Breast cancer researcher warns against online genetic tests

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Paul Thiruchelvam, breast cancer researcher and surgeon, says online tests may be misleading. Credit: Imperial College London

We have never been so fascinated by the secrets inside our cells.

Tests that analyse our genetic make-up—the DNA packaged inside every cell of the human body – are more popular than ever.

Requiring just a swab of saliva, some kits offer a peek into the past, and tell you where your ancestors hailed from. Others provide information closer to home, and reveal whether you carry faulty genes that increase the risk of disease.

DNA analysis firm 23andMe recently gained approval from the U.S Food and Drug Administration to launch a kit that tests for gene mutations that increase the risk of breast and ovarian cancer.

The company hailed the decision as a "milestone in consumer health empowerment".

So do these online tests mark a leap in cancer prevention? Not according to Paul Thiruchelvam, a breast cancer researcher and surgeon from Imperial's Department of Surgery and Cancer. He wrote recently in *The Lancet* medical journal that these tests could be misleading—and potentially harmful. Here he explains why.

What are your concerns about these tests?

Our biggest concern with this direct to consumer test is the potential for false reassurance.

Up to one in five breast cancers are thought to be due to faulty genes.

In many cases, these faults are in two genes—BRCA1 and BRCA2.

Faulty copies of these BRCA genes results in a 40 to 70 per cent lifetime risk of breast cancer, compared to a 12 per cent risk in the general population.

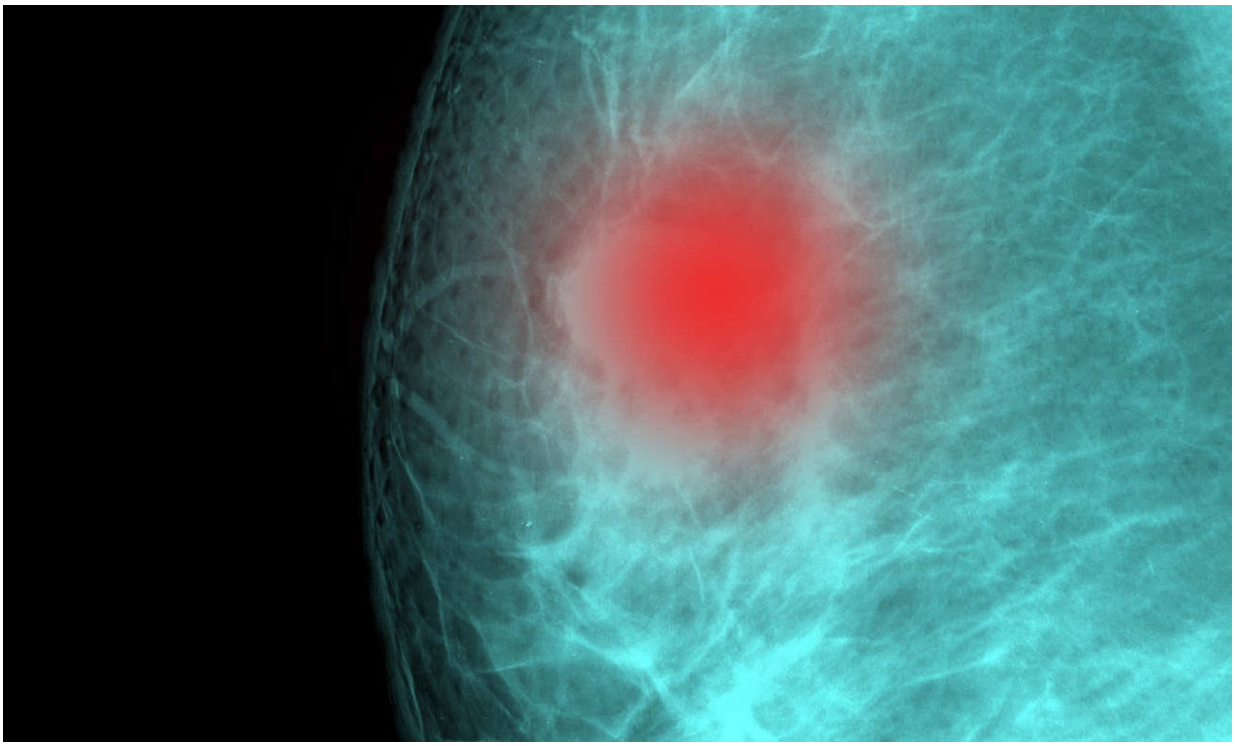
Moreover, the risk of ovarian cancer risk is 20 to 55 per cent with a faulty BRCA gene, compared to 1 to 2 per cent in the population.

Whilst 23andme can detect a BRCA1/2 mutation with high accuracy and precision, a major caveat is that they only test for three possible faults or mutations out of more than a thousand mutations in the BRCA genes that raise cancer risk.

Furthermore, the three mutations it looks for are most commonly found in the Ashkenazi Jewish population, but rarer in other populations.

A negative result means that the individual will not have these three BRCA1/2 mutations, but they may have others.

Additionally, there many other types of faulty genes—aside from BRCA1 and 2—that can increase breast cancer risk such as ATM, CHEK2, PALB2, PTEN and TP53, yet the 23andMe kit doesn't test for these.



Up to one in five breast cancers are thought to be linked to faulty genes. Credit: Imperial College London

So these tests don't provide a complete picture?

Exactly. Imagine the scenario: someone's relative has recently been diagnosed with breast cancer, and they decide to take the online test. The results tell them they don't carry the faulty BRCA gene, and they may be false reassured by the negative result.

Armed with these test results, this individual may not be as vigilant at looking for breast cancer symptoms as they should be. They may have one of the many other faulty cancer genes the online kit doesn't test for and they simply would not know.

However, if they were to avoid this test and go straight to their GP, they could be referred to a specialist. Their doctor may be able to arrange regular checks if required—called surveillance—to monitor for any signs of cancer.

Is genetic testing available on the NHS?

Yes. If a person is worried about their cancer risk a trained healthcare professional will be able to take a thorough family history, and then, if appropriate, refer them for genetic testing. These tests, unlike the online test, test for many mutations in the genes linked to breast cancer.

If a patient is found to have a faulty gene—what next?

This is another concern. In our clinic, when a patient is found to carry a faulty gene, they will see a genetic specialist who will counsel them of their cancer risk before testing and advise them of their options in the event of a positive result.

23andMe customers are signposted on the website to a genetic counsellor but an individual receiving these results online will not have immediate support or guidance.

This is what we're starting to find in clinic—my colleagues are seeing patients who have taken the online tests, are having difficulty interpreting their results, and they don't know whether the implications are positive or negative.

What's your view on these tests?

There is no doubt the online genetic test market will continue to grow as

the price of testing reduces. Genetic tests have transformed cancer treatment—and will continue to improve patients' lives.

There is no doubt that other models of genetic testing need to be explored to overcome the shortcomings of current models of testing.

Many experts believe all breast cancer patients should be tested—regardless of family history. This is because studies have demonstrated that only half of BRCA-positive cancer patients have a strong family history.

At the moment, if an individual doesn't achieve a threshold for testing based on a history of family members with breast or ovarian cancer they may not receive genetic testing. This means a potential BRCA mutation would be missed.

Other cancer specialists have gone one step further and predict whole population testing for faulty cancer genes in as little as ten years.

Whilst online testing enables democratisation of health information and empowers people to take control of their lives and potential risks, it also means that data is being given to individuals who might not be adequately prepared to hear it.

Although people may think they are doing themselves a service by taking these tests, they are actually doing themselves a great disservice.

"Pervasive genetic testing" by Paul Thiruchelvam, Carla Fisher, Daniel Leff and Susan Domchek is published in *The Lancet*

More information: Paul T R Thiruchelvam et al. Pervasive genetic testing, *The Lancet* (2018). DOI: 10.1016/S0140-6736(18)30997-8

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