

Are you a candidate for genetic testing?

- Have you or any family member had breast cancer before age 50?
- Have you or any family member had ovarian cancer at any age?
- Has any male in your family had breast cancer?

If you answered **yes** to **any** of the questions, contact your healthcare provider about discussing genetic testing.

The discovery of the **BRCA1** and **BRCA2** genetic mutations answered many questions about why breast cancer seems to run in families. Genetic testing allows an individual to know whether they are at high risk.

BREAST CANCER GENETIC TESTING



“IT’S TIME TO PLACE YOURSELF IN TRAINED
HANDS . . . YOUR OWN.”

Genetic Testing for Hereditary Breast Cancer

If you have a family history of breast cancer, you are probably very concerned about your risk of developing the disease in the future. You may have heard that there is a blood test that can help identify some women and men who are at higher risk for developing breast cancer. This brochure explains the test and who qualifies for testing.

Breast Cancer Gene Discovery

In the mid 1990s, scientists discovered two genes that, when mutated (altered or changed), greatly increase a person’s risk of developing breast and/or ovarian cancer. The genes are known as **BRCA1** and **BRCA2** (**BR**=breast; **CA**=cancer). These genes do not cause cancer. Both genes protect against developing cancer and are present in both women and men. However, when a mutation in either gene exists, there is less protection and a higher risk of developing cancer. Although breast and ovarian cancers are the most common, other cancers may also result from a mutation in either of the genes.

Testing

Testing for BRCA mutations that increase breast cancer risk has received much attention in the media. This attention has contributed to the incorrect impression that the test indicates breast cancer. In truth, genetic (DNA) testing only indicates that an alteration (change) in a BRCA gene exists. It does not prove the “carrier” will develop cancer. It only suggests that the carrier is at much higher risk for breast and ovarian cancers. The test is only appropriate for a small number of women (and men) and not the general public. It is estimated that currently known BRCA mutations contribute to approximately seven to ten percent of diagnosed breast and ovarian cancers. A healthcare professional with genetic experience is the best person to help determine whether or not genetic testing for BRCA mutations is appropriate for you.

Hereditary Breast Cancer

Everyone is born with two copies of approximately 25,000 genes. One copy comes from your mother, and one copy comes from your father. With regard to breast and ovarian cancer, when a woman inherits a mutated BRCA 1 or BRCA 2 gene from either parent, her risk for developing breast and ovarian cancer is increased. It was once believed that risk for hereditary breast and ovarian cancer came only from the mother’s side of the family. We now know that people are just as likely to inherit BRCA mutations from their father’s family.

A man or a woman may inherit and thus “carry” a BRCA mutation without ever developing cancer. This may cause the disease to look as though it has skipped a generation in the family. In smaller families or families with more men, it may be harder to “see” the hereditary cancer risk because there are fewer women to develop cancer. It was once thought that the number of relatives with cancer was the highest predictor of a hereditary syndrome. We now know that other important factors, such as young age at diagnosis, a history of more than one cancer in the same person or male breast cancer in the family, are also strong clues that may indicate the presence of a BRCA mutation existing in a family.

Mutations in the BRCA genes can be passed equally to the sons and daughters of carriers. This places them at increased risk for developing breast, ovarian and, to a much lesser extent, prostate, colon, and pancreatic cancers. Many people assume that, if they carry a BRCA mutation, their children will inherit it. In actuality, each child has a 50 percent risk of inheriting the BRCA mutation from a carrier parent. It is important to look at both sides of the family to evaluate the likelihood that a BRCA mutation may be present. A genetics professional may use a tool called a three-generation pedigree to record the family history. This helps determine the pattern of cancers in the family and whether genetic testing may be appropriate.

Hereditary Breast Cancer (continued)

Certain ethnic groups have a greater likelihood of carrying a BRCA mutation. For instance, one in 40 Jewish persons of Northern and Central European descent (Ashkenazi) may be carriers of a BRCA mutation. Specific testing of three common BRCA mutations in this population can be ordered at a lower cost than the more extensive test generally needed for other ethnic groups.

What are the Risks?

Women who inherit a BRCA mutation have a 56 to 87 percent risk of developing breast cancer by age 70. They also have a 27 to 44 percent risk of developing ovarian cancer by age 70. Women also face increased risk of developing a second breast cancer if they carry a BRCA mutation. These risks vary depending on which gene has the mutation. Increased risks for other cancers exist but are much lower than the risk of developing breast or ovarian cancer.

Testing for Hereditary Breast and Ovarian Cancer Syndrome

It is important that any person considering genetic testing receive appropriate risk assessment and education. After determining the likelihood of a BRCA mutation within the family, a healthcare professional will discuss the advantages, disadvantages and limitations of genetic testing. In a family that appears to be at increased risk for hereditary cancer, the ideal person to test, although it may not be possible, is the youngest person who has had breast or ovarian cancer.

Following risk assessment and education, you will decide whether you want to pursue testing. This decision is totally up to you. If you should decide to have the test, a small sample of blood will be drawn from your arm and sent to a specific laboratory to determine the presence of a BRCA mutation. Whether you decide to test or not, your healthcare professional will review cancer screening recommendations and possible treatment options based on your family history.

Genetic Testing may be Recommended for:

- A woman with breast cancer before age 50
- A woman with ovarian cancer at any age
- A male with breast cancer
- A woman of any age with breast and ovarian cancer
- A woman of any age with breast and/or ovarian cancer who has one first or second degree* blood relative younger than 50 with breast and/or ovarian cancer
- A woman of any age with breast and/or ovarian cancer who has two or more first or second degree blood relatives (on one side of the family) with breast or ovarian cancer
- A person with bilateral breast cancer or multiple primary sites of cancer at younger than average ages
- A person with a blood relative who is documented as being a BRCA mutation carrier
- A woman who has not had cancer but has a family history of breast or ovarian cancer before age 50

*** first degree relative:**

mother
father
sister
brother
daughter
son

*** second degree relative:**

aunt or uncle
grandparents
grandchildren
niece or nephew
half-brother
half-sister

Potential Benefits of Genetic Testing

A positive test result identifies you as a carrier of a BRCA mutation. Due to the increased risk of developing cancer, it would be recommended that you be carefully monitored so that, if cancer does develop, it would likely be diagnosed at an early stage when treatments are more successful. Knowing that you carry a BRCA mutation could provide the motivation to change potentially harmful lifestyle choices to improve your general health (i.e., smoking, lack of exercise, or alcohol intake). Knowing you have a BRCA mutation can help you and your healthcare provider plan cancer surveillance and risk-reducing options, such as surgery, clinical trials or medications. Other family members may choose to be tested or to pursue increased surveillance based on their awareness of a BRCA mutation in the family.

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When there is a known BRCA mutation in the family, a negative test means the person is at normal risk of developing cancer. This knowledge can relieve anxiety and prevent unnecessary and expensive tests and procedures. Strength of the family history is important when interpreting a negative test in a family without a known BRCA mutation.

Potential Disadvantages of Genetic Testing

A positive test may cause you anxiety stemming from knowledge that you are at "high risk" for developing cancer. Family members may also react unpredictably to positive test results because their own risk of cancer may be increased.

Deciding Whether to Test

The decision to have genetic testing should be made in partnership with a healthcare professional skilled in genetics who will:

- Evaluate your risk
- Determine whether you meet usual criteria for testing
- Answer your questions about the process
- Assist you with financial questions
- Obtain informed consent for testing
- Develop a surveillance/treatment plan whether you test or not
- Protect your confidentiality
- Refer you for psychological counseling as needed
- Provide additional resources on BRCA genes/mutations and genetic testing

The genes are known as **BRCA1** and **BRCA2** (**BR**=breast; **CA**=cancer).