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The Genetic Information Nondiscrimination Act of 2008: Selected Issues

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Abstract. On May 21, 2008, the Genetic Information Nondiscrimination Act of 2008 (GINA) became law (P.L. 110-233). GINA prohibits discrimination on the basis of genetic information in employment and health insurance. This report addresses several issues that the new law, and its future implementation, may raise.

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Summary

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On May 21, 2008, the Genetic Information Nondiscrimination Act of 2008 (GINA) became law (P.L. 110-233). GINA prohibits discrimination on the basis of genetic information in employment and health insurance, and is often described as the first civil rights legislation of the 21st century. The complexity of the U.S. health care system and the continually evolving field of genomics are both reflected in the crafting of GINA. The Department of Health and Human Services recently released a Request for Information (RFI) to assist in formulating its proposed regulations for GINA. This is a good indicator of the level of complexity that will be involved with the implementation of GINA.[1] Some of the issues that the new law may raise are discussed in detail in this report.

Scope of the Law

Broadly, any genetic nondiscrimination legislation had to address, through its determined scope, two key distinctions with significant policy implications. The first distinction is that of manifested, or existing, disease or conditions versus presymptomatic disease or conditions that are not clinically evident. The second distinction is that of genetic information that is predictive of, or indicates a predisposition to, a future disease or condition versus other types of genetic information, including diagnostic information that identifies and confirms a disease or condition. Specifically, lawmakers had to decide whether to include protections for individuals with manifested genetic conditions within the scope of the law, and also had to decide whether to define genetic information to include genetic information beyond predictive genetic information. As passed, GINA *does not* protect against discrimination based on manifested genetic diseases or conditions but *does* provide protection against discrimination based on genetic information beyond predictive genetic information, including diagnostic genetic testing, pharmacogenetic testing, carrier testing, and tumor profiling.[2] Thus, GINA is not limited to protecting only presymptomatic individuals from discrimination based on their future risk of disease, as predicted by their genotype. The policy implications of the scope of the law, thus delineated, are discussed in detail below, and specific examples are provided.

Manifested Genetic Disease

Many individuals may believe that GINA affords protection against discrimination in health insurance and employment on the basis of a manifested genetic disease or disorder (for example, cystic fibrosis), in addition to protecting against discrimination based on genetic information.

[1] See "IRS, DOL, HHS Joint Request for Comments (REG-123829-08) on Implementation of Genetic Information Nondiscrimination Act of 2008 (GINA) (P.L. 110-233)." Accessed at http://pubs.bna.com/ip/BNA/DTR.NSF/4bdb7473996f34e385256b57005ad41a/ 85256453007e2c8d852574de00068c19?OpenDocument on October 31, 2008.

[2] Pharmacogenetic testing may be used to determine both the appropriateness of using a specific drug to treat a given condition in a specific individual and the appropriate dosing regimen of a drug for an individual. A pharmacogenetic test may be defined as a genetic test intended to identify individual variations in DNA sequence related to drug absorption and disposition (pharmacokinetics) or drug action (pharmacodynamics), including polymorphic variation in the genes that encode the functions of transporters, receptors, metabolizing enzymes, and other proteins.A carrier can be defined as an individual who possesses one copy of a mutant allele that causes disease only when two copies are present. Although carriers are not affected by the disease, two carriers can produce a child who has the disease. Carrier testing identifies these individuals. http://www.genome.govTumor profiling may be defined as obtaining and processing complex information from tumors or their precursors that can be used to optimize classification for the purpose of diagnosis, staging, prognosis prediction, and therapy selection. http://www.tumorprofiling.org/methods/tumor_profiling.htm

However, GINA does not appear to shield individuals with a manifested genetic disorder or disease from genetic discrimination in health insurance or employment based on their manifested disease or medical information about their manifested disease.

Title I of GINA specifically clarifies that nothing in the Act prohibits a health insurer or an employer from increasing health insurance premiums, establishing a preexisting exclusion, or establishing rules of eligibility for individual enrollment "based on the manifestation of a disease or disorder of an individual who is enrolled in the plan." In addition, Title II, Section 210, of GINA states that an employer will not be in violation of the Act based on the "use, acquisition, or disclosure of medical information, that is not genetic information, about a manifested disease, disorder, or pathological condition of an employee or member, including a manifested disease, disorder, or pathological condition that has or may have a genetic basis." Taken together, these provisions clarify that the protections set forth by GINA do not apply to manifested disease or medical information, that is not genetic information, about a manifested disease.[3]

Although advocates supported drafting GINA to provide protection against discrimination based on manifested genetic disease, many also argued that GINA should not include protections for such discrimination. Opponents argued that to include such protections would not be in keeping with the spirit of the law, which in significant part is to protect individuals from discrimination in health insurance and employment based on the possibility of future disease, as predicted by their genotypes. With respect to health insurance, a mandate prohibiting insurers from rating policies based on manifested genetic disease would have been fundamentally disruptive to a system predicated upon just such an approach. In addition, commentators believed that it would be unfair to protect individuals with genetic diseases and disorders and not those with diseases or disorders with no known genetic basis. With respect to employment, many believed that existing law, such as the Americans with Disabilities Act (ADA), would provide adequate protection against employment discrimination based on manifested disease, genetic or otherwise. For these reasons, GINA was crafted so as not to extend protection against discrimination based on any manifested disease.

Predictive and Other Genetic Information

During the lengthy debate about genetic nondiscrimination legislation, many argued that it is the predictive and probabilistic potential of genetic information that justifies special regulation of this information's use and collection, and that any law should be limited to predictive genetic information. Specifically, proponents maintained that discrimination against a healthy individual on the basis of the possibility of illness in the future, as uncovered through a genetic test, was unjust. However, GINA's definition of genetic information includes genetic information beyond predictive genetic information, including much diagnostic genetic test information. Advocates supported this broader definition, arguing that genetic information that does not predict susceptibility to disease, such as carrier status or pharmacogenetic testing, should also be protected.

[3] Protection against discrimination based on manifested diseases may be provided by other laws, for example, the Americans with Disabilities Act (ADA). The ADA, 42 U.S.C. sec. 12101 et seq., prohibits discrimination against individuals with disabilities. For a discussion of the ADA, see CRS Report 98-921, *The Americans with Disabilities Act (ADA): Statutory Language and Recent Issues*, by Nancy Lee Jones.

GINA protects against discrimination based on genetic information, and the definition of genetic information in turn rests on the definition of genetic test. Both titles of GINA define a genetic test as "an analysis of human DNA, RNA, chromosomes, proteins or metabolites that detects genotypes, mutations, or chromosomal changes." Title I excepts from its definition of genetic test "an analysis of [human] proteins or metabolites [that detects genotypes, mutations, or chromosomal changes] 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."[4] Biochemical genetic tests provide examples of this type of test, where a protein or metabolite is analyzed, detecting a genotype, mutation or chromosomal change, which may be related to a manifested disease or disorder. In addition, Title I excepts analyses of proteins and metabolites that do not detect genotypes, mutations, or chromosomal changes, such as a cholesterol test, and does not apply to analyses of non-human DNA, RNA, chromosomes, proteins or metabolites (for example, HIV testing).

However, this definition *does not except* any analyses of human DNA, RNA, and chromosomes that detect mutations, genotypes, or chromosomal changes. Therefore, such analyses that produce information that is diagnostic, predictive, or not clearly either in any given situation (such as pharmacogenetic testing, some tumor profiling, or carrier testing) are all protected under GINA. For example, diagnostic genetic test results for Canavan's Disease would be protected genetic information under GINA, as would susceptibility genetic testing results for mutations in the BRCA1 or BRCA2 genes, which could be used to assess future risk of breast or ovarian cancer. This broad definition, as noted previously, also includes pharmacogenetic test results, carrier testing results, and some tumor profiling.

Issues Raised by the Scope of GINA

There are several considerations that the scope of GINA, as discussed in the previous two sections, raises.

First, the second exception to the definition of genetic test in Title I raises questions about why analyses of proteins and metabolites that detect genotypes, mutations, and chromosomal changes, and are directly related to a manifested disease, should be treated differently than similar analyses of RNA, DNA, or chromosomes. This may create a situation where individuals with a manifested disease may face different protections against discrimination based on the type of analysis they undergo to determine therapy, even though both types of analyses technically uncover information about their genotype, mutations, or chromosomal changes. Tumor profiling provides an example of this issue. While tumor profiling based on an analysis of DNA, RNA, or chromosomes to determine therapy or risk of recurrence is to be protected information under GINA, similar information based on the analysis of protein would not be protected. A 2007 study describes the discovery of a protein, S100A2, that is predictive of poor survival in pancreatic cancer and may help select patients who could benefit from surgery.[5] Results from this type of tumor profiling, which may in some cases detect changes at the genetic level, would not be covered under GINA. Both employers and insurers might be interested in such information.

[4] P.L. 110-233, Section 101(d)(7)(B)(ii).

[5] Ohuchida K. et al. "Over-expression of S100A2 in pancreatic cancer correlates with progression and poor prognosis." *J Pathol* 213: 275-282; 2007.

In addition, this exception seems to create the possibility of protecting the results of some newborn screening tests and not others. Specifically, the results of newborn screening tests that are biochemical genetic tests (i.e., they analyze protein or metabolites; detect mutations, genotypes, or chromosomal changes; and are directly related to a manifested disease) may be excepted from protection under GINA, while more classical genetic tests (for example, for cystic fibrosis) would not be excepted. This difference in protection would be based on the target of analysis of the test (i.e., protein in the case of biochemical genetic newborn screening tests or nucleic acid in the case of the classical genetic test) rather than on what the test uncovers: genetic changes in both cases.

Second, the definition of the term "manifested" will affect the scope of the law as implemented. The term is used to create two exceptions in Title I and one in Title II. With respect to the exception to the definition of "genetic test" in Title I, the broader the term is, the more analyses will be excepted from protection under GINA. The earlier the stage of disease defined as "manifested," the broader the exception will be. With respect to the exception allowing insurers to make insurance and employment decisions based on manifested disease, again, the broader the definition, the fewer individuals will potentially be protected under GINA.

Finally, there is the issue of protecting genetic information about a manifested disease, while at the same time not protecting manifested disease status (current health status) or medical information about a manifested disease. Specifically, in the case of diagnostic genetic testing, it is difficult to envision how an employer or insurer would use that genetic information as a basis for discrimination, and how an individual could prove it, because it would occur in the context of manifested disease. At the very least, this may create confusion for the individual in the situation, who may feel protected by virtue of the fact that the disease was diagnosed using a genetic test falling under the scope of the definition of the term in GINA. It is conceivable that health insurers may underwrite hereditary cancers differently than other cancers, if the hereditary cancers were known to have certain properties (e.g., be more aggressive) that might increase costs. In this case, a diagnostic genetic test result, in addition to medical information about a manifested disease, may result in a higher premium, for example, than might simply medical information about a manifested disease.

Selected Examples

The reach and scope of the definitions of genetic test (and therefore, of genetic information) may be more clearly explained through the use of examples. It has been reported that in 2005, a player for the Chicago Bulls franchise experienced cardiac symptoms that may have predisposed him to sudden death during strenuous physical exertion. The team reportedly wanted this player to undergo genetic testing to help determine whether or not he could play safely.[6] In such a case, a positive genetic test result, in the presence of certain cardiac symptoms, would be considered diagnostic (i.e., it would diagnose a manifested disease).[7] It is instructive to consider whether GINA, as passed, would have protected a player in such a case.

GINA would have prohibited an employer from requesting or requiring that an employee undergo genetic testing. Title II, section 202(b) states, "[I]t shall be an unlawful employment practice for

[6] http://www.nytimes.com/2005/10/03/sports/basketball/04curry-wire.html?_r=1.

[7] http://www.nytimes.com/2005/10/06/sports/basketball/06knicks.html?pagewanted=print.

an employer to request, require or purchase genetic information with respect to an employee."[8] However, GINA likely would not have protected an employee from adverse employment action based on any medical information, that is not genetic information, that was available to the employer at the time. For these reasons, it would appear that even with GINA's protections in place, an employer may have taken adverse employment action against an employee, although an employer likely could not have required an employee to take a genetic test. This example also illustrates the lack of a "dangerousness" exception in GINA. The ADA, in contrast, contains such an exception, which has been found to include dangerousness to self.[9] The ADA may provide further protection in this case, but that analysis is beyond the scope of this paper.

In another, fictitious, example, consider an individual who has a manifested disease and undergoes genetic testing to determine either how effective certain drugs, or different doses of the same drug, might be in treating her illness. In this case, it would appear that an employer or a health insurer could, to the extent permitted under other law, discriminate based on the current health status of the individual (i.e., his manifested disease). The same employer or insurer could not use genetic tumor profiling or pharmacogenetic test results, used to guide therapy or drug dosing, as a basis for discrimination. For example, if the results of onco*type*DX® testing determine that an individual's breast cancer is likely to respond well to chemotherapy, this would potentially increase costs to both an employer and a health insurer. However, under GINA, this information may not be factored into either employment or health insurance decisions, although the individual's manifested breast cancer may be considered.

Concluding Remarks

GINA is a complex law, made more so by the constantly shifting state of the science and the fragmented health care system. Issues surrounding whether certain tests are "genetic tests" under the law, or whether certain collections of symptoms are "manifested disease," may be challenging to resolve. These issues may be addressed through regulation; health care providers, as well as advocacy groups, would then face a need to translate this knowledge to the general public.

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[8] P.L. 110-233, Section 202(b).

[9] The ADA specifically lists defenses to a charge of employment discrimination, including that the term "qualification standards" can include a requirement that an individual shall not pose a direct threat to the health or safety of other individuals in the workplace. 42 U.S.C. § 12113. The Supreme Court has interpreted this language to mean that the ADA does not require an employer to hire an individual with a disability if the job in question would endanger that individual's health. *Chevron U.S.A. Inc., v. Echazabal*, 536 U.S. 73 (2002).