



Keeping Pace with the Times — The Genetic Information Nondiscrimination Act of 2008

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LAWS and institutions must go hand in hand with the progress of the human mind. As that becomes more developed, more enlightened, as new discoveries are made, new truths disclosed, and manners and opinions change with the change of circumstances, institutions must advance also, and keep pace with the times.

— Thomas Jefferson, July 12, 1810

When the first federal legislation to prevent the misuse of genetic information was introduced in 1995, many in the health care, research, and policy communities considered the measure to be forward looking. Others called it premature. After all, scientists were just getting ready to start the sequencing of the human genome. Only about 300 genetic tests were available, most of them for rare diseases and usually performed in research settings.

Yet, anticipating an explosion in the clinical relevance of genetic

testing and sensing Americans' growing concern that their genetic information could be used against them by health insurers and in the workplace, we and many others became convinced that reforms were needed as soon as possible.^{1,2} Little did we know that "as soon as possible" would mean a 13-year legislative saga that culminated on May 21, 2008, with President George W. Bush's signing of the Genetic Information Nondiscrimination Act (GINA) of 2008. At last, the United States has a federal law that protects con-

sumers from discrimination by health insurers and employers on the basis of genetic information (see box).

In the years between GINA's inception and its enactment, genomic information has grown exponentially, revolutionizing nearly all areas of biomedical research and, many believe, promising an eventual transformation of health care. Researchers completed the reference sequence of the human genome in April 2003 and went on to produce a map of human genetic variation that has greatly accelerated the search for genes involved in susceptibility to common diseases. Genetic tests now encompass more than 1500 conditions, with most of the growth in the area of common diseases. With many of these tests becoming available in the clinic and some even being offered directly to consumers, GINA's protections could

Quick Guide to GINA

What GINA does

- Prohibits group and individual health insurers from using a person's genetic information in determining eligibility or premiums
- Prohibits an insurer from requesting or requiring that a person undergo a genetic test
- Prohibits employers from using a person's genetic information in making employment decisions such as hiring, firing, job assignments, or any other terms of employment
- Prohibits employers from requesting, requiring, or purchasing genetic information about persons or their family members
- Will be enforced by the Department of Health and Human Services, the Department of Labor, and the Department of Treasury, along with the Equal Opportunity Employment Commission; remedies for violations include corrective action and monetary penalties

What GINA does not do

- Does not prevent health care providers from recommending genetic tests to their patients
- Does not mandate coverage for any particular test or treatment
- Does not prohibit medical underwriting based on current health status
- Does not cover life, disability, or long-term-care insurance
- Does not apply to members of the military

Key terms

"Genetic information" includes information about:

- A person's genetic tests
- Genetic tests of a person's family members (up to and including fourth-degree relatives)
- Any manifestation of a disease or disorder in a family member
- Participation of a person or family member in research that includes genetic testing, counseling, or education

"Genetic tests" refers to tests that assess genotypes, mutations, or chromosomal changes

Examples of protected tests are:

- Tests for *BRCA1/BRCA2* (breast cancer) or *HNPCC* (colon cancer) mutations
- Classifications of genetic properties of an existing tumor to help determine therapy
- Tests for Huntington's disease mutations
- Carrier screening for disorders such as cystic fibrosis, sickle cell anemia, spinal muscular atrophy, and the fragile X syndrome

Routine tests such as complete blood counts, cholesterol tests, and liver-function tests are not protected under GINA

no longer be dismissed as premature; they were rapidly coming to seem essential to Americans' ability to make the most of the much-anticipated era of personalized medicine.

Still, in a policy system that may be better suited to responding to crises than promoting prevention, legislators are rarely in an optimal position to act on the potential effects of emerging technologies. Thanks to the efforts of key lawmakers, their staffs, and

advocates such as the Coalition for Genetic Fairness, GINA eventually garnered overwhelming bipartisan support in the current Congress. One silver lining of GINA's slow progress through Congress is the many opportunities it offered to educate policymakers about the potential of genomic medicine and the challenges that must be addressed if we are to realize that potential.

"GINA is the first major new civil rights bill of the new century,"

said Senator Edward Kennedy (D-MA), who cosponsored GINA in the Senate with Senator Olympia Snowe (R-ME). "Discrimination in health insurance and the fear of potential discrimination threaten both society's ability to use new genetic technologies to improve human health and the ability to conduct the very research we need to understand, treat, and prevent genetic disease," said Kennedy.

To be sure, some protections existed before GINA. The Health Insurance Portability and Accountability Act of 1996, for example, provided some restrictions on the use of genetic information in setting premiums and determining eligibility for benefits in group health plans. GINA, however, will strengthen those safeguards by limiting insurers' ability to use genetic information to raise rates for an entire group and by extending protections to individual health insurance plans. Also, before GINA's passage, many states had enacted laws against genetic discrimination, which varied widely in their scope and degree of protection. GINA now sets a nationwide level of protection but does not preempt state laws that provide even broader safeguards.

Despite the historic protections provided by GINA, we acknowledge that the law is not perfect and does not go as far as many organizations and families had wished. Originally, some had hoped to include protection for people in whom a genetic illness has been diagnosed — not just those whose tests show a genetic susceptibility to disease. Such a provision, however, had two important flaws, one economic and one ethical. First, it would have caused a severe disruption in the individual health insurance mar-

ket in the United States, which currently underwrites on the basis of diagnosed diseases. Second, it would be fundamentally unjust to treat people with genetic diseases differently from those whose diseases are nongenetic or have unknown causes. In the end, lawmakers settled on protecting genetic information that could predict future disease, along with the genetic test results of people who are already affected by a genetic disease.

Along with the benefits it provides to individuals, the new law should have positive effects on the fields of clinical research and health care delivery. Studies have shown the “fear factor” to be a major obstacle to patients’ participation in research studies that involve the collection of genetic information. Fear of genetic discrimination has also put a damper on patients’ willingness to consider genetic tests recommended by their health care providers or to have the results of such tests included in their medical records.³ It must be emphasized that GINA does not in any way limit the ability of health care professionals to do what they are currently doing; they may still use their clinical judgment to decide whether or not to recommend genetic testing to patients under their care.

“This bill unlocks the great promise of the Human Genome Project by alleviating the most common fear about genetic testing,” said Representative Judy Biggert (R-IL), who cosponsored GINA in the House with its leading proponent, Representative Louise Slaughter (D-NY). “It will accelerate research . . . and allow Americans to finally realize the benefits and health care savings offered by gene-based medicine,” noted Biggert.

Now that the President has signed GINA, federal agencies must write the implementing regulations that will provide detailed guidance for health insurers and employers about how to comply with the new law. The health insurance regulations will take effect 12 months from now, and the employment regulations 6 months after that. However, it will take much more than sound regulations to ensure that we reap the full benefits of GINA. We need to make certain that health care professionals and patients understand the new protections — and, equally important, that clinical researchers, research administrators, institutional review boards, and research participants are fully informed about the new law and its implications. Such educational efforts are daunting, given the decentralized nature of our systems of health care delivery and protection of human subjects.

Although safeguarding genetic information from misuse by health insurers and employers is a key prerequisite to more individualized approaches to medicine, many other critical challenges remain. First and foremost, we need to ensure that genetic tests are safe, reliable, and marketed in a clear and truthful manner. There are important gaps in the oversight of genetic tests, and multiple advisory groups have called for regulatory reform to ensure the analytic and clinical validity of genetic tests.^{4,5} Clearly, our country’s substantial investment and innovation in genetic science ought to be matched by innovation in regulation.

Finally, we need to look carefully at other areas of our society in which it might be tempting to use — or misuse — genetic information. GINA addresses only em-

ployment and health insurance, not life insurance, disability insurance, or long-term-care insurance. This is not the result of an oversight: a strategic decision was made early on to recognize the very distinct markets, social purposes, risks of adverse selection, and bodies of relevant law governing these types of insurance. It may well be time for a thoughtful evaluation of these other realms that are likely to be touched by the swift advance of genomic science.

No potential conflict of interest relevant to this article was reported.

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A video interview with Dr. Collins is available at www.nejm.org.

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