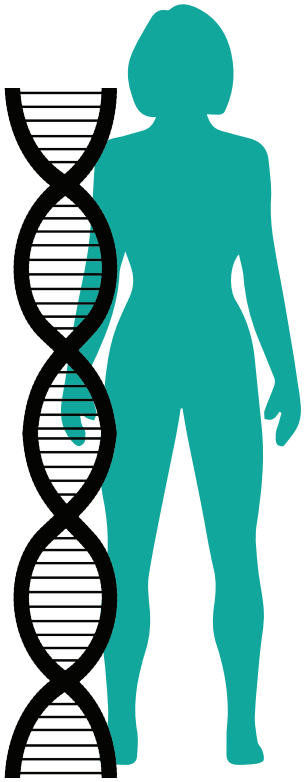
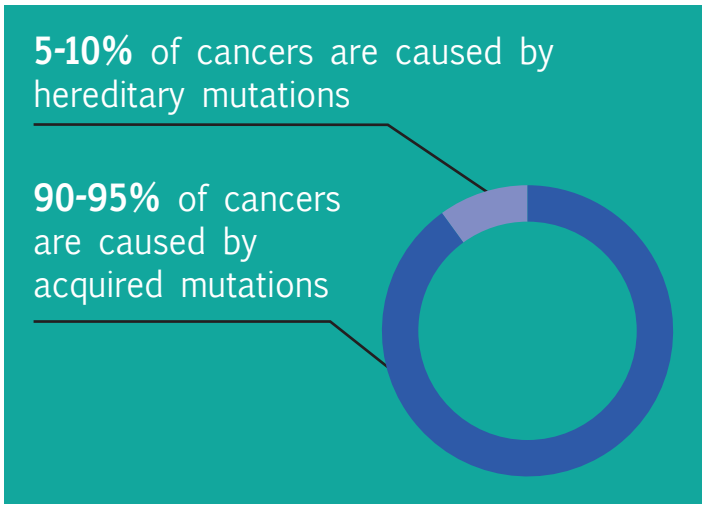
Genetic Cancer Testing

Helpful for some, but not everyone

Think back to the last time you found yourself in a waiting room before an appointment filling out forms about your family's history of disease. Do you ever wonder what exactly your family history means for your health? Today, you have new opportunities to take an active role in understanding your own genetic makeup and disease risk. This is especially true in cancer; mutations on over 50 different genes have been shown to increase a person's risk for developing cancer, sometimes by a significan margin. Genetic testing can show whether one of these known mutations is present, but there are still unanswered questions. Research is underway to determine who is most likely to benefit from gentic testing and how to manage cancer risk in patients who are found to have a harmful genetic mutation.

Until the scientific community learns more, patients are faced with a difficult decision. Genetic testing appears to offer the promise of more informed decision-making. It is natural to wonder if you could benefit from learning more about how your genetic makeup relates to your cancer risk. But the reality is much more complex than it appears at first glance. To understand this, first, we need to understand the fundamental concepts involved in cancer genetics.





Cancer Genetics

DNA is the genetic code that controls all of our characteristics, such as how we look and how our bodies work. A gene is one segment of DNA that performs a specific function. A mutation is a change in the structure of a gene.

Cancer is a disease caused by mutations in our genes. Mutations can be acquired or hereditary. Acquired mutations occur within our lifetimes. These mutations affect only certain cells and cannot be passed from one generation to the next. For example, exposure to tobacco smoke or radiation can cause acquired mutations. Getting older causes acquired mutations too, which is why most cancers occur in those 60 or older.

Hereditary mutations, on the other hand, are inherited from our parents. Hereditary mutations are present in almost every cell in the

body and can be passed down to the next generation.

Harmful hereditary mutations are rare, though. Only around 5-10% of cancers are caused by hereditary mutations, while the rest are caused by acquired mutations.

Benefits and Ambiguity

Some individuals have personal or family histories that suggest a cancer-causing hereditary mutation may be present. If you have multiple first-degree relatives (a parent, sibling or child) who have had cancer; if those family member had rare cancers, more than one type of cancer, or were diagnosed at an unusually young age; or if multiple relatives were diagnosed with the same type of cancer, you are more likely to carry a cancer-causing mutation.

Patients in these situations who do seek genetic testing and learn of a hereditary mutation are sometimes able to take steps to reduce their risk for developing cancer. For example, risk-reducing drugs or surgeries are available for some types of cancer. Increased screening tests (such as a mammogram or colonoscopy) may be appropriate for individuals with certain hereditary mutations.

However, not all results of genetic tests provide clear paths forward. We are learning more about genetics everyday, but there is much that we do not yet understand. Some patients who have genetic testing learn that they posses a harmful genetic mutation for which there are no risk-reducing strategies. Also, many patients seek genetic testing to learn more about their cancer risk and discover that they have a "variant of unknown significance." This means that a mutation is present, but researchers don't know whether it increases cancer risk or not. Ambiguous results do not assist patients and their doctors with making health care decisions.

It's also important to remember that a negative result does not mean a person will not develop cancer. Harmful hereditary mutations

Rafe Swan | Getty Images

are rare, but more than one-third of individuals in the U.S. will be diagnosed with cancer in their lifetime. This is because most cancers are caused by acquired mutations.

The risks and benefits of genetic testing look different for each individual depending on their personal and family health histories. Experts do not recommend genetic testing as a routine screening for everyone. Even for those with a strong family history of cancer, physicians and professional groups agree that patients should receive genetic counseling before and after testing. A genetic counselor can help patients understand the potential outcomes of genetic test results, the advantages of having testing done, and possible difficult outcomes of testing. These conversations help patients make an informed decision about whether they want to have genetic testing and prepare them for all possible results.

Testing at Home

In 2018, 23andMe became the first company authorized by the Food and Drug Administration to offer direct-to-consumer testing for cancer risk. Unlike most genetic testing, this test does not require physician authorization. It might sound like progress to have easy access to genetic testing at your local drug store, but as with other aspects of genetic testing, the reality is not so simple.

23AndMe's approved test is very specific. It tests for three out of the 1,000+ BRCA1 and BRCA2 mutations that are know to increase cancer risk. These three are most likely to be found in individuals of Ashkenazi Jewish descent, so the test could benefit some individuals, but these mutations are

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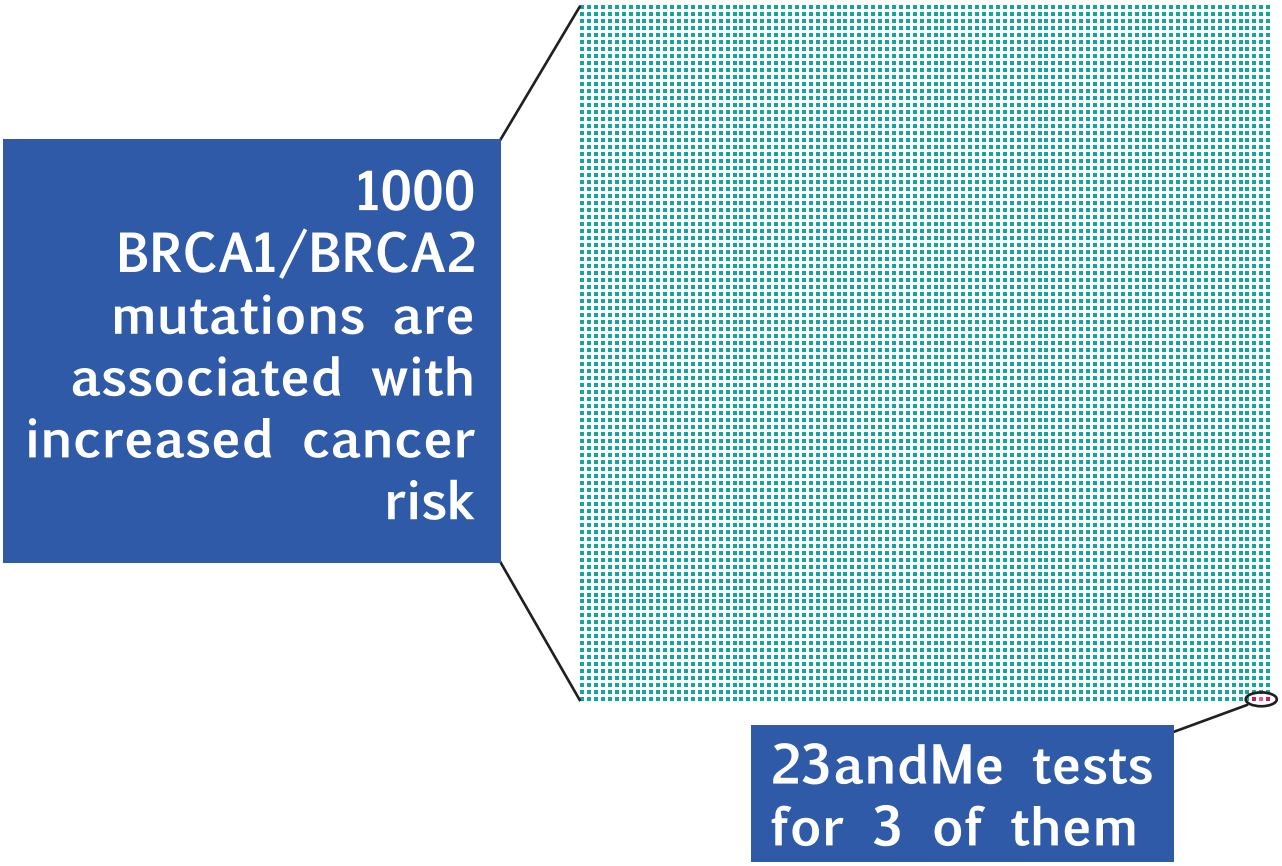
BRCA in the Public Eye

Having a harmful hereditary mutation on the BRCA1 or BRCA2 gene makes a woman up to 50% more likely to develop breast cancer. It also increases risk for other cancers compared to women without the mutation. But BRCA mutations were not familiar to many until, in a 2013 New York Times article, Angelina Jolie publicized her decision to have a preventative double mastectomy after testing positive for BRCA1 and BRCA2. She urged women to consider genetic testing themselves. Jolie's article led to a sharp rise in BRCA testing, but no change in preventative surgery, indicating that the increase in genetic testing was not among those women who are at the highest risk of having a harmful BRCA mutation.



very rare in the general population. One could buy the test thinking it provides conclusive information about BRCA status, but its clinical significance is limited for most women.

We don't yet know which consumers are opting to purchase this test from 23andMe, but we do know that it is most likely to benefit only a small segment of the population. We also know that direct-to-consumer testing increases accessibility, but it comes at a cost of context. Patients who seek tesing without discussing it with a doctor or genetic counselor are likely to miss out on key information related to how their results apply to their overall health and cancer risk.

Next Steps

If you are interested in genetic testing for cancer risk, talk with your doctor. Ask your doctor if you are a good candidate for genetic testing and what type of genetic testing you are most likey to benefit from. Ask for a referral to a genetic counselor. You can also contact your insurance provider to find out about whether any coverage is available and how much the testing is likely to cost.

If you are concerned about your cancer risk, but you don't want to puruse genetic testing right now, your doctor can help. Schedule an appointment to discuss how your family history and lifestyle relate to your risk for cancer. You could talk with your doctor about any changes you can make to reduce your cancer risk. Your doctor can also talk about cancer screening tests that would be appropriate now or in the future.

There is no way to know with certainty whether or not you will develop cancer; all you can do is become informed on your options. Genetic testing is a helpful option for many, but it is just one piece of a complex puzzle.