Information Overload?

As genetic testing kits sold directly to consumers gain popularity, medical professionals and lawmakers wonder if more regulation is needed.

BY KRISTINE GOODWIN

Information is power, the adage goes, and in the brave, new world of direct-to-consumer genetic testing, that information has the potential to help people lower their odds for developing disease.

“In the not too distant future, we expect that physicians will be able to look at patient genetic profiles and assess the best health-care plan,” says Anne Wojcicki, co-founder of the California-based genetic start-up 23andMe.

Today, consumers with curiosity, Internet savvy and a credit card can go online to one of dozens of direct-to-consumer genetic testing companies, and for a few hundred to a couple of thousand dollars find out if they are predisposed to certain cancers, Alzheimer’s, diabetes and the like. The direct-to-consumer umbrella is a large one, with some companies offering genetic tests—like those that determine whether a person is a carrier of a gene for cystic fibrosis, for example—and others offering personal genomic profiles that predict whether a healthy individual has a genetic mutation that puts them at risk for disease. Still others in the market provide nutritional profiles based on a person’s genes and another subset specializes in using genes to determine ancestry.

“There are tests that are beneficial,” says Gail Javitt, a law and policy director at the Genetics and Public Policy Center at Johns Hopkins University, but there is a lack of uniform standards for measuring whether a test is valid or useful. “Right now,” she says, “there is not a process for ensuring that those standards are being met, and as a result, consumers are vulnerable.”

The situation has led to a patchwork of laws around the country and a quandary for policymakers and consumers alike: How to sift through the plethora of companies and technologies and know which tests and services deliver on their promises.

Michael Watson, executive director of the American College of Medical Genetics, worries some direct-to-consumer genetic tests trivialize a complex and nuanced issue. Since many diseases have unknown causes, he says, people might be misled into thinking that a genetic test alone can determine their risk of getting an illness. Factors such as family history, lifestyle and environment also can play a role. Scientists don’t know exactly how genes and environment interact, and there is no genetic smoking gun for many of the conditions being tested.

What’s more, Watson points to “a lack of understanding about the implications of a positive test result.” Put simply, having a higher risk of developing diabetes doesn’t mean that you’re going to get it, and conversely, a person without an elevated risk has no guarantee he’s immune.

“Many of the DTC companies … draw no lines as to how strong the genetic influence needs to be for them to say it is diagnostic versus a low-level risk factor,” Watson argues.

Genetic tests are not new. How they are marketed to the public, however, is.

“Direct-to-consumer is just a method of marketing the test,” says Javitt. The technology is the same whether the test is administered at home or in the doctor’s office. With direct access to testing, there may not be a doctor or genetic counselor involved to “provide a gatekeeping function to make sure the right people get the right tests and they understand what the tests tell them.”

AN EVOLVING INDUSTRY

George Church thinks that in a few years your personal genome will be a household staple, as common as the flood and fire insurance that you hope to never use. Church, a professor of genetics at the Harvard Medical School and founder of the Personal Genome Project, envisions people will pay for this information because it will help them stay healthy and extend their lives.

“Nobody wants to hear that they have cystic fibrosis or Huntington’s disease,” Church argues, but they hope to have the information on file in case they need it some day. “If the price comes down enough, and enough people see some advantage, it will be as common as an insurance policy.”

With the breakneck pace of discovery in the field of human genomics, what costs about $350,000 today—the price for a nearly-complete genome sequence—could drop to about $1,000 by 2009, Church says, though others say it may take longer.

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This fast-moving testing industry picked up where the federally funded Human Genome Project—a $3 billion, 13-year project headed up by the U.S. Department of Energy and the National Institutes of Health—left off. The landmark project resulted in the first complete sequence of the human genome in 2003.

The project’s achievements, combined with significant public and private funding, catalyzed the biotechnology industry and helped to create today’s market of direct-to-consumer testing. The potential demand for these services has investors like Google and Genentech hoping the technology will pay off. The genetic testing market could be worth $12.5 billion by 2009, The Economist reported in 2007.

MURKY REGULATORY ENVIRONMENT

Federal oversight of direct-to-consumer genetic testing is limited. The Food and Drug Administration regulates the safety and effectiveness of medical devices, but has conducted no reviews of at-home genetic tests, according to a 2006 consumer alert from the Federal Trade Commission.

Federal law also requires certification for genetic testing laboratories and addresses personnel qualifications and quality control, among other things. However, Javitt explains, the law “doesn’t look at clinical validity or clinical utility of tests.”

Further complicating matters: Some direct-to-consumer companies offering genomic profiling argue the law doesn’t apply to them because they are not providing medical information.

Twenty-six states and the District of Columbia permit direct-to-consumer testing without restriction, often because the state law is either ambiguous or silent on the matter, according to a 2007 survey by the Genetics and Public Policy Center at Johns Hopkins University. Thirteen states require a physician or other approved provider to order the tests, which effectively bars consumers from ordering the tests on their own. Another 11 states allow consumers to order certain medical tests—such as glucose and cholesterol—but require an authorized health-care provider to order all genetic tests.

Javitt, who headed up the survey, says that most of these laws pre-date the “explosion” of at home genetic testing, and as a result, they are not tailored to today’s hard-to-

Preventing Genetic Discrimination

Lawmakers agree that genetic tests—and the information they reveal—should not come back to bite consumers.

“Genetic medicine holds enormous promise, but the right consumer protections must be in,” says Susannah Baruch, a law and policy director at the Genetics and Public Policy Center at Johns Hopkins University.

To that end, Congress passed—and President Bush signed—the Genetic Information Nondiscrimination Act of 2008, or GINA, that prohibits health plans from requiring genetic testing, and prohibits health plans from using genetic information for underwriting purposes. The law also protects employees from discrimination by their employers based on genetic information, and requires employers to treat any employee’s genetic information as a confidential medical record.

Although details vary, the majority of states already have laws that restrict the use of genetic information by employers and insurance companies, and lawmakers across the country continue to strengthen protections. Thirty-four state and the District of Colombia prohibit employers from discriminating based on genetic information and 44 states and the District prohibit health insurers from basing eligibility on genetic information.

Maryland Senator Jennie Forehand sponsored legislation that was enacted in 2008 to include long-term care insurance, an area that nine other states have addressed in some fashion.

“People are getting tested more often, and that’s a really good thing,” Forehand argues. “You want people who are worried about [a condition that runs in the family] to face it so they can make life decisions.”

Maryland’s law applies to long-term care insurance—GINA does not. Other states—such as Arizona and California—set limits on disability and life insurance companies, areas that also are not addressed under the federal law. Although all the details haven’t been worked out with the federal legislation, “in general, individuals will be able to pursue relief under state laws that are more comprehensive than GINA,” Baruch said. “GINA is considered a ‘floor’ rather than a ‘ceiling’ of protections.”

States With Limits on Direct-to-Consumer Genetic Testing

Maryland Senator Jennie Forehand
Genetic Information ‘Helpful’ for Consumers

When Thomas Goetz decided to have his DNA analyzed by California-based companies 23andMe and Navigenics, he was “curious but not worried.” In the name of research, Goetz, who is deputy editor of Wired magazine in San Francisco, sent off his saliva samples to the companies back in October 2007 and April 2008, along with payment of $1,000 and $2,500, which his company paid. What he got in return: customized reports detailing his odds for developing various diseases.

“It made me alert to certain risks and conditions, which is what I would hope to draw from it,” Goetz says. Unlike some genetic tests that are more definitive, genetic profiling determines a “slightly increased risk—like a 10 percent, 20 percent or 30 percent elevated risk” relative to the average population, Goetz points out. “It’s not a diagnosis; it’s information about ourselves. It’s context.”

Goetz is a well-informed consumer: He has a master’s degree in public health and closely follows the health care and technology issues for his job. However, he says almost any consumer who seeks out these services pays “significant dollars” for their genomic profiles. “By the time you navigate through the website, you have a good grounding” in the subject, he believes. The companies he used were “extremely rigorous about what (consumers) can and cannot extrapolate out of (their) genome.”

When he received his results—which he says were consistent between both companies—Goetz didn’t see anything too troubling. “There were no land mines,” he says. Goetz has a higher-than-average risk of obesity, even though he is not obese and exercises frequently. So even though he’s genetically at risk, “it’s nothing I’m concerned about.” On the other hand, Goetz has an elevated risk of developing prostate cancer and his risk for developing glaucoma is three times higher than for the average American.

Although Goetz doesn’t believe the information is “inherently medical,” it does have “medical implications,” so he thought it made sense to bring it up with his doctors.

USEFUL, NOT PRICELESS

When Navigenics offered Josh Umbehr a free genetic profile last April, he jumped at the chance. As a reviewer for a medical technology blog and second-year family practice resident in Wichita, Kan., Umbehr was curious about the technology and how the results were conveyed.

The process was smooth. “You don’t eat for 15 minutes, you spit into a tube, and that was it,” Umbehr said. “It couldn’t be easier.”

A few weeks later, the company sent him an email with instructions for retrieving his profile online. In a nutshell, he didn’t find “anything particularly scary”—an elevated risk for Crohn’s Disease, heart disease and obesity.

Umbehr was impressed with the quality of the information he received. Consumers receive printouts for their physician, and he said the company “did a good job with education” and the results were “balanced.”

As for the results, Umbehr said that the information is “helpful, but not priceless” since it represents one of many factors that determine someone’s risk for disease—on the same level as family history, Umbehr says.

Still, he sees the information as useful, and he thinks it has the potential to help him “get to the diagnosis sooner” for his patients—and perhaps even for himself. “I’ll start watching for Crohn’s now.”

monitor Internet sales. Companies can hire their own doctor to order tests in the states that require it, and even if the law prohibits direct-to-consumer tests, it is difficult to prevent the sale of a product over the Internet.

What’s more, companies that offer genomic profiles are challenging whether these laws even apply, since they say they are not offering genetic testing per se. “Many of these companies argue that they aren’t even selling a health-care related product,” Michael Watson says. Rather they say they are only “selling information that should be brought to the attention of their health-care provider.”

STATES GET TOUGH

Some states are taking a firm stance. In June, California issued cease-and-desist let-
ters to 13 direct-to-consumer genetic testing companies and demanded proof that the companies are meeting state requirements, such as involving a California-licensed physician, requiring counseling, conducting tests at certified labs, and proving the tests are valid.

This follows similar actions in New York. Jeffrey Hammond, a spokesman for the state health department, says his agency recently sent letters to 26 companies it believes are selling tests directly to consumers, informing them about state regulations and warning that state law “does not provide for direct consumer access to genetic testing.” In addition to requiring an authorized provider to order genetic tests, New York certifies all laboratories doing business in the state, including those located in other states and countries. New York State’s Clinical Laboratory Evaluation Program (CLEP) certifies directors running the laboratories, issues laboratory permits and conducts proficiency testing—a higher standard than federal law.

This extra layer of scrutiny “stems from our concerns for patient safety,” Hammond says. “When it comes to online tests, it raises concerns about what patients will do with this information, and if [that information] is correct.”

CONSUMERS “BE WARY”

Critics cite a number of concerns, but the bottom line is consumers are apt to misinterpret their results, or worse, make potentially life-changing decisions based on the information.

“They’re telling consumers that they might be at an increased risk for disease,” says Robin Grubs, president of the board for the American Board of Genetic Counseling. But what “they’re giving the consumer is not proved.”

According to a 2006 consumer alert from the Federal Trade Commission, “some of these tests lack scientific validity, and others provide medical results that are meaningful only in the context of a full medical evaluation.” Moreover, the commission warns consumers to be “wary of claims about the benefits these products supposedly offer. Some companies claim that at-home genetic tests can measure the risk of developing a particular disease … but the FDA and CDC say they aren’t aware of any valid studies that prove these tests give accurate results.”

Another study by the U.S. Government
Accountability Office found companies that sell nutrigenetic tests—which analyze a consumer’s DNA and make recommendations about nutritional supplements—“may mislead consumers by promising results they cannot deliver.”

Michigan Senator Thomas George, a physician and chairman of the Senate Health Policy Committee, has been closely following the issue. As George sees it, the real issues are “interpretation and reliability—what are we testing for, and what is the value of the test?”

He worries about the impact some of these tests may have on consumers and is concerned that it may further fragment the medical system and waste money. “To market it directly to consumers as though it’s something that will help them make health-care choices may be a false promise.”

On the other hand, not all companies are the same—and even though some companies make unrealistic claims, that’s not across the board. According to Wojcicki from 23andMe, “we recognize that it is possible that people may misunderstand their genetic data if they are given incorrect or incomplete information.” As a result, she says “it is our responsibility to ensure that the information we provide is accurate and understandable.” To that end, 23andMe has developed a star system that reflects the research confidence rating—with one star for preliminary research and four stars for established research.

“We take this responsibility seriously,” Wojcicki says.

SORTING IT ALL OUT

Given the moving target that is the direct-to-consumer testing industry, what’s a policymaker to do? A federal task force on genetics in 2007 identified a handful of recommendations, such as establishing validity and clinical utility of tests, and requiring proficiency testing for labs, to name a few.

The task force also recommended coordination of public and private sector activities. Some testing companies see this as good for business. “We believe that our interests and the government’s interests are aligned. We both want to ensure these services are accurate, reliable and the health and safety of the general public are not compromised,” says 23andMe’s Wojcicki.

To many in the field, education and information are key to protecting consumers and preparing for what’s to come. Watson, of the American College of Medical Genetics, believes a more informed public will separate the wheat from the chaff. “Some of [the regulation] will be through the age-old market place route,” Watson says. “If the public doesn’t find it useful, they won’t use it.”

On the other hand, “as the tests become more validated, they will integrate into health care where follow-up will be better monitored.”

What Consumers Should Know

The American College of Medical Genetics recommends five minimum requirements for genetic testing:

✦ A knowledgeable health professional should be involved in the process of ordering and interpreting a genetic test.

✦ The consumer should be fully informed regarding what the test can and cannot say about his or her health.

✦ The scientific evidence on which a test is based should be clearly stated.

✦ The clinical testing laboratory must be accredited by CLIA, the state or other applicable accrediting agencies.

✦ Privacy concerns must be addressed.

Source: American College of Medical Genetics

CHECK OUT an interview with Anne Wojcicki, co-founder of the California-based genetic start-up 23andMe, and more information on the position of the American College of Genetics on direct-to-consumer testing at www.ncsl.org/magazine.