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## Government moves to oversee genetic tests aimed at consumers

By Judith Graham

Chicago Tribune

CHICAGO — During a stay at a resort two years ago, Carol Arand purchased \$4,000 in genetic tests after a spa physician praised their potential to improve her health.

If ever she got sick, Arand reasoned, she could take the results to her doctor. "I hoped that this genetic fingerprint could be useful," she said.

But that expectation — the promise that many consumers see in genetic testing — hasn't yet been realized. When Arand discovered she had breast cancer and brought the reports to a hospital earlier this year, an expert looked them over with dismay.

The information — vague statements about several dozen genetic variations and their possible health implications — was misleading and the accompanying health claims unsupported, said Tinamarie Bauman, manager of high-risk genetics at Alexian Brothers Hospital Network in the Chicago suburbs.

Many experts agree that health-related genetic testing isn't ready yet to be introduced to a mass audience. Yet several companies have been marketing tests that purport to assess serious concerns, such as the risk of developing diabetes, with remarkably little oversight.

That may be about to change. The Food and Drug Administration moved aggressively last week, putting five companies on notice that it believes personal genetic tests are medical devices and, as such, subject to regulation. Until now, the federal agency has been "conspicuous by its absence," said Dan Vorhaus, editor of the Genomics Law Report.

Three of the companies — 23andMe, Navigenics and deCODE Genetics — sell personal genetic tests over the Internet. Illumina Inc. sells a chip used to scan DNA; Knome Inc. provides complete scans of an individual's genome.

The FDA first signaled a possible shift in direction after another firm, Pathway Genomics, last month moved to sell genetic test kits through Walgreens drugstores. The plan was quickly shelved, but a high-profile congressional committee launched its own investigation.

The tests prompting attention are not the highly specialized DNA scans ordered by physicians. Instead, these consumer-oriented offerings evaluate an individual's risk of developing various illnesses or responding variably to different medications, based on test findings.

That is problematic because most of the genetic contribution to disease remains unknown, as does the manner in which genes interact with each other and the environment, said Darrel Waggoner, associate professor of human genetics at the University of Chicago.

"We're at an early stage in our knowledge of the human genome," said Dr. Muin Khoury, director of the National Office of Public Health Genomics at the U.S. Centers for Disease Control and Prevention. "What we have now is data points, not knowledge."

Yet consumers may make important medical decisions based on test results, such as changing drug regimens or deciding to forgo exams such as mammograms, experts suggested.

Others respond that consumers have a right to their genetic information, however sensitive, incomplete or complicated it may be.

"Generally, I think that people can handle information and that more information is better," said Dr. David Orentlicher, co-director of the Center for Law and Health at Indiana University.

Arand, a Crystal Lake, Ill., landscape designer, says she has no regrets about ordering a panel of genetic tests after attending a workshop on personalized medicine at Canyon Ranch in 2008.

Dr. Jyotsna Sahni, a physician at the Arizona resort, was the speaker. "Canyon Ranch is trying to catch you before you fall into the river of disease," she said. Information about genetic vulnerabilities can alert people to health risks and motivate them to stay healthy, she added.

The "Genovations" tests Arand took offer lifestyle advice while highlighting several dozen genetic markers allegedly linked to disease. For instance, someone with a marker linked with asthma and allergies may be told to reduce stress, get adequate sleep and exercise regularly.

But a 2008 review of a dozen personal genetics test, including Genovations, found "insufficient scientific evidence to conclude that genomic profiles are useful in measuring genetic risk for common diseases or in developing personalized diet and lifestyle recommendations for disease prevention." The paper appeared in the American Journal of Human Genetics.

Dr. Patrick Hanaway, chief medical officer of Genova Diagnostics, a North Carolina company that sells the tests, disputed that assessment, saying scientific studies exploring the value of personalized medicine are published every week. But Khoury and other experts say it's too soon to draw definitive conclusions from those findings.

At Canyon Ranch, Sahni said: "No, I don't think (the tests) are 100 percent accurate, but they give us a clue."

Arand's genetics counselor tried to be diplomatic after Arand was diagnosed with breast cancer and brought in her Genovations reports. "I told her the science isn't really fully developed yet and our genetic makeup alone is a poor predictor of future illness and disease," Bauman said.

Similar concerns have led many experts to suggest a need for tighter regulation of personal genetic tests.

Companies in this field say their primary product is information, not medical diagnoses or advice. The FDA gave no indication it disagreed with that assessment until its recent warning.

Now, the agency seems convinced that personal genetic tests deliver findings with important medical implications and, as such, should be regulated as medical devices. "Consumers may make medical decisions in reliance on this information," Alberto Gutierrez, director of the FDA's office of in vitro diagnostics, wrote in the letters delivered last week.

Previously, companies in this field escaped oversight by claiming they offered a type of laboratory analysis that receives almost no scrutiny â€” so called "laboratory developed tests." The FDA doesn't evaluate this kind of test â€” mostly used by labs to analyze tissue slices or tumors â€” though it claims the authority to do so.

While the Centers for Medicare and Medicaid Services inspects laboratory processes, it hasn't ensured that labs performing genetic tests have special proficiency. The Federal Trade Commission evaluates the legitimacy of companies' marketing claims, but to date the commission has publicly challenged only two firms selling genetic tests.

The result has been the lack of any guarantee that laboratory-developed genetic tests deliver medically valid or meaningful results. This is the gap the FDA now appears ready to address, according to Gutierrez's letters.

Asked for a response to recent developments, 23andMe said it disagreed with the FDA's most recent position but was "open to discussions" about how best to regulate the personal genetics industry. Pathway Genomics acknowledged its responsibility "to ensure public safety," and Navigenics cited a history of working closely with regulators.

Several experts said they hoped regulation would be flexible. The genetics revolution is sure to change the face of medicine, and it's important that innovation continue, they said.

A striking example of the two sides of personal genetic testing comes from Dr. Francis Collins, head of the National Institutes of Health, who sent off saliva samples to three leading companies last year under a pseudonym.

On the positive side, Collins learned he had two common genetic markers for Type II diabetes and has since dropped 20 pounds, he said at a meeting in October. But the companies' interpretations of his genetic material also differed substantially.

Arand says the limitations of current genetic tests don't worry her. She's counting on her genetic profile serving as a helpful tool for doctors in the future. In a way, she's the perfect customer for genetic testing: willing to take some risks and to be patient as the field develops.

"I know we're learning so much more about DNA every day and it's just going to get better and better," Arand said. "The tests I had may not benefit me today, but because my DNA is never going to change maybe they'll make a difference later."

#### **ABOUT GENETIC TESTING**

A few dozen companies offer personal genetic tests directly to consumers. Leaders are 23andMe, Navigenics, deCODE Genetics, and Pathway Genomics.

The companies scan an individual's DNA at 500,000 to 1 million sites, a tiny fraction of 6 billion sites on the human genome.

Each company uses different genetic markers and complex algorithms to calculate a person's genetic risk of developing disease. Some markers are better established than others, and risk estimates can vary.

Some companies, such as Genova Diagnostics, sell these tests through doctors but give the option of having results delivered directly to individuals.

#### **Questions to consider**

If you're thinking about buying a genetic test consider the following questions, compiled by the Human Genetics Commission in the United Kingdom.

#### **Ask yourself:**

â€” Why am I considering taking this test?

â€” What do I hope to do after getting the test result? What if I get an unexpected result?

â€” Should I get advice from my doctor before getting the test?

â€” Will the test results have implications for my relatives? If so, will I tell them? How? Should I discuss this with them beforehand?

#### **Ask the company or person selling the test:**

â€” Is there a proven link between the results of the test and the condition it is intended to detect? How strong is the link? Is it definite, probable or possible?

â€” How large was the population studied in order to determine this link? Has the evidence been published in a peer-reviewed academic journal?

â€” Who will analyze the sample you collect?

â€” If I don't understand what you send me, will someone be available to answer questions? What are this expert's qualifications? Is this included in the price I pay?

â€” What happens to the sample and my personal data after you've completed the test? How will you safeguard the privacy of my sample and data?

â€” Do you share or sell any information to other organizations? Do you analyze my sample for any other purpose than the genetic test I've purchased?