OBAMA-BIDEN TRANSITION PROJECT

THIS DOCUMENT WAS PRODUCED BY AN OUTSIDE PARTY AND SUBMITTED TO THE OBAMA-BIDEN TRANSITION PROJECT.

This document, drafted by the Genetics and Public Policy Center, the Georgetown Health Policy Institute, and the National Workrights Institute, is for the use of interested stakeholders in responding to the Request for Information (RFI) issued October 10, 2008 by the Departments of Treasury, Labor, and Health and Human Services. The RFI may be found at http://edocket.access.gpo.gov/2008/pdf/E8-24194.pdf

Thank you for the opportunity to provide information about issues related to implementation of the Genetic Information Nondiscrimination Act of 2008, in response to the Request for Information issued October 10, 2008 by the Departments of Treasury, Labor, and Health and Human Services. We have organized the discussion by topic area.

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.

[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.

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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.

Example of "family member" as protected under GINA:

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

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

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.

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, which 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 because the test would not detect the "genotypes, mutations, or chromosomal changes" required by GINA's definition.
- An HIV test. Although it is a retrovirus that inserts itself *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"

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.

[2] See, e.g. ERISA §701(b)(1)(B)

- 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 care or testing 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?"
>
> In a case where a health insurer in the individual market asks a 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 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.

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.[3] HIPAA privacy rules provide for several broad exceptions, including one 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

[3] 45 CFR 154.501 (3)

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. For example, medical underwriting questionnaires could be required to prominently state, "In answering these questions, you are protected by federal law from having to reveal any information about your family history or any experience with genetic testing, genetic counseling, or other genetic services not related to diseases you have currently."

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?"

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 practices

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.[4] 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.

[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).

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.

The same study asked underwriters how they would act on information about genetic services. The scenarios presented involved a young woman with a positive BRCA test whose doctor *discussed* or *recommended* the option of prophylactic surgery. In response to the first scenario, five of 13 underwriters said they would take an adverse action – denying her application, surcharging premiums, etc. In response to a doctor's *recommendation* of prophylactic surgery, 10 of 13 underwriters said they would take an adverse action. These findings support the need to clearly specify in regulations that genetic services are protected genetic information and may not be used for medical underwriting purposes.

Group market practices

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:

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

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

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 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.

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.

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.

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.[6] 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.

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

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

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."[7]

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[8]

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.[9] In drafting regulations for GINA, HHS should make 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

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[7] "Genetic Discrimination in Health Insurance: Current Legal Protections and Industry Practices," *ibid.*
[8] 45 CFR 150.205.
[9] 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.

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.

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.