Medical Xpress

Testing for Alzheimer's genes forces difficult decisions

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Last year, Barbara's husband thought it would be fun to learn more about their family's history, so he bought 23andMe gene-testing kits for himself, Barbara and their three daughters for Christmas.

Once she understood it, she realized the information in that report was devastating. She has a much higher risk than most people of developing Alzheimer's disease, a form of dementia with no cure and no good treatment. That's bad news for the whole family. Her husband, who does not have the mutation, may have to take care of her. Her daughters will have lower risk than she does because of her husband's genes, but they have inherited one copy of a gene that raises their risk too.

While she is now "at peace" with the results, Barbara says she probably would not look at them if she had the choice today. "I wish I had known the can of worms I was opening," she said. The A 67-year-old resident of a Philadelphia suburb spoke on the condition of anonymity because of the "stigma" of Alzheimer's. "I'm just not ready," she said.

23andMe launched its "personal genome service," which included health-risk data, in 2007. It stopped revealing health risks under orders from the U.S. Food and Drug Administration in 2013, because it had not received authorization.

For several years, customers got ancestry information and reports about relatively benign genetic traits such as whether they have dry or wet earwax. The company returned to telling people whether they were

carriers for diseases like sickle cell anemia in 2015. This year, the FDA allowed 23andMe, which has more than 3 million customers, to release information about genes that increase their risk for 10 diseases, including two particularly scary ones: Alzheimer's and Parkinson's disease.

New customers can choose whether they want only the ancestry reports or are willing to pay more ($199) for ancestry and health-risk reports. Customers who bought the service in previous years can now choose to see reports that weren't part of the package when they signed up.

Even new customers who purchase the health reports may be blindsided by unexpected information, experts said. They might, for example, be worried about their family history of the eye disease macular degeneration—there's a report on that too—only to find out they're at high risk for something more serious that wasn't on their radar. And particularly savvy consumers can download their raw data and plug it into websites that will tell them more than 23andMe does, another potential source of worry.

On the plus side, the information has the potential to encourage participation in clinical trials that scientists hope will lead to discoveries that will prevent or treat Alzheimer's and other diseases. More than 85 percent of 23andMe customers say they would like to be part of research. The company makes its anonymized data available to academic and drug-company investigators, often for a fee, and it sometimes notifies customers about trials they can join, including two that are seeking people with the gene variant Barbara has, APOE4. The news also gives people time to make lifestyle changes that may lower their risk. And, many people really want this information.

Joyce Tung, vice president of research for 23andMe, said the company's information not only satisfies customers' curiosity, but also has the

potential to help "prevent, understand and treat disease."

But many physicians think it would be better for people to get bad genetic news from experts who can answer their questions immediately. Some are concerned that ill-prepared customers may wish that they had left Pandora's box alone. Every time Barbara forgets why she walked into a room—a common experience for people her age—she wonders: "Oh my gosh, is this it? Is it Alzheimer's?"

Consumers may also have to consider new ethical dilemmas. The Genetic Information Nondiscrimination Act of 2008 protects people with high-risk genes from discrimination on the job or when buying health insurance, but it does not apply to life, disability or long-term care insurance. Companies like 23andMe say their information is secure and well-protected from hackers, but customers may face a quandary if an insurer asks about risk.

Everyone has two copies of the APOE (Apolipoprotein E) gene, which is involved in the transport of fats. It comes in three varieties. APOE2, the rarest, protects against Alzheimer's. Most people have APOE3, which does not affect Alzheimer's risk. But APOE4 is the most common genetic factor associated with late-onset Alzheimer's disease. Not everyone who has it gets Alzheimer's and not everyone who has Alzheimer's has this gene. However, 23andMe tells customers that 40 to 60 percent of people with Alzheimer's have one or two copies of APOE4. Doctors say that patients with two copies tend to get the disease earlier than others and that their lifetime risk is much higher. About a quarter of the population has at least one copy of the APOE4 type, with 2 to 3 percent having two.

According to 23andMe's disclosure information, a woman's chance of having Alzheimer's at age 85 is 6 to 10 percent if she has no APOE4 variant, 27 to 30 percent if she has one and 60 percent if she has two.

The risk is lower for men: 5 to 8 percent with normal genes, 20 to 23 percent with one copy of APOE4 and 51 percent with two copies.

Three Philadelphia-area doctors who specialize in treating dementia—Jason Karlawish, co-director of the Penn Memory Center; Carol Lippa, director of the Cognitive Disorders and Comprehensive Alzheimer's Disease Center at Jefferson University Hospitals, and David Weisman, a neurologist who does research at Abington Neurological Associates—said small numbers of patients have come to them because they were concerned and confused about 23andMe test results. The company asks customers to read information about Alzheimer's genes before they click on their results, but the doctors said some patients still misunderstand whether the results mean they have Alzheimer's or might get it.

"People think of it as the Alzheimer's test, and I think it's misleading if they don't read the small print," Lippa said. One of Weisman's patients, who did not have Alzheimer's but tested positive for APOE4, was already taking Aricept, a dementia drug, when she came to him. (A doctor who specializes in Parkinson's patients at Penn said she has not seen patients who learned they have the much rarer Parkinson's-risk genes.)

Weisman, who knows of two patients who killed themselves after getting an Alzheimer's diagnosis, was so concerned about 23andMe's decision to release information about APOE4 that he wrote to the FDA to urge it to change its "terrible decision."

"This is a travesty," he said in an interview. "The FDA has allowed this private company to release information and potentially drop bombshells into a family with really no control ... or professionalism."

Jessica Langbaum, principal scientist for the Banner Alzheimer's

Institute in Phoenix, said researchers are still in the early stages of understanding the psychological effect of learning that one has APOE4 genes. She said there's less room for misunderstanding when patients get the bad news in person. "I still believe that learning your APOE test results should be done in conversation with a medical health provider, like a genetic counselor," she said.

That's what happens with the Generation Program, two studies Banner is overseeing that are recruiting people with APOE4 genes for clinical trials.

Langbaum had hoped that the new 23andMe testing would at least lead a lot more people to join the trials. That hasn't happened.

Robert Green, a geneticist at Harvard Medical School, and J. Scott Roberts, director of the Genomics, Health and Society Program at the University of Michigan School of Public Health, have been studying the effect of learning about Alzheimer's risk for several years. They analyzed a variety of ways of delivering the news in person. Mostly, they said, people have handled bad news better than many doctors feared. Distress has been brief.

Roberts and Green surveyed 1,648 23andMe customers about genetic results they got before 2013, a group that received both good and bad news. Two percent regretted getting the report and 1 percent said they were harmed A red flag, Roberts said, was that 38 percent hadn't considered the possibility that they could receive "unwanted" news. Thirteen percent of the group chose not to look at APOE results.

Later the researchers analyzed responses from just the people who learned they had one copy of APOE4 (23 percent) or two (2 percent). They were only slightly more distressed than the group as a whole.

Green said increasingly easy access to genetic information will force people to grapple with what they want to know. "An awareness of what you might find and whether you want it or not has got to creep out into all of society," he said.

There are, of course, people who want the whole truth. A 77-year-old Philadelphia man, who spoke on the condition of anonymity, found out through 23andMe that he has two APOE4 genes and went for testing. So far, his results are good. His mother died of Alzheimer's and his older sister is in the end stages now. He didn't care about his ancestry. He took the 23andMe test because he wanted to know about Alzheimer's. "To me, knowledge is power," he said. "If somebody tells me that, in a year, my mind's going to be wiped out, at least I can have a pretty good year."

Julie Gregory signed up for 23andMe in its early days hoping for information about an immunological syndrome but was instead "devastated" to learn that she is APOE4 positive. She is now grateful for the information and is trying to prevent dementia with a healthy lifestyle and anti-inflammatory diet. She started ApoE4.info, an online support group for people who have the APOE4 variant. It has 2,000 members, most of whom are 23andMe customers, she said, and is growing "by leaps and bounds" this year.

The group has a Thinking about Testing page. "We wanted people to be aware this is a big decision," Gregory said. "Don't just blindly click buttons like I did."

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