

1,100 genetic tests for depression to cardiovascular disease

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Gene-Test Marketing Raises Public Health fears: A simple cheek swab can tell you about your ancestry. A bit of blood can tell you how likely you are to get cancer or heart disease. You can find out if you're destined to be a high-performance athlete or a Sunday morning quarterback. Welcome to the wonderful world of direct-to-consumer genetic testing.

A simple computer search turns up dozens of companies offering DNA tests at a wide range of prices. Some are based on the latest cutting edge science, others offer legitimate if limited tests, and still others are out and out frauds. And that worries some health care professionals.

"The field is ever-evolving, it's not regulated, and it's potentially fraught with difficulty for patients," said Erin Tracy, M.D., of Massachusetts General Hospital. "My fear is that if people have tests and don't get appropriate counseling, they might make their own determinations" about treatment, Dr. Tracy said.

In a Personal Perspectives article in the December issue of the *Journal of the American College of Obstetrics and Gynecology*, Dr. Tracy argued that physicians need to be concerned about the rising tide of "unsubstantiated medical claims" on the Internet.

Even when tests are valid, Dr. Tracy said, "patients need a better understanding of the pitfalls" — the nuances of the results.

The natural source for that understanding is the physician, she said, but "we're so busy — so fully stretched at every visit" that adding genetic counseling may be difficult, if not impossible.

Dr. Tracy's article was twinned in the journal with a policy statement on the issue of direct-to-consumer genetic tests from the American Society of Human Genetics.

According to the statement, about 1,100 genetic tests are available clinically, most only through a health care provider. But increasingly, tests are being offered to the consumer "often without any health care provider involvement or counseling," the authors wrote.

The offerings range from tests for single-gene disorders, such as cystic fibrosis, to tests for predisposition to complex, multifactorial diseases, such as depression and cardiovascular disease.

The latest entrants offer whole-genome scans for dozens of markers associated with various ailments, including heart disease and diabetes.

Some companies also urge changes in diet or use of nutritional supplements on the basis of results. For instance, GeneLink, of Jersey City, N.J., offers "powerful DNA-based predictive assessments" that measure single nucleotide polymorphisms (SNPs) associated with such things as cardiovascular health or oxidative stress.

What's more, the company says, it has developed "customized formula systems" linked to the tests. "GeneLink can now provide Nutrigenetic and Dermagenetic product formulations that truly address individual needs," according to the firm's Web site.

Such claims worry Gail Javitt, J.D., of the Genetics and Public Policy Center of Johns Hopkins University and one of the authors of the human genetics society statement.

"We lack empirical data, but certainly there's potentially a big public health concern," she said. There's great potential, she says, for "consumers being misled and making bad health decisions."

Dr. Javitt said it's a "challenge" figuring out what's false and misleading and what's valid science, responsibly presented.

She ticked off several key issues:

Does a lab get the right answer? The answer is soon clear when the test is for a baby's sex, for example, but may be much less obvious with the results of others tests.

- How do the testers know that their work is clinically valid?
- How does anyone know if a test is actually useful?

Even if the tests are available only through a health care provider, patients have begun asking for them — despite a sometimes whopping cost and little reason, Dr. Tracy said.

She's had a number of patients ask for the *BRCA1* test for breast cancer linked mutations, even when there's no indication that they should have the \$3,000 test.

"If it's not indicated, I will tell them why," Dr. Tracy said. But if a woman doesn't like the answer, there's nothing to stop her from finding another doctor.

And what if the result is a false positive? Lacking context and fearing cancer, many women may simply opt to have their breasts and ovaries removed, Dr. Tracy said.

Dr. Javitt's last two questions become even more important when you consider the latest entries in the field, Iceland's DeCodeMe and California's 23andMe and Navigenics.

The three companies offer — for between \$985 and \$2,500 — a whole-genome scan of all known SNPs, as well as some thoughts on what they mean.

They are, Dr. Javitt said, direct-to-consumer gene testing "on steroids."

The companies offer some cutting-edge science and have platoons of high-powered genetic researchers on their advisory boards. DeCodeMe, for instance, is spearheaded by Kari Stefansson, M.D., whose work unraveling the genome has put him in the forefront of the science.

There's probably little question that these labs will get the right answer, but Dr. Javitt's other questions remain. Is there any evidence to show that knowing your own pattern of SNPs will be of any benefit?

David Agus, M.D., of Cedars-Sinai Medical Center in Los Angeles, the founder of Navigenics, says the answer is Yes.

If his company can tell a client he or she has an increased risk of colon cancer, Dr. Agus told a reporter, the customer is likely to get more and earlier colonoscopies. "That's going to save lives," he was quoted as saying.

Less definitive is the message from 23andMe. The company's goal, stated in an open letter to the medical community, "is to help our customers understand their own genetic information and how the current biomedical literature pertains to it."

"What we do not and will not do is provide medical advice to our customers, the company continued. "Though our service delivers personalized data, the information it provides is tailored to genotypes, not to individuals."

That said, there are clearly individual data and exactly how individuals will choose to use them is not yet clear. It may be that Dr. Agus is correct and customers will use the information to improve their lifestyles in order to lessen risks.

Equally, of course, others may use a lower genetic risk of, for instance, heart disease, as an excuse to keep smoking.

The medical community is not totally against direct-to-consumer testing, however. In fact, it has clear potential benefits, Dr. Javitt and colleagues at the human genetics society said in their policy statement, including "increased consumer awareness of and access to testing."

But they called for a number of steps, including greater regulation of labs and action against companies that make false or misleading statements, to help patients avoid some of the pitfalls of the testing revolution.

One example of a potential pitfall — albeit an unlikely one for most of us — came early in December, at the expense of Nobel laureate James Watson, Ph.D.

Dr. Watson was on the hot seat for remarks he allegedly made about the intelligence of blacks. Coincidentally, his entire genome — one of the few individual genomes yet sequenced — was available on the Internet.

DeCodeMe's Dr. Stefansson analyzed the data and found, he told the *New York Times*, that Dr. Watson's genome had about 16 times the number of genes thought to have an African genesis than would be the case for an average white European.

Put another way, Dr. Watson would have much the same genetic profile if one of his great-grandparents had been African.

Dr. Stefansson said his company had not developed the original genetic data and he did not want to place too much weight on the analysis. But, he told the *Times*, "on my face, [the result] would elicit smiles."

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