Coping with Bad Genetic News

New research suggests that most people can cope with learning that they are at high genetic risk for disease.

By Emily Singer

As direct-to-consumer genetic testing spreads, a major concern expressed by ethicists and physicians has been whether the average person will be able to understand the results of these somewhat subtle tests. Rather than giving an answer in black and white, the tests predict whether someone has an elevated risk for developing common diseases, such as Alzheimer's. Even if consumers do understand the results, it has been unknown how they might react to news that they have a sequence of DNA that raises their risk of developing a disease.

Two new studies suggest that most patients cope easily with such negative genetic information. People who learn that they carry a high-risk genetic variant for Alzheimer's disease, called APOE4, have no greater anxiety over their long-term prospects than do those who don't know their risk, according to research published today in the New England Journal of Medicine. Another recent study of smokers revealed that those who found out that they had a lower genetic risk for developing lung cancer were just as interested in stopping smoking as those determined to be at higher cancer risk.

"The findings may help us to subdue paternalistic concerns that we have to protect people from this information," says Colleen McBride, chief of the Social and Behavioral Research Branch at the National Human Genome Research Institute, in Bethesda, MD, and senior author on the smoking study. "People given the option to take these tests can protect themselves, and they find it useful to know the results, even if the test hasn't been proven to make a difference in what they do."

In the past few years, a number of companies have sprung up to offer genetic testing directly to consumers. "Studies like this are important because we are clearly going to see testing like this make its way routinely into mainstream medicine," says Michael Christman, president of the Coriell Institute for Medical Research. Because the results of this type of testing are much more complex than the genetic tests currently used...
most commonly in medicine--largely single-gene testing for rare, severe disorders, such as cystic fibrosis--physicians worry about how people will react. Some have speculated that someone at high risk for neurological disease might give up on long-term relationships, or someone at low genetic risk for type 2 diabetes might indulge in a diet of doughnuts and cheeseburgers.

To date, most sociological studies of genetic testing have focused on rare inherited diseases rather than on more common ones, such as Alzheimer's. Robert Green and his colleagues at Boston University are among just a handful of researchers examining this issue: Green's team has spent the past decade studying the impact of genetic testing for APOE4, which raises the risk of developing Alzheimer's disease threefold in those who inherit one copy and tenfold in those who have two copies. No proven treatments exist to reduce Alzheimer's risk in APOE4 carriers, and testing for the risk variant is not currently recommended. But surveys indicate that 15 percent of primary-care physicians who treat patients with Alzheimer's have already been asked about the test.

In the newly published study, Green and his colleagues offered APOE4 testing to adult children of people with Alzheimer's disease and then revealed the results to half of the group. The team found that people clearly understood their results, and that six weeks after learning them, those who were told that they had the high-risk variant seemed more stressed than the other participants. But that spike in anxiety had faded by the time participants were tested again both six months and one year afterward.

"We were astounded by how many people wanted to know: more than 20 percent wanted to receive it," says Green. "Even though patients clearly understood there was nothing they could do to stave off the disease, they had nonmedical reasons to learn about it: to prepare their children, to think about the longevity of careers."

For example, "people do in fact change insurance purchasing behavior based on this information," says Green. "We should be cautious as medical professionals not to dismiss those personal reasons, as long as we can convince ourselves it's not harmful to offer this information."

Green and his collaborators have also found that people who know they have the high-risk gene are more likely to take vitamins. "That's fine, except that some types of supplements are highly unregulated and can be harmful," he says. "You can easily imagine people trying to link results of genetic tests to the purchase of unproven vitamins that could at best take their money and distract them, and at worst could be harmful."
Researchers caution that results from the study are not necessarily indicative of the general population. For example, Green's team weeded out people who scored high on measures of anxiety and depression at the start of the study. And the study does not examine all of the potential drawbacks of testing. In an editorial accompanying the paper, Rosalie Kane, a public-health specialist, and Robert Kane, a physician, both from the University of Minnesota, in Minneapolis, say that people who test positive for high-risk genetic variants might be denied some types of insurance. The Genetic Non-Discrimination Act, passed last year, prohibits such discrimination in employment and health insurance, but not in life, disability, or long-term care insurance.

One of the other major concerns for the new generation of genetic testing is how best to deliver the results. In Green's APOE4 study, participants learned of their risk through genetic counselors--but this may not always be possible as genetic testing becomes more widespread. "I would be interested going forward to see how people who received this information without counseling deal with it," says Christman. "Some of the direct-to-consumer companies are doing this right now."

In the lung-cancer study, McBride and her collaborators offered smokers who had a family member with lung cancer genetic screening for a variant associated with a higher risk of developing lung cancer. Information about the risks and benefits of the test, provided to help people decide whether to take it, as well as the results were delivered online.

The researchers found that all of the people in the study who tested high risk understood the meaning of the results, while only about 60 percent of those who scored low risk understood them. "That kind of defies expectation," says McBride. "Psychological theories predict that people protect themselves from threatening information, and one way to do that is by not understanding it."

The researchers found no difference between the high- and low-risk participants' interest in getting additional tools to quit smoking. "Telling someone they are low risk doesn't undermine their motivation to seek out cessation materials, and being told you are high risk didn't increase motivation," McBride says. "All smokers were motivated enough to log on and consider testing and availed themselves of cessation materials."

McBride says that she doesn't think genetic testing itself will motivate people to quit smoking or lose weight or make whatever changes might help their health. Instead, she says that the tests' utility may be to motivate people to take initial steps--"to get someone engaged in a smoking-cessation program or dietary-change intervention."
McBride is now studying the impact of genetic tests that analyze many spots on the genome and assess risk for multiple diseases, such as those offered by a number of online gene-testing companies. "There the story is much more complicated," she says. "The results might conflict with each other, and people might be at risk for many conditions."

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**Upcoming Events**

**Lab to Market Workshop**
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**EmTech 09**
Cambridge, MA
Tuesday, September 22, 2009 - Thursday, September 24, 2009
http://www.technologyreview.com/emtech

**Nanotech Europe 2009**
Berlin, Germany
Monday, September 28, 2009 - Wednesday, September 30, 2009
http://www.nanotech.net

**2009 Medical Innovation Summit**
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Monday, October 05, 2009 - Wednesday, October 07, 2009
http://www.ClevelandClinic.org/innovations/summit

**Optimizing Innovation 2009**
New York, NY
Wednesday, October 21, 2009 - Thursday, October 22, 2009