As part of an ongoing collaboration funded by The Pew Charitable Trusts, the Genetics and Public Policy Center at Johns Hopkins University, the Georgetown Health Policy Institute, and the National Workrights Institute are pleased to submit the following comments in response to the Request for Information on the Genetic Information Nondiscrimination Act, issued October 10, 2008 by the Departments of Treasury, Labor, and Health and Human Services.

The Genetics and Public Policy Center

Established in 2002 with a grant from The Pew Charitable Trusts, the Genetics and Public Policy Center at Johns Hopkins University is a multi-disciplinary center working to help policy makers and the public better understand and respond to the challenges and opportunities arising from rapid advances in human genetics and their application to health care. The Center has conducted in-depth policy analysis and social science research, including public opinion and attitude research, on genetic testing and genetic technologies. We have provided technical assistance to Members of Congress and Congressional staff throughout consideration of GINA. We have had the opportunity to testify in front of several Congressional committees considering GINA, and our research on public attitudes toward the use of genetic information was cited during final consideration of the bill on the House and Senate floors. Individual members of the Center's staff have been integrally involved in aspects of genetic nondiscrimination legislation since the first versions of legislation were proposed in 1995.

Our interest in genetic discrimination policy is inexorably linked to our belief that genetic research will lead to a clearer picture of the role of genetics in health and disease and help drive the development of new diagnostic tools and treatments. Researchers now have powerful tools to dissect the genetic, environmental, and lifestyle factors that contribute to health and disease, and our nation's robust biotechnology and pharmaceutical industries are translating those findings into new diagnostics and medicines to preserve health and prevent disease. We have long been concerned that genetic discrimination, and the fear expressed by many that genetic test results could be used against them, inhibits volunteers from participating in genetic research and deters individuals from pursuing recommended genetic testing in their own health care. The regulations that implement GINA must carry out the intent of Congress by clearing the way for new scientific and clinical advancements aimed at improving the health of all Americans.

The Georgetown Health Policy Institute

The Georgetown Health Policy Institute is a multi-disciplinary group of faculty and staff dedicated to conducting research on key issues in health policy and health services research.

Institute members are engaged in a wide diversity of projects, focusing on issues relating to health care financing, the uninsured, federal health insurance reforms, quality of care and outcomes research, mental health services research, and the impact of changes in the health care market on providers and patients.

Karen Pollitz directs the Institute's research on private health insurance markets and regulations. She and her colleagues co-authored a recent study, "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices" in the journal *Inquiry*. Ms. Pollitz also provided expert testimony on health insurance genetic discrimination to Congress during consideration of GINA.

The National Workrights Institute

The National Workrights Institute (NWI) was founded in 1988 by the American Civil Liberties Union to extend protection for human rights into the American workplace – rights such as freedom of expression, privacy, due process, and freedom of association. In the two decades since, NWI has become the nationally recognized leader in workplace human rights. In 2000 the Institute became an independent organization.

NWI has been intimately involved with the issue of genetic privacy and discrimination from the beginning. NWI staff served on the groundbreaking advisory panel to the Congressional Office of Technology Assessment whose 1990 report first pointed out the potential for widespread genetic discrimination and the need for legal protection. NWI staff also served on the Commission on Genetics and Employment for the National Conference of State Legislators and worked on many of the state genetic nondiscrimination laws now in effect. Institute Legal Director Jeremy Gruber is a founder and co-chair of the Coalition for Genetic Fairness, the primary advocacy organization for the Genetic Information Nondiscrimination Act. He has spent the last twelve years advocating for the enactment of the Genetic Information Nondiscrimination Act, working with all stakeholders as well as Congressional members and their staffs.

I. DEFINITIONS IN GINA:

Summary

The definitions that appear in the final law are the core of the law and reflect a compromise reached after many years of negotiations.[1] Regulations should clarify for health plans and issuers what does and does not fall under the definitions. It may be useful for federal agencies to provide a non-exclusive list of examples under some of the definitions.

The definition of key terms related to genetics historically has presented a challenge for policymakers. For example, a problematic definition promulgated under HIPAA included, as part of the definition of genetic test, any information derived from "physical medical examinations," (29 CFR 2590.701-2) which created far too broad a scope. Conversely, state law definitions sometimes have been far too narrow, excluding family history or other aspects of genetic information. Rapid advances in genetic research and new technologies add to the challenge; some laws reflect an early understanding of genetics, but actual scientific progress quickly outpaces statutory language.

At the federal level, regulations must strive to respond to the latest scientific and medical advances, reflecting the best possible understanding of what Congress intended a term to encompass.

The key terms in GINA are "genetic information," "genetic test," and "genetic services."

GENETIC INFORMATION

The term "genetic information" means information about an individual's genetic tests, the genetic tests of that person's family members, and the manifestation of a disease or disorder in an individual's family members (sometimes referred to as "family history.") It also includes any request for, or receipt of, genetic services, or participation in clinical research that includes genetic services, by an individual or family members. "Genetic services" is defined separately and addressed below.

The definition of "genetic information" specifically includes the manifestation of a disease or disorder in a family member. "Family member" is defined as a first-, second-, third-, or fourth-degree relative. Individuals may become family members by birth, marriage, adoption, or intent to adopt; thus, the following family members are included whether the relationship is biological or legal:

- First-degree relatives: parents and siblings
- Second-degree relatives: grandparents, grandchildren, aunts, uncles
- Third-degree relatives: great-grandparents, first cousins, great-aunts and great-uncles.

[1] For a history and analysis of the compromises reached during GINA negotiations, see Baruch, S., and K. Hudson. 2008. Civilian and Military Genetics: Nondiscrimination Policy in a Post-GINA World. *The American Journal of Human Genetics* 83: 435-444 and Hudson, K.L, M.K. Holohan, and F.S. Collins. 2008. Keeping Pace with the Times — The Genetic Information Nondiscrimination Act of 2008. *New England Journal of Medicine* 358: 2661-2663.

- Fourth-degree relatives: great-great grandparents, first cousins once removed.

"Genetic information" does not include information about sex or age.

Regulations should clarify that all genetic information that meets the definition is protected. For example, genetic information of an individual (whether from family history or genetic testing) obtained by a health insurer before GINA's effective date is the current genetic information of that individual and is protected by GINA.

GENETIC TEST

The definition of "genetic test" in GINA is fairly technical. The law says that "genetic test" means an analysis of human DNA, RNA, chromosomes, proteins, or metabolites, to detect genotypes, mutations, or chromosomal changes.

According to the definition in Title I, the health insurance provisions of the law, "genetic test" does *not* include

''(i) an analysis of proteins or metabolites that
does not detect genotypes, mutations, or chromosomal
changes; or

''(ii) an analysis of proteins or metabolites that
is directly related to a manifested disease, disorder,
or pathological condition that could reasonably be
detected by a health care professional with appropriate
training and expertise in the field of medicine involved."

It may be useful for regulations to provide examples of protected tests and those that are not included. For example, results of the following tests would clearly be protected under the definition of "genetic test" in GINA:

- Tests for the Huntington disease mutation or BRCA1/BRCA2 (breast cancer) or HNPCC (colon cancer) mutations. These are examples of tests of human DNA to detect mutations.
- Carrier screening of adults using genetic analysis to determine the risk of conditions such as cystic fibrosis, sickle cell anemia, spinal muscular atrophy, and fragile X syndrome in future offspring. Carrier screening provides information to prospective parents about the risk of a future child having the disease. These tests generally are performed on human DNA to detect genotypes.
- Amniocentesis or Chorionic Villus Sampling to detect abnormalities in a fetus during pregnancy. These are tests of the fetus's human DNA or chromosomes to look for genotypes, mutations or chromosomal changes. Under GINA, the pregnant woman and

her family members explicitly are protected from discrimination on the basis of this genetic information.

- Newborn screening tests. These tests use either DNA or RNA analysis or protein or metabolite analysis to detect genotypes, mutations, or chromosomal changes. Tests for conditions such as PKU allow preventative treatment to begin before disease manifests in a newborn.
- Preimplantation genetic diagnosis performed on embryos created using *in vitro* fertilization. These are tests of the embryo's DNA or chromosomes to look for genotypes, mutations or chromosomal changes. Under GINA, the individuals and family members who "legally hold" the embryos explicitly are protected from discrimination on the basis of this genetic information.
- Pharmacogenetic tests. Tests to detect genotypes/mutations that are associated with how a person will react to a particular drug or drug dosage.
- DNA testing to detect genetic markers that are associated with information about ancestry.
- DNA testing that reveals family relationships, such as paternity.

While the last two examples are unlikely to be of interest or relevance to health insurers or employers, we include them to illustrate that Congress wrote definitions that do not rely on the purpose or intended use of the test.

Regulators should consult with scientific experts in this field such as the National Human Genome Research Institute or the American Society for Human Genetics to ensure that all genetic techniques are included.

The following tests would not be covered under GINA, as they do not meet the definition's requirements.

- Complete blood counts (CBC, or blood panels) which do not detect genotypes, mutations, or chromosomal changes.
- Cholesterol tests do not meet the requirements of the definition of genetic tests because they do not detect genotypes, mutations, or chromosomal changes. During consideration of GINA, the question arose whether a standard cholesterol test could be considered a genetic test because in rare cases it would reveal an extremely high cholesterol level associated with a genetic disease known as "hypercholesterolemia." However, in a case where a standard cholesterol test reveals such an extremely high cholesterol level, the test still would fail to meet the definition of genetic test. This issue was raised several times during negotiations of GINA and it is clear that Congressional intent was not to include cholesterol tests in GINA's definition.
- An HIV test. Although it is a retrovirus that inserts itsel*f into* human DNA, HIV is not itself human DNA, and measuring the presence of infectious agents such as bacteria, viruses, and fungi does not constitute a genetic test under the law's definition.

The exceptions stated in (i) and (ii) do not add much meaning to GINA that is not already present in the definition.

Exception (i) simply restates part of the rule in the definition, that unless a test of proteins and metabolites measures genotypes, mutations, or chromosomal changes, it does not meet the definition.

The text of (ii) may best be read as clarifying what is meant by "manifest disease." Exception (ii) states a three-pronged test to be outside the protections of GINA:

- The test must be an analysis of proteins or metabolites [not an analysis of DNA, RNA, or chromosomes]
- The test must be directly related to a manifest disease, disorder, or pathological condition.
- The *disease* could reasonably be detected by a health care professional with appropriate training and expertise in the field of medicine involved.
    - This prong clarifies that in order to be considered "manifested" the disease has to have signs (other than a genetic test) and symptoms beyond a genetic marker that would allow the disease to be detected by a health care provider. Regulations should specify that "manifestation" should be linked to the presence of "signs" (other than a genetic test) and "symptoms" of the disease, disorder, or pathological condition.
    - Regulations should specify that a genetic test result is not, by itself, enough to diagnose a manifest disease. If it were, any genetic test result could be declared a "diagnosis" of future disease that has not actually manifested itself in a detectable way -- gutting the protections afforded by GINA and undermining Congressional intent. There is legal precedent for ensuring that a test result cannot by itself be used as the basis of making a diagnosis. HIPAA states "Genetic information shall not be treated as a condition described in subsection (a)(1) [a pre-existing condition] in the absence of a diagnosis of the condition related to such information."[2]

Examples of tests that would meet this three part test would include tests related to both genetic and non-genetic disease such as:

- blood sugar of a diabetic
- cholesterol levels of someone with heart disease

ADDITIONAL ISSUES RELATED TO "MANIFEST DISEASE"

[2] See, e.g. ERISA §701(b)(1)(B)

GINA does not prevent discrimination based on a manifest disease. For example, if an individual already has breast cancer, GINA does not prohibit an individual market insurer from refusing to sell her a policy (subject to state law). However, regulations should clarify the following points:

- Under GINA, the manifestation of a disease in family members of an individual also constitutes genetic information about the individual. Health insurers are not allowed to discriminate against the relatives of a person with manifest disease based on this family history, even if they are dependents on the original individual's health plan or members of the same group health plan.

- The genetic information of an individual with a manifest disease is protected under GINA and cannot be used for underwriting.

    - Example: an individual with breast cancer might undergo genetic testing and learn that because she tests positive for a BRCA mutation, she is at increased risk for ovarian cancer. Although her rates may go up because of her breast cancer, the insurer cannot raise her premiums based on the increased risk for ovarian cancer in the future.

- Enforcement of GINA must include mechanisms for ensuring that underwriting is not based on genetic information but is reasonably based on information (such as manifest disease or claims history) not prohibited for such use by GINA.

GENETIC SERVICES

"Genetic services" includes any of the following: a genetic test, genetic counseling (including obtaining, interpreting, or assessing genetic information), or genetic education.

The definition of genetic test is addressed above.

Genetic counseling and genetic education may take a variety of forms.

Example: A woman who seeks BRCA testing (genetic testing for breast and ovarian cancer risk). Typically, this woman would seek and receive genetic counseling and/or education before and/or after the genetic testing.

- Before testing, a counselor or doctor would explain the risks and benefits of testing and what the test results mean.
- Before and after testing, a counselor or doctor would explain her lifetime risks of developing breast or ovarian cancer.
- Whether or not the woman decides to have the genetic test to learn about her risks, a counselor or doctor would review with her clinical options that can reduce her risks, and perhaps make recommendations. Options in the case of BRCA might include earlier and more frequent mammograms and preventive

measures such as taking tamoxifen or having preventive surgery to remove the ovaries or breasts.

The regulations should specify that GINA protects all of the above examples as counseling. Information about these events cannot be requested by an insurer or used as the basis of underwriting.

Insurers should explicitly inform prospective enrollees that they are not seeking information related to genetic services. The federal agencies should develop model language as guidance for state insurance regulators and for insurers and health plans and ensure that the forms, such as enrollment forms or health risk assessments, comply.

> For example, an acceptable question to ask would be, "Has a doctor or health care provider recommended any medical care in the future for diseases or conditions you currently have? In answering this question you should not include genetic testing or care related to genetic testing, genetic counseling, or genetic diseases for which you are believed to be at risk." An unacceptable question would be, "Has a doctor or health care provider recommended any medical care in the future?"
>
> Thus in the example above, if a health insurer in the individual market asks the prospective enrollee whether she has discussed any future medical care or prospective surgery with a physician, the prospective enrollee should be explicitly informed that she is not required to disclose genetic information which would includes counseling related to the BRCA test and discussion or recommendation of additional preventive strategies. In addition, once the individual is enrolled, to the extent the health insurer generally covers the medical services that were discussed in the genetic counseling, the insurer must cover the cost of the services without subjecting them to a pre-existing condition exclusion.

- Payment of claims for genetic services is subject to a showing of medical necessity, discussed below.

- Information about claims for genetic services may reveal genetic information to health insurers who thereby would obtain genetic information without violating GINA. However, in enforcing GINA, regulators should consider implementing requirements for insurers to *isolate* the information obtained through claims processing from the underwriting process, *notify* enrollees that they have received this information but will not use it, and *certify* to the Secretary that they will not use this information for underwriting.

The regulations should specify that GINA protects all of the above examples as *genetic services*. Information related to genetic services cannot be requested by an insurer or used as the basis of underwriting.

In addition, implementing regulations should specify that genetic services include information about preventive therapies and screenings that patients may *consider or undergo* to reduce their risks revealed by genetic information. During consideration of GINA, many Members of Congress stressed that ending genetic discrimination in health insurance is essential in order that new preventive measures can be developed and pursued without fear.[3] Most patients today undergo genetic testing for the express purpose of learning their risk status and available preventive options. If GINA were to protect only patient's test result, but nothing that might subsequently be done to reduce risk, its protections would be hollow.

Of note, a survey of state health insurance regulators asked whether current state law prohibitions on genetic discrimination in health insurance would also protect applicants who explore or pursue preventive or risk-reducing therapies because of their genetic information. Most regulators responded that their state laws would also protect patients in these circumstances. As one explained, "This information is fruit from the same poison tree." However, a few state regulators did not think their regulatory protections were this broad. Further, most insurance company medical underwriters surveyed believed GINA-like state law protections do not apply to the exploration or pursuit of preventive or risk-reducing therapies. In response to a doctor's recommendation of prophylactic surgery, 10 of 13 underwriters said they would take an adverse action. In light of such differing interpretations of the reach of state genetic nondiscrimination laws, it is imperative that federal regulations make clear that GINA protections apply to all genetic services, including the consideration and pursuit of preventive screening and therapies to reduce inherited risk of disease.[4]

UNDERWRITING

GINA prohibits the use of genetic information by health plans and Medigap and health insurance issuers for "underwriting purposes." The statute defines underwriting as

> "rules for, or determination of eligibility (including enrollment and continued eligibility) for benefits under the plan or coverage; the computation of premium or contribution amounts under the plan or coverage; the application of any pre-existing condition exclusion under the plan or coverage; and other activities related to the creation, renewal, or replacement of a contract of health insurance or health benefits."

Regulations should take note that this definition relies heavily on language found in HIPAA privacy regulations.[5] HIPAA privacy rules provide for several broad exceptions, including one

[3] See, for example, floor remarks of Senators Enzi (page S 3365), Levin (S 3372), and Reid (S 3372), *Congressional Record*, April 24, 2008.
[4] K. Pollitz, B. Peshkin, E. Bangit, and K. Lucia, "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices," *Inquiry*, 44: 350-368 (Fall 2007). A copy of the article is enclosed with this submission.
[5] 45 CFR 154.501 (3)

for “health care operations,” under which covered entities (health plans and health insurance issuers, etc.) may use and disclose protected medical information. Underwriting is included in the list of activities that comprise the definition of health care operations.

In drafting regulations to implement the prohibition on collection of genetic information for underwriting purposes, the agencies should coordinate with the Department of Health and Human Services to ensure that GINA and HIPAA privacy rules governing underwriting are consistent. Although the use of protected health care information for underwriting purposes may be permitted under HIPAA privacy rules, the use of genetic information for underwriting is prohibited under GINA.

II. GINA’S IMPACT ON PRACTICES

There are several areas in which regulations should clarify that health insurers and health plans will need to alter current practices. Overall we believe the burden on these entities will be minimal.

Prohibition on Collection of Genetic Information

GINA prohibits group health plans, group and individual health insurance issuers, and Medigap insurers from requesting, requiring, or purchasing genetic information (1) at any time for underwriting purposes, and (2) for any purpose prior to enrollment in coverage. Thus:

- Insurers and plan sponsors may not ask prospective enrollees for information about genetic testing, genetic services, or family history in initial enrollment or medical underwriting questionnaires.
- As described above, GINA protects information about recommendations for future preventive care as part of the genetic services definition. Thus, prospective enrollees who are asked about anticipated future care could not be required to reveal information from past genetic counseling or other genetic services.
- Insurers may not consider genetic information (including genetic tests, genetic services, or family history) in the course of any other underwriting practices, such as renewal or experience rating or post-claims underwriting investigations.

Incidental Collection

GINA includes an exception to the prohibition on requesting, requiring, or purchasing genetic information, which applies to the collection of genetic information which is incidental to the request, requirement, or purchase of other information concerning an individual. The genetic information collected must not be used for underwriting purposes.

In general the prohibition on collection of genetic information is meant to ensure that it is not used for underwriting. To help guard against the possibility of unlawful *use* of information that was incidentally obtained, regulations should underscore the duty of insurers and group health

plans to take affirmative steps to *avoid* requesting, requiring, or purchasing genetic information. Overly broad requests for health information that are likely to also gather some genetic information should be prohibited. We believe that the burden should rest with the collector to show why broad requests are necessary and to take steps to ensure that genetic information is not accidentally collected. The regulation should provide additional guidance on the definition of "incidental."

Regulations should specify that group health plans and group and individual health insurance issuers are not allowed to ask for, seek, or obtain genetic information about applicants before they enroll in coverage. For example, as discussed below, although questions about laboratory tests legitimately may be asked in some circumstances, they must be narrowly framed. It should be made clear and explicit to the enrollee that the insurer does not intend to ask for information about genetic tests or any other genetic information, including family history of disease, and that such information should not be revealed in answering questions. Regulators should develop model language for insurers and issuers to inform people that they should not reveal genetic information.

Federal agencies should develop model language for insurers to use and ensure that the forms comply. For example:

- Acceptable question: "Has a doctor or health care provider recommended any medical care in the future for diseases or conditions you currently have? In answering this question you should not include care or testing related to genetic testing, genetic counseling, or genetic diseases for which you are believed to be at risk. In addition, do not include information about genetic services, including counseling by a doctor or other health practitioner about genetic test results or options to reduce your risk of onset of genetically based conditions in the future."
- Unacceptable question: "Has a doctor or health care provider recommended any medical care that you should receive in the future?"
- Acceptable question: "Have you had any laboratory tests in the past two years? In answering this question you should not provide any information about genetic tests."
- Unacceptable question: "Have you had any laboratory tests in the past two years?"
- Acceptable question: "Have you taken any prescription drugs in the past year? In answering this question, you should not provide any information about medications that your doctor has recommended you take to reduce the risk of onset of a genetically based condition in the future (such as a preventive dose of tamoxifen to reduce your inherited risk of breast cancer.)"
- Unacceptable question: "Have you taken any prescription drugs in the past year?"

Regulators also should ask insurers to *certify* what steps will be and are taken to *isolate*, *protect*, and *destroy* genetic information that may inadvertently be collected. Regulations should require plans and insurers to *notify* the enrollee if information was inadvertently collected. Such a requirement would encourage plans and insurers not to collect such information in the first place. In addition, we strongly advise requiring periodic summary reporting to regulators by health plans and health insurance issuers of instances of incidental collection of genetic information. This will inform oversight and compliance audit efforts by regulators.

Individual market

A study of medical underwriting practices in the individual insurance market asked chief medical underwriters how they would respond to hypothetical applicants, some of whom had undergone genetic testing that detected a mutation predisposing the applicants to various health conditions in the future.[6] In seven of the 92 decisions tracked by this study, underwriters responded that they would use genetic information as the basis for a decision to decline, postpone, or limit coverage or surcharge premiums.

This study noted that medical underwriting questionnaires for individual health insurance policies generally do not ask directly for information about genetic tests. However, other types of broad questions that appear on applications and other investigations into an applicant's health status and health history may result in the incidental or inadvertent collection of genetic information. For example, patient medical records typically are requested on approximately 20 percent of applications. In the course of investigating an applicant's medical history, genetic information is likely to be uncovered. Of 23 senior medical underwriters surveyed, 16 reported they had encountered genetic information about an applicant at least once before.

Group market

Group health insurance policies purchased by employers are not medically underwritten in the same manner as individual policies. While less has been published about group market underwriting practices, industry sources indicate that small employer group applicants often are medically underwritten for purposes of determining risk-related premiums. Questions asked of small group applicants may not be as extensive as those asked of applicants in the individual market. However, the same protections against both deliberate and inadvertent collection of genetic information must apply to policies sold in the group market.

In addition, group health insurance premiums often are experience rated. Group issuers may use various methods to gather data for experience rating purposes. For example:

[6] K. Pollitz, B. Peshkin, E. Bangit, and K. Lucia, "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices," *Inquiry* 44:350-368 (Fall 2007).

- For very small groups, some carriers simply review all claims submitted in a year in order to determine the subsequent year's premiums.
- Some group carriers review total claims only for small group policies with a loss ratio that exceeds a certain threshold.
- Some carriers review only a sample of claims that are associated with certain diagnostic codes or procedure codes. Selected codes (for example, for MRI) would tend to signal risk of higher utilization in the future.

Group health insurance issuers should review their rating practices carefully and take steps to avoid the collection or use of genetic information. In addition, as noted above, federal agencies should require insurers and plans to notify individuals when incidental collection of genetic information occurs, and to provide periodic summary reports to regulators on the occurrence of incidental collections.

Wellness Programs and Health Risk Assessments

Many issues related to wellness programs will arise during consideration of regulations related to Title II of GINA.

Regulations implementing Title I must specify that wellness programs that are part of or related to the health insurance offered by an employer must comply with Title I's prohibition on the collection or use of genetic information, including family history.

Health risk assessments are questionnaires designed to identify preventable health risks on an individual and group level. Typically they cover all areas of behavior such as seatbelt use, tobacco use, alcohol use, and frequency of exercise. They also ask about family history of disease and illness. Eighty-three percent of employer-based wellness programs use health risk assessments; sometimes the program consists exclusively of such an assessment.[7] They are generally administered immediately after enrollment in the wellness program.

Regulations should clarify that wellness programs covered by Title I because they are part of or related to the health insurance offered by an employer may not include questions about family history on their initial risk assessment questionnaires and may not use family history to make decisions about what benefits or rewards to offer enrollees.

To best protect individuals from being coerced into revealing their family history to an entity that controls their health insurance costs, regulators should interpret broadly when a wellness program is part of or related to an employer's health insurance plan. In some cases, health risk assessments are administered by the same health insurers or issuers that administer an employer's group health insurance plan and thus clearly are reached by Title I of GINA. In other cases, regulators may consider factors such as whether the employee's health premiums vary depending on either participation in the program or results of the program. Wellness plans

[7] Forrester Research, "What Consumers do with Health Risk Assessments." Oct. 2007.

that provide medical care or services may be considered separate ERISA plans and thus also would be subject to Title I.

III. RESEARCH EXCEPTION

Under the "research exception" in GINA, a group health plan or a health insurance issuer in the group, individual, or MedSupp market may request (but not require) a participant or beneficiary to undergo a genetic test if five conditions are met. These conditions are intended to establish that the test results are part of a legitimate research endeavor with adequate protections both to protect patients and to prevent genetic information from "research" from being used for underwriting by a plan or issuer. Research conducted by health plans and issuers that involves their own enrollees must be scrutinized to ensure that so-called "research" cannot become a broad exception to the GINA rule that plans may not request and collect genetic information. GINA's prohibition of the request for genetic information is a critical provision that both protects patients from feeling unduly pressured to take a genetic test and prevents insurers from obtaining genetic information that they might use unlawfully.

It is worth noting that this section was added at the request of Kaiser Permanente, which has undertaken a well-designed research project that appears to meet the requirements laid out here and which we believe adequately protects patient-participants. It is not known if other plans and issuers anticipate such research for the future.

In general, we believe regulations should state that this section applies to any research conducted by *or supported* (partially or fully funded by) a group health plan or health insurance issuer. Kaiser Permanente has a unique structure that allows it to design a protocol involving its own patients. However, it is more likely that a plan or issuer would *fund* such research and the paid researcher would recruit among the health plan's enrollees. In order to ensure that patients do not feel coerced into taking genetic tests as part of research funded by plans and issuers, and to implement a strategy of preventing insurers from acquiring genetic information that might then be misused in underwriting, we believe all aspects of this section must be applied rigorously to any research that a plan or issuer conducts or supports financially.

Comments on the five requirements, A-E:

> *`(A) The request is made, in writing, pursuant to research that complies with part 46 of title 45, Code of Federal Regulations, or equivalent Federal regulations, and any applicable State or local law or regulations for the protection of human subjects in research.'*

- Regulations should clarify that research conducted by or supported by a group health plan or health insurance issuer must comply with either 45 CFR 46 (for federally funded research), or the substantially equivalent regulations that must be met for research leading to FDA-approved products. Current FDA regulations are summarized in 21 *Code*

of Federal Regulations part 50 (Informed Consent), part 56 (IRB Standards), part 312 (rules on Investigational New Drugs) and parts 812 and 813 (Investigational Devices). The regulations should clarify that FDA regulations are the only federal regulations "equivalent" to 45 CFR 46 and that research must comply with one of these rules as well as any additional state or local laws.

- In general, the federal Office of Human Research Protection has determined that research involving coded samples -- that is, research in which a code exists linking the sample to the donor, but where the link to the code is not available to the investigator using the sample -- is *exempt* from human subjects regulation and the requirement for informed consent under 45 CFR 46 and the equivalent regulations under FDA. Thus some such research has moved forward with a requirement that patient-subjects must affirmatively opt out of participation rather than a protocol that requires voluntary written informed consent before researchers perform tests on their blood or tissue samples. In some cases, there has been no notice to research participants of the planned use of their samples. We believe that because of the particular risks of misuse of genetic information obtained through research conducted by the same entity that conducts underwriting and sets premium and eligibility rates, *all* research conducted under this section, whether or not the protocol involves coded samples, should involve written voluntary informed consent from every participant. Regulations should specify that research cannot be exempt simply because it involves coded samples.

- In addition, 45 CFR 46 allows Institutional Review Boards (IRBs) to waive the requirement to obtain informed consent if "the research could not practicably be carried out without the waiver or alteration." GINA regulations should specify that because of the particular risks inherent in research conducted by plans and issuers, this waiver option is not available for research carried out under this section.

  `(B) The plan or issuer clearly indicates to each participant or beneficiary, or in the case of a minor child, to the legal guardian of such beneficiary, to whom the request is made that--

  `(i) compliance with the request is voluntary; and

  `(ii) non-compliance will have no effect on enrollment status or premium or contribution amounts.'

- As stated above, we believe written voluntary informed consent of every participant must be obtained.

  `(C) No genetic information collected or acquired under this paragraph shall be used for underwriting purposes.'

- Plans and issuers should describe their plans for ensuring that any genetic information collected through research they are conducting or funding is isolated from their underwriting activities. This description should be included in their institutional review board (IRB) application and in the notice they provide to the secretary of the Department of Health and Human Services (DHHS).

  `(D) The plan or issuer notifies the Secretary in writing that the plan or issuer is conducting activities pursuant to the exception provided for under this paragraph, including a description of the activities conducted.'

- The regulations should specify what should be included in the notice the plans provide to the Secretary, such as a copy of the protocol submitted to the IRB and the IRB approval. The plan should be submitted to the Secretary and certified within a short time period or permission to proceed with subject recruitment should be considered granted.

  `(E) The plan or issuer complies with such other conditions as the Secretary may by regulation require for activities conducted under this paragraph.'

- This section provides the authority needed for regulators to create the specific requirements described above.

Finally, the RFI asks (1) whether a model notice would be helpful to facilitate disclosure to plan participants and beneficiaries regarding a plan's or issuer's use of the research exception and what information would be most helpful to participants and beneficiaries, and (2) whether a model form would be helpful for reporting to the departments by a plan or issuer claiming the research exception, and what information should plans and issuers report.

- While we believe that a model notice and model form would be useful, we do not believe that notice is a substitute for written voluntary individual informed consent, which is required under this section.

- The regulations should specify what should be included in the notice the plans provide to the Secretary, such as a copy of the protocol submitted to the IRB and the IRB approval. The plan should be submitted to the Secretary and certified within a short time period or permission to proceed with subject recruitment should be considered granted.

IV. REQUEST OR REQUIRE A GENETIC TEST

GINA prevents an insurer or issuer or their representative from requesting or requiring that an enrolled individual take a genetic test. This provision was designed to prevent enrolled individuals from feeling pressured by their insurer or an insurer's representative and to prevent

the insurer from usurping the legitimate role of the health care provider in advising patients about their health care.

- Regulations should specify that plans and issuers may not contact patients directly to request or require that they take a genetic test.

However, GINA allows health plans to provide information to both doctors and patients about availability and appropriate use of genetic testing in medical care and permits health care providers to continue to offer and recommend genetic testing to their own patients.

- Regulations should specify that GINA does not prohibit a plan or issuer from providing information to enrolled or covered individuals about genetic testing. For example, a plan may send written information about carrier screening or cancer predisposition genetic testing to all covered individuals or to subgroups based on appropriate demographic factors.

- Regulations should emphasize that, as is stated clearly in 101 (c) (2) and 102 (c) (2), GINA does not "limit the authority of a health care professional who is providing health care services with respect to an individual to request that such individual or a family member of such individual undergo a genetic test." This rule of construction clarifies that it must be the health care professional who is directly treating the individual who makes the request.

- Regulations should clarify that plans and issuers may provide information to health care providers in their networks about available professional resources and guidelines on genetic testing and encourage them to follow them in making recommendations to their patients.

V. PAYMENT

GINA does not prohibit a group health plan from obtaining or using the results of a genetic test in making a determination regarding payment. GINA does, however, require the plan to request only the minimum amount of information necessary to accomplish the intended purpose. The regulations should provide clarity about this aspect of GINA.

GINA does not prohibit a health insurer or issuer from requiring that an enrollee show that a service is medically necessary. In some cases, an enrollee may reveal genetic information to prove medical necessity.

For example, a patient who has had breast cancer and tested positive for BRCA mutation is at heightened risk for ovarian cancer. She may seek a prophylactic oophrectomy and may be asked to justify the medical necessity of the surgery. The patient may reveal the positive BRCA test result, or she and her doctor may argue that her own history of breast cancer puts her at heightened risk and is thus enough to prove medical necessity.

Another patient may have no personal history of breast and ovarian cancer, no positive BRCA genetic testing, but a very strong family history of breast and ovarian cancer. She may seek prophylactic surgery based on her family history (which is itself her genetic information) and the insurer may determine whether that information meets their standards for medical necessity. The insurer may not request or require that she take a genetic test as a condition of payment.

Regulations should require that if an insurer makes a determination that only disclosure of a genetic test result will suffice to prove medical necessity, that determination must be in writing and must cite the specific evidence or guidelines on which it is based. Insurers should be required to report periodically the number of times they make such determinations.

VI. ENFORCEMENT

Federal enforcement of GINA requirements occurs differently depending on the federal agency involved.

DHHS has fallback authority to enforce GINA requirements against health insurance issuers (group and individual) when there is a finding that States have not enacted the necessary legislation to bring its laws into compliance with federal requirements or when a state does not otherwise substantially enforce those requirements. Similar fallback enforcement authority rests with DHHS for Medigap insurance policies. The following comments will focus mainly on non-Medigap health insurance issuers.

The Department of Labor (DOL) has direct authority to enforce GINA requirements against both group health plans and group health insurance issuers. In addition, DOL may refer group health plan violations to the Internal Revenue Service (IRS) which has authority to levy an excise tax on group health plans.

DHHS Fallback Enforcement and Coordination with States

No state health insurance laws today are fully in compliance with GINA requirements.[8] In particular, no state law definitions of genetic information completely conform to the federal law definition. No states currently have adopted GINA's definition of genetic test. Many do not include family history in the definition of genetic information. No state definitions specifically reference genetic services. In addition, not all states provide for as comprehensive protection against health insurance discrimination based on genetic information. To make their laws conform to GINA, all states will need to revise their definitions, many will have to add a prohibition on collection of genetic information, and all will need to adopt a prohibition on requiring individuals to take a genetic test.

[8] "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices," *ibid.*

Under HIPAA, DHHS enforcement is triggered with regard to the group health insurance market when a State fails to substantially enforce any "provision or provisions." This means DHHS can and must enforce any specific requirement that States fail to substantially enforce. By contrast, DHHS enforcement is triggered with regard to the individual health insurance market whenever a State fails to substantially enforce "requirements of this part." This suggests that States might enforce most, but not all, federal requirements for individual health insurance and still not trigger DHHS enforcement.

GINA specifies that the Secretary of HHS shall have the same authority to enforce GINA requirements with respect to the individual health insurance market as s/he has with respect to the group market. Accordingly, when regulations are drafted, DHHS should emphasize that States should take care to adopt and enforce *each and every* health insurance provision of GINA.

There is no reason to expect that States will not act to conform their laws to GINA. In fact, prior to GINA, 43 states already prohibited (at least to some extent) discrimination by individual market insurers based on genetic information. Further, a survey of state health insurance regulators indicates that most take a broad view of their enforcement authority and would prohibit certain acts of genetic discrimination that are now prohibited by GINA even if these are not specifically stated in statute. For example, when presented with research findings that many insurers would underwrite based on genetic services, most State regulators said they would interpret their state law to also protect consumers who explore or pursue preventive or risk-reducing therapies because of their genetic information. As one explained, "This information is fruit from the same poison tree."[9]

According to the HHS HIPAA enforcement regulation, sources of information that would trigger an investigation of State enforcement include (but are not limited to)

- A complaint received by DHHS
- Information learned during informal contact with State officials
- A report in the news media
- Information from governors and commissioners of insurance regarding the status of their enforcement of federal requirements
- Information obtained during periodic review of State health care legislation and regulations
- Any other information that indicates a possible State failure to enforce federal requirements[10]

Recently, however, an official from the Centers for Medicare and Medicaid Services testified that DHHS would only investigate a State failure to enforce federal minimum HIPAA standards upon receipt of an individual complaint.[11] In drafting regulations for GINA, HHS should make

[9] "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices," *ibid.*
[10] 45 CFR 150.205.
[11] See testimony of Abby Block before the House Committee on Oversight and Government Reform, July 17, 2008. Available at http://oversight.house.gov/story.asp?ID=2089.

available and publicize methods for individuals to register a complaint about GINA health insurance protection violations, including State failure to enforce such protections. A variety of methods, including telephone complaints to a toll-free number, written complaints, and complaints filed via an Internet site, should be available. HHS should also develop and describe plans to maintain regular communication with state officials, health insurance brokers, industry officials, consumer advocates, reporters, researchers, and others who might have information about the status of GINA consumer protections in health insurance.

HHS should also develop and describe plans for periodic review of State health insurance laws and regulations to ensure GINA protections have been adopted in all States. Further, HHS should develop guidance for states on what constitutes "substantial" enforcement. For example, substantial enforcement of GINA rules on collection of genetic information should include forms reviews by State regulators to ensure that insurance policy applications do not ask overly-broad questions that would regularly lead to the incidental collection of genetic information. In addition, State market conduct examinations should include review of medical underwriting manuals and rating policies and procedures to ensure that genetic information is not being used inappropriately by insurers.

The Secretary of DHHS also may wish to conduct periodic "look behind" investigations to gather independent information about the status of State enforcement of GINA protections.

DOL Enforcement

GINA gives the Secretary of Labor new enforcement authority under GINA. The Secretary has authority to impose civil money penalties against health plans for violation of GINA protections. In addition, DOL's GINA enforcement authority also extends directly to group health insurance issuers.

Accordingly, regulators should develop and describe procedures by which DOL will exercise its enforcement authority and gather information that would form the basis of a determination of noncompliance. DOL should describe procedures by which it would accept complaints from individuals. In addition, DOL should develop and describe plans for periodic review of GINA compliance by group health plans and group health insurance issuers. To the extent DOL opts to work cooperatively with State health insurance regulators, procedures for gathering and sharing information about practices in this market should also be designed.

Public Outreach and Education

Finally, all of the relevant federal agencies should issue guidance on notice requirements for group health plans and health insurance issuers to alert consumers to their new protections under GINA. The Secretaries of DHHS and DOL also should engage in outreach to State officials

to educate them about GINA requirements and determine what assistance States may need in order to adopt and enforce these in a timely and effective manner.

VII. CONCLUSION: GINA'S BENEFICIAL IMPACT

Throughout Congressional consideration of GINA, health insurers and issuers argued that the legislation was not necessary because they did not use, and did not plan to use, genetic testing in underwriting or other aspects of their business. We believe that few policies, procedures, or practices of group health plans and health insurance issuers will be affected by GINA. The primary change will be the prohibition on use of family history in the individual health insurance market. Procedurally, for entities that are already compliant with ERISA and HIPAA, GINA imposes minimal additional requirements.

Ultimately we believe GINA provides benefits to both health insurers and employers. In overall costs, the fear of genetic discrimination interfering with individuals' willingness to pursue testing has negatively affected health insurers (who must pay more to treat conditions that are not prevented or caught early) and employers (who bear the economic costs if employees require more sick days and medical leave). These entities – and all of us – will benefit if people can pursue the best preventive medical care available, and it is that promise that GINA regulations must seek to fulfill.

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What Affects Health Insurance Coverage?

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Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices

Most states have enacted genetic nondiscrimination laws in health insurance, and federal legislation is pending in Congress. Scientists worry fear of discrimination discourages some patients from participating in clinical trials and hampers important medical research. This paper describes a study of medical underwriting practices in the individual health insurance market related to genetic information. Underwriters from 23 companies participated in a survey that asked them to underwrite four pairs of hypothetical applicants for health insurance. One person in each pair had received a positive genetic test result indicating increased risk of a future health condition—breast cancer, hemochromatosis, or heart disease—for a total of 92 underwriting decisions on applications involving genetic information. In seven of these 92 applications, underwriters said they would deny coverage, place a surcharge on premiums, or limit covered benefits based on an applicant's genetic information.

Scientific breakthroughs in the identification of genetic markers for disease are developing at a rapid pace, yielding great promise—but also risk—for individuals and our health care system. Genetic research holds the key to understanding the cause and potential cure for many health conditions. Predictive genetic testing can help individuals understand their risk of disease and, in some cases, take steps to prevent its onset or manage its course. Accompanying the promise of human genetic research, however, is fear of potential unintended consequences. In particular, experts repeatedly have warned that genetic information also might be used by health insurers to deny or limit access to coverage.

In 1991, the Human Genome Project Working Group on Ethical, Legal, and Social Issues (ELSI) appointed a Task Force on Genetic Information and Insurance. The final report issued by the task force expressed concern that discrimination based on genetic information would make it difficult for individuals and their relatives to obtain health coverage and needed health care; further, fear of health insurance discrimination could discourage patients from seeking genetic testing that might benefit them

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medically and inform scientific research.[1] The task force recommended;

> Information about past, present or future health status, including genetic information, should not be used to deny health care coverage or services to anyone... Until [health insurance coverage] is universal, alternative means of reducing the risk of genetic discrimination should be developed. As one step, health insurers should consider a moratorium on the use of genetic tests in underwriting. In addition, insurers could undertake vigorous educational efforts within the industry to improve the understanding of genetic information (Task Force on Genetic Information and Insurance 2003).

Legal Protections Against Genetic Discrimination in Health Insurance

In response to the task force recommendations, numerous federal and state laws have been adopted, though none comprehensively protects against genetic discrimination in health insurance. In 1996, Congress enacted the Health Insurance Portability and Accountability Act (HIPAA), setting federal minimum standards for private health insurance, including a requirement that employer-sponsored group health plans may not exclude participants based on health status, including genetic information. HIPAA also prohibited group health plans from imposing pre-existing condition exclusion periods based on genetic information. However, HIPAA did not prohibit individual market health insurers from underwriting on the basis of genetic information, nor did it limit insurers in any market from varying premiums on that basis.

Since HIPAA, 43 states have prohibited use of genetic information by individual market health insurers. Most have enacted statutory prohibitions, which vary. Some state laws, for example, prohibit medical underwriting based on genetic test results, but not on family history. A few states prohibit insurers from denying coverage based on genetic information, but permit premiums to be surcharged. Interestingly, most state insurance regulators would enforce a broader prohibition on genetic discrimination than plain statutory language might otherwise indicate. For example, most state regulators say insurers cannot underwrite based on family history, even when this is not specifically included in the state law definition of genetic information. However, no state laws apply to group health benefits offered by self-insured employer plans because a federal law, the Employee Retirement Income Security Act of 1974 (known as ERISA), pre-empts state regulation in this area (Table 1).

Scientific and public policy leaders, including the Secretary of Health and Human Services' Advisory Committee on Genetics, Health, and Society, continue to call for comprehensive legal prohibitions on health insurance discrimination:

> [The Committee] heard from many Americans who are concerned about the misuse of genetic information by third parties, such as health insurers and employers, and the potential for discrimination based on that information. Many stated that fear of genetic discrimination would dissuade them from undergoing a genetic test or participating in genetic research studies. Others stated they would pay out of pocket for a genetic test to prevent the results from being placed in their medical record. Such concerns are a deterrent to advances in the field of genetic testing and may limit the realization of the benefits of genetic testing (Secretary's Advisory Committee 2001).

Federal legislation to prohibit genetic discrimination in health insurance was first introduced in the 104th Congress in 1995. It was considered initially as a provision of broader "patient bill of rights" legislation that was never enacted. In 2003, a stand-alone bill was approved by the Senate by a vote of 95-0 and endorsed by President George Bush, but it was not acted on in the House of Representatives. In February 2005, the Senate again approved legislation to prohibit health insurance discrimination based on genetic information, this time by a vote of 98-0; however, the 109th Congress adjourned without further action on the bill. New legislation was introduced in the 110th Congress with bipartisan support, and it

Table 1. State prohibitions on use of genetic information in medical underwriting, individual market

	Prohibited underwriting action											
	Application asks about:			Deny coverage based on:			Raise premium based on:			Exclusion rider based on:		
State	Family history	Received genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results
AL[a]						✓			✓			✓
AK												
AZ[b]					x	✓		x	✓		x	✓
AR[c]		x	x		x	x		x	x		x	x
CA[d]			✓			✓			✓	✓	✓	✓
CO		x	✓	x	x	✓	x	x	✓	x	x	✓
CT	x	x	x	x	x	✓	x	x	✓	x	x	✓
DE				x	✓	✓	x	✓	✓	x	✓	✓
DC	x	x	x	x	✓	✓	x	✓	✓	x	✓	✓
FL		✓	✓	x	✓	✓	x	✓	✓	x	✓	✓
GA						✓						✓
HI	x	x	✓	x	✓	✓	x	✓	✓	x	✓	✓
ID			✓		x	✓		x	✓	✓	✓	✓
IL[e]	x	x	✓	x	x	✓				x	x	✓
IN		x	✓	x	✓	✓	x	✓	✓	✓	✓	✓
IA												
KS		✓	✓	x	✓	✓	x	✓	✓	x	✓	✓
KY			✓		✓	✓		✓	✓	✓	✓	✓
LA	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
ME				✓	✓	✓	✓	✓	✓	✓	✓	✓
MD	x	x	✓	x	✓	✓	x	✓	✓	x	✓	✓
MA	x	x	x	✓	✓	✓	✓	✓	✓	✓	✓	✓
MI		✓	✓	x	x	x	x	x	x	✓	✓	✓
MN		✓	✓		✓	✓		✓	✓	✓	✓	✓
MS												
MO[f]		x	x	x	x	x	x	x	x	x	x	x
MT			✓		x	✓		x	✓	x	x	✓
NE												
NV		✓	✓	x	✓	✓	x	✓	✓	x	✓	✓
NH		✓	✓		✓	✓		✓	✓	x	✓	✓
NJ	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓	✓
NM				✓	✓	✓	✓	✓	✓	✓	✓	✓
NY				✓	✓	✓	✓	✓	✓	✓	✓	✓
NC		x	x	x	x	✓	x	x	✓		x	x
ND					x			x			x	
OH			✓			✓			✓			✓
OK[g]				x	x	x	x	x	x	x	x	x
OR	✓	✓	✓	x	x	✓	✓	✓	✓	✓	✓	✓
PA	**	**	**	**	**	**	**	**	**	**	**	**
RI	x	✓	✓	x	✓	✓	x	✓	✓	x	✓	✓
SC				x	✓	✓	x	✓	✓	x	✓	✓
SD												
TN		x	✓		✓	✓		✓	✓		✓	✓
TX				x	x	✓	x	x	✓	x	x	✓
UT		✓	✓	x	✓	✓	x	✓	✓	x	✓	✓
VT				✓	✓	✓	✓	✓	✓	✓	✓	✓
VA				x	✓	✓	x	✓	✓	x	✓	✓

Table 1. (continued)

	Prohibited underwriting action											
	Application asks about:			Deny coverage based on:			Raise premium based on:			Exclusion rider based on:		
State	Family history	Received genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results	Family history	Referral for genetic services (incl. counseling or testing)	Positive genetic test results
WA	√	√	√	√	√	√	√	√	√	√	√	√
WV												
WI		√	√		√	√		√	√		√	√
WY				x	x	x	**	**	x	**	x	

Source: Statutory research by Georgetown University and responses of state insurance regulators to Georgetown survey conducted May–June 2006.

Notes: Regulators in five states did not respond to the survey: California, Mississippi, New Mexico, New York, and Vermont. In these states, table only indicates prohibitions found in statutory language. Blank spaces indicate there is no prohibition—either in statute or via other regulatory authority.

√ indicates prohibition found in state statute.

x indicates state regulator confirms practice is prohibited, but practice is not specified in statute.

** Regulator did not answer this question. No statutory prohibition found.

[a] Alabama prohibitions only apply to genetic information about risk of cancer.

[b] Arizona prohibits practices unless "applicant's medical condition and history and either claims experience or actuarial projections establish that differences in claims are likely to result from the genetic condition."

[c] Arkansas prohibitions apply "except to the extent and in the same fashion as an insurer limits coverage or increases premiums for loss caused or contributed to by other medical conditions presenting an increased risk."

[d] California prohibits insurers from denying "enrollment or coverage to an individual solely due to a family history of breast cancer, or who has had one or more diagnostic procedures for breast disease but has not developed or been diagnosed with breast cancer."

[e] Illinois allows an insurer to "consider the results of genetic testing...if the individual voluntarily submits the results and the results are favorable to the individual."

[f] Missouri prohibits insurers from inquiring "to determine whether a person or blood relative of such person has taken or refused a genetic test or what the test results of any test were..." except with approval of the applicant to consider this type of information.

[g] Oklahoma prohibitions apply "except to the extent and in the same fashion as an insurer limits coverage or increases premiums for loss caused or contributed to by other medical conditions presenting an increased risk."

passed the U.S. House of Representatives on April 25, 2007, by a vote of 420-3 and with the support of President Bush (*Congressional Record* 2007). At this writing, a vote in the U.S. Senate was still pending.

For years, a debate over the need for federal legislation contributed to congressional inaction. The insurance industry has testified there is no evidence that insurers engage in genetic discrimination and, therefore, a federal prohibition is unnecessary.[2] In addition, results from a survey of actuaries, genetic counselors, insurance agents, and regulators in seven states, published in 2000, found "almost no well-documented cases of health insurers either asking for or using presymptomatic genetic test results in their underwriting decisions" (Hall and Rich 2000). The report cited a tendency of medical underwriters to focus on near-term risk of disease because of the high rate of enrollment turnover in health insurance policies. Other studies undertaken to document instances of genetic discrimination in health insurance have been criticized as anecdotal.[3]

Debate over the potential problem of genetic discrimination in health insurance is not likely to be resolved based on documented instances. The science of genetic testing is still young, and relatively few individuals have undergone predictive genetic testing in the United States. For example, since genetic testing for hereditary breast/ovarian cancer became clinically available via *BRCA1* and *BRCA2* testing in the mid-1990s, about 75,000 individuals have been tested through the commercial lab that holds the patents on these genes, and approximately 9,000 have

received positive test results (Myriad 2006). Many, if not most of those patients with positive test results likely were insured by employer-sponsored group health plans, where discrimination based on health status is already largely prohibited. Therefore, health insurers that do medically underwrite coverage likely have had very few opportunities to act on genetic information.

Even so, as causative genes associated with increased susceptibility to common diseases such as asthma, heart disease, and cancer are identified, the number of tested individuals will grow considerably. It is therefore important to understand how health insurers would respond to genetic information about applicants for coverage when they encounter this information in the medical underwriting process.

Background on Medical Underwriting

This study focuses on individual health insurance, which plays a small but important role in our nation's system of health coverage. People often turn to this market when they cannot get health benefits from an employer or when they are ineligible for public programs such as Medicare or Medicaid. In 2005, more than 17 million people in the United States were covered by individual health insurance, or 6.6% of the nonelderly population (U.S. Bureau of Census 2006). On average, over a three-year period, one in four adults buys or seeks individual coverage (Duchon et al. 2001).

Individual health insurance is medically underwritten in most states. This means applicants for coverage must submit information about their current and past health status (for example, whether they have been diagnosed with medical conditions such as diabetes, dates of and reasons for recent physician visits, and names and dosages of recently prescribed medications). Health insurance applications typically do not include specific questions about genetic test information nor about family health history.

Underwriters make a decision to issue or decline coverage based solely on health status information provided on the application for insurance as often as 50% of the time (Thomas and Chaput 2007). For other applications, additional information may be required. All applications for medically underwritten health insurance policies also require written consent to release any medical records and to submit to further medical examinations that may be requested. Most often, additional medical information will be sought directly from the applicant (for example, a telephone interview to determine results of a recent Pap test) or her physician. Less frequently, applicants may be required to take a physical examination or submit samples of urine, blood, or saliva for testing. A 2001 report on medical underwriting practices found that in the course of 420 applications for coverage studied, underwriters requested further specific medical histories 179 times, attending physician statements and/or copies of patient medical records 140 times, samples of blood, saliva, or urine for laboratory testing 46 times, and paramedic physical examination of the applicant 21 times (Pollitz, Sorian, and Thomas 2001). Other experts on individual health insurance market underwriting suggest patient medical records are typically requested on 20% of applications, while a very small portion of insurers (estimated at fewer than one in 10) may request records on more than 40% of applications (Thomas and Chaput 2007). It is in this additional investigation of an applicant's medical history and health status that information about genetic testing is likely to be discovered. Underwriters can come across medical information they did not specifically seek. Once disclosed, however, they are obliged to consider, evaluate, and act upon all available information.

Background on Predictive Genetic Testing

Some genetic tests are more predictive than others. Mutations such as those associated with Huntington's disease mean the patient has a virtual 100% chance of developing that health condition. Other genetic tests are less predictive, with gene mutations suggesting healthy patients may develop a health condition in the future. This study focuses on three types of predictive genetic testing:

- ☐ Hereditary breast cancer is one of the most commonly requested predictive genetic

Table 2. Lifetime cancer risks associated with *BRCA1/2* mutations

Cancer site	Risks in *BRCA1/2* carriers	Risks in the general population
Breast	40% to 85% (high incidence of premenopausal breast cancers)	8%
Second breast cancer after an initial diagnosis	40% to 60% risk in opposite breast (~30% risk within first 10 years)	Opposite breast: .5% to 1% per year following diagnosis, leveling off at 10% to 20%
	Long-term elevated risks of cancer in affected breast after lumpectomy and radiation	Same breast: risk is variable
Ovarian	10% to 40% (higher in *BRCA1*)	< 2%
Other	Elevated risks of cancers of the prostate, male breast, pancreatic, possibly others	Rare, except for prostate cancer

Source: Peshkin and Isaacs 2005.

tests and it has received a lot of attention in the popular media. As subsequently described, the interpretation and management implications subsequent to *BRCA1* and *BRCA2* testing are complex.

- ☐ Hemochromatosis, or iron overload, is a more common condition, but it is frequently undiagnosed, and genetic testing is arguably underutilized because of the availability of excellent biochemical tests to diagnose the condition.
- ☐ Genetic tests for heart disease are marketed directly to consumers despite poor predictive power. An increasing number of genetic tests are becoming available via the Internet in this manner. This is relevant to the insurance industry because knowledge obtained by consumers through the Internet may translate to increased demand for services or medications that could be unnecessary or of unproven efficacy.

Predictive Genetic Testing for Breast Cancer

Each year, more than 210,000 American women are diagnosed with breast cancer. Between 5% and 10% of these women have a hereditary form of the disease (American Cancer Society 2005). Hallmarks of hereditary breast cancer include an early age at diagnosis (before age 50 or in premenopausal women); associated higher rates of other types of cancer (especially ovarian) in the family history; an increased incidence of multiple cancers within the same woman (such as bilateral breast cancer or breast and ovarian cancer); and a positive family history of breast and/or ovarian cancer, often extending over two or more generations. Most cases of hereditary breast cancer are attributable to mutations in the *BRCA1* or *BRCA2* genes and these are the genetic tests that are most often requested by women with breast cancer and/or a family history of breast cancer.

In the general population, *BRCA1/2* mutations occur in approximately one in 500 individuals (Eng, Hampel, and Chappelle 2001), although prevalence is much higher among women of Ashkenazi Jewish descent, an estimated one in 40 of whom carry a mutation in *BRCA1* or *BRCA2* (Struewing et al. 1997). These mutations are inherited in a dominant fashion, meaning that the children of a parent with a *BRCA1* or *BRCA2* mutation have a 50% chance of testing positive for the mutation. In most cases, individuals who test negative for a mutation present in their family can be reassured that their cancer risks are the same as those in the general population.

The lifetime risk of developing breast and/or ovarian cancer is substantially higher (though not certain) in women who inherit mutations in *BRCA1/2*. Risk levels vary depending on the type of cancer and the patient's personal cancer history. Women who have had breast cancer face a significantly elevated risk of second breast cancers and ovarian cancer (Metcalfe et al. 2005) (see Table 2).

For women who do receive positive *BRCA1/2* genetic test results, there are a number of different options available to potentially detect cancer in its early stages and to reduce the risk of developing cancer altogether. Options for early detection include multimodal screening starting at age 25. This regimen includes clinical breast exams every six to 12 months, mammography every six to 12 months, and consideration of annual magnetic resonance imaging (MRI) of the breasts. There are no data as of yet to show whether such an approach reduces breast cancer mortality in mutation carriers.

In terms of risk reduction options, the hormonal drug tamoxifen has been shown to reduce the risk of second breast cancers by at least 50% in breast cancer survivors with *BRCA1/2* mutations (Gronwald et al. 2006). Mutation carriers who have their ovaries removed prior to age 50 also obtain the benefit of reducing their breast cancer risk by half (Rebbeck et al. 2002). The most effective means for reducing the risk of breast cancer is prophylactic mastectomy, or removal of the breast/s before cancer develops. This procedure reduces breast cancer risk by over 90% (Rebbeck et al. 2004). Experts predict high-risk, newly diagnosed breast cancer patients may opt increasingly for genetic testing to inform their decisions about surgery (e.g., to opt for bilateral mastectomy instead of breast conservation treatment) (Schwartz et al. 2004). Today, the utilization rate of prophylactic mastectomy by unaffected women in the United States is low, at about 15% (Wainberg and Husted 2004).

Unlike breast cancer, ovarian cancer is much more difficult to detect in its early stages. Most ovarian cancers are diagnosed at an advanced stage with a high rate of mortality (Modugno 2003). Therefore, mutation carriers are urged to undergo prophylactic bilateral salpingo-oophorectomy (PBSO), or removal of the ovaries and fallopian tubes by age 40 or when childbearing is complete. Such surgery reduces the risk of ovarian cancer by at least 80% (Rebbeck et al. 2002; Kauff et al. 2002). For women who do not opt for oophorectomy, surveillance for ovarian cancer is available and consists of pelvic exams, transvaginal ultrasounds, and CA-125 blood tests every six to 12 months beginning by age 35 (Peshkin and Isaacs 2005). In light of these considerations, the utilization rate of prophylactic oophorectomy is much higher than that observed for prophylactic mastectomy, generally ranging from about 30% to 50% in the United States (Wainberg and Husted 2004).

Predictive Genetic Testing for Hemochromatosis

Hereditary hemochromatosis (HHC) is the most common genetic condition in the United States, with an estimated prevalence of one in 200 to 500 individuals or 1 million people affected (Centers for Disease Control and Prevention 2006). This disease involves increased absorption of dietary iron. Excess iron accumulates in multiple organs resulting in early, often vague, symptoms such as abdominal pain, weakness, lethargy, joint pain, and skin hyperpigmentation. If undiagnosed or untreated, more serious complications include hepatic cirrhosis, diabetes, congestive heart failure or arrhythmias, and arthritis. The average age of onset is between age 40 and 60 in men and after menopause in women (Kowdley et al. 2006).

The gene associated with HHC is called *HFE*, and two common mutations in this gene, referred to as C282Y and H63D, were identified in 1996. The condition is inherited in an autosomal recessive fashion, which means that if both parents carry an *HFE* mutation, their child has a 25% chance of having two *HFE* mutations. Even if an individual has two mutations in the *HFE* gene, it is not guaranteed that he or she will develop elevated iron levels or, if iron levels do increase, that more serious complications are inevitable. However, precautionary steps are warranted. Unlike the complex management options available for carriers of *BRCA1/2* mutations, individuals can avoid the complications of hemochromatosis with simple phlebotomy, or blood-letting. In addition, they are advised to avoid iron supplements, alcohol, raw seafood, and other dietary sources of iron. If end-stage disease is avoided and total body iron depletion is successful via phlebotomy, then life expectancy is not significantly altered (Kowdley et al. 2006).

Genetic testing for HHC is considered to be of moderate use because hemochromatosis also can be easily and inexpensively identified through simple blood tests once the condition progresses to the point of causing iron overload. At that point, phlebotomy treatments—also simple and inexpensive—can control the condition effectively. However, because genetic testing can identify mutations in the *HFE* gene long before any symptoms of iron overload appear, patients can be closely monitored through blood tests to detect early signs of elevated iron levels in the blood and take corrective action.[4]

Predictive Genetic Testing for Heart Disease and Direct-to-Consumer Marketing

Among the leading causes of death in the United States, heart disease ranks number one, claiming more than 685,000 lives per year (Jemal et al. 2006). Family history appears to play an important role in many cases of heart disease. However, the genetic basis of these conditions is very complex and not yet well understood. Although dozens of gene alterations that play a role in heart health have been identified, most of these associations are not well established, and some genetic alterations may be very common in the general population.

The availability and marketing of genetic testing directly to consumers through the Internet is a recent development that capitalizes on the well-informed consumer's fear of developing common, potentially preventable conditions. However, the clinical value of many of these tests has not been established, particularly in the area of gene analysis to create individualized dietary and lifestyle recommendations (sometimes called "nutrigenetic" testing). The Government Accountability Office (GAO) recently released a report regarding nutrigenetic testing, in which it concluded that many medical claims purported by several Internet sites could not be substantiated (U.S. GAO 2006). Moreover, there were problems with laboratory quality control and researchers found that the nutritional supplements marketed on the Web sites not only might not be beneficial, but could in fact be harmful.

One company studied provides an analysis of 13 genes presumed to play a role in heart health. These genes are involved in functions such as homocysteine removal, vitamin metabolism, regulation of the inflammatory response, and cholesterol and triglyceride metabolism. The extent to which the various gene alterations contribute to heart disease risk is unclear; moreover, many of the studies about the risks associated with these gene variants have not been replicated or are applicable only to specific populations. Nevertheless, for a fee of $269, the company combines the results of the genetic tests with its analysis of diet and lifestyle questionnaires to provide individuals with an action plan, consisting of advice regarding dietary changes and supplementation, as well as physical activity and lifestyle recommendations (e.g., smoking cessation).

Although many of the genetic tests available over the Internet are of uncertain clinical utility, consumers also can go online to obtain validated tests, including *BRCA1/2* and *HFE* analyses (DNA Direct 2006). Why would individuals opt for testing in this manner? And who would be most likely to pursue this type of testing? Studies have not been performed to examine these questions, but it is likely that direct-to-consumer testing will find a niche with those who are concerned about privacy or insurance discrimination, as well as by individuals who are basically healthy, insured, and financially secure (Wolfberg 2006). While online testing offers convenience and anonymity, experts in the field of genetics are concerned other unintended consequences also might result. In particular, online genetic test results may be obtained without first undergoing genetic counseling. Experts worry that individuals could misinterpret the results of tests offered through the Internet, resulting in requests for health care services that might not be necessary. There is also concern that direct-to-consumer testing may be used by those who are not appropriate candidates for it, and may result in unnecessary worry or reassurance about risk, as well as expensive follow-up that may or may not be clinically indicated (Wolfberg 2006).

Finally, from an insurance underwriting perspective, direct-to-consumer Internet marketing of genetic testing could increase

substantially the number of individuals who seek such testing. This could result in larger numbers of health insurance applications from individuals who are concerned (appropriately or not) that they are at increased risk for developing serious health conditions.

Study Methodology

Researchers from Georgetown University partnered with private risk management consultants to design and implement this study. Senior medical underwriters from 23 insurers—some local and some multistate—participated. The study was conducted in three phases. First, a written survey asked participants to underwrite eight hypothetical applicants for coverage. Then researchers attended a national meeting of health insurance underwriters to present scientific background information about predictive genetic testing and engage underwriters in a group discussion of industry practices. Finally, a follow-up written survey was administered to probe an additional issue raised in the course of the group discussion. Participants and insurers were promised anonymity.

The Hypothetical Applicants

The eight hypothetical applicants were arranged in pairs that were almost identical except one person in each pair had received a positive genetic test result. For each pair of applicants, medical information was provided that likely would prompt further investigation by underwriters. The survey noted when genetic test result information was discoverable via patient medical records or other follow-up inquiry. The applicants follow:

- ☐ Ann and Brenda are healthy 29-year-old women who receive regular annual mammograms well before the age of 40, when such screening is recommended for the general population. Upon review of medical records, it is clear that both Ann and Brenda have a family history of breast cancer. In addition, Brenda has inherited a *BRCA1* mutation, meaning her lifetime risk of breast and ovarian cancer is significantly elevated, though not certain.
- ☐ Clarice and Donna are 48-year-old women and 10-year breast cancer survivors. Both women recently had preventive surgery to remove their ovaries. Upon review of medical records, it is clear that Donna's reason for undergoing surgery was a genetic test result from 2003 which was positive for mutation in the *BRCA1* gene, meaning her lifetime risk of a second breast cancer is significantly elevated, but not certain.
- ☐ Evan and Fritz are 52-year-old men in good health. Both receive regular blood tests to monitor blood iron levels. In follow-up telephone interviews, both men acknowledge a close family history of hemochromatosis, though blood tests for both men consistently have been negative for elevated blood iron levels. Fritz also has undergone genetic testing with a positive result, meaning his blood iron levels eventually may increase and need to be managed.
- ☐ Galen and Howard are 44-year-old men in excellent health. Both of their insurance applications disclosed a recent consultation with a cardiologist, and both take several nutritional supplements daily. Medical records indicate Galen sought his checkup after a neighbor his age died suddenly of a heart attack. Howard's visit was prompted by an online genetic testing company report that said he has gene variants that put him at risk for heart disease. The cardiologist questioned the validity of the tests and assured him the gene variants found are commonly observed in most people.

Questions Posed

Participants were asked first about their familiarity with the types of genetic test information explored in this survey, including their own knowledge level about each genetic test studied and the number of times they actually had encountered genetic test information on individual health insurance applications. The survey then asked the following questions:

1. Would you issue coverage to this applicant?

2. What additional information would you request? (e.g., medical history, doctor's records)
3. Would an exclusion rider[5] be applied?
4. Would a premium surcharge be applied?
5. Would any other benefit modifications be applied? (e.g., increased deductible)

The initial written survey was administered via email in spring 2006. Several weeks later, members of the research team attended the annual professional meeting of health insurance underwriters to present an informational background briefing on genetic test information and medical management of risk, and to engage in a discussion of underwriting approaches to this information. More than 100 underwriting personnel from approximately 50 companies attended this meeting and participated in the discussion. The next month, a follow-up survey was administered to probe underwriting actions that might be taken in response to medical interventions to reduce the risk of hereditary breast cancer. Participants were asked to consider additional information about one of the hypothetical applicants, Brenda. Specifically, they were asked:

1. If Brenda's medical records indicated her doctor had *discussed* options to reduce her risk of getting breast cancer (e.g., prophylactic mastectomy), what underwriting action would you take on her application?
2. If Brenda's medical records indicated her doctor had *recommended* options to reduce her risk of getting breast cancer (e.g., prophylactic mastectomy), what underwriting action would you take on her application?

Survey Participants

Survey participants were not selected at random. Officials of the professional association of health underwriters identified 40 chief underwriters whom they knew to be experts in their field, who supervise other underwriters, have at least five to 10 years experience, and who regularly attend their association's annual professional meeting where the latest industry practices and trends are discussed. All 40 were asked to participate in this survey and 23 agreed to do so. All of the participants worked for companies that sell individual health insurance. Sixteen worked for national, commercial insurers that write coverage in multiple states; seven worked for nonprofit Blue Cross Blue Shield plans. The size of participating insurers varied, though according to state insurance regulator data, three of the participating insurers rank among the top 10 health insurance companies based on national market share, and eight rank among the top 25 companies (National Association of Insurance Commissioners 2005).

States Surveyed

Participants from national health insurers were asked to consider the hypothetical applications from the perspective of different states, including one that has strict prohibitions on the use of genetic information in medical underwriting and one that does not.

Limitations of Methodology

The small number of survey respondents self-selected from a convenience sample means results cannot be interpreted as representative of the entire health insurance industry. In addition, because the survey asked questions about only three genetic tests, results provide no information about how underwriters might respond to other types of genetic information or inherited conditions. Other study design aspects may have biased results. For example, survey respondents came from a sample of those who participate in a professional underwriting study group and who tend to be more senior, expert, and informed about issues. In addition, the survey clearly identified the issue being studied, potentially biasing respondents to answer "correctly." On the other hand, survey vignettes also made obvious applicants' genetic information. Therefore, results do not shed light on how well underwriters recognize, or overlook, this information when they encounter it in practice. Nevertheless, the responses of so many mainstream insurers provide important insights into industry underwriting practices related to genetic information. They are also helpful in identifying issues and questions for further study.

Study Findings

Underwriters considered the following background medical information about four pairs of hypothetical applicants. One member of each pair was described as having positive genetic test information. In seven instances, an adverse underwriting action was taken on applicants based on their genetic test result; in two others, participants indicated uncertainty as to how to underwrite an applicant with genetic test information.

Ann and Brenda

This hypothetical applicant pair tested underwriting responses to healthy women who have no personal cancer history, but one (Brenda) who has a mutation in the *BRCA1* gene, placing her at higher risk of developing breast cancer at some point during her lifetime.

All of Ann's applications received a standard offer of health insurance, meaning she was offered a policy at the most favorable premium rate with no special coverage restrictions. In three instances, however, a carrier's underwriting action for Brenda differed from that for Ann. One insurer denied Brenda's application for coverage; another offered a policy with a rider excluding coverage for all breast diseases and disorders; a third offered coverage with a 25% premium surcharge (see Table 3).

Table 3. Underwriting actions on 23 applications for coverage by Ann and Brenda

Insurer	Underwriting action on Ann	Underwriting action on Brenda
1	*	*
2	*	*
3	*	*
4	*	*
5	*	*
6	*	*
7	*	Premium surcharge (25%)
8	*	Deny
9	*	*
10	*	*
11	*	*
12	*	*
13	*	*
14	*	*
15	*	*
16	*	*
17	*	*
18	*	*
19	*	*
20	*	*
21	*	*
22	*	*
23	*	Rider disease/disorder of breast

*Indicates standard offer with no premium surcharge, rider, or other special coverage restriction.

Clarice and Donna

This applicant pair tested underwriting responses to 10-year breast cancer survivors who successfully completed all treatment, and later had additional surgery to reduce their future risk of ovarian cancer. One of the applicants (Donna) underwent genetic testing and learned she has a mutation in the *BRCA1* gene.

Both Clarice and Donna received adverse underwriting actions on 12 of their 23 applications for health insurance. On the other 11 applications, both women received standard offers of coverage. These actions were taken based on the applicants' cancer history, not on the results of genetic testing. However, one insurer that offered Clarice coverage for a surcharged premium denied Donna's application. This insurer was not one of those that had taken an adverse action on Brenda based on her genetic test results (see Table 4).

Evan and Fritz

This applicant pair tested underwriting responses to genetic test information about hereditary hemochromatosis. Both men have a close family history of this condition, but neither currently exhibits any symptoms of having the condition. Fritz has undergone genetic testing and learned he has inherited gene mutations that make it likely, though not certain, he will develop HHC at some point during his lifetime.

Evan received 22 standard offers of coverage on 23 applications. One underwriter "pended" his application with an explanation that no further action could be taken without a definitive diagnosis. Insurers will sometimes pend (or postpone consideration of) an application when a patient anticipates

Table 4. Underwriting actions on 23 applications for coverage by Clarice and Donna

Insurer	Underwriting action on Clarice	Underwriting action on Donna
1	Deny	Deny
2	*	*
3	*	*
4	Premium surcharge (50%)	Premium surcharge (50%)
5	*	*
6	Rider disease/ disorder of breast	Rider disease/ disorder of breast
7	Premium surcharge (60%)	Premium surcharge (60%)
8	Deny	Deny
9	*	*
10	*	*
11	Premium surcharge (60%)	Deny
12	Premium surcharge (60%)	Premium surcharge (60%)
13	*	*
14	Premium surcharge (60%)	Premium surcharge (60%)
15	*	*
16	*	*
17	*	*
18	*	*
19	*	*
20	Deny	Deny
21	Deny	Deny
22	Deny	Deny
23	Premium surcharge (150%) + Rider disease/disorder of breasts	Premium surcharge (150%) + Rider disease/disorder of breasts

*Indicates standard offer with no premium surcharge, rider, or other special coverage restriction.

a change in health or risk status—for example, if a diagnostic test or a medical procedure is imminent. The patient may reapply for insurance later, once pending care is concluded and a clearer assessment of risk can be made. In essence, the application is denied with an invitation to try again in the future. The underwriter who pended Evan's application wrote that "coverage could not be offered without a definitive diagnosis." This underwriter did not seem to understand that so far Evan has not been diagnosed with, nor does he have any signs or symptoms of, hemochromatosis. This underwriter took the same action on Fritz's application. During the group discussion, most underwriters agreed that a diagnosis of iron overload would be grounds for automatic denial of coverage.

Table 5. Underwriting actions on 23 applications for coverage by Evan and Fritz

Insurer	Underwriting action on Evan	Underwriting action on Fritz
1	*	Pend. Unable to offer without diagnosis.
2	*	*
3	*	*
4	*	*
5	*	*
6	Pend. Unable to offer without diagnosis.	Pend. Unable to offer without diagnosis.
7	*	*
8	*	Deny
9	*	*
10	*	*
11	*	*
12	*	*
13	*	*
14	*	*
15	*	*
16	*	*
17	*	*
18	*	*
19	*	*
20	*	*
21	*	*
22	*	*
23	*	*

*Indicates standard offer with no premium surcharge, rider, or other special coverage restriction.

Fritz received 20 standard offers of coverage. His application was pended by one other insurer who appeared not to understand that Fritz does not have HHC. A third insurer denied Fritz's application (Table 5).

Galen and Howard

This pair of applicants tested underwriting responses to genetic information about risk of heart disease obtained from an online genetic testing company. Though experts have criticized the scientific validity of such online "nutrigenetic" testing, it is readily and inexpensively available to the general public. An online genetic testing company told Howard he has gene variants that put him at risk for heart disease.

All of Galen's 23 applications, but only 20 of Howard's received standard offers of

Table 6. Underwriting actions on 23 applications for coverage by Galen and Howard

Insurer	Underwriting action on Galen	Underwriting action on Howard
1	*	*
2	*	*
3	*	*
4	*	*
5	*	*
6	*	*
7	*	Unsure. Would refer to medical director.
8	*	Pend until further evaluation completed.
9	*	*
10	*	*
11	*	*
12	*	*
13	*	*
14	*	*
15	*	*
16	*	*
17	*	*
18	*	*
19	*	*
20	*	*
21	*	Unsure. Would refer to medical director.
22	*	*
23	*	*

*Indicates standard offer with no premium surcharge, rider, or other special coverage restriction.

coverage. One of Howard's applications was pended. Two other underwriters were unsure how to handle Howard's application and said they would need to refer it to the medical director (Table 6).

Underwriting Actions in Different States

Survey participants working for national health insurance companies that write coverage in multiple states were asked to consider the hypothetical applications from two different states—one that prohibits medical underwriting based on genetic information and one that does not. However, none of the underwriters varied their actions based on the applicant's state of residence.

Underwriter Views on Genetic Information

In general, underwriters said they have limited knowledge about the science of genetic testing and its implications for risk management. In the written survey, most underwriters described their own knowledge level as limited (Table 7). In group discussion, one underwriter said she thought *BRCA1/2* testing was more predictive, with a genetic mutation signifying a 100% chance of getting breast cancer. Another said he was surprised to learn that so few people have undergone predictive genetic testing to date. At the same time, most underwriters indicated they do occasionally encounter genetic information about people in the course of evaluating health insurance applications. Sixteen of those surveyed said they had encountered such information at least once before.

In group discussion after the survey, underwriters talked about why they would or would not take action on genetic test results discovered during the underwriting process. Most said they would not because their company policy is to underwrite on the basis of a definitive diagnosis and treatment, and they do not underwrite on the basis of family history or genetic information in the absence of a diagnosis. Underwriters also cited the consumer complaints and adverse publicity as reasons, although most understood their company policy to have been adopted pursuant to laws prohibiting this practice. (Those from multistate insurers said

Table 7. Underwriter knowledge about genetic testing

Condition/test	Level of knowledge			
	None at all	Some	High	No answer
BRCA (in cancer survivors)	5	10	2	6
BRCA (in unaffected patients)	6	13	1	3
Hemochromatosis	8	10	2	3
Testing for cardiovascular disease	15	3	2	3

Note: Table reflects responses from 23 underwriters.

their company policy would apply even in the minority of states that have not yet enacted legislation.)

When asked whether they would underwrite based on genetic information in the absence of legal prohibitions, many answered "yes." However, several expressed concerns. One underwriter noted the variation in the predictive power of different genetic tests. She said it might make more sense for underwriters to act on a positive test result indicating a person will certainly develop a health condition versus, for example, a *BRCA1/2* mutation where the increased lifetime risk of breast cancer is elevated but variable (40% to 85% chance). Another participant wondered how the evolution of genetic science may change views on this issue in the future. Experts in the field expect a battery of predictive genetic tests may become available within the next decade and, eventually, everyone may be able to learn his or her genetic predisposition to a number of diseases and disorders. Some underwriters also wondered who would be left to cover if everybody eventually were uninsurable.

Underwriting Based on Risk Reduction Interventions

The discussion with underwriters also raised the issue of how to distinguish between genetic test results, which many underwriters would not act upon, and related clinical information, which might be actionable. Discussion focused on risk reduction measures that women may consider when they learn they carry a mutation in *BRCA1/2* genes. These include prophylactic surgery and/or hormone therapy, and more frequent and intensive cancer screening. Underwriters generally stated they would be much more likely to act on information in an applicant's medical records indicating that such interventions had been recommended or even discussed.

This discussion prompted a follow-up survey question in which underwriters were asked to re-consider "Brenda's" application for coverage with additional information showing her doctor *discussed* with her the option of prophylactic surgery to reduce future risk of breast cancer. In addition, underwriters were asked to respond to information showing Brenda's doctor had *recommended* prophylactic surgery to reduce future risk of breast cancer.[6]

Table 8. Underwriting actions for Brenda based on interventions to reduce breast cancer risk (counseled vs. recommended)

	Underwriting action	
Insurer	Doctor discussed prophylactic surgery to reduce risk	Doctor recommended prophylactic surgery to reduce risk
1	*	Postpone
2	Probably rider	Probably rider
4	Rate	Rate
6	Rider	Rider
7	*	Rider or deny
10	*	Deny
11	Deny	Deny
12	*	Rider
14	*	*
15	*	*
16	*	Postpone
17	Deny	Deny
20	*	*

*Indicates standard offer with no premium surcharge, rider, or other special coverage restriction.

Only 13 underwriters answered the follow-up survey. Among them, however, more were inclined to underwrite based on risk reduction interventions. Underwriters viewed the possibility of future testing or surgery as distinct from genetic test information; they saw action on contemplated surgery as a clear and appropriate instance of protecting the insurer from adverse selection. Based on Brenda's doctor having *discussed* prophylactic surgery, two underwriters would deny Brenda's application, two others would (or likely would) apply a rider excluding coverage for the preventive surgery, and one would surcharge Brenda's premium. If Brenda's medical records indicated her doctor had *recommended* prophylactic surgery, only three of the underwriters would have made a standard offer of coverage. The other 10 said they would deny the application, postpone consideration until surgery had been completed, or issue coverage with a rider excluding coverage for the prophylactic surgery (Table 8).

We presented insurance regulators with this survey finding and asked whether their state laws that prohibit discrimination based

Table 9. State prohibitions on use of genetic services in medical underwriting, individual market

State	Prohibited underwriting action					
	Deny coverage based on:		Raise premium based on:		Exclusion rider based on:	
	Physician discusses risk reduction options	Physician recommends risk reduction options	Physician discusses risk reduction options	Physician recommends risk reduction options	Physician discusses risk reduction options	Physician recommends risk reduction options
AL[a]	x	x	x	x	x	x
AK						
AZ[b]						
AR[c]	x	x	x	x	x	x
CA[d]					✓	✓
CO	x	x	x	x	x	x
CT	x	x	x	x	x	x
DE	x	x	x	x	x	x
DC			x			
FL	✓	✓	✓	✓	✓	✓
GA	x	x				
HI	x	x	x	x	x	x
ID	x	x	x	x	✓	✓
IL[e]	x	x			x	x
IN	x	x	x	x	✓	✓
IA						
KS	x	x	x	x	x	x
KY	x	x	x	x	✓	✓
LA	x	x	x	x	x	x
ME	✓	✓	✓	✓	✓	✓
MD	x	x	x	x	x	x
MA	✓	✓	✓	✓	✓	✓
MI	x	x	x	x	✓	✓
MN	x	x	x	x	✓	✓
MS						
MO[f]						
MT	x	x	x	x	x	x
NE						
NV	x	x	x	x	x	x
NH	x	x	x	x	x	x
NJ	✓	✓	✓	✓	✓	✓
NM						
NY	✓	✓	✓	✓	✓	✓
NC	x	x	x	x	x	x
ND						
OH	x	x	x	x	x	x
OK[g]	x	x	x	x	x	x
OR	x	x	✓	✓	✓	✓
PA	**	**	**	**	**	**
RI	x	x	x	x	x	x
SC	x	x	x	x	x	x
SD						
TN						
TX	x	x	x	x	x	x
UT	x	x	x	x	x	x
VT	✓	✓	✓	✓	✓	✓
VA	x	x	x	x	x	x
WA	✓	✓	✓	✓	✓	✓
WV						
WI	x	x	x	x	x	x
WY	**	**	**	**	**	

Table 9. (continued)

Source: Statutory research by Georgetown University and responses of state insurance regulators to Georgetown survey conducted in May–June, 2006.

Notes: Regulators in five states did not respond to the survey: California, Mississippi, New Mexico, New York, and Vermont. In these states, table only indicates prohibitions found in statutory language. Blank spaces indicate there is no prohibition—either in statute or via other regulatory authority.

√ indicates prohibition found in state statute.

x indicates state regulator confirms practice is prohibited, but practice is not specified in statute.

** Regulator did not answer this question. No statutory prohibition found.

[a] Alabama prohibitions only apply to genetic information about risk of cancer.

[b] Arizona prohibitions apply unless "applicant's medical condition and history and either claims experience or actuarial projections establish that differences in claims are likely to result from the genetic condition."

[c] Arkansas prohibitions apply "except to the extent and in the same fashion as an insurer limits coverage or increases premiums for loss caused or contributed to by other medical conditions presenting an increased risk."

[d] California prohibits insurers from denying "enrollment or coverage to an individual solely due to a family history of breast cancer, or who has had one or more diagnostic procedures for breast disease but has not developed or been diagnosed with breast cancer."

[e] Illinois allows an insurer to "consider the results of genetic testing...if the individual voluntarily submits the results and the results are favorable to the individual."

[f] Missouri prohibits insurers from inquiring "to determine whether a person or blood relative of such person has taken or refused a genetic test or what the test results of any test were..." except with approval of the applicant to consider this type of information.

[g] Oklahoma prohibitions apply "except to the extent and in the same fashion as an insurer limits coverage or increases premiums for loss caused or contributed to by other medical conditions presenting an increased risk."

on genetic information also would protect applicants who explore or pursue preventive or risk-reducing therapies because of their genetic information. Most regulators responded their state prohibition also would protect patients in these circumstances. As one explained, "This information is fruit from the same poison tree." However, eight state regulators did not think their regulatory protections were that broad (Table 9).

Policy Implications

Industry experts and others have insisted that health insurance discrimination based on genetic information happens rarely today, if at all, and there is evidence to support this contention. The low incidence of predictive genetic testing in the general population is one key reason. In addition, prohibitions in more than 40 states may discourage insurers from actively seeking out information about applicants' genetic status or from acting upon such information when it is discovered in the course of underwriting. Most carriers surveyed said they do not underwrite based on genetic information. Responses did not vary when applicants were moved from states that prohibit underwriting on genetic information to states that do not.

However, findings showed that some individual market insurers would act on genetic information if they discovered it. In seven of the 92 decisions tracked by this study, an insurer used genetic information as the basis for its action to decline/postpone and limit coverage or surcharge premiums. These seven decisions were limited to five of the 23 insurance carriers and were spread across all four applicants with genetic information. One of these five respondents expressed uncertainty as to the meaning of one of the genetic tests. Experts in the field of genetics have long called for "vigorous educational efforts" within the insurance industry to improve understanding about genetic information. Findings from this study suggest such education could be beneficial. Comprehensive federal legislation also could reinforce and strengthen state restrictions and promote a uniform standard within the health insurance industry to never use genetic information in medical underwriting.

The study findings raise additional, new questions. When genetic testing is performed in research or clinical settings, patients are almost sure to receive counseling about risk reduction options. To date, statutory prohibitions on genetic discrimination focus narrowly on genetic information in the absence of, or prior to, a diagnosis of the inherited condition. However, therapeutic options to reduce inherited risk may be an overlooked, though important, gray area. Insurers appear more likely to underwrite based on significant

medical interventions (such as surgery) that doctors recommend or even discuss with patients after genetic test results are delivered. On the other hand, most state insurance regulators believe their genetic nondiscrimination laws also protect information about medical treatment patients may consider or pursue to reduce risk of hereditary disease.

These questions remain largely theoretical today, when relatively few patients undergo genetic testing. However, in the not-too-distant future, real-life patients may confront them more often. At this point, the limits to public policy may be tested. From the insurer perspective, medical underwriting in individual health insurance is based on a key premise: the insurer promises to cover an individual's future health care risks, but only if the applicant discloses known risks today. Public policy has insisted on an exception for genetic information—that is, to protect this information, at least partially, because the clinical significance and promise of this science is so profound. Policymakers will have to decide how comprehensive and uniform protections should be. In so doing, they will have to consider the problem of health insurance discrimination in light of what genetic testing means for patients today and what it is likely to mean in the future. Advances in genetic science may make possible dramatic improvements in medicine and public health that can reduce or prevent the incidence of many serious and expensive health conditions. For that day to come, patients will need assurances that they can both learn their genetic status and take appropriate actions to reduce their risk and improve their health without endangering their insurability.

Notes

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1 Task Force concerns have since been documented. See for example Apse et al. 2004. See also Armstrong et al. 2000.

2 See, for example, HIAA 1998. See also Rowe 2002.

3 See for example, Geller et al. 1996, and criticism of this study by the American Council of Life Insurance, "Statement Regarding the Council for Responsible Genetics 'Study' on Genetic Discrimination," April 11, 1996.

4 According to the American College of Physicians, there is currently insufficient evidence to recommend for or against routine *HFE* screening in the general population. However, biochemical testing is appropriate for patients with other complications (e.g., those with type 2 diabetes or cardiomyopathies) and relatives of individuals with HHC. Genetic testing for HHC also may help establish a diagnosis in individuals with elevated iron levels. See Qaseem et al. 2005.

5 An exclusion rider is an amendment to the insurance policy that specifically excludes coverage for a named health condition. Sometimes exclusion riders also eliminate coverage for body parts or systems that a health condition might affect.

6 This type of counseling and medical follow-up related to genetic testing is sometimes referred to as "genetic services."

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