What Genes *MAY* Tell Us

About Breast Cancer Risk

Investigation into current knowledge related to genetic influences for Breast Cancer reveals far more and different information than even five years ago. Unchanged is the cautionary note from researchers that genetic predispositions are rarely a guarantee, and that environment and lifestyle factors remain important in most cases.

Current Guidelines to Indicate Genetic Testing for Breast Cancer

Among other criteria, current guidelines recommend genetic testing for someone diagnosed with breast cancer if:

- there is a known mutation in the family
- the person was diagnosed at age 50 or younger
- the person has been diagnosed with two primary breast cancers
- a first-degree female relative (mother, sister) was diagnosed with breast cancer at age 50 or younger or was diagnosed with ovarian cancer
- two or more first-degree relatives were diagnosed with breast, prostate, and/or pancreatic cancer
- a man in the family has been diagnosed with breast cancer

Individuals who have the following situations are more likely to have an inherited genetic mutation:

- there are blood relatives (grandmothers, mother, sisters, aunts) on either the mother's or father's side of the family who had breast cancer diagnosed before age 50
- there is both breast and ovarian cancer on the same side of the family or in a single individual
- there is a relative(s) with triple-negative breast cancer
- there are other cancers in the family in addition to breast, such as prostate, melanoma, pancreatic, stomach, uterine, thyroid, colon, and/or sarcoma
- women in the family have had cancer in both breasts
- the individual is of Ashkenazi Jewish (Eastern European) heritage
- the individual is African American and has been diagnosed with breast cancer at age 35 or younger
- a man in the family has had breast cancer
- there is a known genetic mutation linked to breast cancer in the family

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In the general population, approximately 12% of women will be diagnosed with breast cancer over a lifetime. Of all those diagnosed, approximately 5-10% will have some genetic predisposition. That's a small percentage, but with a genetic issue, the chances of getting breast cancer may be significantly greater, so it's important information. In the case of men getting breast cancer, the incidence is stated as 1 in 1,000 - less than 1% of all diagnosed cases, but men also can carry gene mutations related to breast cancer. However, since men's breast cancer risk is so low, even with a genetic mutation elevating risk, it remains low, and is unlikely to manifest as breast cancer. Still, if men carry the problem gene mutation, they can pass it on to their children, so it is highly significant in that respect.

Most of those diagnosed with breast cancer wonder if there is some genetic influence. Medical practitioners look at family history of cancer to see if genetic testing is indicated. (See adjacent chart)

A recent study published in the Journal of Clinical Oncology looked at a group of nearly 1000 women diagnosed with breast cancer and administered genetic testing to all. Approximately 10% were found to have a genetic mutation that would influence their diagnosis or treatment. But, assessing each individual's history in the context of existing guidelines for testing, it was shown that nearly half of those with mutations would NOT have met the criteria to be recommended for genetic testing. This study is leading questions about the utility of the guidelines. In February of 2019, the American Society of Breast Surgeons (ASBrS) recommended giving every person diagnosed with breast cancer a genetic test with a multi-gene panel. Included was a recommendation that there be retesting for those who were tested prior to 2014.

Another area of growth related to genetic testing is the proliferation of DTC (direct to consumer) genetic testing. DTC genetic testing started as a way to make it easier to track down long lost relatives and learn more about family history and heritage. Individuals submit saliva samples, and analysis is completed by the genetic testing service. DNA variations can provide clues about ancestry and family relationships. Genetic ancestry testing, or genetic genealogy, allows users a deeper investigation. As well as geographic and demographic information about ancestors, they may find information about various genetic quirks and sometimes the likelihood of contracting certain health conditions. While various companies offer the service, only the test company 23andMe is currently granted US FDA approval to provide genetic information, including the BRCA-1 and BRCA-2 mutations and certain other medical information.

The medical community advises caution about making any health decisions based on DTC genetic testing. A recent study of the accuracy of DTC testing results identified a 40% false positive rate for findings related to cancer risk genes. Also, an identified mutation or other risk factor does not guarantee that someone will develop the disease. As well, the absence of the mutation does not guarantee that the person will not develop the disease.

Finally, the study of breast and other cancers is ongoing and many non-hereditary causative relationships are becoming understood. They will not show up in genetic testing but should not be underestimated.

Health professional organizations and patient advocacy groups strongly recommend that individuals explore their health concerns with their primary care providers first and seek referrals to genetic counseling if warranted. If conditions warrant, based on medical and family history, or an existing diagnosis, a genetic counselor will recommend testing and health insurance is likely to cover the cost.

Sources and Resources

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