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Perspective

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GINA, Genetic Discrimination, and Genomic Medicine

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In 2008, after 13 years of advocacy by the genetics community and U.S. lawmakers, the Genetic Information Nondiscrimination Act (GINA) was signed into law. GINA is the first U.S. federal antidiscrimination statute crafted to address an area where there was no well-documented history of widespread discrimination and no stigmatized group to protect. The statute's language is unusual, proposing not only "to fully protect the public from discrimination" but also to "allay their concerns about the potential for discrimination, thereby allowing individuals to take advantage of genetic testing, technologies, research and new therapies."

GINA was initially lauded as a huge legislative success, though some critics argued that it didn't go far enough in providing comprehensive protection. For example, GINA defined genetic information to include genetic test results and family history but excluded from its protections people with manifest disease. This exclusion established a legislative gap between employment protections afforded by GINA for asymptomatic people, as well as those offered by the Americans with Disabilities Act (ADA) for people with substantially limiting impairments. People with manifest disease who are not yet disabled were left unprotected. In addition, whereas GINA prohibits discrimination in employment and health insurance, it does not address life, disability, or long-term care insurance. And it does not apply to employers with fewer than 15 employees or to the U.S. military, the TRICARE military health system, the Indian Health Service, the Veterans Health Administration, or the Federal Employees Health Benefits Program, though other policies protect people in those programs.

Has GINA nevertheless succeeded in its twin missions of preventing discrimination and alleviating public fears about the potential for discrimination? That's a difficult question to answer. In the past 6 years, genetic testing has dramatically increased, yet there have been very few cases of discrimination in which GINA's authority could be tested. The Equal Employment Opportunity Commission (EEOC) estimates that in fiscal year 2013 there were 333 GINA-related charges of employment discrimination (vs. more than 90,000 in other areas), and most of them also included ADA-related claims. In some cases, according to Peggy Mastroianni of the EEOC, the GINA claim was added only after the investigation of an ADA charge uncovered evidence that an employer had asked about an employee's family history.

Although GINA does not explicitly prohibit discrimination in the provision of life, disability, or long-term care insurance — and in selling such products, companies can legally utilize genetic information and do routinely ask about family history — there is little evidence that even those companies are requesting and utilizing genetic test results in their underwriting, despite recent increases in genetic testing. Perhaps GINA sent a powerful symbolic message to all insurers that society would not tolerate genetic discrimination, and that message was heard.

> ***Fear of discrimination may be preventing some people from participating in translational research studies that are exploring the real-world consequences of utilizing genomic information, with its potential for unanticipated or incidental findings.***

Whether GINA has allayed fears of discrimination as intended is even more difficult to assess, especially since the law is not well known. In a multistate survey conducted in 2010, 2 years after GINA was enacted, less than 20% of adult respondents said they were aware of it.[1] In June 2014, we conducted an online survey of 1479 people in the United States using Mechanical Turk, a marketplace for Web-based surveys run by Amazon.[2] Of the respondents, 79% were still unfamiliar with GINA, and of those who claimed to be familiar with it, only 44% knew that it protected against genetic discrimination in health insurance, 31% knew that it provided protection in employment, and 23% incorrectly thought it provided protection in life, disability, and long-term care insurance. Surprisingly, awareness did not necessarily translate into reassurance: after reading a description of GINA, 30% of respondents reported that they were actually more concerned about discrimination.

In fact, there's some evidence that fear of discrimination may be preventing some people from participating in translational research studies that are exploring the real-world consequences of utilizing genomic information, with its potential for unanticipated or incidental findings. For example, in the ongoing MedSeq Project,[3] a randomized trial in which the results of whole-genome sequencing are electronically stored in participants' medical records and patient and physician outcomes are tracked, 25% of prospective participants who declined participation cited fear of insurance discrimination as the primary reason, after a consent process in which they were specifically educated about GINA. It's hard to know whether such refusals reflect a failure of GINA to sufficiently allay concerns about discrimination — or simply represent the appropriate exercise of informed consent.

The Affordable Care Act (ACA) changes the prism through which GINA may be viewed. The ACA prohibits discrimination by health insurers on the basis of preexisting conditions, including genetic test results, thereby closing the gap in health insurance protection for persons with manifest disease. However, the ACA does not address that gap in terms of employment discrimination, and it encourages employers to offer wellness programs that could tie health insurance costs to employee participation — creating a potential conflict with GINA if such programs utilize family history or predispositional genetic testing.[4]

It is tempting to argue that protections against genetic discrimination should simply be expanded to cover life, disability, and long-term care insurance. But these products are perceived as more optional and commercial than health insurance, and companies selling them practice a type of open discrimination, classifying people in risk categories in order to charge different premiums to different groups. The tension between what GINA does and does not cover highlights fault lines in the efficiency–equity tradeoffs of the insurance marketplace. Thus far, we have collectively decided that it is inequitable to discriminate against employees or in the pricing of health insurance but more acceptable to discriminate for products such as life, disability, and long-term care insurance. In defending the right to such discriminatory underwriting, insurers have claimed that if applicants have relevant information that isn't available to insurers, such as robust genetic risk information, low-risk consumers will drop out of the mix and higher-risk consumers will disproportionately purchase coverage, forcing companies to raise prices and causing a "death spiral" of adverse selection.

This concern was largely theoretical until we showed that healthy people with higher-risk results on predictive genetic testing were more likely to use that information to make decisions about purchasing long-term care insurance.[5] If this finding is generalizable, then for insurance products that remain outside GINA's scope, the status quo is unlikely to last. As more people obtain their own genetic risk information, companies selling such products may feel forced to test customers genetically in order to stratify customer risk. Alternatively, we may eventually have to abandon risk-based underwriting and adopt a more unitary pricing system that pools risk.

The standard argument for regulating risk classification is that it's unfair for employers to discriminate or insurers to charge different rates because of immutable risks. GINA's exceptionalism may, in part, reflect a genetic determinism and therapeutic nihilism that were prevalent in 1995, when Congress first considered this issue, but that will be far less salient in the future. Although genetic determinism with regard to highly penetrant mendelian conditions may persist, it's now clear that everyone carries genetic variants that will influence, but in most cases not exclusively determine, one's health status. The science of genomic medicine is moving rapidly toward multiscale network and systems biology by elucidating the complex interactions of genomics, physiology, and environmental influences. In a future informed by this science, we may be able to personalize risk stratification and then tailor diet, exercise, and pharmaceuticals and even edit genes to promote wellness by preventing and minimizing illness. Eventually, the notion of immutable genetic risks may become obsolete, and it may be less important to grant genetic information special protection than to protect everyone from all forms of medical discrimination. As all medicine in a sense becomes genomic medicine, perhaps the genetic nondiscrimination secured by GINA will translate into nondiscrimination in all of medicine.

Disclosure forms provided by the authors are available with the full text of this article at NEJM.org.

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Using Drugs to Discriminate — Adverse Selection in the Insurance Marketplace

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Eliminating discrimination on the basis of preexisting conditions is one of the central features of the Affordable Care Act (ACA). Before the legislation was passed, insurers in the nongroup market regularly charged high premiums to people with chronic conditions or denied them coverage entirely. To address these problems, the ACA instituted age-adjusted community rating for