

Response to comments submitted 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

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 have analyzed the comments submitted to the recent Genetic Information Nondiscrimination Act (GINA) Request for Information (RFI). We submit this additional analysis to address a number of issues raised in others' responses to the RFI.

1. Health Risk Assessments, Wellness Programs, and Disease Management Programs

Summary: GINA prohibits health insurers from requesting or requiring genetic information, including family history. There is no exception in Title I for wellness programs, health risk assessments (HRAs) used as part of wellness programs, or disease management programs implemented by health insurance issuers for their enrollees. Thus, genetic information including family history may not be collected through HRAs and wellness programs that are part of the employer-sponsored health plan, nor collected by health insurance issuers prior to enrollment.

Regulations implementing Title I must specify that 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.

HRAs are questionnaires designed to identify preventable health risks on an individual and group level. Typically they cover 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 HRAs; sometimes the program consists exclusively of such an assessment.¹ They have also been used by a health plan prior to enrollment to determine whether an individual should participate in a disease management program targeting behaviors that trigger or worsen a particular condition such as diabetes or heart disease.

Regulations should clarify that programs covered by Title I because they are part of or related to the health plan 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. Regulators should interpret broadly when determining whether a program is part of or related to an employer's health benefit plan. For example, wellness programs that relate directly to the premium contribution required of group

¹ Forrester Research, "What Consumers do with Health Risk Assessments." Oct. 2007.

health plan participants and beneficiaries, or to the benefits or cost sharing available to group health plan participants and beneficiaries, should be considered part of a group health plan. If HRAs are administered by the same health insurance issuers that administer an employer's group health plan, the wellness program should be considered part of the group health plan or related to the group health plan and thus clearly reached by Title I of GINA. Regulators should also consider factors such as whether the employee's health plan premiums vary based on either participation or health status achievements in the program. In such cases, the wellness program should also be considered part of the group health plan and subject to Title I requirements. Regulations also should specify a process for finding that a wellness program is *not* related to or part of a group health plan. A wellness program would be considered separate from the group health plan if it is administered separately from that plan and if no information from the wellness program can be shared with the group health plan or its administrators.

Many insurers and plans appear to be opposed to making changes to their HRAs and other forms based on the prohibitions of GINA. We urge federal regulators to state clearly that HRAs used in conjunction with a group health plan or related wellness, disease management, or other programs may not include questions about family history or other genetic information once GINA is in effect. We recommend heightened oversight on this topic, including outreach and education to issuers and group plan administrators as well as compliance audits.

Regulators should develop – and HRA forms should include – a notice to individuals that they need not and should not provide genetic information in answering any questions on the form. We recommend that if and only if no genetic information is requested on the form and such notice is provided, information that an individual volunteers may be considered “incidental” information and treated as such.

Some comments summarized concerns that will arise under Title II of GINA. Department of Labor (DOL), Internal Revenue Service (IRS), and Department of Health and Human Services (HHS) regulators will need to provide guidance. In particular, these agencies should work closely with the Equal Employment Opportunity Commission (EEOC) to provide guidance on the issues overlapping between Title I and Title II, as these issues relate to wellness programs that impose substantial financial penalties (e.g., higher deductibles) on participants based on results of HRAs.

We believe the HIPAA nondiscrimination rules currently permit a great deal of discrimination based on health status and should be revisited by DOL, HHS, and the Department of Treasury. The EEOC should also consider whether wellness programs currently authorized under HIPAA qualify as “voluntary” wellness programs under federal employment law. However, should current HIPAA nondiscrimination rules stand, GINA regulations should emphasize that GINA's new prohibition on collection and use of genetic information for underwriting purposes supersedes HIPAA nondiscrimination rules. That is, regardless of whether a group health plan may provide different benefits or charge different premiums based on participation in or health status outcomes of wellness programs under current HIPAA nondiscrimination rules, GINA

prohibits the collection or use of genetic information for all underwriting activities defined in GINA, including determination of eligibility for benefits; computation of premiums; or other activities related to the creation, renewal, or replacement of a contract of health insurance or health benefits.

Several responses suggested that wellness programs and/or disease management programs fall under the Rule of Construction 102(a)(2), that allows a health care professional providing health care services to an individual to request that the individual take a genetic test (or ask if the individual has a family history of a condition or for other relevant genetic information). Regulations should clarify what constitutes a treatment setting. It is clear that Congress did not intend for a doctor or nurse's mere involvement in a wellness program or disease management program to exempt a program under this rule of construction.² Thus, a plan representative who happens to be a health care provider and plays a role in a wellness program or disease management program, but has no or a very limited treatment relationship with the individual, does not qualify. However, regulations should clarify that all plans and all plan representatives may inform and educate patients and providers alike about the availability of genetic testing and plan policies for covering such care, so long as no request or requirement is conveyed.

We believe the following examples, based on those offered in the RFI responses, are not prohibited by GINA:

- A health care provider who is providing health care to an individual may make a recommendation about BRCA testing to a patient.
- A health care provider in a staff model HMO where the doctor works for the insurer similarly may request that a patient take a genetic test. He or she is providing health care to the individual.
- A health care provider may request that a patient take a pharmacogenetic test before beginning a particular course of treatment. The health care plan may not directly request that the individual take that test, but the plan may inform the individual of a plan policy that they will not pay for the treatment unless the pharmacogenetic test shows that it will be effective. Pharmacogenetic tests usually refer to diagnostic tools designed to help patients avoid therapies with little chance of success, reduce the number of adverse events, and/or determine what drug or what dose will be most effective for a patient.
- A health care plan may send all enrollees information about carrier screening.
- A health care plan may send targeted enrollees information about genetic testing based on their age, ethnicity, or other indicators that the genetic test is appropriate for them.
- A health care plan may identify people with a manifested specific genetic disease and provide information about a Center for Excellence for that disease.

² See also the RFI response of Congresswoman Louise Slaughter and Judy Biggert, specifying that GINA was intended to prevent insurers and plans and their representatives from contacting patients to request or require them to take a test.

- A health care plan may send reminders to individuals to be sure to get the recommended screenings for conditions for which they are at risk.
- A wellness program that is part of an employer-sponsored group health plan may offer mammography on-site to all employees.
- If family history collected by a personal physician indicates that an individual is a candidate for colorectal cancer screening, a health plan can advise the provider and patient about how the plan covers related services, including genetic testing and increased surveillance through colonoscopy.

Finally, one comment questions whether a voluntary personal health record that includes genetic information and family history can be used by a plan to identify an individual for disease management. Some of the legal rules and practices governing personal health records are in formation and we believe federal regulators should be cautious so as not to inadvertently open loopholes that relate to other areas of law. The privacy of genetic information must be protected and the plan may not request, require, or simply commandeer genetic information in a personal health record. Underwriting on the basis of genetic information in all forms is prohibited, and disease management programs, like wellness programs, should not be underwriting under a new guise.

Blue Cross Blue Shield Association of America (BCBS) states that some plans currently ask about family history and that there will be a cost to changing these forms. They assert that GINA's restrictions on use of genetic information to adjust premiums or establish eligibility do not apply to wellness programs and other HRAs because health promotion and disease management are different from underwriting. They also assert that wellness programs are an example of incidental collection. BCBS notes that health care professionals are specifically permitted to notify an individual about the availability of a genetic test.

Response: As described above, there is no exception in Title I for wellness programs, disease management, and related programs, and thus HRAs for these programs may not include questions about family history or other genetic information once GINA is in effect. Forms should include notice to individuals that they need not and should not provide genetic information in answering any questions on the form. If no genetic information is requested on the form *and* such notice is provided, then any information that an individual nevertheless volunteers may be considered "incidental" information and treated as such.

BCBS is correct that health care professionals may notify or inform an individual about the availability of a genetic test. Health care providers who are providing health care to an individual (i.e., not plan representatives who also happen to be providers) may, in the course of providing treatment or health care, request that an individual take a genetic test or provide family history.

The **Society for Human Resource Management (SHRM)** argues that that HIPAA rules permit HRAs for group health plans as long as eligibility to participate is not conditioned on results of

the HRA. SHRM also states that typically any information such as family history is provided to a third-party service provider, not directly to the group health plan, who does not seek or want the HRA information, only verification that the employee has completed the assessment. However, SHRM acknowledges elsewhere in its comments that employers may obtain genetic information incidentally through implementation of on-site wellness programs.

Response: GINA clearly prohibits the collection of genetic information prior to enrollment in a group health plan. Therefore, HRAs administered prior to group health plan enrollment must be revised to eliminate questions about family history or other genetic information. Some other concerns raised by SHRM concern employers and Title II of GINA. Federal agencies should clarify how they will determine when a wellness program is not a group health plan and thus is subject to Title II of GINA rather than Title I.

The **American Benefits Council** (ABC) recommends that regulations clarify that collection and use of genetic information is permissible for administration of wellness programs. ABC states that it is “typical” for large employers to offer a wellness program, including one that rewards participants for achieving a particular health status, and that it is common for wellness programs to collect data through HRAs that ask about family history. ABC states that the information is used to match participants’ risk factors to wellness program or educational material, and to design future plan benefits or wellness programs. ABC asks for a clarification that although GINA prohibits adjustment of premiums based on genetic information, any adjustment of premiums based on data (including family history) collected through a wellness program or HRA that would be permitted under the HIPAA nondiscrimination rules will still be permitted. ABC notes that in some cases plans provide rewards based on mere completion of HRAs and argues that any information collected this way should be considered “incidental,” not underwriting, so long as any rewards are not based on a particular response.

Response: ABC’s comments contradict SHRM’s arguments, as ABC acknowledges the interest of group health plans and employers in using genetic information to determine benefits and adjust premiums based on the risk factors of the participant population. As described above, HRAs for these programs may not include questions about family history or other genetic information once GINA is in effect and we recommend that forms notify individuals that they need not and should not provide genetic information.

America’s Health Insurance Plans (AHIP) asks for clarification that wellness programs may use genetic information, stating that wellness programs run by health plans may look at genetic predisposition to diseases that may then be prevented or managed. AHIP provides examples of how this information might be used, such as to provide an alternative standard for a patient who has familial hypercholesterolemia who is supposed to meet a particular cholesterol level standard as a wellness plan participant. AHIP also argues that GINA should not apply when a wellness program does not require participants to meet a standard in order to receive a benefit. AHIP also notes that a medical provider who is part of a “bona fide” wellness program may tell an enrollee about the availability of a genetic test.

Response: Wellness programs that fall under Title I may not collect or use genetic information including family history information on HRAs or other forms prior to enrollment in the group health plan or at any other time for underwriting purposes. Although a health care provider who is part of a wellness program may inform an enrollee about the availability of a genetic test, only a health care provider who is providing care to the individual may request that the individual take a genetic test (or provide genetic information such as family history). Regulations should clarify that a plan representative who happens to be a doctor or a nurse does not qualify and such plan representatives cannot collect such information prior to enrollment. The wellness program exception is part of GINA Title II and does not apply to group health plans with wellness program components under Title I.

National Business Group on Health (NBGH) describes the growing employer practice of requesting that employees complete HRAs that include family medical history questions. NBGH states that these HRAs may come prior to employer plan enrollment, and there may be financial incentives for completion of HRAs, such as eligibility for specific medical treatments. According to NBGH, employers require HRAs to “secure participants’ early focus, attention, and participation” and to “alert vendors who evaluate health risk.” NBGH argues that these practices are not prohibited by GINA.

Response: NBGH’s comments suggest that the use of HRAs before and during enrollment in a health plan is widespread. Under GINA, the programs that fall under Title I may not collect or use genetic information, including family history information, on HRAs or other forms. Federal agencies should clarify a process for determining when an HRA is not part of a group health plan and thus is instead subject to Title II of GINA and the HIPAA rules regarding wellness plans.

2. Genetic Information in Payment Determinations, and the “Minimum Necessary” Standard

SUMMARY: 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 establish the medical necessity of the requested care.

In some cases, an enrollee’s own medical history or other non-genetic information may suffice to prove medical necessity. Some enrollees may decide independently to reveal genetic information to prove medical necessity; in other cases family history or a genetic test may be *required* to show medical necessity.

Regulations should provide additional guidance, emphasizing that plans should follow widely accepted clinical practice guidelines for determining when genetic information, including family history, is needed to justify a test, treatment, or procedure. AHIP and BCBS suggest adoption of the HIPAA standard for what information is the “minimum necessary.” However, this standard

is very general, requiring only “reasonable efforts” to limit the use or disclosure of protected health information. Given that the prohibition on plans’ ability to request or require information is one of GINA’s core protections, we suggest that more specificity is needed. We also recommend that requests for test results by insurers should be accompanied by a statement that the test result is the minimum necessary information required and notifying the patient and doctor of their right to appeal if they disagree. We also recommend that federal regulators establish a process for enrollees and plans to handle such appeals.

BCBS states that genetic test results may be needed for coverage decisions or claims determination, and that the “minimum necessary” information will need to be determined on a case-by-case basis.

Response: The examples offered in BCBS’ response suggest that in determining whether a test is medically necessary the BCBS rules will typically default to requiring family history and/or a genetic test result. However, in some cases this collection and use of genetic information should not be necessary. For example, if an insurer were to always require a positive BRCA test result before approving coverage for preventive surgery or heightened breast cancer screening, this rule would be overly broad, exceeding the minimum necessary standard for some patients. A patient’s own history of previous cancer could be offered as proof that preventive surgery or heightened screening is medically necessary to reduce her risk of future breast and ovarian cancer. We suggest that, at a minimum, federal agencies should develop a process to adjudicate whether the minimum necessary standard was met in cases of disputed claims. If a plan denies payment for a claim based on what an individual provides, evidence must be cited for why the information provided is insufficient, and the patient and doctor should have the opportunity to submit alternative information if they disagree.

NBGH says that in some cases the use of genetic information is necessary for payment purposes.

Response: See discussion above of minimum necessary rule.

SHRM comments that genetic information may be necessary to verify the type of a test administered and the fact that it was conducted. This allows the plan to determine whether the test itself is a covered service and the amount the plan will pay.

Response: In these cases no genetic information need be revealed. The fact of the test being conducted should be the information necessary to allow the plan to determine payment.

AHIP states that plans may request and review the result of an individual’s genetic test to determine if a particular treatment (such as prophylactic mastectomy) is medically necessary. AHIP also states that plans should be able to use pharmacogenetic testing in order to predict response to treatment. AHIP asks for the ability to obtain the results of genetic tests to make payment determination “without federal requirements or restrictions.”

Response: Although plans may collect and use genetic information in determining payment, GINA requires the plan to request only the minimum amount of information necessary to accomplish the intended purpose. In most cases plans should not tell patients *a priori* that a genetic test result is necessary to obtain payment, unless they can provide an evidence-based policy showing the appropriateness of that rule in those particular circumstances. We believe GINA does not interfere with a plan's ability to require a patient to reveal the results of a scientifically validated and clinically accepted pharmacogenetic test before the plan agrees to pay for a particular drug treatment. Ideally this testing will occur through the patient's treating health care professional.

3. *Incidental Collection of Genetic Information*

Summary: GINA's prohibition of collection of genetic information helps ensure that the information is not used for underwriting. However, it is not a violation of GINA if a plan obtains genetic information *incidentally* through the request, requirement, or purchase of other information. We recommend that regulators interpret "incidental" collection narrowly and provide clear guidance on what plans must do to avoid it.

Regulations should specify that health plans and issuers are not allowed to ask for, seek, or obtain genetic information about applicants before they enroll in coverage. For example, although questions about laboratory tests legitimately may be asked on an application for a medically underwritten individual health insurance policy, 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 for any other genetic information, including family history of disease, and that such information need not and should not be revealed in answering questions. As described above, we recommend that regulators develop model language for plans to notify individuals that they need not provide genetic information on any questionnaires or applications prior to enrollment in health insurance or a group health plan. Similar notification should be provided in other verbal and written interactions between plans and enrollees.

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, such as a request for all medical records, should be prohibited. 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.

We also recommend that if plans do obtain genetic information incidentally, regulations should require 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. Plans and

insurers should keep records of inadvertent collection that show information was not used for underwriting and make the records available for auditing and compliance efforts.

HIPAA rules that protect the use and disclosure of health information by insurers have permitted its use for health care operations, including underwriting. However, GINA now prohibits the use of genetic information for underwriting. Thus, plans must institute strict firewalls within their companies to make sure genetic information that is incidentally collected and used for permitted purposes such as claims payment or quality review never reaches the plan's medical underwriting office, where its use is prohibited.

Our responses to specific examples follow:

BCBS describes examples of situations in which incidental collection may arise:

- A patient volunteers information in course of claims or service inquiry
 - **Response:** As described above, we recommend that regulators develop model language for plans to notify individuals that they need not provide genetic information in verbal or written interactions with the plan.
- A plan reviews past claims files for purposes of quoting a new business rate/ A plan acquires claims data from another insurer in the course of a merger or acquisition
 - **Response:** Claims review may result in a plan incidentally acquiring genetic information. As described above, we recommend that regulations require insurers to have a written and enforceable policy as to what steps to take when genetic information is inadvertently collected. Regulations should require plans and insurers to notify the enrollee if information was inadvertently collected. Plans and insurers should keep records of inadvertent collection and make them available for auditing and compliance efforts.

SHRM provides examples of situations in which incidental collection may arise, although several of the examples would fall under Title II of GINA.³ SHRM requests clarification on how genetic information must be safeguarded internally so that employers will not be liable for inappropriately having or using such information.

Examples offered by SHRM that seem to fall clearly under Title II include:

- In a voluntary Weight Watchers plan, unrelated to group health plan, sponsored by employer; at a meeting one employee tells another about inherited health condition; information “flows” into employee population.
- Employer sponsors on-site health facility and hires the doctor directly. During medical emergency, doctor learns of employee genetic information.
- Employer requires death certificate to justify bereavement leave, cause of death listed is genetic.

Response: Regulations should provide clarity about how plans will be held responsible for information that is collected incidentally through legitimate activities. Regulations should explain the interaction between GINA Title I and GINA Title II as well as GINA's interaction with existing laws and regulations that establish how to keep information needed for administration of a health benefit plan separate from underwriting or from an employer's hiring and firing functions.

SHRM also gives an example of rating by a self-insured group health plan (presumably for setting COBRA premiums). We suggest the plan should be required to certify what claims data were used and attest to the fact that genetic information did not affect rate. SHRM also requests additional clarification of employer liability if actuaries or other agents use information for claims data analysis and group ratings. We agree that regulators should clarify the liability of agents.

AHIP suggests the following scenarios under which incidental collection could occur:

- An underwriter reviews an applicant's medical records and incidentally notices genetic test results.
- An individual applies for coverage with an insurer that previously insured her (and therefore had claims data).
- An individual applies for coverage with an insurer that previously insured one of her family members (and therefore had family history claims data).
- An applicant voluntarily divulges genetic information without being asked.
- An individual voluntarily enters genetic information on her Personal Health Record
- One insurer purchases or merges with another.

AHIP also asks for guidance on a plan's review of a provider's use of genetic testing

- to assess compliance with standards of clinical practice and evidence-based medicine or
- to review billing practices and medical records including genetic information to assess whether fraud and abuse has occurred.

Response: We agree that clarification is needed on these scenarios. The scenario involving an underwriter combing "medical records" underscores the need for regulations that emphasize the importance of narrow requests for information. The underwriter may not use and should not receive genetic information and should take care to request only information that is permitted for underwriting. Broad dumps of "medical records" almost inevitably will include genetic information. Regulations should be clear that plans should protect genetic information obtained by the health plan from falling into the underwriter's possession and keep records showing that inadvertently collected information was not used for underwriting.

The scenario involving an individual seeking coverage from an insurer who covers her relative

raises concerns. The insurer should not be reviewing or probing the relative's claims while processing the new application for insurance. Such a practice would be a direct seeking of family history and is prohibited under GINA.

ABC suggests that genetic information (family history) should be considered collected incidentally when it is not the "sole purpose" of an HRA to collect such information. ABC argues that because plans provide rewards based on mere completion of HRAs, rather than on the basis of the answers or any subsequent health behavior, any information collected should not be considered underwriting because no rewards would be based on a particular response.

Response: As described in greater length above, Title I of GINA does not permit health insurance plans to collection genetic information including family history through health risk assessments, wellness programs, or disease management programs – doing so would not be considered "incidental" collection, and it is simply prohibited.

ABC states that processing information for quality improvement may result in incidental collection. ABC argues that regulations should define as "incidental" any request for or collection of data where genetic information is "not the primary focus, using examples of the collection of genetic information for wellness programs, HRAs, on-site clinics, or quality improvement activities."

Response: ABC's proposal is far too broad. The question of the "primary focus" of a program or form is irrelevant. GINA's intent is to prevent the collection of genetic information that may then be used to discriminate against an individual. Group health plans and group and individual health insurance issuers should not be permitted ask for, seek, or obtain genetic information about applicants before they enroll in coverage. Permitted questions should be narrowly framed and regulators should develop model language for plans to notify individuals that they need not provide genetic information on HRAs and other health forms. Regulations should underscore the duty of insurers and group health plans to take affirmative steps to *avoid* requesting, requiring, or purchasing genetic information.

As described above, we recommend that if plans do obtain genetic information incidentally, a variety of regulatory approaches should ensure that the information is not used to discriminate in underwriting.

ABC also argues that because GINA uses HIPAA's definition of "group health plan," there is an exception for on-site health centers, which they argue should be defined broadly to include health fairs.

Response: HIPAA regulations from 1997 do not provide any elaboration on the definition of the exemption for "coverage for on-site medical clinics." See ERISA 733(c)(1)(G) It would be important to provide such guidance, to distinguish what is meant by "coverage for" health care clinics and the more common health fairs. If an employer provides on-site medical clinic for

employees to receive personal medical attention, it is reasonable to assume these providers are subject to the same rules and permissions as other health care providers and may inquire about family history and discuss or recommend genetic testing. But it must be made clear that the provider and the clinic may not share information with the group health plan or the employer. And if this clinic is exempt from Title I it must fall under GINA Title II.

4. *Research Exception*

SUMMARY: Under GINA’s “research exception” a group health plan or a health insurance issuer 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 that protect patients and prevent genetic information from “research” from being used for underwriting by a plan or issuer. It is critical that regulations ensure that written voluntary informed consent is obtained from every participant in research.

Federal regulators should develop a process by which research is appropriately scrutinized so that it cannot become an overly-broad exception to the GINA rule that plans may not request and collect genetic information. Regulations should state that the rules apply to any research conducted by *or supported* (partially or fully funded by) a group health plan or health insurance issuer. Regulations also should make clear that plans claiming GINA’s research exception must obtain written voluntary informed consent. In order to be sure that this occurs, federal regulators should not simply accept IRB approval but institute a regulatory process by which plans show that they are obtaining consent. IRB approval is not necessarily adequate in all circumstances because although GINA specifies that research should comply with 45 CFR 46 or equivalent regulations under the Food and Drug Act, there are circumstances under which research is exempt from these regulations or under which requirements are waived.⁴ 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

⁴ 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. 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.”

eligibility rates, *all* research should involve written voluntary informed consent from every participant.

Plans and issuers should also 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 application and in the notice they provide to the secretary of the Department of Health and Human Services.

We believe 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.

The **Pharmaceutical Care Management Association (PCMA)** notes that genetic tests keyed to drug therapies are being developed and "when such tests are available health plans and insurers may wish to avail themselves of the research exception to enable their plan participants and beneficiaries to participate in clinical studies measuring the effectiveness of such tests in managing drug therapies."

Response: Research relating to genetic testing and drug therapy would qualify for the research exception under GINA if it meets the other requirements described above.

Kaiser Permanente (KP) recommends that model notice (to participants) should not be duplicative of other required disclosures or informed consent. KP asks regulators to clarify that this is not a "separate approval process" for projects but rather "a process that is already conducted by IRBs." KP also states that "rulemaking should clarify that the research exception is limited to notification."

KP states that a model form for plans wishing to claim the research exception would be helpful and recommends the form include certification that written notice was given to participants, research is compliant with requirements of research and informed consent, and participation is voluntary. KP seeks specific guidance about the timing and content the form and suggests that notification to regulators should be on a regular periodic basis rather than for each individual project.

Response: We agree that a research exemption notice to participants would be helpful and should be written to be neither duplicative nor inconsistent with other disclosures or informed consent required under all applicable laws and regulations. Regulators should ask plans claiming the research exception to show that written voluntary informed consent is obtained from each research participant. IRB approval alone may not be sufficient if the IRB waived consent requirements or found research to be exempt from IRB review, thus regulators should confirm that consent was obtained.

AHIP cites Kaiser Permanente and Aetna as examples of plans conducting research. AHIP states that regulations should permit plans to sponsor, continue, and support valuable research projects, programs and protocols using genetic tests.

AHIP supports a model notice for participants in research but states it “should not be based solely on GINA’s research requirements” and that HHS should work with other federal, state, and local agencies to provide information that is not duplicative. AHIP also supports a model form for plans claiming the research exception and asks for clarity about which agency would receive such a form.

Response: Written voluntary written informed consent should be obtained from every patient. Plans should be able to provide proof of this to federal regulators and should be required to show proof in the form submitted in requesting the exemption. In some cases, IRB approval alone may not suffice because of the possibility of waiver or exemption. We agree that model notice to patients would be helpful. We agree that the information provided should not be duplicative of other legal notice requirements.

BCBS seeks clarification whether research using claims databases which may include the fact that a claim for a genetic test was paid or that multiple claims were paid for same family should not be considered research requiring the exception.

Response: This is not research subject to the exemption. We believe these circumstances would fall under the rules for review of claims. GINA would prohibit the use of any information inferred or directly collected from such review.

5. Definitions

SUMMARY: Definitions in GINA should be clarified in the regulations. We recommend that regulators provide examples and remain flexible as technology changes.

Genetic information and genetic tests

MIB group describes the definition of genetic information as “construed by GINA proponents” as including family history and argues that it could be construed more narrowly.

Response: The definition of genetic information in the text of GINA includes family history. This issue is settled in the legislative language.

The **American Society for Human Genetics** (ASHG) asks for clarification that

- Genetic information collected before GINA’s effective date is protected.
- New techniques such as assessing methylation and evaluating metabolites (as in newborn screening) should be included in the definitions.

Response: We agree with ASHG’s recommendations.

AHIP recommends that regulations should clarify that genetic tests are not genetic information simply because testing involves genetic material or precedes an individual's application for health insurance or actual insurance coverage. They state clarification is needed that if the test is directly related to a manifested disease or condition, it does not meet the definition of a genetic test.

Response: We agree that clarification is needed that provides examples of testing that does and does not meet the GINA definition. However, it is not the case that any test related to a manifested disease or condition does not meet the definition. For example, a woman with breast cancer might undergo BRCA testing. The BRCA test is protected by GINA. The information that the woman has breast cancer is not. Regulations should clarify that the genetic information of an individual cannot be used simply because manifest disease is present.

Genetic services

AHIP asks that regulations clarify that GINA does not prohibit insurers from using actual claims experience to set initial and renewal premiums for groups. They state that the costs of genetic tests or genetic services do not meet the definition of "genetic information" and so insurers can set or renew premium rates or contribution amounts for the group as a whole using this information.

Response: As indicated in the Senate report, the costs of genetic information can be included, along with costs of all other claims, for purposes of experience rating in group health plans. However, this is different from saying that the fact that a patient has used genetic services (including genetic counseling, testing, increased intensive screening, or prophylactic surgery) can be used to justify a rate increase for that patient based on his/her genetic risk for future health care needs. GINA would not permit that in either group or individual coverage. Further, history of claims for genetic services (or for recommended genetic services) cannot be used by health insurers in any market to disqualify a patient from coverage, or subject a patient to a pre-existing condition exclusion. This approach is consistent with Congressional intent to allow and encourage access to life-saving preventive care.

Manifest disease

Numerous comments agree that GINA regulations should provide additional guidance on the definition of "manifest disease" and should define this term as meaning the existence of signs and symptoms, not merely a genetic marker or genetic test result. For example, **BCBS** requests that regulations make a "clear distinction" between manifest disease and genetic information.

AHIP requests clarification on how agencies will view an example where an individual receives a genetic test that should have shown a manifest disease or condition, but the disease was not detected because of human error. Presumably AHIP believes this should not be considered a

genetic test, and not be protected by GINA because it falls into the (ii) exception to the definition of 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”

Response: It is difficult to think of an example that would arise under this scenario, but it would be appropriate for regulations to clarify that a reasonableness standard applies and “could reasonably be detected” would apply whether or not the particular provider actually did detect the disease in that particular case.

AHIP also argues that GINA does not prohibit insurers from setting or adjusting premiums for a group based on an “individual’s health information, including information about the manifestation of a disease or disorder...”

Response: This description of what GINA does not prohibit is not quite accurate. GINA does not prohibit underwriting on the basis of health information that is not genetic information. Health information that *is* genetic information may not be the basis for setting or adjusting premiums.

AHIP argues that when claims data are used to set initial or renewal premiums for a group, information about the claims data of family members covered under the same policy can be used.

Response: As long as one family member’s health information is not used to further increase the premium of another relative in the group, GINA protection is satisfied. Because there is other applicable federal and state law in this area we recommend that the regulations specify that GINA *does not prohibit* this practice rather than saying insurers *can set premiums* based on manifested disease.

6. *Scope*

AHIP requests clarification of the application of GINA to FEHBP, state and local government health plans, SCHIP, Medicare, and Medicaid, and the intersection of GINA with state laws and NAIC model regulations. AHIP also requests clarification that HIPAA “excepted benefits” are not included in GINA such as disability insurance, other insurance products for specific aspects of health care. AHIP argues that long term care should be clearly outside scope of GINA.

Response: We agree that clarification would be helpful in these areas. We anticipate that to the extent some benefit plans (such as disability or life insurance) are offered through employers, GINA Title II will apply.

7. *Enforcement*

Many responses to the RFI requested clarification of how GINA Title I will be enforced and how the “firewall” between Title I and Title II remedies will operate.

We agree, and believe it would be helpful for federal regulations to explain, that NAIC’s adoption of GINA modifications to their model Medigap act constitute a “safe harbor”: states that adopt the model are not preempted by federal law. It is likely that come May 21, there will be state laws that do not conform to GINA and clarity is needed about what will happen then. To the extent state laws depart from GINA, federal guidance in regulations is necessary to describe how federal preemption will work.

8. HIPAA Privacy Rules

AHIP proposes specific language to ensure that changes to HIPAA privacy rule are limited to GINA requirements and states importance of there being no restrictions or interference with delivery of health care or HIPAA Privacy Rule’s definitions for treatment or payment functions.

Response: Before GINA, HIPAA permitted the use of protected health information for health care operations including underwriting. Now, GINA prohibits underwriting on the basis of genetic information. Regulations need to ensure that the underwriting function of plans is kept separate from other health care operations where genetic information may be used. When legitimately obtained or used, genetic information should be flagged to prevent its collection by underwriters and use in underwriting. Health insurers should certify to federal agencies that any genetic information they may possess was not used for underwriting.