr. Weber said that she was confronting a new ramification of women's fears of discrimination. Women who were tested as part of her research program are refusing to participate in long-term studies that would assess their risk of developing cancer and the success of interventions, like frequent mammograms, or hormone treatments, or surgery to remove their breasts or ovaries.

The women, Dr. Weber said, fear that it would be a red flag on their medical records to participate in the study. Their mammogram results and any biopsies, for example, would have to be sent to the university's research program.

The only possibility of learning if there is a way to prevent cancer in women who are genetically predisposed, Dr. Weber said, is to do long-term studies. If no one will participate, she said, what is the point in testing women to see if they have the cancer genes?

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
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(INCLUDING COVER)

HHS draft bill - Insurance & genetic discrimination

REMARKS

Changes to version circulated yesterday are in bold print. Has been sent to OMB. Still waiting for DOL insert, page 4.

Judy

A BILL

To prohibit discrimination against individuals and their family members on the basis of genetic information, or a request for genetic services.

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 Information Nondiscrimination in Health Insurance Act of 1997".

SEC. 2. AMENDMENTS TO EMPLOYEE RETIREMENT INCOME SECURITY ACT OF 1974.

(a) PROHIBITION OF HEALTH INSURANCE DISCRIMINATION ON THE BASIS OF REQUEST FOR OR RECEIPT OF GENETIC SERVICES.

(1) NO ENROLLMENT RESTRICTION FOR GENETIC SERVICES.—Section 702(a)(1)(F) of the Employee Retirement Income Security Act of 1974 (29 U.S.C. 1182(a)(1)(F)) is amended by inserting before the period "or request for or receipt of genetic services".

(2) NO DISCRIMINATION IN GROUP RATE BASED ON GENETIC INFORMATION.—Section 702(b)(1) of such Act (42 U.S.C. 1182(b)(1)) is amended—

(A) by striking "may not require any individual"

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1 and inserting "may not—

2 (A) require any individual";

3 (B) by moving the remainder of the paragraph two

4 ems to the right;

5 (C) by striking the period and adding "; or"; and

6 (D) by adding at the end the following new

7 subparagraph:

8 **"(B) adjust premium or contribution rates for a**

9 **group or the individuals in a group on the basis of**

10 **genetic information concerning or a request for or**

11 **receipt of genetic services by any member of such group**

12 **or a covered dependent of such member.".**

13 (b) LIMITATION ON COLLECTION AND DISCLOSURE OF GENETIC

14 INFORMATION.—Section 702 of such Act (29 U.S.C. 1182) is amended

15 by adding after subsection (b) the following new subsection:

16 "(c) WITH RESPECT TO COLLECTION AND DISCLOSURE OF GENETIC

17 INFORMATION.—

18 "(1) COLLECTION.—Except as provided in paragraph (3), a

19 group health plan, or a health insurance issuer offering

20 health insurance coverage in connection with a group health

21 plan, shall not request or require disclosure of genetic

22 information concerning an individual who is a participant or

1 beneficiary (or an applicant for coverage as a participant
2 or beneficiary).
3 "(2) DISCLOSURE.—Except as provided in paragraph (3)—
4 "(A) a group health plan, or a health insurance
5 issuer offering health insurance coverage in connection
6 with a group health plan, shall not disclose genetic
7 information about an individual who is a participant or
8 beneficiary (or an applicant for coverage as a
9 participant or beneficiary) to any other health plan or
10 health insurance issuer; and
11 "(B) a physician, hospital, or other person that
12 provides health care items or services to an individual
13 shall not disclose genetic information about such
14 individual to any group health plan, or health
15 insurance issuer offering health insurance coverage in
16 connection with a group health plan.
17 "(3) EXCEPTIONS.—
18 "(A) The provisions of paragraphs (1) and (2)
19 shall not apply to a request by or disclosure to a
20 health plan or issuer that provides reasonable
21 assurances that it provides health insurance coverage
22 to such individual and requires such information for

1 payment of claims or coordination of benefits.

2 "(B) Notwithstanding paragraphs (1) and (2), a

3 health plan or issuer that provides health care items

4 and services and provides reasonable assurances that it

5 requires such information for diagnosis or treatment of

6 such individual may—

7 (i) request (but not require) disclosure of

8 such information by the individual; and

9 (ii) request or require such information from

10 another health plan, or health insurance issuer,

11 or provider of health care items and services.

12 "(C) The provisions of paragraphs (1) and (2)

13 shall not apply if the individual, or the individual's

14 legal representative, provides prior written

15 authorization of such disclosure.".

16 [NOTE: DOL IS DRAFTING LANGUAGE THAT WILL PROHIBIT PLAN

17 FIDUCIARIES FROM DISCLOSING GENETIC INFORMATION TO

18 EMPLOYERS/PLAN SPONSORS.]

19 (c) DEFINITIONS.—Section 706(d) of such Act (29 U.S.C.

20 1191b(d)) is amended by adding at the end the following new

21 paragraphs:

22 "(5) FAMILY MEMBER.—The term 'family member' means,

with respect to an individual, a spouse or adopted child of that individual, or another individual related by blood to that individual or to a spouse or adopted child of that individual.

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

"(7) GENETIC SERVICES.—The term `genetic services' means health services provided to obtain, assess, and interpret genetic information for diagnostic and therapeutic purposes, and for genetic education and counselling.".

(d) EFFECTIVE DATE.—The amendments made by this section shall apply with respect to group health plans and group health insurance coverage for plan years beginning after 1 year after the date of the enactment of this Act.

SEC. 3. AMENDMENTS TO THE PUBLIC HEALTH SERVICE ACT.

(a) AMENDMENTS RELATING TO THE GROUP MARKET.—

(1) PROHIBITION OF HEALTH INSURANCE DISCRIMINATION ON THE BASIS OF REQUEST FOR OR RECEIPT OF GENETIC SERVICES.—

(A) NO ENROLLMENT RESTRICTION FOR GENETIC SERVICES.—Section 2702(a)(1)(F) of the Public Health

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1 Service Act (42 U.S.C. 300gg-1(a)(1)(F)) is amended by
2 inserting before the period "or request for or receipt
3 of genetic services".
4 (B) NO DISCRIMINATION IN GROUP RATE BASED ON
5 GENETIC INFORMATION.—Section 2702(b)(1) of such Act (42
6 U.S.C. 300gg-1(b)(1)) is amended—
7 (i) by striking "may not require any
8 individual" and inserting "may not—
9 (A) require any individual";
10 (ii) by moving the remainder of the paragraph
11 two ems to the right;
12 (iii) by striking the period and adding ";
13 or"; and
14 (iv) by adding at the end the following new
15 subparagraph:
16 **"(B) adjust premium or contribution rates for a**
17 **group or the individuals in a group on the basis of**
18 **genetic information concerning or a request for or**
19 **receipt of genetic services by any member of such group**
20 **or a covered dependant of such member.".**
21 (2) LIMITATION ON COLLECTION AND DISCLOSURE OF GENETIC
22 INFORMATION.—Section 2702 of such Act (42 U.S.C. 300gg-1) is

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1 amended by adding after subsection (b) the following new
2 subsection:
3 "(c) WITH RESPECT TO COLLECTION AND DISCLOSURE OF GENETIC
4 INFORMATION.—
5 "(1) COLLECTION.—Except as provided in paragraph (3), a
6 group health plan, or a health insurance issuer offering
7 health insurance coverage in connection with a group health
8 plan, shall not request or require disclosure of genetic
9 information concerning an individual who is a participant or
10 beneficiary (or an applicant for coverage as a participant
11 or beneficiary).
12 "(2) DISCLOSURE.—Except as provided in paragraph (3)—
13 "(A) a group health plan, or a health insurance
14 issuer offering health insurance coverage in connection
15 with a group health plan, shall not disclose genetic
16 information about an individual who is a participant or
17 beneficiary (or an applicant for coverage as a
18 participant or beneficiary) to any other health plan or
19 health insurance issuer; and
20 "(B) a physician, hospital, or other person that
21 provides health care items or services to an individual
22 shall not disclose genetic information about such

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individual to any group health plan, or health insurance issuer offering health insurance coverage in connection with a group health plan.

"(3) EXCEPTIONS.—

"(A) The provisions of paragraphs (1) and (2) shall not apply to a request by or disclosure to a health plan or issuer that provides reasonable assurances that it provides health insurance coverage to such individual and requires such information for payment of claims or coordination of benefits.

"(B) Notwithstanding paragraphs (1) and (2), a health plan or issuer that provides health care items and services and provides reasonable assurances that it requires such information for diagnosis or treatment of such individual may—

(i) request (but not require) disclosure of such information by the individual; and

(ii) request or require such information from another health plan, or health insurance issuer, or provider of health care items and services.

"(C) The provisions of paragraphs (1) and (2) shall not apply if the individual, or the individual's

1 **legal representative, provides prior written**
2 **authorization of such disclosure.".**
3 (3) DEFINITIONS.—Section 2791(d) of such Act (42 U.S.C.
4 300gg-91(d)) is amended by adding at the end the following
5 new paragraphs:
6 "(15) FAMILY MEMBER.—The term `family member' means,
7 with respect to an individual, a spouse or adopted child of
8 that individual, or another individual related by blood to
9 that individual or to a spouse or adopted child of that
10 individual.
11 "(16) GENETIC INFORMATION.—The term `genetic
12 information' means information about genes, gene products,
13 or inherited characteristics that may derive from an
14 individual or a family member.
15 "(17) GENETIC SERVICES.—The term `genetic services'
16 means health services provided to obtain, assess, and
17 interpret genetic information for diagnostic and therapeutic
18 purposes, and for genetic education and counselling.".
19 (b) AMENDMENT RELATING TO THE INDIVIDUAL MARKET.—Subpart
20 3—Other Requirements of part B of title XXVII of such Act is
21 amended by inserting after section 2751 (42 U.S.C. 300gg-51) the
22 following new section:

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1 **"SEC. 2752. PROHIBITION OF HEALTH INSURANCE DISCRIMINATION ON**
2 **THE BASIS OF GENETIC INFORMATION.**
3 "(a) IN GENERAL.—Except as provided in subsection (b), the
4 prohibitions of—
5 "(1) discrimination based on genetic information or
6 request for or receipt of genetic services, and
7 "(2) collection of genetic information by a health
8 insurance issuer, or disclosure of genetic information by or
9 to such an issuer,
10 shall apply to a health insurance issuer offering coverage in the
11 individual market to the same extent as they apply pursuant to
12 section 2702 to a health insurance issuer subject to section
13 2702.
14 "(b) EXCEPTION.—The provisions of subsection (a) shall not
15 prevent a health insurance issuer subject to subsection (a) from—
16 "(1) requesting or requiring disclosure of genetic
17 information about an individual who is a participant or
18 beneficiary (or an applicant for coverage as a participant
19 or beneficiary) relating to a disease or condition for which
20 the individual has been positively diagnosed or has received
21 treatment at any time;
22 "(2) requesting an individual, as a condition for

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1 **initial enrollment, to undergo a physical examination or**
2 **related tests to determine whether the individual has a**
3 **disease or condition; or**
4 "(3) using information specified in paragraph (1) or
5 the results of an examination or test specified in paragraph
6 (2) to deny or vary the terms and conditions of health
7 insurance benefits or coverage.".
8 (c) AMENDMENT CONCERNING MEDIGAP.—Section 2791(c)(4) of such
9 Act (42 U.S.C. 300gg-91(c)(4)) is amended by inserting "except
10 for purposes of sections 2702(a)(1)(F), 2702(c), and 2752" after
11 "Social Security Act)".
12 (d) TECHNICAL AMENDMENT.—Title XXVII of such Act is amended
13 in the subpart heading following section 2744 (42 U.S.C. 300gg-
14 44) by striking "Subpart 3" and inserting "Subpart 2".
15 (e) EFFECTIVE DATE.—The amendments made by subsections (a)
16 through (d) shall apply with respect to—
17 (1) group health plans and group health insurance
18 coverage for plan years beginning, and
19 (2) health insurance available or in effect in the
20 individual market,
21 after 1 year after the date of enactment of this Act.
22 **SEC. 4. AMENDMENTS TO INTERNAL REVENUE CODE OF 1986.**

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1 (a) PROHIBITION OF HEALTH INSURANCE DISCRIMINATION ON THE
2 BASIS OF REQUEST FOR OR RECEIPT OF GENETIC SERVICES.—
3 (1) NO ENROLLMENT RESTRICTION FOR GENETIC
4 SERVICES.—Section 9802(a)(1)(F) of the Internal Revenue Code
5 of 1986 is amended by inserting before the period "or
6 request for or receipt of genetic services".
7 (2) NO DISCRIMINATION IN GROUP RATE BASED ON GENETIC
8 INFORMATION.—Section 9802(b)(1) of such Code is amended—
9 (A) by striking "may not require any individual"
10 and inserting "may not—
11 (A) require any individual";
12 (B) by moving the remainder of the paragraph two
13 ems to the right;
14 (C) by striking the period and adding "; or"; and
15 (D) by adding at the end the following new
16 subparagraph:
17 **"(B) adjust premium or contribution rates for a**
18 **group or the individuals in a group on the basis of**
19 **genetic information concerning or a request for or**
20 **receipt of genetic services by any member of such group**
21 **or a covered dependent of such member.".**
22 (b) LIMITATION ON COLLECTION AND DISCLOSURE OF GENETIC

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1 INFORMATION.—Section 9802 of such Code is amended by adding after
2 subsection (b) the following new subsection:
3 "(c) WITH RESPECT TO COLLECTION AND DISCLOSURE OF GENETIC
4 INFORMATION.—
5 "(1) COLLECTION.—Except as provided in paragraph (3), a
6 group health plan, or a health insurance issuer offering
7 health insurance coverage in connection with a group health
8 plan, shall not request or require disclosure of genetic
9 information concerning an individual who is a participant or
10 beneficiary (or an applicant for coverage as a participant
11 or beneficiary).
12 "(2) DISCLOSURE.—Except as provided in paragraph (3)—
13 "(A) a group health plan, or a health insurance
14 issuer offering health insurance coverage in connection
15 with a group health plan, shall not disclose genetic
16 information about an individual who is a participant or
17 beneficiary (or an applicant for coverage as a
18 participant or beneficiary) to any other health plan or
19 health insurance issuer; and
20 "(B) a physician, hospital, or other person that
21 provides health care items or services to an individual
22 shall not disclose genetic information about such

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1 individual to any group health plan, or health
2 insurance issuer offering health insurance coverage in
3 connection with a group health plan.
4 "(3) EXCEPTIONS.—
5 "(A) The provisions of paragraphs (1) and (2)
6 shall not apply to a request by or disclosure to a
7 health plan or issuer that provides reasonable
8 assurances that it provides health insurance coverage
9 to such individual and requires such information for
10 payment of claims or coordination of benefits.
11 "(B) Notwithstanding paragraphs (1) and (2), a
12 health plan or issuer that provides health care items
13 and services and provides reasonable assurances that it
14 requires such information for diagnosis or treatment of
15 such individual may—
16 (i) request (but not require) disclosure of
17 such information by the individual; and
18 (ii) request or require such information from
19 another health plan, or health insurance issuer,
20 or provider of health care items and services.
21 "(C) The provisions of paragraphs (1) and (2)
22 shall not apply if the individual, or the individual's

1 **legal representative, provides prior written**
2 **authorization of such disclosure.".**
3 (c) DEFINITIONS.—Section 9805(d) of such Code is amended by
4 adding at the end the following new paragraphs:
5 "(6) FAMILY MEMBER.—The term `family member' means,
6 with respect to an individual, a spouse or adopted child of
7 that individual, or another individual related by blood to
8 that individual or to a spouse or adopted child of that
9 individual.
10 "(7) GENETIC INFORMATION.—The term `genetic
11 information' means information about genes, gene products,
12 or inherited characteristics that may derive from an
13 individual or a family member.
14 "(8) GENETIC SERVICES.—The term `genetic services'
15 means health services provided to obtain, assess, and
16 interpret genetic information for diagnostic and therapeutic
17 purposes, and for genetic education and counselling.".
18 (d) EFFECTIVE DATE.—The amendments made by this section
19 shall apply with respect to group health plans and group health
20 insurance coverage for plan years beginning after 1 year after
21 the date of the enactment of this Act.
22

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Genetic Discrimination: Perspectives of Consumers

E. Virginia Lapham,* Chahira Kozma, Joan O. Weiss

In a study of the perceptions of 332 members of genetic support groups with one or more of 101 different genetic disorders in the family, it was found that as a result of a genetic disorder 25 percent of the respondents or affected family members believed they were refused life insurance, 22 percent believed they were refused health insurance, and 13 percent believed they were denied or let go from a job. Fear of genetic discrimination resulted in 9 percent of respondents or family members refusing to be tested for genetic conditions, 18 percent not revealing genetic information to insurers, and 17 percent not revealing information to employers. The level of perceived discrimination points to the need for more information to determine the extent and scope of the problem.

The rapid advances in human genetics, largely fueled by the Human Genome Project (HGP), have resulted in the expansion of the number and range of genetic tests (*1*). These tests are capable of providing carrier and presymptomatic information including risk of future disease, disability, and early death. In addition, these tests may reveal genetic information not only about the health of the individual, but also about his or her family members (*2*).

Concern about access to genetic information by health insurers has historical support (*3*, *4*). In the early 1970s, several insurance companies discriminated against individuals who were carriers of sickle cell anemia, even though they were quite healthy (*5*). The use of genetic information to deny life insurance to individuals leaves their dependents more vulnerable to economic consequences than is the case with the 70% of adults who are covered (*6*). The use of genetic screening to identify workers who may be particularly sensitive to noxious environments has been the principal focus of concern about workplace genetic testing even when done for benevolent reasons (*7*). Issues of genetic discrimination in employment and insurance have become more urgent as a result of the genome project (*8*).

Overall concerns about privacy and confidentiality have led the Ethical, Legal, and Social Issues (ELSI) Branch of the National Center for Human Genome Research to identify this issue as a top priority with the goal of proposing legislation specifically designed to protect people against genetic discrimination (*9*). Additionally, several working groups and scholars are focusing on this issue and have developed background papers and policy recommendations about the use of genetic information in health insurance as well as other areas such as life insurance and employment (*10*, *11*). Despite these concerns about potential genetic discrimination and documentation of individual cases, there is little information about the incidence and range of the problem (*12*).

This report provides information on the experiences of 332 individuals with one or more family members with a genetic disorder who are affiliated with genetic support groups. The study was part of the Human Genome Education Model (HuGEM) Project of the Georgetown University Child Development Center and the Alliance of Genetic Support Groups. It was the first phase of the HuGEM Project with the aim of getting input from 300 consumers in order to develop, implement, and evaluate a collaborative education model for consumers and health care providers.

Participants were recruited primarily through the national, regional, and local genetic support groups affiliated with the Alliance of Genetic Support Groups. Notices were put in two issues of the monthly *Alliance Alert* and letters were sent to the directors of 101 genetic support groups (representing an estimated 585,800 members). The notices contained information about the study and requested volunteers that were at least 18 years old and with one or more persons in the family with a genetic disorder who would be willing to participate in a 30-min telephone interview to provide opinions on the ethical, legal, and social issues of the HGP as well as priority topics for education. Volunteers were assured confidentiality of their responses. Random sampling was considered and ruled out because of time, cost, and the primarily educational focus of the project. Thus, the findings are applicable only to this group. Support group leaders were requested to distribute the letter requesting volunteers at meetings and in newsletters. Persons interested in participating were to complete a form at the bottom of the letter or call a 1-800 number for more information.

As a result of information provided by the support groups to the members or through the *Alliance Alert*, a group of 483 persons (from 73 different groups) contacted the Alliance of Genetic Support Groups about the study. They were sent information about the study and about the Human Genome Project. Of these, 336 (70%) returned consent forms (*13*). From this group, four persons decided not to participate after the interviews started, 306 persons completed telephone interviews, and 26 requested and completed the questionnaire by mail, for a total return of 332 respondents from 44 states and the District of Columbia (*14*).

Respondents were primarily female, highly educated, married, and Caucasian (*15*)—characteristics believed to be typical of genetic support groups (*16*). Age categories ranged from the twenties through seventies with a median age in the forties. A range of religious preferences was reported (*17*). There was an average of 2.1 affected family members per respondent with a range of 1 to 12 affected members reported.

The study questionnaire was composed primarily of questions with multiple choice responses. Telephone interviews were conducted by four social workers, a genetic counselor, and a consumer administrator (*18*) and lasted an average of 40 min with a range of 29 to 90 min. The content covered five areas: demographic information; knowledge of the Human Genome Project (61% had heard about the HGP before volunteering to participate in the study, 74% considered the HGP very important to their families, and 81% considered it very important to society); personal and family experience in areas related to genetic testing and research; opinions on a range of ethical, legal, and social issues; and priority topics for education. The education priorities were used to develop and implement educational forums in the mid-Atlantic and Pacific Northwest regions and will be described elsewhere.

Respondents were asked whether they or other family members had encountered problems with health insurance, life insurance, and employment (*19*). The term "genetic discrimination" was not used in the survey. It is used in this report to describe prejudicial actions as perceived by the respondents that resulted from insurers' or employers' knowledge of an individual's genetic condition, carrier status, or presumed carrier status, based on observation, family history, genetic testing, or other means of gathering genetic information (*20*).

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Respondents reported 101 different primary genetic disorders. The 18% of families with two or more disorders were asked to select one for purposes of the study. Of the primary disorders 68% were single-gene disorders, 10% were chromosome disorders, 11% were multifactorial disorders, 11% were major malformation syndromes, and less than 1% were mitochondrial and endocrine diseases.

Data analysis included frequency responses and comparison of responses to the questions on genetic discrimination by education, religious preference, and health of respondent and they showed no statistically significant differences (Pearson value of $P < 0.05$ was considered significant). Gender and ethnicity showed no significant differences when controlled for sample size.

Consumer experiences with health insurers were deemed important because the availability of affordable health insurance often determines who does and who does not have access to health care (*4*). For many people with genetic disorders, health insurance may mean the difference between life and death (*21*).

Although considerable genetic information may already be available to insurers in medical records, 40% of the respondents recalled being specifically asked about genetic diseases or disabilities on their applications for health insurance (Table 1). It cannot be assumed that the remaining 60% had not been asked questions about genetic diseases and disabilities. Many of them volunteered the information that they had never applied for health insurance. Some were able to maintain the coverage they had prior to diagnosis of a genetic disorder. Others had not applied because they assumed the genetic condition in the family would result in being turned down. Whether or not this information was then used to deny insurance to these people based on their genetic condition is not known.

Twenty-two percent of the respondents (Table 1) said that they or a family member were refused health insurance as a result of the genetic condition in the family. Since insurers do not need to provide reasons for turning down applications, it might be argued that respondents may have subjectively assumed that the denials were made because of the genetic condition. In this study, however, 83% of those who were refused health insurance had also been asked about genetic diseases or disabilities on their applications. Looked at in another way, nearly half (47%) of those who were asked about genetic diseases or disabilities on an application for health insurance were subsequently turned down. As health and life insurers are primarily regulated by states and most states are just beginning to address genetic issues in legislation (*22*), it is not known how many insurers actually ask genetic questions on applications.

The 31% of respondents with health insurance coverage who were denied reimbursement for some service or treatment indicated reasons such as the treatments were considered experimental, and services such as physical or occupational therapy were not considered a medical necessity. Time limits for submitting claims were also an issue, with insurers not paying claims that were more than a year old even when they had been submitted within the year and returned for more information. In several instances, payment was denied even though preapproval for a treatment or service had been given.

The large majority (83%) of respondents (Table 2) said they would not want their insurers to know if they were tested and found to be at high risk for a genetic disorder. The rate decreased to 78% when a similar question was asked that added the condition, "if the insurer pays for the tests." Some of the respondents noted that they would pay for genetic tests themselves or not be tested if they wanted to keep their genetic information confidential. The fear of genetic discrimination, as shown in Table 3, resulted in 9% of the respondents or a family member refusing to be tested for a genetic condition. This fear eliminates the opportunities of individuals to learn that they are not at increased risk for the genetic disorder in the family or to make life-style changes to reduce the risks or seriousness of the condition. It may also affect the number of people willing to participate in scientific research (*10*). Fear also prevented 18% of the respondents from revealing genetic information to an insurance company.

Approximately 70% of adults in the United States have some form of life insur-

Table 1. Questions and responses about experiences of consumers in areas of health insurance, life insurance, and employment. The total number of respondents is 332.

Question	Responses (%)		
	Yes	No	Don't know
As a result of the genetic condition in your family, have you or a member of your family been—			
Asked questions about genetic diseases or disabilities on an application for health insurance?	40	55	5
Refused health insurance?	22	76	2
Refused insurance coverage of some service or treatment?	31	67	2
Refused life insurance?	25	70	6
Asked questions about genetic diseases or disabilities on a job application?	15	83	2
Denied a job or let go from a job?	13	85	2

Table 2. Questions and responses to opinions about genetic information in insurance and employment.

Question	Responses (%)		
	Strongly agree or agree	Disagree or strongly disagree	Not sure or don't know
Genetic testing should be part of pre-employment physical exams.	4	94	2
Health insurers should be able to get genetic information if they pay for the tests.	16	78	6
	Yes	No	Not sure
If you were tested and found to be at high risk for a genetic disorder with serious complications, which of the following would you want to know the results of the test?			
a. Your employers?	6	87	7
b. Your insurance company?	11	83	6

ance (23). It is widely available, and only 3% of those who apply for coverage are declined. Of the 97% accepted, 5% are required to pay higher than standard premiums (24). This may be compared to the respondents in this study in which 25% (Table 1) of the respondents or affected family member have been refused life insurance (25).

Two questions were asked about the employment experiences of the study population. As noted in Table 1, 15% of the respondents said that they or affected family members had been asked questions about genetic diseases or disabilities on job applications. This increased to 20% of affected respondents ($P = 0.006$). It is not clear how often this information was used to subsequently deny the job to the applicants but the possibility exists and was of concern to respondents. In this study, 87% of respondents (Table 2) would not want their employers to know if they were tested and found to be at high risk for a genetic disorder with serious complications.

Thirteen percent of all respondents (Table 1) reported that they or another family member had been denied a job or let go from a job because of the genetic condition in the family. This was true for 21% of affected respondents and 4% of unaffected respondents ($P = 0.00001$). The percent was reduced to 9% ($P = 0.006$) for those with an affected child, even though a higher proportion of these respondents were in the workforce than the total population.

During the course of the analysis, a question was raised as to whether the perceived problems encountered in job application or denial or dismissal emanated from an employer's perception of a visible disability. To approach this question, analysis was done for the 77 unaffected respondents whose only affected family member was a child of less than 16 years of age. It was found that 7% of this population was asked about genetic diseases or disabilities on a job application and 3% were denied or let go from a job. These numbers should only be used as a starting point for future analyses.

For the affected respondents, some specific examples highlight the kinds of problems experienced. A man with a sex chromosome disorder reported that he had been denied a job following a pre-employment physical exam after the doctor wrote the name of the possible disorder on his medical report. The employer, in this case, knew it was illegal to use the diagnosis in the hiring decision and told the applicant that he would deny the conversation in the future if asked. A woman with a skeletal disorder reported that she was given termination notice the day after she informed her employer of a genetic diagnosis. The notice was withdrawn after she sought legal counsel. Examples provided by other respondents focused on effects of the genetic condition that could come under the protection of the 1995 interpretations of the Americans with Disabilities Act (26). The dilemma for persons with genetic disorders is that they must show not only that they have a genetic defect but also that they were regarded as "disabled" by an employer and discriminated against because of that perception. This raises concerns about the privacy and confidentiality of genetic information in the workplace.

A total of 17% have not revealed genetic information to their employers (Table 3) for fear of losing their jobs or insurance coverage. This increased to 25% of affected respondents ($P = 0.00001$). Overall, 43% of the respondents reported that they or members of their family have experienced genetic discrimination in one or more of the three areas. This included health insurance only (9%), life insurance only (11%), employment only (6%), and more than one category (17%).

Additional studies of persons with genetic disorders are indicated to confirm or deny the perceptions of the consumers in this study. It is possible that members of genetic support groups who have experienced genetic discrimination may have been more motivated to volunteer for this study. On the other hand, persons with these resources of higher education and membership in support groups traditionally have the skills and means to work with and influence social systems and may have experienced less discrimination than other groups. With adequate funding, a random sampling of respondents from support group or clinic populations could be selected with probability methods and objective as well as subjective information could be gathered.

Another goal would be to design more detailed questions to elicit information on genetic discrimination from respondents. Distinctions between the implications of overt genetic disease and conditions on each person and the effects on unaffected family members, or persons who are carriers or do not overtly express the consequences of the genetic condition will require further study. Consumers may be willing to participate if confidentiality is assured and trust is established. In this study, it was also found important for the interviewers to have clinical as well as technical skills in interviewing to facilitate the comfort level of discussing sensitive issues. This would also be recommended for future studies.

Although the goal of the HGP (and other genetic testing and research) is to help people, it could also cause harm if the level of perceived discrimination is in fact true. Neither the authors nor the respondents (as indicated in earlier responses) are suggesting that the HGP should not continue. On the contrary, there is strong support to continue research and to find ways to deal with genetic discrimination including federal or state legislation, guidelines, and standards among insurers, employers, researchers, and health professionals, and citizen advocacy to establish protections.

Table 3. Percentage of respondents withholding information or refusing to be tested for a genetic condition as result of fear.

Question	Responses (%)		
	Yes	No	Don't know
As a result of a genetic condition, have you or a member of your family—			
Refused to be tested for a genetic condition for fear of your insurance coverage being dropped.	9	89	2
Not revealed genetic information to an insurance company.	18	79	3
Not revealed genetic information to an employer.	17	81	2

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12. See, for example, P. R. Billings et al., Am. J. Hum. Genet. 50, 476 (1992).
13. As the interviews progressed and it was apparent that few members of ethnic or racial minorities were volunteering, additional telephone outreach was made to support groups to increase awareness of the project and opportunities for volunteering. This was only partially successful as seen in the demographic data.
14. There were no statistically significant differences in the responses on the telephone and written interviews so they are reported here together.
15. Of the 332 respondents, 80% were female, 57% have at least a bachelors degree, and 90% were Caucasian. Other characteristics were: 75% were married and living with their spouses, 76% have children, and 63% work outside the home. Family relationship, whether or not the respondent or family members were affected, age of diagnosis, and current age were also recorded.
16. Precise data on the demographics of genetic support groups are not available. Impressions are from staff of the Alliance of Genetic Support Groups based on their conversations and communications with the member organizations and attendance at national, regional, and local meetings.
17. Religious preferences were Roman Catholic, 26%; Protestant, 41%; Christian-other, 9%; Jewish, 11%; other, 2%; and none, 12%.
18. The interviewers were trained in interview techniques by the principal investigator and participated in pretesting the questionnaire.
19. The questions on possible genetic discrimination were taken from a questionnaire developed by Dr. Dorothy C. Wertz, The Shriver Center, Waltham, MA, entitled, Ethical Issues in Genetics, Part I, p. 33, No. 34, and used with permission of Dr. Wertz (letter of 16 December 1993).
20. This definition of genetic discrimination differs from the one used by Billings et al. (12) as they did not include actions against persons who were symptomatic or visibly affected by their genetic disorders. The design of our questionnaire does not permit analysis according to the definition of Billings et al. (1993). Because the questions on discrimination ask about all family members at once, the questions do not distinguish among: (i) the direct consequences of ongoing genetic disease or conditions, (ii) the effect of genetic disease on other family members, and (iii) the consequences of genetic information gained through testing.
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22. K. H. Rothenberg, J. Law Med. Ethics 23, 313 (1995).
23. The Ad Hoc Committee on Genetic Testing/Insurance Issues, Am. J. Hum. Genet. 56, 328 (1995).
24. "Report of the ACLI-HIAA Task Force on Genetic Testing," The American Council of Life Insurance and The Health Insurance Association of America (1991).
25. Many respondents said they had never applied for life insurance because they assumed they would be turned down.
26. Since 1990, the Americans with Disabilities Act (ADA) has provided protection for persons with disabilities in the workplace. The ADA prevents employers from openly denying employment or firing an individual solely on the basis of a "disability" if there are "reasonable accommodations" that can be made in the work setting to allow the person to perform his or her job. In April 1995, the ADA was interpreted by the U.S. Equal Employment Opportunity Commission to include healthy people who are carriers of genetic disorders. Implementation in general relies on employers and employees knowing and similarly interpreting the law as well as having good faith efforts to comply.
27. We wish to thank the HuGEM Advisory Committee: N. A. Holtzman, F. Neal-Smith, E. O. Nightingale, K. H. Rothenberg, and L. Walters, for assistance in planning and carrying out the study. We also wish to thank L. Palincsar, M. A. Wilson, S. Rennert, and M. Volner, for their assistance with the telephone interviews and M. J. Verdieck and M. O'Reilly for research assistance. Most of all, we wish to thank the consumers who provided the data. Funded by the National Institutes of Health at the National Center for Human Genome Research in the branch of Ethical, Legal and Social Issues, grant number RO1-HG00786-03. This support does not constitute an endorsement of the views expressed in this article.

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Kap104p: A Karyopherin Involved in the Nuclear Transport of Messenger RNA Binding Proteins

John D. Aitchison, Günter Blobel,* Michael P. Rout

A cytosolic yeast karyopherin, Kap104p, was isolated and shown to function in the nuclear import of a specific class of proteins. The protein bound directly to repeat-containing nucleoporins and to a cytosolic pool of two nuclear messenger RNA (mRNA) binding proteins, Nab2p and Nab4p. Depletion of Kap104p resulted in a rapid shift of Nab2p from the nucleus to the cytoplasm without affecting the localization of other nuclear proteins tested. This finding suggests that the major function of Kap104p lies in returning mRNA binding proteins to the nucleus after mRNA export.

Transport across the nuclear envelope occurs through nuclear pore complexes (NPCs) and is governed by the interaction of soluble transport proteins (karyopherins) with the transport substrate and the NPC (*1–12*). Most of our understanding of the mechanism of translocation comes from studying protein import in semipermeablized cells (*1*) of model karyophilic proteins that carry a nuclear localization signal (NLS) from either the SV40 large T antigen or nucleoplasmin (*2*). These classical NLSs are recognized by karyopherin α in a dimeric cytosolic complex with karyopherin β (*3–8*). The complex docks at the NPC through its interaction with nucleoporins that contain characteristic repeated peptide motifs (*6–11*). The small guanosine triphosphatase, Ran, and p10 are required for the subsequent translocation of the substrate (and karyopherin α) through the NPC (*11*, *12*).

Distinct saturable and noncompeting pathways for the import of different karyophiles have been uncovered through the use of microinjection studies in oocytes (*13–15*). Similarly, saturable noncompeting pathways exist for the export of macromolecules from the nucleus (*14*, *16*, *17*). The signals that mediate many of these processes are different from classical NLSs (*14*, *15*, *17–19*) and thus may use recognition factors other than karyopherin α and karyopherin β for nuclear transport. Here we characterize the first such factor, which we term Kap104p and which is required for the import of at least two yeast nuclear mRNA binding proteins.

The *Saccharomyces cerevisiae* proteins Kap60p and Kap95p are homologs of mammalian karyopherin α and karyopherin β (*20*). Sequence comparisons of Kap95p with the complete yeast genome database uncovered three additional proteins that are structurally similar to Kap95p; two of these, which we term Kap123p and Kap104p, have not been previously characterized (*21*), and the third, Pse1p, was identified as a multicopy enhancer of protein secretion (*22*). The sequence alignment of Kap104p with Kap95p is shown (Fig. 1A). The proteins bear substantial similarity over their entire lengths, and secondary structural predictions suggest that Kap95p and Kap104p share the same overall domain structure of HEAT motifs (*23*).

Deletion of *KAP104* resulted in a severe growth defect and temperature sensitivity (*24*). Immunofluorescence microscopy (*25*) with antibodies specific for Kap104p (in wild-type cells) showed that Kap104p was mainly cytosolic and was apparently absent from the nucleus (Fig. 1B). However, in *nup120Δ* cells, which cluster their NPCs to a region of the nuclear envelope opposite the nucleolus (*26*), Kap104p colocalized with the nucleoporin Nsp1p (*27*) (Fig. 1C). The ability to detect coincident staining of the nucleoporins and Kap104p under these conditions likely was due to an interaction of Kap104p with NPCs.

Subcellular fractionation (*28*) was consistent with the distribution of Kap104p detected by immunofluorescence. Kap104p was present mainly in the cytosolic fraction,

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Flunk the Gene Test and Lose Your Insurance

THE GORRELLS
Jane (front) and her daughter Danielle know they share a gene associated with colon cancer

DNA probes may offer dazzling insights. But they have helped foster a pernicious new form of discrimination.

BY GEOFFREY COWLEY

JAMIE STEPHENSON HAS SEEN FIRSThand what modern genetic science can do for a family. When her son David was 2 years old, a pediatrician noticed developmental delays and suspected fragile X syndrome, a hereditary form of mental retardation. A lab test confirmed the diagnosis, and the Stephensons spent several years learning to live with it. When David was 6, he visited a neurologist, who scribbled "fragile X" on an insurance-company claim form. The company responded promptly—by canceling coverage for the entire family of six. There is no medical treatment for fragile X, and none of David's siblings had been diagnosed with the condition. "The company didn't care," Stephenson says. "They just saw a positive genetic test and said, 'You're out'."

From the dawn of the DNA era, critics have worried that genetic testing would create a "biological underclass"—a population of people whose genes brand them as poor risks for employment, insurance, even marriage. The future is arriving fast. Med-

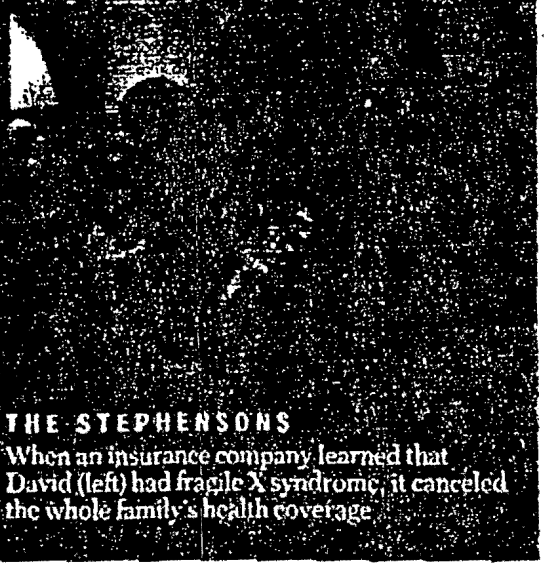
KEN SCHLES

ical labs can now test human cells for hundreds of anomalous genes. Besides tracking rare conditions, such as Huntington's disease and cystic fibrosis, some firms now gauge people's susceptibility to more common scourges. By unmasking inherited mutations in p53 (main story) and other genes, the new tests can signal increased risk of everything from breast, colon and prostate tumors to lymphoma and leukemia. Many of the tests are still too costly for mass marketing, but that will change. And as the Stephensons' story suggests, the consequences won't all be benign. "This is bigger than race or sexual orientation," says Martha Volner, health-policy director for the Alliance of Genetic Support Groups. "Genetic discrimination is the civil-rights issue of the 21st century."

No one would argue that genetic tests are worthless. Used properly, they can give people unprecedented power over their lives. Prospective parents who discover they're silent carriers of the gene for a disease like Tay-Sachs, which causes death by the age of 3, can make better-informed decisions about whether and how to have kids. Some genetic maladies can be managed through medication and lifestyle changes once they're identified. And while knowing that you're at special risk for cancer may be an emotional burden, it can also alert you to the need for intensive monitoring. Jane Gorrell knows her family is prone to colon cancer. Her father developed hundreds of precancerous polyps back in the 1960s, and both she and her sister had the same experience during the '70s. Their condition, known as familial adenomatous polyposis, has since been linked to a mutation in the p53 gene—and Gorrell has learned that one of her two children inherited it. Though the child has suffered no symptoms, she gets frequent colon exams and is helping researchers test a drug that could help save lives.

The catch is that no one can guarantee the privacy of genetic information. Outside of large group plans, insurance companies often scour people's medical records before extending coverage. And though employers face some restrictions, virtually any company with a benefits program can get access to workers' health data. So can schools, adoption agencies and the military. Employees of Lawrence Berkeley Laboratory (LBL), a large research institution owned by the Department of Energy and operated by the University of California, recently discovered that the organization had for three decades been quietly testing new hires' blood and urine samples for evidence of various conditions. They claim blacks were screened for the sickle-cell trait, Latinos for syphilis and women for pregnancy. "I can't say the information was put to some incredibly harmful use, because we don't know what happened," says Vicki Laden, a San Francisco lawyer who has tried unsuccessfully to sue the lab for civil-rights violations. LBL recently stopped the testing.

> "Genetic testing has the potential to revolutionize medicine. But revolutions can have casualties."
>
> —FRANCIS COLLINS
> Director of the Human Genome Project



THE STEPHENSONS
When an insurance company learned that David (left) had fragile X syndrome, it canceled the whole family's health coverage

How often is genetic information used against people? No one knows, but there are signs that discrimination is fairly common—even in the absence of sophisticated tests. In one recent study, researchers led by Georgetown University sociologist Virginia Lapham surveyed 332 families belonging to genetic-disease support groups; 22 percent of the respondents said they'd been refused health insurance, and 18 percent claimed their perceived risks had cost them jobs.

In another 1996 study, a team led by Lisa Geller of Harvard Medical School documented more than 200 instances in which healthy people experienced genetic discrimination. As in Lapham's study, many participants told of losing their health and life insurance. But that wasn't their only problem. One respondent was denied a job *selling* insurance after he disclosed that he had hemachromatosis, a hereditary iron-storage problem that can be treated for about $1,200 a year. A social worker who had excelled during her first year at a Wisconsin health-care company was quickly forced out after mentioning that her uncle had Huntington's, a degenerative brain disease that victims' offspring have a 50 percent chance of developing. Carol Isaacson Barash, the bioethics consultant who administered the Geller study, recalls another case in which a woman in the early stages of Huntington's disease was unable to place her child with a private adoption agency. A public agency took the child, eventually matching her with a couple who had previously been rejected themselves—because one partner carried the Huntington's gene. "It's a stark commentary on how society regards people at risk," says Barash.

Not surprisingly, people from high-risk families have come to fear tests almost as much as disease. Lori Andrews, a Chicago law professor and former head of a federal task force on the social implications of genetic research, notes that only 15 percent of people with a Huntington's afflicted parent choose to learn their own status. Discrimination isn't their only concern: virtually everyone testing positive for the Huntington's gene develops debilitating symptoms during middle age, and doctors can do nothing to help. But people who might benefit from genetic tests are almost as leery. When researchers at the Georgetown University Medical Center surveyed 279 people from families plagued by breast and ovarian cancer, only 43 percent wanted to be tested for hereditary mutations in BRCA1, a recently discovered gene that is often implicated in those diseases. Many said the prospect of discrimination scared them off. Kendra McCarthy, a 47-year-old administrator at the Virginia Department of Mental Health, saw her mother die of breast cancer at 34, her father of esophageal cancer at 41. When scientists developed tests that might have gauged her own susceptibility, she always declined to take one. McCarthy doesn't regret that decision. She has developed

can use this study

Information, Please

One of the wonderful things about science is that there are no stupid questions. Nobel Prize winners ask the same ones you do. Who cares if they are working to cure deadly diseases while you're only trying to make sense of the science pages. A primer:

What is a gene?

The basic unit of heredity, a gene consists of an ordered sequence of chemicals called nucleotides and designated by the letters A, T, C or G. A gene is situated on one of the 23 pairs of human chromosomes. It carries instructions for the production of proteins. The sequence of nucleotides serves as a chemical code, determining which protein the gene makes. Genes take the form of a double helix.

Where are genes?

Every cell in the body contains exactly the same genes (roughly 100,000). We have two copies (identical or just similar) of every gene. The only exceptions are sperm and egg cells, which contain one for each chromosome pair rather than both, and red blood cells. What makes one cell different from another (liver from muscle, for instance) is not their genes—which are identical—but which of the genes are turned on.

What is a mutation?

Any change in the normal sequence of nucleotides in a gene. A mutation can be the deletion of a nucleotide or the change of one nucleotide to another.

What causes a mutation?

A mutation can be inherited; if the egg or sperm cell that produced the child contained a mutation, so will every one of the child's 30 trillion cells. A mutation can also be caused by radiation, by "mutagens" (chemicals, such as benzopyrene, found in cigarette smoke) or by some viruses.

What happens if a gene is mutated?

It no longer produces the protein it was designed to. Since a cell has two copies of each gene, often it can get by with only one healthy version, and a mutation has no medical consequences. But if the mutation occurs in an egg or sperm cell, it can be passed on to children. And if both copies are mutated, or if the cell needs both copies to function, then the cell is no longer able to carry out its job.

KATHERINE LAMBERT

KENDRA MCCARTHY
Though cancer stalks her family, she shunned genetic testing. "If prevention was possible, I would say it makes sense."

breast cancer, but she still has her health coverage, and a life-insurance plan to protect her two sons.

Besides depriving people of potentially useful information, the fear of discrimination can hamper scientific progress. Barbara Weber, a geneticist with the University of Pennsylvania Cancer Center, often asks women who learn they have BRCA1 mutations to take part in confidential follow-up studies. She wants to know which strategies are most effective for preserving their health. "We have the tools to answer these questions," she says. "All they have to do is tell us how they're doing every six months." Yet three fourths of the women she approaches say no.

The federal government is now taking steps to make genetic information less threatening. The recently enacted Health Insurance Portability and Accountability Act (better known as Kennedy-Kassebaum) bars insurers from treating genetic mutations as "pre-existing conditions" unless they're causing illness. The insurance act also guarantees coverage to anyone leaving one group plan for another, whatever his pre-existing conditions. That will make switching jobs easier for many people, but ethicists say the act is only a first step. Because it covers only group plans and doesn't deal with disability insurance, it won't do much for folks like Theresa Morelli, an Ohio lawyer who applied for independent coverage several years ago. Morelli was 28 and in perfect health when she met with an insurance agent and paid her first premium. A month later, she got her check back, along with a letter saying her application had been denied because her father had Huntington's disease. Morelli's dad had in fact received that diagnosis—erroneously, it turned out—and her doctor had made a note of it in her chart.

The restrictions on employers are as porous as those on insurers. The 1990 Americans with Disabilities Act bars companies from discriminating against people with disabilities—and it defines that term broadly enough to include genetic mutations that have yet to cause symptoms. But the act does nothing to keep employers from gathering medical information. "As long as employers have access to genetic information," says Lewis Maltby of the ACLU, "they'll have an irresistible incentive to use it."

Lawmakers are now racing to strengthen the protections. At least 15 states have recently placed restrictions on insurers or employers, and Congress will consider several bills in the new session. The insurance industry argues that it should be free to charge people rates that reflect their risks, at least when dealing with individuals and small companies. That way, says Richard Coorsh of the Health Insurance Association of America, each applicant pays the fairest possible price. But most ethicists contend that where health coverage is concerned, people shouldn't be penalized for risks they can't modify.

Discrimination isn't the only potential downside to genetic testing. Some of the new susceptibility tests are only vaguely predictive of illness—and no one forces the companies that offer them to counsel patients about what the results actually mean. Even when the tests are sound and the results secure, the knowledge they create can dash hopes and divide families. Unfortunately, principled ignorance doesn't always make life easier.

With ANNE UNDERWOOD *in New York,* KAREN SPRINGEN *in Chicago and* MARY HAGER *in Washington*

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Advent of Testing for Breast Cancer Genes Leads to Fears of Disclosure and Discrimination

C-1 NYT 2/4/97

By GINA KOLATA

DR. BARBARA WEBER is still reeling from the experience of a patient who told her insurance company that she had a mutated gene that could cause breast cancer.

The woman had been tested for the mutation at Dr. Weber's clinic at the University of Pennsylvania, and Dr. Weber told her the results. Since some studies indicate that women with the mutated gene have a 90 percent chance of developing breast cancer, the woman wanted both of her breasts removed right away. Before she had the operation, she submitted a claim to her insurance company, Dr. Weber said, not disclosing that she had had the genetic test but reporting a strong family history of breast cancer.

The company turned her down, Dr. Weber said, on the ground that it did not pay for preventive medicine. So, at the woman's request, Dr. Weber submitted the woman's genetic test results. At that point, Dr. Weber said, the company told the woman that it would not pay for the surgery because she had a preexisting condition — a genetic defect — when she took out her health policy.

"It was absolutely unbelievable," Dr. Weber said. She said that the woman, who did not return telephone calls requesting an interview, had the surgery *anyway*. Afterward, Dr. Weber said, when pathologists examined the woman's breast tissue, they found a cancerous tumor that had been missed by mammograms.

Gene Wilkinson for The New York Times

Dr. Barbara L. Weber says patients who get gene testing are refusing to enroll in studies for fear of discrimination.

It is cases like this and fears of similar treatment that are convincing some women and researchers that it might be too dangerous to put genetic testing results on medical charts and in clinical records, where privacy cannot be assured.

Women worry that insurers will raise their rates, or refuse to insure them, that employers will not hire them or promote them, and even that friends and family members might treat them differently if they knew that they were tainted with a deadly gene.

Fears of genetic testing have been expressed by some people for several years, starting with tests for rare genetic diseases like Huntington's. But the recent discovery of two genes that can cause breast and ovarian cancer when mutated has made the issue more pressing.

Not only is breast cancer the disease that women fear the most, but the mutations, in genes known as BRCA1 and BRCA2, are relatively common in Ashkenazi Jewish populations (of Eastern European descent), affecting as many as 1 in 58 women.

In addition, many women who are not Jewish but have a strong family history of breast or ovarian cancer also inherited mutated genes. In the last year, several commercial laboratories have begun offering tests for breast cancer gene mutations and major medical centers across the country have set up their own testing programs.

Some legal experts say the fears of discrimination may be exaggerated. Federal legislation that

Continued on Page C3

Please See Next Page

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will go into effect on July 1 addresses part of the issue, focusing on people who are covered under group medical plans. The law states that if an individual was in such a plan for at least 12 months and had a genetic condition diagnosed in the past 6 months, a new insurer cannot use that genetic information to deny or limit coverage.

Alexander Morgan Capron, an ethicist and law professor at the University of Southern California, said insurers could charge people with genetic conditions more for insurance. But, Mr. Capron added, 29 states including, New Jersey, have recently passed laws preventing health maintenance organizations and health insurance companies from charging people more because they have a gene mutation. In addition, the Americans With Disabilities Act prevents employers from discriminating against people who have disease-causing genetic mutations. Employers who insure their employees are exempt from the state laws, Mr. Capron said.

Richard Coorsh, a spokesman for the Health Insurance Association of America, said that people's fears that insurance companies would discriminate against them might be overblown. Many people are insured through their employers as part of large groups, and they would be covered even if they did have a cancer-causing gene.

But Mr. Coorsh said that those who change jobs or lose their jobs and have to seek insurance as individuals "will probably end up having to pay a lot more money" for health insurance if they test positive for a cancer gene. There have been few systematic studies to assess how real the risk of genetic discrimination is, but one survey, published in October in the journal Science, showed that the fear, at least, is widespread.

The study, by Dr. E. Virginia Lapham of the Georgetown University School of Medicine and her colleagues, involved 332 people who belonged to support groups for families with a variety of genetic disorders. Of them, 25 percent believed they were denied life insurance because of their disorder, 22 percent thought they were denied health insurance, and 13 percent believed they were not hired for a job or that they lost a job because of the disorder.

Whether or not these suspicions are justified, they are having an effect. Some researchers at major medical centers are worried enough to work under a cloak of secrecy. They test for the breast-cancer genes in under the aegis of a research program, which allows doctors to keep the results secret, coded and encased in locked files.

Some researchers advise women who are tested not to tell even their private doctor of their test results if the doctor insists on putting all relevant information in a woman's medical record. Some doctors agree not to write down test results, relying instead on their memories.

At treatment centers, which, unlike research centers, are not permitted to hide test results, some women have used aliases to protect their privacy.

"If we ever needed proof that the system is broken, this is it," said Dr. Francis Collins, director of the National Center for Human Genome Research in Bethesda, Md. "The system forces people to take drastic steps to protect themselves. It is putting a terrible burden on patients."

Doctors, too, are placed in an untenable situation, Dr. Collins said. "You are forced sometimes to have interactions with insurance agents or with other physicians or with H.M.O.'s where you would have to pretend you don't have the information," Dr. Collins said. "It's a very strange dilemma — to choose between patient confidentiality and telling the truth."

It is, said Dr. Thomas Murray, the director of the Center for Biomedical Ethics at Case Western Reserve University in Cleveland, "a classic ethical quandary," and one with no obvious solution.

Moreover, some cancer researchers note, there is a legitimate reason why insurers and employers might want to avoid people with genes that can cause cancers: the insurers because of the extra cost of treating them and the employers because of the extra insurance cost and because of lost productivity.

"I'm not sure you can totally blame insurance companies or employers," said Dr. C. Kent Osborne, a cancer researcher at the University of Texas in San Antonio. "It's a bigger problem than that. It's something we have to deal with as a society."

In the meantime, doctors and patients are trying to act expediently.

Dr. Funmi Olopade, who directs the breast cancer genetic testing program at the University of Chicago, was interviewed just after an appointment with a 25-year-old woman, who came to learn her test results. The woman was found to have a genetic mutation that can cause breast and ovarian cancer. The university keeps these records secret.

But, Dr. Olopade said, the woman asked her if she could tell her private doctor of her test and what it disclosed.

Dr. Olopade suggested that the young woman start by asking her doctor if he felt he had to put everything he knew about a patient into the medical records. If so, Dr. Olopade said, the woman might not want to tell.

And with good reason, said Dr. Mark Siegler, who directs the ethics program at the University of Chicago. More than a decade ago, before the proliferation of electronic records, before the merging of health maintenance organizations that both pay for and care for patients, he investigated the complaint of a woman who had come to the hospital for elective gall bladder surgery and had said that too many people were looking at her medical records.

"I started to ask how many people had legitimate access to her chart," Dr. Siegler said. "I stopped counting at 75." He concluded, he said, "that confidentiality really didn't exist." That is even more true today, he added.

Some doctors, like Dr. John Glick, the director of the University of Pennsylvania's cancer center, say they will not put genetic testing information in a woman's medical records if she asks that it be kept out. Instead, Dr. Glick said, he simply remembers what the woman said about her test results.

Other doctors refuse to go along with such requests. One woman, who tested positive for the breast cancer gene at the University of Pennsylvania, said her private doctor told her it would be dishonest not to include the test result in her medical records. So, the woman said, "I asked that my medical records be closed and I changed doctors."

The woman said she was worried about health insurance. "The company my husband works for just got bought out, so things aren't as steady as they used to be," she said. "You can survive breast cancer, but you can't survive if you don't have insurance."

Dr. Henry Lynch, who directs the

Some doctors won't put results of genetic tests in a woman's records...

cancer center at Creighton University in Omaha, tests women for the breast cancer gene as part of a research program in which he can keep the results secret, and as part of a clinical program, where he cannot. But some women in his clinical program use aliases. Others pay for tests out of their own pockets, hoping to keep the information out of the hands of their insurance companies.

Dr. Weber said that she was confronting a new ramification of women's fears of discrimination. Women who were tested as part of her research program are refusing to participate in long-term studies that would assess their risk of developing cancer and the success of interventions, like frequent mammograms, or hormone treatments, or surgery to remove their breasts or ovaries.

The women, Dr. Weber said, fear that it would be a red flag on their medical records to participate in the study. Their mammogram results and any biopsies, for example, would have to be sent to the university's research program.

The only possibility of learning if there is a way to prevent cancer in women who are genetically predisposed, Dr. Weber said, is to do long-term studies. If no one will participate, she said, what is the point in testing women to see if they have the cancer genes?

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Individual, Family, and Societal Dimensions of Genetic Discrimination: A Case Study Analysis

Lisa N. Geller, Joseph S. Alper, Paul R. Billings, Carol I. Barash, Jonathan Beckwith, and Marvin R. Natowicz*

Keywords: genetic discrimination, genetic testing, Medical Information Bureau, state insurance commissions.

ABSTRACT

Background. *As the development and use of genetic tests have increased, so have concerns regarding the uses of genetic information. Genetic discrimination, the differential treatment of individuals based on real or perceived differences in their genomes, is a recently described form of discrimination. The range and significance of experiences associated with this form of discrimination are not yet well known and are investigated in this study.*

Methods. *Individuals at-risk to develop a genetic condition and parents of children with specific genetic conditions were surveyed by questionnaire for reports of genetic discrimination. A total of 27,790 questionnaires were sent out by mail. Of 917 responses received, 206 were followed up with telephone interviews. The responses were analyzed regarding circumstances of the alleged discrimination, the institutions involved, issues relating to the redress of grievances, and strategies to avoid discrimination.*

Results. *A number of institutions were reported to have engaged in genetic discrimination including health and life insurance companies, health care providers, blood banks, adoption agencies, the military, and schools. The alleged instances of discrimination were against individuals who were asymptomatic and sometimes impacted on other*

can we cite this study

* All authors contributed equally to this work.

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asymptomatic relatives. Few surveyed respondents knew of the existence of institutions such as state insurance commissions or the Medical Information Bureau, Inc., which may play roles in redress of grievances or correction of misinformation.
Conclusions. *Genetic discrimination is variable in form and cause and can have marked consequences for individuals experiencing discrimination and their relatives. The presence of abnormal genes in all individuals makes each person a potential victim of this type of discrimination. The increasing development and utilization of genetic tests will likely result in increased genetic discrimination in the absence of contravening measures.*

The growing knowledge of human genetics, stimulated in part by the Human Genome Project, has engendered a societal need to understand potential hazards as well as benefits of this knowledge. With the increased ability to identify genetic differences, it is important to elucidate appropriate uses of genetic information from the perspective of both individuals and the public. At the same time, safeguards must be developed to minimize inappropriate uses of this information[1-4]. One area of concern is genetic discrimination.

The term "genetic discrimination" has been used to describe the differential treatment of individuals or their relatives based on actual or presumed genetic differences as opposed to discrimination based on phenotype[5-7]. Sources of genetic information that enable such discrimination include genetic and sometimes non-genetic medical tests, family histories, and information obtained from clinical examinations. Each of these sources has important limitations in terms of its reliability in predicting whether a particular genetic condition will occur and, if so, the clinical course of the associated disease. These limitations include but are not restricted to the sensitivity and specificity of genetic tests, the intrinsic clinical variability of many hereditary conditions, and the importance of environmental factors.

A pilot study of genetic discrimination showed this problem to involve more disorders than was previously revealed by isolated reports. The reported incidents involved a variety of social institutions such as life and health insurance organizations, and suggested that genetic discrimination may become a significant social policy problem[5]. Based on this work and reports of genetic discrimination by others, there is now serious concern regarding the importance of this problem[3-5, 8-11]. This concern is intensified by the proliferation and increasing utilization of genetic tests made possible in part by technological advances made by the Human Genome Project and by the application of these technologies by commercial interests[12-14].

Previous reports of genetic discrimination involved studies of relatively small numbers of individuals and, consequently, would not be expected to reveal a full range of the discriminatory experiences faced by affected individuals and their families. Here we report on case studies obtained in 1992-1993 of individuals living throughout the United States at-risk for or related to people at-risk for the following disorders: hemochromatosis, phenylketonuria (PKU), mucopolysaccharidoses (MPS), and Huntington disease. We describe the spectrum of discrimination reported by some of these individuals and discuss its implications through analysis of selected informative cases.

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METHODS

Study Design

Individuals at-risk for genetic discrimination were sent questionnaires during 1992-1993. The definition of genetic discrimination has been previously presented and distinguishes genetic discrimination from discrimination based on phenotype[15]. Only those cases in which individuals appeared to have no symptoms (i.e. no apparent phenotype at the time of the reported discrimination) were included in this study.

Questionnaires and Interviews

Two questionnaires and an interview instrument were developed based on findings from our pilot study. They were reviewed by consultants experienced in qualitative research methods and questionnaire design and were approved by the Shriver Center's Institutional Review Board. The two questionnaires were distributed through the mailing lists of genetic disease organizations selected according to the criteria described below, and were accompanied by a letter describing the research group and the goals of the study. One questionnaire was directed at individuals who had or were at-risk to develop a genetic condition. The other questionnaire was directed at parents who had a child with a genetic condition. Both were designed to acquire information about whether an individual believed that she/he or a relative had experienced discrimination because of a genetic diagnosis or assumption of genetic predisposition to the disorder, as well as a brief description of the discriminatory event(s) (Table 1, see p. 85). The descriptions of the alleged discriminatory events were used to screen the returned questionnaires for cases that appeared to fit the definition of genetic discrimination used in this study. That is, the person alleging discrimination was not symptomatic for a genetic disorder (or any other disease which might confound the claim of discrimination) nor did the complaint appear to involve legitimate actions by companies or individuals that were construed as "unfair" by the individual claiming discrimination. Some cases were included where an apparent conflict between the perception of the individual and the apparent point of view of an institution illustrate areas of ambiguity concerning the "fairness" of the situation.

Telephone interviews were conducted with individuals who, from their responses to the questionnaire, appeared to have experienced genetic discrimination as described above. An attempt was made to interview all individuals whose questionnaire answers met the above criteria and who indicated in their returned questionnaires that they were willing to be contacted by telephone. The script (available by request) used to conduct the interviews was designed to obtain more detailed information about the perceived discriminatory event(s) (Table 2, see p. 86-87). For example, in cases of alleged discrimination by an insurance company, the interviewee was asked about how the company found out about his/her genetic status, what correspondence and conversations were held with individuals at the company, what reasons were given by the company for actions taken, whether outside support or counsel was sought, whether the case was reconsidered after communication with the company, length of time of response, who within the company handled the case, whether alternative policies were offered, etc. In addition, general information was elicited regarding the economic and educational status of the individual, as well as pertinent medical

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information including whether the individual was asymptomatic. We also sought to determine the origin of the genetic diagnosis or supposed genetic risk factor, perceived ability to redress a grievance, the extent to which an individual challenged adverse decisions, and the impact of personal genetic information on the individual and her/his family. The Medical Information Bureau, Inc. (MIB) and state insurance commissions are institutions which may be useful to individuals seeking redress regarding insurance. After conducting a number of interviews, it became apparent that the respondents differed widely in their knowledge concerning avenues for seeking redress for complaints involving insurance. Consequently, questions concerning whether the individual knew of the MIB and state insurance commissions were added to subsequent interviews. Consenting interviewees were anonymously tape recorded to aid in the transcription of information. In addition, documentation of discriminatory events was sought, including such items as letters from insurance companies and medical and personal notes.

Study Groups

The specific disorders targeted for this study were Huntington disease, phenylketonuria (PKU), hemochromatosis, and mucopolysaccharidoses (MPS). They were chosen because they met the following criteria: (1) the genetic basis of the condition is known and unambiguous; (2) discrimination directed against individuals with these conditions would most likely be due to the genetic bases of these conditions, rather than due to physical symptoms; and (3) support groups for persons with these conditions exist so that individuals could be contacted easily. These conditions were also selected because they cover a spectrum of situations including dominant and recessive disorders, treatable and untreatable disorders, relatively common disorders for which screening programs exist, and rare disorders for which screening programs are not indicated. Note that the individuals with recessive disorders included both those with the genotype for the disorder and those who are simply carriers.

Huntington disease is a fatal, untreatable, autosomal dominant disorder whose symptoms generally appear in middle age. There is currently a molecular genetic test available to diagnose this condition. Hemochromatosis is an iron storage disorder with a variable phenotype, some individuals being completely asymptomatic. This autosomal recessive disorder is treatable by phlebotomy (drawing blood). PKU is an autosomal recessive disorder for which all newborns in the United States are tested. If left untreated the disorder results in mental retardation. However, PKU is successfully treated by placing the child on a special diet. MPS disorders are usually associated with mental retardation and organomegaly.

A total of 27,790 questionnaires were mailed by the following groups to their members: The Huntington Disease Society of America, The Hemochromatosis Research Foundation, The National M.P.S. Foundation, and the PKU Clinic at Children's Hospital, Boston, MA. These mailing lists included donors and interested individuals who were not appropriate respondents to the questionnaire, a factor which was expected to result in a low response rate. However, contacting individuals through a national organization was more likely to get a broader response than that obtained by contacting individuals through a few clinics. Respondents to the questionnaire

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included individuals who were at-risk to develop a genetic disorder but who were not informed of their genotype, individuals who were presymptomatic for a specific genetic condition, and individuals who were asymptomatic because of receipt of a therapy or were heterozygotes and thus only "carriers" for an autosomal recessive condition.

RESULTS

Of the 917 returned questionnaires, 455 respondents asserted that they had experienced genetic discrimination and 437 that they had not. The remainder gave ambiguous answers that could not be specifically classified. Some respondents reported experiencing genetic discrimination in more than one setting. After screening the questionnaires for cases that appeared to fit the definition of genetic discrimination used in this study, we were able to contact for interviews 206 individuals who reported that they experienced genetic discrimination. Detailed breakdowns of the responses of all respondents to the questionnaire, categorized by disease group, are given in Table 3 (see p. 88).

A variety of different institutions allegedly discriminated against the respondents. The majority of cases involved discrimination by health and life insurance companies but there were a number of cases involved employers, adoption services, and blood banks. The cases reported below are grouped according to agents/institutions allegedly engaged in the genetic discrimination, followed by results of the impact of genetic discrimination on individuals and family members, responses and counter-measures taken to mitigate the effects of genetic discrimination, and information pertinent to the underlying bases of this phenomenon.

<u>Agents/institutions engaging in discriminatory practices</u>

- Health and Life Insurance Corporations

Four aspects of discrimination are illustrated by the cases involving health and life insurance: (1) discrimination against individuals who were asymptomatic; (2) differential treatment of asymptomatic individuals or families once a genetic diagnosis was established, thus treating the genetic diagnosis as a preexisting condition; (3) the failure of some group insurance plans to provide coverage for qualified individuals with a genetic diagnosis; and (4) the loss of insurability suffered by relatives of an individual with a presumed genetic disease. These issues are illustrated by the following cases.

Case a: A health maintenance organization (HMO) had covered the medical expenses of a child since birth but refused to pay for occupational therapy after she was diagnosed with MPS-I, claiming that the condition was pre-existing. All bills relevant to the condition had been paid up to the time of diagnosis. The occupational therapy had been pre-approved by the managed care corporation. The situation was remedied after the family complained to a customer service representative of the HMO.

Case b: A private insurer in Colorado notified the parents of a three year old who had been recently diagnosed with an MPS syndrome that the child's policy was terminated

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although the family had been on the policy for nine months before the diagnosis. After an extended negotiation that included retention of a lawyer and the threat of a lawsuit, the insurance policy was reinstated. However, a rider was added to the policy excluding coverage for two common MPS-related complications.

Case c: A 24 year old woman was denied life insurance due to her "strong family history of Huntington's Chorea" and the fact that she has never been tested to determine if she is "currently a carrier." The rejection letter stated that if she "should be tested and if found to be negative," the company would issue a standard contract.

Case d: A mother submitted applications for employment-based life insurance policies simultaneously for her two children, one of whom had Hurler syndrome, a form of MPS. Both were rejected. The rejection letter indicated that the child with Hurler syndrome was denied a policy because the condition is fatal; no reason was given for the denial of a policy to the other child. She was later able to obtain coverage for the healthy child through a different employer.

• Clinical Professionals

In several cases medical professionals reportedly pressured patients or clients at-risk for having children with serious genetic conditions, to undergo prenatal diagnostic testing or to forsake having children.

Case e: A PKU gene carrier reported that during a routine pediatric visit, her child's doctor advised her that it would be unwise to have more children and that she should consult a genetic counselor to understand "the implications of PKU".

Case f: A couple in which one member was at-risk for Huntington disease reported that physicians tried to compel them to undergo prenatal genetic testing and reportedly coerced them to sign a document agreeing to abort an affected pregnancy. They also reported being required by a health care provider to undergo genetic counseling despite their belief that they had comprehensive knowledge about the genetic risks and their decision to continue any pregnancy irrespective of Huntington disease status.

• Adoption

Three issues are illustrated by the cases of alleged discrimination by adoption agencies. They are: (1) a misunderstanding of the nature of the presumed genetic condition with consequent unfair treatment of the prospective parents; (2) the requirement that individuals "pass" a genetic test before being allowed to adopt a child; and (3) the assumption that individuals with genetic diagnoses should adopt only children at-risk of having a disability.

Case g: One respondent, a carrier for MPS, was required by an adoption agency to repeat the blood and urine tests routinely required of prospective parents. It was reported that agency personnel found it "inexplicable" that the original test results were normal in someone who was a carrier for a genetic disorder.

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Case h: A married woman learned that she was at-risk for Huntington disease when she was 25 years old. A year later she and her husband decided to adopt a child on the advice of her physician. The latter told her it would be better for her not to have her own children and that she could easily adopt. She therefore underwent a tubal ligation and the couple began the adoption process. The adoption agency application asked why the couple was not able to have children biologically, inquired about the presence of hereditary disorders, and required certification from a doctor that the couple was sterile. Shortly after filing the application, the couple received a letter from the adoption agency refusing them the opportunity to adopt based on the woman's risk of Huntington disease.

Case i: A birth mother with Huntington disease was refused the opportunity to place her child up for adoption through a state adoption agency but the child was accepted by a private agency. A couple with one member at-risk for Huntington disease had been unsuccessful in trying to adopt a child who was assumed to be genotypically normal. However, that at-risk couple was permitted to adopt the at-risk infant.

• Armed Services

The case described below involving the armed services shows that even institutions as structured as the military may not have a consistent policy with regard to people at-risk for genetic conditions.

Case j: An individual enlisted in the Air Force and revealed his (approximately 50%) risk status for developing Huntington disease. When applying for reenlistment, he was discharged due to his risk status, although he was asymptomatic. The brother of this individual served in the Marines (who were aware of his risk status) until he became symptomatic for Huntington disease at which time he received a medical discharge and treatment at a V.A. hospital.

• Employers

In many of the cases involving employment, individuals believed that they were not hired or were fired because they were at-risk for genetic conditions. In other cases, individuals who were employed reported that they were reluctant to seek either a more desirable job or a job in a different location because they feared that they would be unable to obtain health insurance in their new position.

Case k: A 24 year old woman was fired from her job as a social worker shortly after her employers learned that she was at-risk to develop Huntington disease. In the eight-month period prior to her termination she received three promotions and outstanding performance reviews. However, while conducting an in-service training on admitting and caring for Huntington disease patients, she revealed that she had a family member with Huntington disease. Shortly afterwards, she was given a poor performance review. Her employers declined to give examples of poor performance. She was soon fired and told by a co-worker that the employer was concerned about her risk to develop Huntington disease.

Case l: A 53 year old man was interviewed for a job with an insurance company. During his first interview he revealed that he had hemochromatosis but was asymptomatic. During the second interview, the company representative told him that the company would be interested in hiring him but would not be able to offer him health insurance because of his hemochromatosis. He agreed to this condition. During the third interview he was told that although they would like to hire him, they were unable to do so because of his hemochromatosis.

• Educational institutions

Our study elicited a few reports of genetic discrimination occurring in educational institutions. As is the case for the examples described above, the examples of genetic discrimination by educational institutions involved the denial of opportunities to apparently qualified individuals because of a perceived genetic abnormality in those individuals.

Case m: In a small town, two healthy children attended the same school as their disabled brother. That brother had MPS II and attended a special education class. When in second grade, one of the healthy children was judged to have poor penmanship. A teacher decided that this indicated the onset of MPS II and sent the child back to first grade without consulting the parents or a physician. The parents protested and the child was placed in the appropriate grade.

• Blood Banks

Twenty-two respondents with hemochromatosis reported that they were not able to donate blood. The American Red Cross has a policy of rejecting blood donations from all individuals with hemochromatosis arguing that the donations are treatments, not gifts[16]. A significant number of respondents stated that they donated blood because their health insurance would not pay for phlebotomy treatments. In some cases, blood banks were willing to perform phlebotomies as treatment for a fee. Several of these cases have previously been discussed[11]. In the case below, prejudice or ignorance of a medical condition apparently played a role in inappropriately denying a potential donor the opportunity to give blood.

Case n: A man who had regularly donated blood for a number of years was refused phlebotomy when the nurse responsible for scheduling at the local blood bank learned that he had been diagnosed with Huntington disease. Donating blood was important to this man as a way of making a contribution to society. His neurologic findings, if any, were apparently not an issue since the director of the blood bank invited him to resume his blood donations once that particular nurse retired.

Personal reactions among people at-risk for genetic discrimination

Not unexpectedly, experiencing one or more episodes of genetic discrimination engenders a gamut of personal/psychological reactions for both the affected individual and, often, for other family members. These involve loss of self-esteem, alienation from family members and others, and alterations in family dynamics. For example, some individuals reported that they felt stigmatized and unworthy of marriage or that

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they should only marry disabled individuals. Others behaved as if they had a genetic condition even though they had not been diagnosed. In some instances, family members blamed each other for problems caused by a genetic diagnosis. The cases excerpted below illustrate some of these complex feelings and interpersonal dynamics.

Case o: A woman at-risk for Huntington disease chose her profession (school teacher) because it provided good benefits, especially disability benefits. Although some of her friends know of her risk she has kept the information from her employers as she "doesn't trust" them.

Case p: A woman at-risk for Huntington disease reported that friends and co-workers continually pressure her to undergo genetic testing arguing that if information can be obtained, she should not remain ignorant. As a result, she feels less free to discuss her risk status with friends and is resentful of the intrusion.

Case q: A woman learned of her risk of Huntington disease in adolescence. She reported feeling that she could not have a "normal" life, had no interest in marriage or children, and chose a career which had good disability benefits available. Later, genetic testing revealed that she was at low risk to develop Huntington disease and since then has actively pursued changes in career and marriage.

Strategies to avoid genetic discrimination

Many individuals utilized strategies to avoid experiencing genetic discrimination. These strategies included purchasing insurance policies prior to genetic testing, being tested anonymously, paying out-of-pocket for tests so that insurance companies would not obtain the results, providing partial disclosure of relevant information and, sometimes, providing incorrect information (see also reference 17).

Individuals reported avoidance of genetic testing or situations where genetic information could be used against them. For example, several of these respondents reported that they had never been rejected for insurance because they had not applied for insurance: they stated the belief (true in the case of Huntington disease) that their applications would necessarily be denied.

Case r: The parent of an individual died of Huntington disease. Fearing adverse consequences at work if the diagnosis became known, the individual arranged for the diagnosis "asphyxiation" to be reported as the cause of death so as to avoid mention of the disease in an obituary.

Case s: An 18 year old man, at-risk for Huntington disease, wished to enlist in the Marines in order to serve in the Persian Gulf War. He believed it unlikely that he would become symptomatic during his tour of duty but that his risk status would disqualify him from service. He therefore answered "no" to questions regarding hereditary disorders on his application and did not include Huntington disease in his family medical history.

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Knowledge of the Medical Information Bureau

The Medical Information Bureau, Inc. (MIB) is a private, non-profit corporation which provides insurance companies with medical and certain non-medical information about potential insurees[18]. Member organizations (mostly insurance companies) have access to its computerized data bank of information about individuals. Thus, genetic information provided by the MIB could result in inappropriate discrimination in obtaining health or life insurance, particularly if recorded data are incorrect or misleading[18,19]. The MIB does offer the opportunity for individuals to examine their MIB records and request corrections. However, doing so requires knowledge of the existence and function of the MIB. We therefore questioned individuals concerning their awareness of the MIB. Only 10 of the 55 (18%) respondents asked about the MIB knew of its existence and none had asked for access to their MIB records.

Knowledge and use of state insurance commissions

State insurance commissions are charged with regulating the insurance industries in their states. Consequently, appeal to a state insurance commission is one mechanism for challenging a perceived discriminatory decision regarding procurement of insurance. Therefore, study participants were sampled regarding their knowledge of state insurance commissions. Nineteen out of 58 respondents (33%) knew of the existence of state insurance commissions. Many of the nineteen thought the purview of these commissions was limited to automobile insurance.

DISCUSSION

The purpose of this study was to determine the varieties and impact of genetic discrimination using the case studies of individuals having, or who were at-risk for, abnormal genotypes. The results extend the findings of the pilot study: genetic discrimination exists and has significant impacts on individuals and their families. The pilot study provided evidence of genetic discrimination in several social arenas: health insurance, life insurance, and adoption. This study confirmed these findings, providing many more instances of genetic discrimination in each of these areas and in identifying additional institutions engaged in discrimination.

This study also revealed several additional ramifications of the phenomenon of genetic discrimination. First, some cases were reported in which clinical professionals appeared to give judgmental and possibly coercive counsel to persons who were at-risk for abnormal genotypes and/or who were at-risk of having offspring with abnormal genotypes. While all individuals, professional or otherwise, have personal positions on ethical issues, the imposition of a clinician's values with respect to reproductive decisions is regarded as inappropriate[20]. It is not possible to determine whether the alleged incidents occurred exactly as they were related to us. Nonetheless, even if some of these cases reflect a misunderstanding between the health care provider and patient, they suggest that poor communication can give rise to the perception of discrimination.

Second, while this study was not designed to evaluate the psychological effects of genetic discrimination, the pervasiveness and importance of these effects became

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apparent. The cases that we have summarized illustrate typical feelings expressed by respondents. Individuals reported stigmatization by relatives, friends, and other members of their communities. Some respondents reported that a genetic diagnosis resulted in feeling a loss of self-worth; others reported feeling powerless to challenge adverse decisions. These responses were common and indicate the significant impact that discrimination can have on people's lives.

Third, many of the individuals reported that they had not experienced genetic discrimination. However, comments on their questionnaires revealed that many of them had adopted strategies to ensure that others would not learn of their genetic backgrounds. Apparently they perceived a possibility of genetic discrimination and took action to avoid it.

Fourth, most respondents lacked either the information or the inclination to deal with the discrimination they encountered. For example, although it might appear that state insurance commissions are the appropriate avenues for redress of grievances against insurance companies, our study found that only 19 out of 58 individuals who reported experiencing discrimination knew of the existence of state insurance commissions. A recently published study found that state insurance commissioners were unaware of incidents of genetic discrimination[21]. Our results indicate that this lack of awareness arises not because genetic discrimination does not exist as was suggested by that study, but rather because consumers did not appeal to the insurance commissions.

Even individuals who were aware of regulatory agencies often did not avail themselves of opportunities to redress their grievances. Some of these individuals felt they had little hope of successfully challenging discrimination. In one case, an attorney who had been denied health insurance because of diagnosis of (asymptomatic) hemochromatosis, did not reapply for insurance or fight the denial because she "didn't want to be reminded" that she had a genetic diagnosis.

In order to increase our understanding of the causes of genetic discrimination and thus suggest strategies for minimizing discriminatory incidents, we attempted to determine whether instances of genetic discrimination occurred as a result of ignorance on the part of the discriminating institution or as a result of institutional policy. The study revealed instances of both causes of discrimination and, in some instances, the causes were difficult to distinguish. For example, in several reported cases involving a few specific companies, life insurance agents in a branch office were unaware of company policy that individuals with asymptomatic hemochromatosis should not be denied an insurance policy. This pattern suggests not only ignorance on the part of the agents, but may suggest that the company is not troubled by and may even condone such ignorance.

Other cases provide additional examples of ignorance giving rise to genetic discrimination. Representatives of an adoption agency reportedly thought that parents of children with MPS could not be healthy. Some teachers and school officials apparently believed that unaffected siblings of affected relatives must themselves be at-risk for becoming ill and in need of special treatment.

The single clear example of genetic discrimination due to institutional policy is that of the American Red Cross's refusal to accept blood donations from people with hemochromatosis. Because the standard treatment for hemochromatosis, phlebotomy,

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is not covered by many health insurers, some individuals donate blood as an alternative to treatment. There is apparently no medical reason to restrict the use of this blood; the policy of the American Red Cross to refuse blood from hemochromatosis patients is based on the non-altruistic nature of the donation[16].

The instances of differential treatment based on genetic tests described in this paper raise questions concerning the legality of this treatment. Under what circumstances is it legal to limit an individual's opportunities for employment, insurance, education, or for adopting a child on the basis of genetic information? Existing federal legislation such as the Rehabilitation Act of 1973 and the Americans with Disabilities Act of 1990 (ADA) provide that much differential treatment against those with disabilities is unlawful. The question of whether the ADA provides protective coverage for individuals who have abnormal genes but are asymptomatic or presymptomatic has been the subject of considerable debate[22-23]. Recent guidelines from the Equal Employment Opportunity Commission (EEOC) regarding the definition of "disability" under the ADA specifically address the issue of genotypically abnormal individuals who are asymptomatic or presymptomatic and states that those individuals are covered under the definition of disability in the ADA if they are regarded as disabled[24].

Although the ADA provides broad legal protections against genetic discrimination, it is apparent that, for the most part, its provisions do not apply to insurance underwriting[6,7,18,24]. There is great concern that insurance companies are abandoning community rating, in which all people in a given geographical area pay the same premiums, in favor of underwriting, that is, setting premiums on the basis of an individual's risk. In principle, there is no need for underwriting since the rates set in community rating reflect the known incidence of morbidity and mortality in that geographical area. However, insurance companies are concerned that other companies will skim, i.e., insure only those individuals with the lowest risks. In addition, they fear that individuals who know that they are at risk for increased morbidity or mortality will buy an excessive amount of insurance[18]. Our study showed that this possibility is real; several individuals who were at-risk for Huntington disease reportedly attempted to buy life insurance or increase the amount of their coverage when they first learned of their at-risk status or when symptoms of the disease appeared. Given these pressures on insurance companies to resort to underwriting, it is likely that there will be increasing use by insurance companies of genetic as well as other sophisticated medical tests.

Since federal regulation of insurance companies is extremely limited, any meaningful restrictions of the use of genetic tests by insurance companies will need to arise from state legislation. States vary widely in their regulation of genetic testing by insurance companies[18,25]. Some states have already enacted legislation prohibiting or limiting the use of genetic information in insurance underwriting. Although at this time there is little prospect of a universal health care policy in the United States, any health care policy will have to deal with the increasing use of genetic tests in medicine and by insurance companies.

This study of genetic discrimination is limited in several respects. The respondents in this study are members of genetic disease support groups who do not represent the entire population of affected individuals. They are also a self-selected subgroup of the

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membership of those groups. In addition, documentation of discrimination was difficult to obtain in many instances. Some types of discrimination such as employment discrimination cannot be documented easily. In other instances, records of discrimination were not kept, especially if the individuals believed that there was no recourse for appeal of an adverse decision. Thus, it is difficult to determine to what extent reports of genetic discrimination are of actual rather than perceived discrimination.

The low response rate to the questionnaires is due to several factors besides the actual incidence of genetic discrimination. No follow-up mailings of the questionnaire were done, a practice which greatly increases the response rate. In addition, the mailing lists of the disease organizations include many individuals who are not personally affected by the disorders and so would not respond to the questionnaire. This is reflected in the fact that some questionnaires were returned by people who noted that they were not appropriate respondents. It is also possible that the response rate reflects a real result; that while genetic discrimination exists at this time it is not a widespread phenomenon.

Finally, we emphasize that this study is not a survey, but rather and attempt to collect case studies in order to examine the varieties of genetic discrimination. Consequently any statistical analysis of the cases would be both inappropriate and unnecessary.

This first extensive study of genetic discrimination extends and confirms the results of earlier ones. The cases from this study are consistent with the interpretation that although not systematic, genetic discrimination does occur in a wide variety of contexts and can cause hardship to affected individuals and their families. Many instances of genetic discrimination described in this study are similar to other types of discrimination. However, the distinctive nature of this type of discrimination lies in its effect on individuals who are asymptomatic and may never become symptomatic. Because the number and use of genetic tests is expanding rapidly and will continue to increase, it is vital that standards be developed in the near future to ensure that genetic information be used fairly. As our society struggles to be more equitable in its treatment of people regardless of race, age, or gender, it cannot ignore or justify inequities based on genotype.

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Table 1. Questionnaire

1.	What is your genetic diagnosis?
2a.	Do you think that you may have been refused social benefits or denied opportunities because of your diagnosed condition?
2b.	If no, do you have any concerns for the future, or other comments?
3.	In what year did the event/s occur?
4.	Around what issue did you experience difficulties? (Health insurance, life insurance, adopting children, military, social services, church/synagogue, community/neighbors, other (please specify).
5.	Please describe your experiences, explaining why these are discriminatory.
6.	Are there any other comments that you would like to make?
7.	Please note which institution sent you this questionnaire.
8.	May we contact you for more information? (name, address, telephone)

The above questions were distributed in a questionnaire to individuals associated with genetic disease support groups who were likely to have a genetic diagnosis (see Methods, pp. 73-75). In addition to the questions listed above, the questionnaire had a brief definition of genetic discrimination and an assurance of confidentiality. A nearly identical questionnaire was distributed to individuals likely to be carriers for a genetic disorder.

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Table 2. Partial List of Questions from the Telephone Guide Used for Interviews on Genetic Discrimination

Number of children/family members affected by the condition
Pedigree
Presence or absence of confounding disabilities
Context of the occurrence or the event
(Insurance, employment, public entities & accommodations/housing, education, government, community, other)

Insurance
Type? (Health, life, disability, automobile, home/mortgage, commercial loan)
Obtaining, renewing, or switching insurance?
Company name?

Employment
(Hiring, promotion, transfer, job responsibilities, compensation, eligibility for benefits, provision for disability, association with someone disabled, other)
Company name?
Employer/title?
Type of job and relevance of the condition to job performance
Was physical accessibility to an activity curtailed in any way?
Were reasonable accommodations requested? If so, were they provided?
After the incident, where did you work?
If you changed jobs, why?
Describe educational background and qualifications for job
Jobs held before and after (title/duties/length of time/ why left)

Public entities and accommodation
(Adoption agency, public housing, obtaining a loan, professional licensing, other licensing, transportation services, place of education, day care center, recreational facility, other)

Education
(Admission, activity restriction, termination, health service, other)

Government
(Military, benefits, social security entitlement, federal, state, local, other)
Military
(Entrance, transfer, job responsibilities, activity restrictions, termination, promotion, other)
Army, Navy, Air Force, Marines, National Guard, other

Benefits
(type: eligibility criteria and dates)

Community
(Neighborhood, religious community, recreational facilities)

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Table 2 continued: Partial List of Questions from the Telephone Guide Used for Interviews on Genetic Discrimination

Details of Discrimination

How was information about your condition revealed?
Did the institution get information from the Medical Information Bureau? What information did they get? How did you find out?
What did you request and why? (describe all events, contacts/correspondence which precede the institution's denial)
Who did you first contact? (include job title)
Type of correspondence (letters, phone, person)
What, if any, additional medical information were you required to disclose? (May we receive a copy of their request?)
Were you given a reason why this information was required?
Did you voluntarily submit additional information or medical letters of support? (May we receive documentation?)
Did you seek help from an outside source, such as personnel, other people you know, or a disease support group, etc.?
What was the nature of their reply?
(refusal to consider case, request for additional information—if so, what was requested?)
Were you or your physician requested to submit information? Other?
Did the person making the decision explain to you what they thought and why? How was this communicated to you?
How long did the institution take to respond to your initial inquiry?
Was the response made in person? By phone? By letter? (Personalized form or letter? May we obtain a copy? If not, why?)
Who replied? (job title)
Did this person continue to handle your case, or was it referred to a supervisor? If so, how high up within the organization did consideration of your case go? Did you ever request that a supervisor take charge?

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Table 3. Tabulation of questionnaires

	Questionnaires (returned / sent)	Reporting genetic discrimination yes	no	ambiguous
Huntington disease	623 / 25,924	276	329	18
Hemochromatosis	138 / 1,250	53	85	0
MPS	57 / 420	44	10	3
Phenylketonuria	22 / 200	12	8	2
Other*	77 / none	70	5	2
Total	917 / 27,790	455@	437	25

* This category includes questionnaires with information regarding disorders other than those listed above and those where it was not possible to ascertain the disorder the individual had. Questionnaires returned blank are not included.

@ The 206 interviewees were from the 385 individuals in this category reporting an association with Huntington disease, hemochromatosis, MPS, or phenylketonuria. See Methods (pp. 73-75) for a more detailed description of the selection of individuals interviewed.

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Attachment 2

NATIONAL COALITION FOR HEALTH PROFESSIONAL EDUCATION IN GENETICS

Catalyzed by the American Medical Association, the American Nurses Association, and the National Human Genome Research Institute, the National Coalition for Health Professional Education in Genetics is an effort to provide an organized, systematic, and national approach to the provision of genetics education for all health care professionals.

The Coalition is comprised of leaders from approximately 100 diverse health care professional organizations, consumer and voluntary groups, government agencies, industry, managed care organizations, and genetics professional organizations (please see attached list of participant organizations).

Recognizing the rapid pace of human genetics research and its impact on an increasing number of health care disciplines, the mission of the National Coalition for Health Professional Education in Genetics is to ensure that our nation's health care providers have the knowledge, skills, and resources to effectively and responsibly integrate new genetic knowledge and technologies into the prevention, diagnosis, and management of disease.

The goals of the Coalition are: to stimulate and formalize interest in establishing genetics education as a top priority for health care professional organizations and their memberships; to create mechanisms for collaboration between member organizations; to identify and coordinate existing and future genetics activities for health care professionals; and to develop new initiatives to meet identified needs.

The first meeting of the Coalition's full membership was held March 10, 1997, at the National Institutes of Health. Top priorities for the Coalition as identified by members at that meeting are: the development of a comprehensive, web-based genetics information center; the development of a core curriculum in genetics for health professionals to serve as a template that can be modified according to discipline; and the provision of incentives for providers (such as integration of genetics questions into certification and licensure exams) to learn about genetics. Interdisciplinary working groups are currently being formed to begin tackling these tasks.

THE NATIONAL COALITION FOR
HEALTH PROFESSIONAL EDUCATION IN GENETICS
Participant Organizations
Meeting of March 10, 1997

May 22, 1997

**** = Steering Committee Member Organization**

1 Agency for Health Care Policy Research
2 **Alliance of Genetic Support Groups ****
3 American Academy of Family Physicians
4 American Academy of Insurance Medicine
5 American Academy of Nursing
6 American Academy of Oral Medicine
7 American Academy of Pediatric Dentistry
8 American Academy of Pediatrics
9 American Academy of Physician Assistants
10 American Association for Dental Research
11 **American Association of Colleges of Nursing ****
12 American Association of Colleges of Osteopathic Medicine
13 American Association of Colleges of Pharmacy
14 American Association of Critical Care Nurses
15 American Association of Dental Schools
16 American Association of Occupational Health Nurses
17 American Association of Orthodontists
18 American Association on Mental Retardation
19 American Board of Genetic Counselors
20 American Cancer Society
21 American Cleft Palate-Craniofacial Association
22 American College of Cardiology
23 **American College of Medical Genetics ****
24 American College of Obstetricians and Gynecologists
25 American College of Physicians
26 American College of Preventive Medicine
27 American Dental Hygienists Association
28 American Dietetic Association
29 American Federation for Medical Research
30 **American Medical Association****
31 American Medical Informatics Association
32 **American Nurses Association ****
33 American Occupational Therapy Association
34 American Organization of Nurse Executives
35 American Osteopathic Association
36 American Pediatric Society
37 American Psychological Association

38 American Public Health Association
39 American Society for Clinical Laboratory Science
40 American Society of Clinical Oncology
41 **American Society of Human Genetics ****
42 Armed Forces Institute of Pathology
43 Association for Behavioral Science and Medical Education
44 Association of Academic Health Centers
45 **Association of American Medical Colleges ****
46 Association of Asian Pacific Community Health Organizations
47 Association of Professors of Human and Medical Genetics
48 Association of Professors of Medicine
49 **Association of Schools of Allied Health Professions ****
50 Association of Schools of Public Health
51 Association of Teachers of Preventive Medicine
52 Association of Women's Health, Obstetric, and Neonatal Nurses
53 **Biotechnology Industry Organization ****
54 Centers for Disease Control and Prevention
55 College of American Pathologist
56 Council of Medical Genetics Organizations
57 **Council of Medical Speciality Societies ****
58 Council of Regional Networks of Genetic Services (CORN)
59 Council on Social Work Education
60 DANA Alliance for Brain Initiatives
61 Department of Veterans Affairs
62 Federation of Special Care Organizations
63 Foundation for Blood Research
64 **Health Resources and Services Administration, Maternal and Child Health Bureau ****
65 Howard Hughes Medical Institute
66 International Patient Advocacy Association
67 **International Society of Nurses in Genetics ****
68 Joint Commission on Accreditation of Healthcare Organizations
69 **March of Dimes Birth Defects Foundation ****
70 Medical College of Virginia
71 National Action Plan on Breast Cancer
72 National Association of Community Health Centers, Inc.
73 National Association of Neonatal Nurses
74 National Association of Pediatric Nurses Associates and Practitioners
75 National Association of School Nurses
76 National Association of Social Workers
77 National Board of Medical Examiners
78 **National Cancer Institute, National Institutes of Health ****
79 National Center for Genome Resources
80 National Heart, Lung, Blood Institute, National Institutes of Health

81 **National Human Genome Research Institute, National Institutes of Health ****
82 National Institute of Child Health and Human Development, National Institutes of Health
83 National Institute of Dental Research, National Institutes of Health
84 National Institute of Diabetes and Digestive and Kidney Diseases, National Institutes of Health
85 National Institute of Environmental Health Services, National Institutes of Health
86 National Institute of General Medical Sciences, National Institutes of Health
87 National Institute of Mental Health, National Institutes of Health
88 National Institute of Neurological Disorders and Stroke, National Institutes of Health
89 **National Institute of Nursing Research, National Institutes of Health ****
90 National Institute on Aging, National Institutes of Health
91 National Institute on Alcohol Abuse and Alcoholism, National Institutes of Health
92 National Institute on Deafness and Other Communication Disorders, National Institutes of Health
93 National Institute on Drug Abuse, National Institutes of Health
94 National Marfan Foundation
95 National Medical Association
96 **National Organization for Rare Disorders ****
97 National Organization of Nurse Practitioner Faculties
98 **National Society of Genetic Counselors ****
99 **Nursing Organization Liaison Forum ****
100 Office of Rare Disease Research, National Institutes of Health
101 Office onWomens Health, Department of Health and Human Services
102 Oncology Nurses Society
103 Pharmaceutical Research and Manufacturers of America
104 Sigma Theta Tau, International, Inc.
105 Society of Gynecologic Oncologist
106 Society of Medical College Directors of CME
107 Society of Pediatric Nurses
108 Society of Pediatric Nurses Board
109 Uniformed Services Academy of Family Physicians

Attachment 3

Appendix 1

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SCIENCE

Genetic Discrimination and Health Insurance: An Urgent Need for Reform

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

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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.

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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 recessive disorder such as cystic fibrosis that might affect their children or future children.

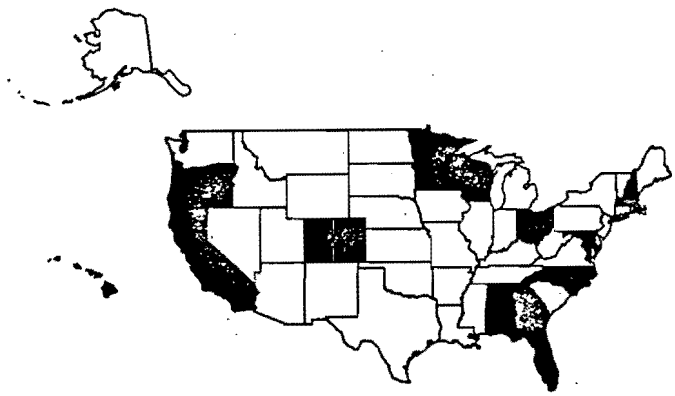

Fig. 1. State laws on the use of genetic information in health insurance (7). States shown in purple were the first states to enact legislation addressing genetic issues in insurance. Florida and Alabama laws prohibit insurers from denying coverage on the basis of the sickle cell trait. North Carolina 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, Arlyne Draper, Nancy Evans, Harmon Eyre, Leslie Ford, Janyce N. Hedetniemi, 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 Stopfer.
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 Kitcher, Joseph McInerney, Jeffrey Murray, Dorothy Nelkin, Rayna Rapp, Marsha Saxton, 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. Tilgham, and David Valle.

For the opportunity to participate in a discussion of the issues raised in this Policy Forum, go to the following URL (http://sci.aaas.org/aaas/policy).

Attachment 4

Appendix 3

GENETIC INFORMATION AND HEALTH INSURANCE ENACTED LEGISLATION*

STATE	CITATION	DESCRIPTION
Alabama (1982)	AL ST: 27-5-13	Prohibits health insurers from denying coverage because applicant has sickle cell anemia.
Arizona (1997)	H 2144	Prohibits disability insurers (includes health insurers) from rejecting an application or determining rates, terms or conditions on the basis of a genetic condition - in the absence of a diagnosis of the condition. Prohibits a person from requiring the performance of or performing a genetic test without written informed consent. Prohibits the release of the results of a genetic test without consent. Prohibits employers from failing or refusing to hire, from discharging or from otherwise discriminating on the basis of the results of a genetic test.
California (1994)	Insurance Code: §10123.3; §10140; §10148; §10149; §10149.1; §11512.95 Health & Safety Code: §1374.7	Prohibits health insurance plans from refusing to enroll or accept persons based on genetic characteristics; Prohibits health insurers from requiring a higher rate or charge on the basis of genetic characteristics; Provides for privacy protection of genetic information.

*This chart reflects legislation enacted as of May 15, 1997. These state laws were designed to address discrimination and/or privacy issues specifically regarding genetic information and health insurance. In addition to these laws, at least eight states have enacted health insurance portability laws in 1997. These portability laws contain provisions preventing health insurers from basing eligibility on genetic information.

This chart supplements K. H. Rothenberg, *Genetic Information and Health Insurance: State Legislative Approaches*, 23 JOURNAL OF LAW, MEDICINE & ETHICS 312 (1995). Chart prepared by Professor Karen Rothenberg, University of Maryland School of Law, and Ms. Barbara Fuller, NAPBC.

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California (1995)	Insurance Code: §10123.3; §10140; §10147; §11512.95; §10123.31; §10123.35; §10140.1; §10140.5; §11512.96; §11512.965 Health & Safety Code: §1374.7; §1374.9	Prohibits health insurance plans from offering or providing different terms, conditions or benefits on the basis of genetic characteristics.
California (1996)	Civil Code: §56.17 Health & Safety Code: § 1374.7 Insurance Code: §742.24; §742.405; §742.407; § 10123.3; §10123.35; §10140; §10140.1	Prohibits health insurers from seeking, using or maintaining genetic information for any nontherapeutic purposes. Prohibits health insurers from discriminating in the renewal of policies on the basis of genetic characteristics. Revises the definition of genetic characteristics to include family history. Applies prohibitions on genetic discrimination by health insurers to "multiple employer welfare arrangements."
Colorado (1994)	Title 10, Art. 3, Part II: §10-3-1104.7	Prohibits the utilization of information derived from genetic testing from being used to deny access to health care insurance. Provides for privacy protection of genetic information.
Florida (1978)	FL ST: 626.9707	Prohibits insurers from refusing to issue and deliver any policy of "disability" insurance, which "affords benefits and coverage for any medical treatment or service," solely because a person has the sickle cell trait. Prohibits a "disability" insurance policy from charging a higher rate solely because a person has the sickle cell trait.
Florida (1992)	FL ST: 760.40	Provides for informed consent and privacy protection of genetic information. Provides for mandatory reanalysis if the utilization of genetic information results in a denial of insurance.

This chart supplements K. H. Rothenberg, *Genetic Information and Health Insurance: State Legislative Approaches*, 23 JOURNAL OF LAW, MEDICINE & ETHICS 312 (1995). Chart prepared by Professor Karen Rothenberg, University of Maryland School of Law, and Ms. Barbara Fuller, NAPBC.

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Georgia (1995)	Title 33, Chapter 54	Prohibits the use of genetic testing except to obtain information for therapeutic or diagnostic purposes. Provides for written consent prior to genetic testing. Provides for privacy protection of genetic information. Prohibits health insurers from seeking information derived from genetic testing.
Indiana (1997)	H 1684	Prohibits insurers, other than life insurers, from obtaining the results of any genetic screening or testing without a separate written consent. Provides that an insurer is not liable if they inadvertently receive the results of genetic testing or screening. Provides that an insurer that inadvertently receives testing or screening results may not use the genetic testing or screening results in violation of other sections of the law. Prohibits health insurers from requiring an individual to submit to genetic screening or testing when processing an application for coverage or in determining insurability. Prohibits health insurers from considering any information obtained from genetic screening or testing in a manner adverse to the applicant or an individual already covered. Prohibits health insurers from inquiring, directly or indirectly, into the results of genetic screening or testing, or from using such information to cancel, refuse to issue or renew, or limit benefits. Prohibits health insurers from making a decision adverse to an applicant based on entries related to the results of genetic testing or screening in medical records or other reports of genetic screening or testing. Prohibits health insurers from developing and asking questions regarding the medical history of an applicant that reflect the results of or are questions designed to ascertain the results of genetic screening or testing. Prohibits health insurers from canceling, refusing to issue, refusing to renew, or refusing to enter into a contract based on the results of genetic screening or testing.

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Indiana (1997) - continued	H 1684 - continued	Prohibits health insurers from delivering, issuing for delivery, renewing or executing a contract that limits benefits or establishes premiums based on the results of genetic screening or testing. Provides for health insurers to consider the results of genetic screening or testing if the results are voluntarily submitted by the applicant seeking renewal of coverage AND if the results are favorable to the applicant.
Maryland (1986) Maryland (1996)	Insurance Code: Art. 48A, §223(b)(4) S 276 (Ch. 24)	Prohibits health insurers from making or permitting differentials in rates based on any genetic trait, unless there is actuarial justification. Prohibits health insurers from using a genetic test or the results of a test to reject, deny, limit, cancel, refuse to renew, increase the rates of, affect the terms or conditions of, or otherwise affect a health insurance policy or contract. Prohibits health insurers from requesting or requiring a genetic test for the purpose of determining whether or not to issue or renew health benefits coverage. Prohibits the release of the results of a genetic test without the prior written authorization of the individual.
Minnesota (1995)	S.F. No. 259	Prohibits health insurers from utilizing information from genetic testing to determine eligibility, establish premiums, limit coverage, or renew coverage. Prohibits health insurers from requiring a genetic test and from inquiring or determining whether or not an individual has had a genetic test.
New Hampshire (1995)	NH ST: Chapter 141-H	Prohibits health insurers from conditioning the provision of health insurance coverage on the results of genetic testing. Prohibits health insurers from considering genetic testing in the determination of rates or benefits. Prohibits health insurers from requiring a genetic test and from inquiring or determining whether or not an individual has had a genetic test.

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New Jersey (1996)	S 695 A 1411	Prohibits health insurers from discriminating in the issuance, withholding, extension, renewal, or establishment of rates, terms or conditions on the basis of genetic information. Prohibits any person from obtaining or retaining an individual's genetic information without first obtaining authorization from the individual. Prohibits the disclosure of genetic information in a manner that permits identification of the individual without the authorization of the individual.
New York (1996)	S 4293-D A 5796-C	Prohibits authorized insurers from requesting or requiring an individual proposed for insurance coverage to be the subject of a genetic test without receiving the written informed consent of such individual prior to such testing.
North Carolina (1975)	NC ST: §58-65-70	Prohibits health insurers from refusing to issue health insurance because an individual has sickle cell trait or hemoglobin C trait. Prohibits health insurers from charging higher premiums based on sickle cell trait or hemoglobin C trait.
Ohio (1993)	OH ST: §1742.42; §1742.43; §3901.49; §3901.491; §3901.50; §3901.501	Prohibits health insurers from canceling, refusing to issue, or renewing coverage or limiting benefits based on genetic screening or testing. Prohibits health insurers from requiring a genetic test or making an inquiry as to the results of genetic screening or testing. Provides for consideration of genetic testing if the results are favorable to the applicant and voluntarily submitted. Establishes the Task Force on Genetic Testing in Health Insurance.

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Oklahoma (1996)	HCR 1113	Creates the Task Force on Prevention of Genetic Discrimination. The duties of the Task Force include: reviewing House Bill 2478; reviewing legislation from other states and any model legislation related to genetic discrimination; and, making recommendations to the legislature by January 1, 1997 concerning proposed legislation which the Task Force deems necessary to prevent genetic discrimination particularly with regard to insurance and employment.
Oklahoma (1997)	HCR 1012	Creates the Task Force on Prevention of Genetic Discrimination. The duties of the Task Force include: - reviewing H.B. 2478 of the 2nd Session of the 45th Oklahoma Legislature which proposes the creation of a Genetic Nondiscrimination Act for the State of Oklahoma - reviewing legislation from other states and any model legislation related to genetic discrimination; and - making recommendations to the Legislature by January 1, 1998, concerning proposed legislation which the Task Force deems necessary to prevent genetic discrimination, particularly with regard to insurance and employment.
Oregon (1995)	OR ST: 659.036; 659.227	Prohibits health insurers from utilizing genetic information to reject, deny, limit, cancel, refuse to renew, increase the rates of, or affect the terms and conditions of health insurance policies. Provides for informed consent and privacy protection of genetic information.
Tennessee (1997)	H 413	Prohibits insurers from denying or canceling health insurance coverage on the basis of genetic information or on the basis that the individual or a family member of an individual has requested or received genetic services. Prohibits insurers from varying the premiums, terms or conditions for health insurance on the basis of genetic information or on the basis that the individual or a family member of an individual has requested or received genetic services. Prohibits insurers from requesting or requiring the disclosure of genetic information. Prohibits insurers from disclosing genetic information without prior written authorization.
Virginia (1995)	S.J.R. 372	Establishes a subcommittee to study the legal and policy ramifications of breast cancer susceptibility gene research, including the ethical and legal issues of health insurance coverage and reimbursement.

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