23andMe's test for a breast-cancer gene can reveal life-changing information, but can also lead down an expensive and complicated path

- 23andMe received approval from the Food and Drug Administration earlier this year to test for three BRCA mutations with its at-home ancestry tests earlier this year.
- 23andMe screens for only three BRCA mutations that are most common among people of Ashkenazi Jewish descent.
- Color Genomics is offering confirmation testing to people who have received a positive result using 23andMe.

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A reporter examines a 23andMe DNA genetic testing kit in Oakland, California.

Lara Diamond took 23andMe's genetic test to complete her family tree.

Then she found out she had a genetic mutation that could increase her risk for breast cancer.

The test flagged a faulty BRCA2 gene: When functioning properly, it helps create a protein that acts as a tumor suppressor. When it's not, a person has a greater risk of developing some types of cancer, including breast cancer in men and women and ovarian cancer in women.

Diamond identified her mutation five years ago, shortly before the Food and Drug Administration (FDA) instructed 23andMe to drop BRCA testing from its ancestry tests. Earlier this year, the agency cleared 23andMe to again offer this service, meaning more women may find themselves in Diamond's position.

The FDA's decision has ignited controversy over whether direct-to-consumer genetic testing that screens for just three cancer-causing mutations (out of thousands) is empowering or reckless. Either way, the option exists — but now consumers and health-care companies will need to address what happens next.

"I think this is the world we live in," said Dr. Robert Green, a professor at Harvard Medical School and a geneticist at Brigham and Women's Hospital. "It's becoming a much more genomically integrated world, and we can't hold that back. We have to create systems that manage it responsibly."

The cost of confirming results

About 12 percent of women will develop breast cancer at some point in their lifetime.

One study estimates about 70 percent of women who inherit a BRCA mutation will develop breast cancer by the the time they're 80.

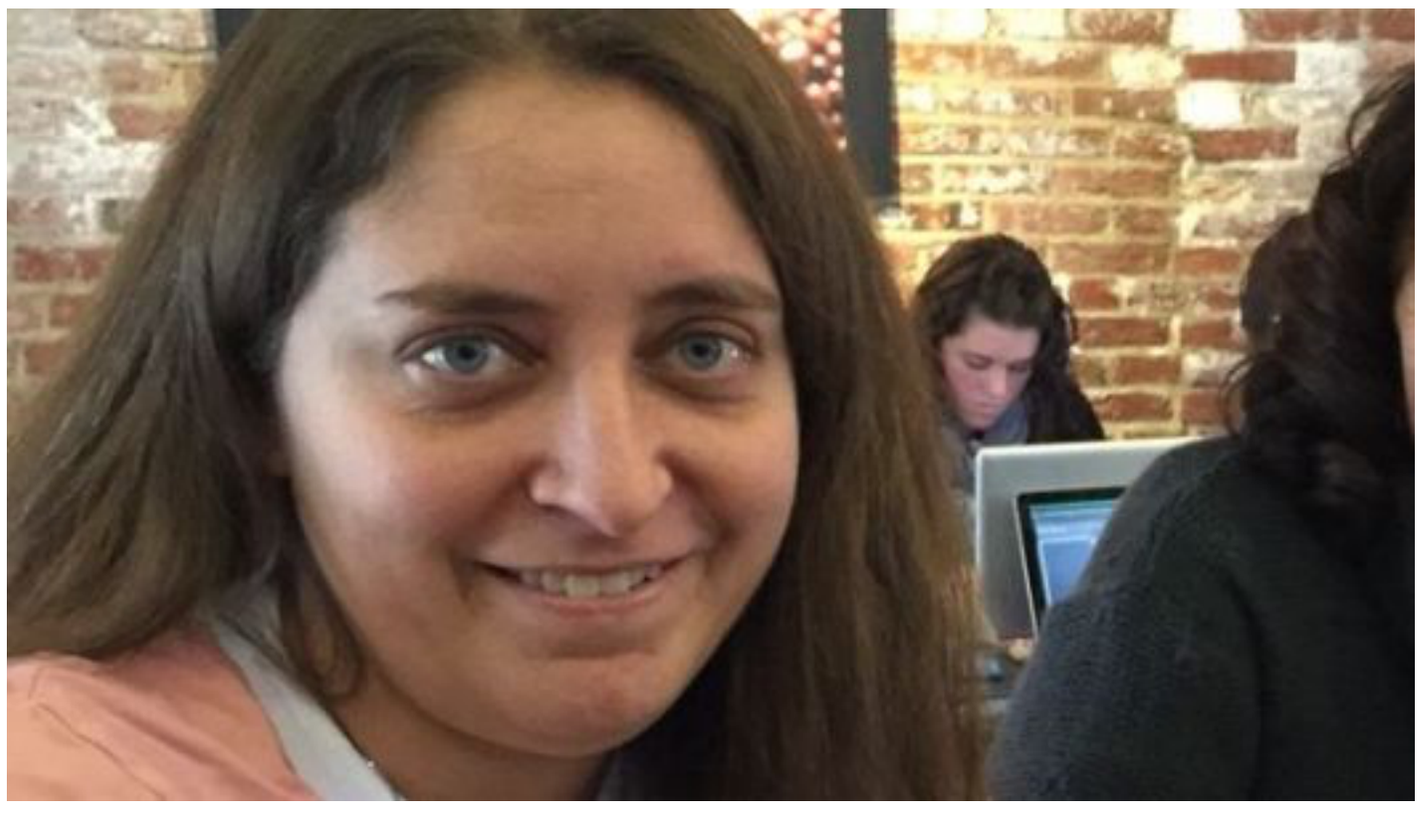
The diagnosis has become less deadly over the past few decades as treatment has improved and doctors are catching it earlier. Still, about 40,920 women in the U.S. are expected to die from breast cancer this year.

23andMe charges $199 for its ancestry and health kit, which includes the BRCA1 and BRCA2 testing. However, both the company and the FDA caution that a positive result should not be used to make medical decisions.

That's because 23andMe, as a direct-to-consumer company, does not have a physician involved in the process, said Stacy Detweiler, a genetic counselor and medical affairs associate at 23andMe. She stressed the company's tests are more than 99 percent accurate, but the tests aren't performed in a clinical setting.

People's next steps should include following up with a health-care provider or reaching out to a genetic counselor to pursue confirmatory testing, she added.

'Very fortunate'



Source: Lara Diamond

Lara Diamond, 42, took 23andMe's ancestry kit in 2013 and learned she had a BRCA2 mutation.

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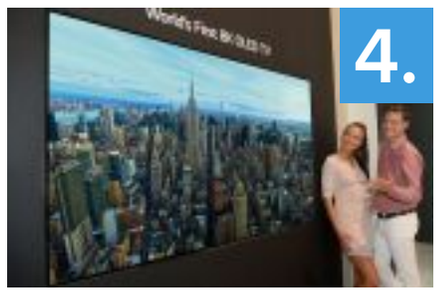

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Genetic testing can cost as little as $100. But can also go up to more than $2,000, depending on the nature and complexity of the test, according to the National Human Genome Research Institute.

Meanwhile, insurance typically won't cover the cost if someone simply wants it done. Plans usually require a doctor's note, recommendation from a genetic counselor, or a detailed family history.

For Dia d, covering the costs herself paid off. She tested positive again, a continued with screening. An MRI revealed she had Stage 1 breas cer: Thanks to the initial 23andMe test, she caught it early enough to treat with surgery.

Diamond contacted all the genetic counselors she could find in the Baltimo ea to get tested again so she could confirm her positive result, but most couldn't see her for three or four months. She managed to snag an appointment within two weeks, because she called every day until she found an office that had a cancellation.

Her insurance wouldn't cover the test, because none of her immediate relatives had breast or ovarian cancer, as typically required. She only needed one particular location within the gene tested, so it was less expensive than sequencing the entire gene.

It still cost Diamond $450, plus a $100 co-pay for the visit.

"I was very fortunate. If it was just a little big bigger, I would've needed chemo," said Diamond, now 42. "23andMe saved me from chemo."

Unfortunately, not everyone can afford to pay the out-of-pocket costs. Color Genomics spotted an opportunity. The company sends people kits to swab their saliva and sends the sample to Color's certified and accredited lab where it's tested for genetic mutations.

Color's confirmation testing looks at 30 genes, including BRCA1 and BRCA2, that are linked to eight hereditary cancers. The company normally offers the service for $250.

It partnered with the BRCA Foundation in 2016 to offer the tests to parents, siblings and adult children of people with mutations for $50. Earlier in June, Color expanded the program to people who tested positive for a BRCA mutation in 23andMe's test.

Color co-founder and CEO Othman Laraki penned a blog post titled, "Supporting patients left in limbo by (direct-to-consumer) BRCA genetic testing" to announce the move. In an interview, he said it's not a shot at 23andMe, but a means to help people take the next steps in an easier and more affordable way.

2019

"It's about reducing the cost and friction for the people that have the highest likelihood of mutation where if they find out about it, they have a dramatic increased likelihood of being able to detect early cancer," Laraki said.

23andMe declined to comment on Color's program. A spokesman said 23andMe has a handful of examples where people have come to the company for either its health or ancestry services, not expecting a result that can be life changing.

Explaining the results

Color helps connect users with genetic counselors who can explain the results. Diamond, who tested positive for a BRCA2 mutation in 2013, took Color's test once it became available. She had already had results confirmed and but wanted to test the service.

She thinks Color offering a reduced price for people who have tested positive for a BRCA mutation using 23andMe's test would be "wonderful," because it's so inexpensive compared to what she paid.

Looking back, Diamond said 23andMe's BRCA screening worked for her because it helped identify a genetic mutation she would have likely not have found until she learned she had developed breast cancer. She doesn't recommend it for everyone, though.

23andMe screens for only three BRCA mutations that are most common among people of Ashkenazi Jewish descent. Testing negative doesn't mean someone is not at any risk for breast, ovarian or any other type of cancer.

"I think 23andMe for genealogy purposes is great, but it's more for people who don't have the history but who had a mutation that was caught by accident. It's extra powerful," Diamond said.

Sue Friedman, founder and executive director of FORCE, or Facing Our Risk Of Cancer Empowered, said direct-to-consumer genetic testing is a complicated topic. It can help people identify risk they might not have otherwise have found, but it can also leave some people confused.

Her group, which advocates for topics around heredity breast and ovarian cancer, recommends people see a genetic counselor before and after testing. However, wait times can be months long and it can be hard to find an office nearby.

Telemedicine, or virtual visits, is emerging as one way to overcome these challenges. Harvard's Green chairs the scientific advisory board for a company called Genome Medical that offers these services.

No matter how people see a genetic counselor, experts agree it's crucial to see one — regardless of the result.

"The key point is that this is powerful information," said Allison Kurian, associate professor at Stanford University School of Medicine and director of the Stanford Women's Clinical Cancer Genetics Program. "When people get a test result that's positive, they should go to a genetic counselor. That's key to making sure the result is interpreted correctly and getting good advice."

Kurian added: "When people test negative, this may not be the last word. It may be, but they should consult with a genetic counselor or another clinician with similar expertise in genetics, so they can determine whether the right testing has been done."

Clarification: *Color Genomics sends people kits to swab their saliva, then sends the sample to its own certified and accredited lab, where it's tested for genetic mutations.*

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