

Legacy Health

Cancer genetics: Breast cancer

Breast cancer affects almost 1 in 8 women during their lifetime, mostly at older ages (>60). Most breast cancers (about 90 to 95%) are “sporadic” cancers that can happen to anyone. The remaining 5 to 10% are due at least in part to an inherited predisposition, that comes down from one generation to the next. (A “pre-disposition” means a higher chance of getting a disease.)

We now realize that all cancer is genetic. We each start as a single cell, and grow to two, four, eight etc. Each time a cell divides, there is a small chance for a gene mutation to arise. As we get older, we build up more mutations, which may lead to tumors and to cancer. About 5 to 10% of cancers arise because there is a mutation present from the beginning, so all future cells are “one step ahead” towards becoming cancerous. When this happens, there are more cancers, and at younger ages than average, both in individuals and in their families.

Women in families with a mutation in particular genes, such as BRCA1 and BRCA2, have an increased chance of developing breast, ovarian and other forms of cancer. However, inheriting a predisposition is not enough to cause a cancer to develop. All breast cancers result from many factors, including age, lifestyle, gene changes acquired over a lifetime, environment, etc.

BRCA1 and BRCA2 are called “cancer-predisposing genes”; when they are damaged they may lead to development of cancers in those tissues where the affected genes normally work. Mutations in BRCA1 and 2 account for about 1/3 of the hereditary cases of breast cancer in the United States. (This is about 1/3 of the 5 to 10% of breast cancers that are due to a hereditary mutation.) In addition, BRCA1 and BRCA2 are likely involved in up to half of hereditary ovarian cancers.

Mutations in BRCA1 and 2 are associated with high lifetime cancer risks of up to 70 or 80%. Other mutations in more recently discovered genes are associated with more moderate lifetime risks of up to 50%. This means that most people with mutations in these genes will not develop a cancer, BUT their children each still have a 50% risk of inheriting the same mutation. Not all the genes that may lead to cancer have been identified, and although the sensitivity of clinical testing is superb, it is not perfect; some mutations are still beyond the detection capabilities of these tests. Therefore, genetic testing cannot identify all mutations that may contribute to breast and/or ovarian cancer.

In addition, individuals with inherited mutations who have had breast cancer have a higher chance of a recurrence of breast cancer; they also are at higher risk of developing new breast, ovarian and/or other cancers.

While men with these mutations are not at risk for ovarian or endometrial cancers, they are at increased risk for breast cancers – up to 9% for BRCA2. *Just like women, their children each have a 50% chance to share any mutation they have.*

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Genetic testing process and options

The gene testing used today includes checking for high-risk and moderate-risk genes, including BRCA1 and 2. This testing is very sensitive and accurate, but (like any test) it is not perfect. If a cancer-predisposing mutation is found, it indicates an increased chance for developing cancer. After testing, an office visit with a cancer genetics counselor will help you understand the test results; the counselor may offer screening and early detection recommendations, as well as preventative surgery possibilities, if appropriate.

Receiving a negative (“normal”) test result does not guarantee that a gene mutation is not present. Testing is not 100 percent accurate, and testing is not yet available for all cancer-predisposing genes. Sometimes test results are considered “uninformative”; either a gene change is detected but it is not known if it may lead to cancer, or no mutation is found in a family but the family history strongly suggests an inherited predisposition.

Of course, choosing not to be tested is always an option.

One alternative to individual testing is to collect DNA samples from one or more family members and store them frozen in a banking laboratory. In this way, the family preserves the option of testing in the future. With DNA samples from family members who have had cancer, future generations will have family genetic information available, if needed, to help interpret their genetic testing.

Another alternative to testing is for a person to assume he/she has inherited the predisposition running in the family. They could then take steps to decrease the risk of developing cancer by more frequent screenings and/or preventative surgery when appropriate. These measures, of course, do not guarantee a 100 percent reduction in the risk of developing cancer.

For more information or questions, call Legacy Genetic Services at 503-413-6534.

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