

Do consumers who order gene testing discuss results with their doctors? Not likely, study finds

By Deborah Kotz September 26, 2013

If you could pay \$100 to gaze into a crystal ball and determine any diseases that may be lurking in your future, would you do it? More than 400,000 people have already done this with [23 and Me](#) and other direct-to-consumer gene testing kits, but few actually discuss the results with their doctors or genetic counselors, according to new research from Brigham and Women's Hospital.

They do, though, share freely those DNA scan results with their family and friends.

"This is preliminary data but it suggests that 70 percent of people discuss their results but only 5 to 10 percent said they'd talk to a genetics counselor about them," said study leader, Dr. Robert Green, a medical

geneticist at Brigham and Women's Hospital.

The results, presented at the Consumer Genetics Conference in Boston, were based on a survey of more than 1700 people who ordered 23 and Me and other direct-to-consumer gene tests; it was funded by the National Institutes of Health. Green and the other researchers have no financial interest in any testing companies.

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Another surprising find from the survey: A significant number of people who send their DNA samples away have already been diagnosed with diseases like diabetes or colon cancer. "It's human nature to want to gain an understanding into why you have a disease," Green said. "But it's somewhat strange since these tests are all about predicting your risk of developing certain diseases."

The tests work by detecting markers rather than

specific gene mutations themselves; more expensive genetic testing ordered by doctors can identify mutations for certain hereditary diseases like Huntington's, early Alzheimer's, and hereditary forms of breast and colon cancers. It's tough to know how much these markers really can assess risk, Green said, since rigorous studies haven't been done to validate them.

Some new add-on's to the consumer tests include identifying markers to determine how the body responds to certain medications and a host of genetic traits that increase the likelihood of everything from lactose intolerance to male pattern baldness.

That's a lot of information for a person to digest without a doctor's help. It could also be why another company, called [Pathway Genomics](#), requires a consumer's physician to order the test.

The US Food and Drug Administration has taken a cautious approach to these tests, largely frowning on their use, but not going as far as to ban them from the market.

"Marketing genetic tests directly to consumers can increase the risk of a test because a patient may make a decision that adversely affects their health, such as stopping or changing the dose of a medication or continuing an unhealthy lifestyle, without the intervention of a learned intermediary," FDA device

director Dr. Jeffrey Shuren said in [2010 testimony](#) before a Congressional committee.

If the survey results are telling, the FDA does have reasons for its concern; only 10 percent of the respondents said they had consulted doctors after receiving their tests results and were planning on following those up with further testing.

That means they could be making changes on their own — or opting not to lose the extra weight if it turns out they're not at added risk for, say, heart disease.

Green pointed out that on the positive side, some people may be inspired to eat more nutritiously or start walking more if they learn they have, say, an increased diabetes risk. On the other hand, the test only takes into account genetic markers, so a "300-pound person eating jelly doughnuts may be told he has the same diabetes risk as a marathon runner even though they clearly don't based on their lifestyle," he added.

Shuren also pointed out that "none of the genetic tests now offered directly to consumers has undergone premarket review by FDA to ensure that the test results being provided to patients are accurate, reliable, and clinically meaningful."

That's a message consumers may not hear if they're just perusing the websites of these companies before plunking down their money to get tested.