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Keeping Secret Codes

How a law designed to prevent genetic discrimination may not protect the patients who need it most.

BY HEATHER BOERNER



WHEN BLOOD SAMPLES sent from the University of Chicago Medical Center arrive at a clinical genetic testing laboratory, geneticists spend weeks carefully extracting the blood's most elemental components: wispy strings of DNA. Isolated using heat and an alkaline and processed through a machine designed to isolate just the right section of DNA, the results come out as little pink, red, green, and blue spikes on a slip of paper. This red peak may mean a mutation of the gene that governs blood clotting. That green one may mean a genetic predisposition to breast cancer. Whatever the case, the geneticist analyzes them all against a standard for that part of the DNA helix, creates a report on the results, and sends it back to the University of Chicago, where it might land in the hands of Melody White-Perpich.

White-Perpich, a genetic counselor at the Cancer Risk Clinic at the University of Chicago Medical Center, will review the findings and then, instead of filing them down in the basement where patients' regular medical records are kept, she will head toward a special cabinet in her office. There, she'll slip the document into a separate, very thick folder.

For 10 years, that file has housed her patients' genetic profiles and, by keeping that data outside of their regular charts, protected the hun-

dreds who pass through her doors every year from genetic discrimination by insurers and employers. It is, she says, a very important file—and a very important part of her job is to defend the information within it.

"We call that file a shadow file," said White-Perpich. "Though we are not aware of any of our patients experiencing genetic discrimination, we've been conscientious about protecting our patients."

Only after she and her patient discuss the ramifications of the genetic results will the patient make the choice: Sign a medical release form, add that slip of paper to her official medical files, and report the results to her other healthcare providers so they can begin a specialized regimen of observation, medication, testing, or prophylactic surgery; or return that paper to the shadow file to protect it—and the patient—from the prying eyes of medical underwriters who might use that information against the patient by, for example, excluding coverage for a disease the tests show she has a propensity to develop.

White-Perpich, however, foresees a day in the not-so-distant future when shadow files may disappear.

It's not that medical underwriting has stopped. Indeed, the healthcare industry continues to use people's illnesses as an excuse to discontinue coverage or raise premiums. But starting this year in

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November, White-Perpich and her fellow genetic counselors shouldn’t need to keep secret patient files because, under a law that President George W. Bush signed in May 2008, people’s genetic code can’t be used to discriminate against them. Sen. Ted Kennedy hailed the law, called the Genetic Information Nondiscrimination Act (or GINA), as “the first major new civil rights bill of the new century.”

Supporters see the law as potentially ushering in a new age of medicine, imagining a world in which practitioners can harness breakthroughs and understand a person’s genetic makeup to create truly personalized care plans, saving their patients from the pain and suffering caused by some of the country’s most common illnesses.

But while the law offers promise, it also endured a 13-year legislative battle that some say left its ideals in place but its enforcement capacity battered. And after eight years of the Bush administration, the agencies

charged with regulating employers and health insurers are hobbled as well. So though the law may be the most airtight healthcare discrimination law ever passed, observers worry that it could be nothing more than a feel-good platitude from a lame-duck president.

“It will raise an interesting question,” said Karen Pollitz, director of the Health Policy Institute at Georgetown University. “What happens if you throw a law and no one comes?”

Because Joanna Rudnick discovered through genetic testing that she carries certain disease markers, she receives routine MRIs as part of surveillance for breast and ovarian cancer.

GENETIC MEDICINE HAS taken off only in recent decades. In the 1950s, researchers sought to confirm obscure and inheritable illnesses through what they then knew of the genetic code. Those tests were limited to single-gene mutation illnesses or chromosomal disorders, such as sickle-cell anemia or Down syndrome.

But in 1990, the National Institutes of Health and the U.S. Department of Energy began a vast undertaking: They aimed to sequence all 20,000 to 25,000 genes on the human genome, to discover mutations, and spur new research. Then the field broke open. In 1995, NIH scientists discovered specific alterations in the genetic code associated with breast and ovarian cancer. Suddenly, genetic testing went from a rare endeavor to a potential savior for thousands of women and some men diagnosed with the disease every year.

“For a long time with GINA, lawmakers thought it was a solution in search of a problem,” said Joann Boughman, executive vice president of the American Society for Human Genetics, a professional organization for geneticists. “But a lot changed their mind when they discovered the BRCA [breast cancer] genes. Just about everyone knows someone with breast cancer.”

Today, 4.1 million newborns receive genetic tests for up to 28 genetic diseases, and thousands of adults every year get genetic tests to determine everything from whether one is a carrier for hemophilia to whether one carries the gene that increases risk for Alzheimer’s disease. Others are tested to confirm medical diagnoses.

But genetic medicine and genetic testing are tricky undertakings fraught with ethical dilemmas. While science has now uncovered up to 900 genetic abnormalities that could lead to medical conditions, scientists haven’t developed cures or treatments for all of those disorders. So it may not always be in a patient’s best interest to get tested, said White-Perpich. It also may not be in the best interests of the patient to keep information that is relevant to her care out of her permanent medical file.

The Council for Responsible Genetics worries about such “predictive” genetic tests because its members aren’t sure the benefits outweigh the potential stigma associated with knowing you may develop a disease.

Likewise, simply having a genetic mutation is not a guarantee of illness. For instance, this year, the National Cancer Institute at the National Institutes of Health expects 192,000 American women to be diagnosed with breast cancer. Only about 5 to 10 percent of those women will carry the BRCA 1 or 2 mutations. And not all women with the mutation will develop breast cancer.

For Joanna Rudnick, learning she had the BRCA1 gene mutation seven years ago didn’t seem to change her life terribly at first. She got the test after her sister, who’d already been tested, suggested it. She and her sister didn’t think there was much chance they would have it even though their mother had ovarian cancer at 43. They didn’t see it elsewhere in their family tree, and just wanted to confirm their assumption.

Later, they both discovered what no one of her mother’s or grandmother’s generation talked about: Ovarian cancer was all over her family tree, on both sides of the family, as well as later-onset breast cancer on her mother’s side of the family. Rudnick is also Ashkenazi Jewish, a group that’s genetically predisposed to such mutations.

Then the test results came back: positive.

“When I first found out, it felt extremely stigmatizing, very isolating,” said Rudnick, a Chicago native and documentary filmmaker.

She didn’t talk about it. She kept the news in a “separate corner” of her mind. She didn’t really confront what it meant for her until she started a documentary on other people who had also tested positive for the gene mutation. They call themselves “previvors,” she said: people who are facing the risk of cancer every day.

For Rudnick, that has meant she takes her body in for a heavy rotation of medical tests every six months: twice-yearly transvaginal ultrasounds to observe her ovaries and breast MRIs to check for lumps. She hopes that by being vigilant she can catch any cancer in its

beginning stages. She hasn't ruled out eventually having prophylactic surgery to remove her breasts, eliminating almost all of the tissue at risk for developing breast cancer.

"My body is under heavy surveillance," she said. "I'm watched in a way I never would have been if I hadn't had the test."

EVEN AS GENETIC TESTS were being developed to help people like Rudnick monitor their health, observers were noticing some distressing trends: Clinical trials that required test subjects to have their genes sequenced refused to participate, fearing their health insurers would drop them. Worried patients began to pay for the tests out of pocket or through risky mail-order companies to keep the results off the books. They hesitated to let their doctors know their results and couldn't explain to their health insurers why they needed more regular monitoring.

Before GINA, some piecemeal protections against genetic discrimination did exist. The Health Insurance Portability and Accountability Act of 1996 included genetic privacy protections for people covered under group health plans. And 47 states have some form of genetic discrimination law, ranging from prohibiting employers from having access to their workers' genetic test results to prohibiting medical underwriters from using genetic test results to raise premiums, deny coverage, or drop someone from a plan. But up until now, the law has been spotty at best, and none of them have been tested with a genetic discrimination case resulting in a court decision.

In a 2001 suit between the U.S. Equal Employment Opportunity Commission and Burlington Northern Santa Fe Railroad Company, union members who submitted workers compensation claims for carpal tunnel syndrome complained that the company was requiring them to submit to a blood test—a test that unbeknownst to them was being used to determine if any of the employees had a genetic predisposition for the condition. When one worker refused to submit to the test, he claimed the company threatened to fire him.

Eventually the case settled out of court, and Burlington Northern agreed to eliminate genetic testing from its practices.

"Regardless of its outcome, this case serves as a beacon illuminating a troubling future," warned Michael Stein, then an assistant law professor at the College of William and Mary and now director of the Harvard University Project on Disability, in a journal article on genetic discrimination. "It warns people that they may have to absorb liability for injuries to themselves if [employers] can show that they are less than normally resistant to being injured."

Then rumors started swirling. Had anyone lost their health insurance because of a genetic test? None of the genetic counselors, geneticists, or experts at the National Institutes of Health who talked to *Registered Nurse* had witnessed any genetic discrimination. But the Council for Responsible Genetics claims it has documented more than 500 cases of such discrimination.

It certainly is possible, said Georgetown's Pollitz. Her department surveyed chief medical underwriters at a number of health insurers, asking them what they would do if they were presented with a potential customer who had tested positive for a genetic abnormality related to a medical condition.

"Several said they wouldn't take the person with genetic test

results," she said. "Others said they'd offer coverage but charge more, or permanently exclude from coverage the condition for which they were genetically predisposed. So we documented that it could happen."

And so, the shadow file in White-Perpich's office was born.

It's no wonder, then, that when Rudnick decided to get tested seven years ago, she did so anonymously, paying out-of-pocket the several hundred dollars it cost to extract her DNA and sequence it.

"I was terrified I'd be dropped by my insurance," recalled Rudnick. "I was so afraid my insurance company would say, 'You're uninsurable' that I kept it out of all my medical records and had to really fight to get the preventive care I needed. I was always saying, 'Please don't tape-record our discussion.' I filled out health forms and never put anything down about it. The irony is that when you have this [genetic] information, nothing is more important than health insurance."

Indeed, a cottage industry of direct-to-consumer mail-order genetic tests has sprung up to cater to that fear. The industry, regulated by the Federal Trade Commission, has been cited a few times for offering misleading and inappropriate testing. For instance, some companies promise to send you personalized skincare, vitamin, and other "nutrigenomic" healthcare guidance based on your genetic results. The FTC warns consumers that mail-order genetic tests "aren't a substitute for traditional healthcare" because results can be difficult to analyze.

AWARE OF THE PROBLEMS, geneticists and privacy advocates began campaigning for GINA 13 years ago. That's the same length of time it took to sequence the entire human genome. So when GINA passed, Rudnick found herself planted in front of C-SPAN, watching, and crying.

"I was thinking of all the people who didn't get tested because they were afraid of losing their job or insurance and crying," she said. "That barrier just isn't there anymore."

GINA has some promising characteristics: It extends genetic discrimination coverage to individual health insurance. It creates a federal mandate of genetic privacy over health insurance and employment. It does not, however, prevent health insurers from asking about your family history on insurance applications. It doesn't prevent insurers from dropping patients or raising their premium once a genetic illness begins to manifest. And the law doesn't cover disability or life insurance.

One of the primary things it does do is consolidate and strengthen state laws. Many contain language that make them all but worthless, said M.K. Holohan Quattrocchi, senior health policy analyst at the National Human Genome Research Institute of the National Institutes of Health. For instance, some laws exclude chemical tests from coverage—but all genetic tests are chemical tests.

Not only does GINA, which is set to fully go into effect in November, standardize the laws, it also sets a higher standard for state regulation than any previous law, said Pollitz.

"The standard GINA sets out for states is that any time the state fails to enact or enforce the law, the federal government can come in and enforce it," she said. "It's not 'close enough.' It's really, 'You better adopt this as we wrote it.'"

But the primary breakthrough of this law is its clarity on the point of genetic discrimination. Health insurers and employers

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cannot know genetic information about people they cover or employ, except in rare instances when employers use genetic tests to ensure that their employees aren’t being exposed to unsafe chemicals at work.

“It becomes quite a promising model because Congress has now in one law been quite thorough,” said Pollitz. “It says you just can’t discriminate at all. So many federal reforms—HIPAA being a prime example—outlawed some discrimination but left some alone. This one is an important precedent.”

Then she added, “It’s important to really get the regulation right everywhere. Otherwise, there will be lingering concerns on the part of people that maybe it won’t protect them completely.”

But there’s a problem—or there may be. Remember, GINA bounced around the halls of Congress for 13 years. Desperate to pass it, supporters finally agreed to some considerable watering down of the law, said Michael Watson, a geneticist and executive director at the American College of Medical Genetics, a trade and professional group for geneticists.

“When GINA started, there was a world more enforcement in the bill,” he said. “Over the past eight years, there’s been a constant reduction in enforcement [provisions]. When it was up for a vote this last round, it was getting serious and those involved recommended that we accept some reductions in enforcement to get enough Congress members to sign on.”

The biggest reduction in enforcement came in the legal arena: The right to sue is gone. Under versions of the law proposed in 2003, state attorneys general and individuals had the right to sue if one lost his job or health insurance, or if his health insurance premium increased, due to disclosure of genetic information. In the version proposed by Rep. Tom Daschle, there was no limit to how high the reward could be.

The new law strips that right and replaces it with a series of penalties, to be enforced by the federal Equal Employment Opportunity Commission, the Department of Health and Human Services, Department of Labor, and Department of the Treasury: \$100 per day per patient when an insurer or employer doesn’t comply with the law; between \$2,500 and \$15,000 yearly for each violation.

But the law also provides several limitations to soften the bite of regulation, including forgiving discrimination when it’s corrected within 30 days of first occurrence, and forgiving violations that are corrected before the date on which the insurer receives a violation notice from regulators. The law also limits the overall amount of penalties an insurer can be charged if the violations of the law were

unintentional to 10 percent of its taxable income during the previous tax year or \$500,000.

“We made the penalties much less draconian,” said Rep. Judy Biggert, one of the Republicans who cosponsored the bill. “We wanted to make sure they [insurers and employers] followed the law but we weren’t going to make a nightmare out of it.”

What will enforcement look like once it’s put into place? No one knows, said Pollitz, who recalls seeing draft language of changes to the law that would have “gutted it.” Regulators at the applicable agencies are writing the regulations now.

“Between passage and implementation is interpretation,” she said.

The Bush administration has been notoriously lax in regulating insurers and other industries. Take, for example, HIPAA. When it passed in 1996, the Department of Health and Human Services created the Health Care Financing Administration and the Office of Private Health Insurance. The Clinton administration hired state insurance regulators to run the office, and created a structure in which regulators could ensure that the law was being implemented equally in all 50 states. The regulators regularly traveled across the country, answering questions and assuring that people were protected from discrimination.

“And then Clinton left town and Bush came in and it’s just gone,” Pollitz said of HCFA. “There’s not an office of private insurance anymore. Someone might have that title, but they don’t do anything. A lot of the state regulators quit and left. A lot of the people in the federal government got reassigned to work on Medicare Part D or whatever else. It’s been pretty thoroughly dismantled.”

The office has been so dismantled, in fact, that when Pollitz attended a meeting of state insurance regulators recently and mentioned the passage of GINA, state regulators “had no idea” what she was talking about.

Likewise, when HIPAA first passed, three states—California and two small states—refused to implement it. The federal HCFA was able to step in and force enforcement in the two smaller states. But with the large size of California, the federal government wasn’t able to enforce it.

“People in California didn’t have HIPAA rights for the first two years,” Pollitz said. “With GINA again, it raises the question of what happens when someone is in trouble? Even though Congress says you have these rights, that you have these protections, if there’s no enforcement, you have nothing.” ■

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