Other Factors (>90% of all cancers)

Mutations in BRCA genes (5-10% of all cancers)

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Breast and Ovarian Cancer Susceptibility Genes

What are breast and ovarian cancer susceptibility genes?

Genes are the instructions that guide your body’s growth and development. Each cell in the human body has about 20,000 genes, and there are two copies of most genes. You inherit one copy of each gene from your mother and a second copy from your father.

There are some genes that protect the breast and ovary cells from cancer. When one of these genes is not working right, you are more likely to get cancer. These are called breast and ovarian cancer susceptibility genes. The most common ones are BRCA1 and BRCA2. Women with a mutation (genetic change) in a BRCA gene are much more likely to get breast and ovarian cancer. Men with a BRCA mutation also have a higher risk for breast cancer. The two BRCA genes account for most cases with inherited susceptibility to breast and ovarian cancer, but there are other genes that can also raise your cancer risk.

How do these genes cause cancer?
The breast and ovarian cancer susceptibility genes do not cause cancer. Normally, these genes help fix cells that get genetic damage or break down cells that cannot be fixed. If one of these genes is not working right, a damaged cell in the body may not get fixed. This can eventually lead to cancer. But not everyone with a BRCA mutation will get cancer.

Does my family history of breast or ovarian cancer mean that I have a BRCA mutation?
Not necessarily. Cancer is so common that many people have a history of cancer in the family. Most cancer can be explained by common risk factors like aging, lifestyle choices, and exposures in the environment. Only about 5-10% of breast and ovarian cancers are linked to inherited mutations in a BRCA gene or another susceptibility gene.

Could I carry a BRCA mutation?
BRCA mutations have been identified in families worldwide. There are certain risk factors that help identify who might carry a BRCA mutation. You are more likely to carry a BRCA mutation if you answer “YES” to one or more of the following statements:

- I have had invasive breast cancer and ovarian cancer
- I have had invasive breast cancer in both breasts
- I have had ovarian cancer
- I had invasive breast cancer before age 45
- I had invasive breast cancer before age 65 and my ancestry is Jewish
- I have had invasive breast cancer and I have one or more close relatives with invasive breast cancer or ovarian cancer
- I am a male who has had invasive breast cancer
- I have a strong family history of invasive breast cancer and/or ovarian cancer

What if I do not have any risk factors?
Testing is most useful for individuals who are at high risk. If your cancer history does not include any special risk factors, then you do not need genetic counseling or testing. However, you should still follow the routine screening recommendations for breast cancer.
What if I have one or more risk factors?
Talk with your medical provider about your concerns. Your provider can review your cancer history and may refer you to a genetic counselor. Genetic counseling is the first step to find out if a BRCA mutation is in your family. A genetic counselor reviews your medical records, your health history, and your family history of cancer. This information is used to estimate the chance that you could have a mutation in one of the cancer susceptibility genes. Your genetics visit helps determine whether or not genetic testing is indicated.

How is a BRCA mutation identified?
A blood test can check the cancer susceptibility genes for mutations, but not all mutations have the same risk for cancer. It is easier to understand a result when there is a strong pattern of cancer in the family.

What do I need to think about before having BRCA testing?
Deciding to test the cancer susceptibility genes is very personal. One important part of genetic counseling is exploring what testing means for you and your family. There can be complex, and sometimes unexpected, emotional effects. Your genetic counselor will discuss the pros and cons of genetic testing; including emotional aspects, impact on the family, and cancer screening options.

What does it mean if the BRCA test is positive?
A positive BRCA test means a mutation with a risk for cancer has been found in one of the BRCA genes. A woman with a BRCA mutation has a lifetime risk of developing breast cancer that may be as high as 85%. It also means she has a higher risk of ovarian cancer. A man with a BRCA mutation may be at higher risk for breast cancer and other cancers compared to men without a BRCA mutation. And any person with a BRCA mutation could pass the mutation to their offspring.

What are some of the benefits of BRCA testing?
BRCA testing may be able to help explain the cancer history in your family and clarify your cancer risk. When a mutation is found, it can help guide your medical care. Cancer screening is recommended more often and starts at a younger age. There are also treatment options to lower the risk of cancer.

What are some of the drawbacks to BRCA testing?
When no mutation is found, some women may believe they are no longer at risk for cancer. They may not follow routine screening recommendations. For someone who learns they have a mutation, it can be stressful, especially if no clear plan of action is in place to deal with the results.

Screening for Breast Cancer
All women are at risk for breast cancer. Screening helps find breast cancer early, when it is most treatable and curable. There are three important steps you can take for yourself, regardless of your family history:

1. Monthly breast self-exam
2. Breast exam by a doctor or nurse
3. Regular mammograms.

Mammograms are strongly recommended for all women who are between the ages of 50 to 69. If you are age 40 to 49, talk with your health care provider about when to begin regular mammograms. If you are considered high risk, your provider may suggest that you begin mammograms earlier.

Are there ways to lower my risk of breast cancer?
Some risk factors for breast cancer cannot be changed, like gender, age, and genetic risks. However, choosing a healthy, low-fat diet, getting regular exercise, limiting alcohol, and not smoking may lower your chance of developing cancer in general.