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BRCA Genetic Testing

In the mid 1990s, scientists discovered two genes that, when mutated (altered or changed), greatly increase the risk of a person developing breast and/or ovarian cancer. The genes are known as BRCA1 and BRCA2 (BR=breast; CA=cancer). Both genes serve a protective function against cancer and are present in women and men. When a mutation in either gene exists, there is a high risk of developing breast or ovarian 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, which 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, only 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 mutations cause approximately 7 to 10 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 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 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 cancer. Many people assume that, if they carry a mutation, their children will inherit it. In actuality, each child has a 50 percent risk of inheriting the mutation from a carrier parent. It is important to look at both sides of the family to evaluate the likelihood that a mutation may be present. A genetics professional uses 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.

Certain ethnic groups have a greater likelihood of carrying a BRCA mutation. For instance, 1 in 40 Jewish persons of Northern and Central European descent (Ashkenazi) may be carriers of a mutation. Specific testing of three common mutations in this population can be ordered at a lower cost than the more extensive test generally needed for non-Ashkenazi Jews.

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. Women also face increased risk of developing a second breast cancer if they carry a 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 counseling. After determining the likelihood of a mutation within the family, a genetics professional will discuss the advantages, disadvantages and limitations of genetic testing. In a family that appears to be at increased risk for hereditary cancer, although it may not always be possible, the ideal person to test is the youngest person who has had breast or ovarian cancer.

Following counseling, 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 or an oral swish rinse will be used to collect a saliva sample. The sample will be sent to a special laboratory to determine the presence of a BRCA mutation. Whether you decide to test or not, your counselor will review cancer screening recommendations and possible treatment options based on your family history.

Genetic Testing May Be Recommended For:

- Breast cancer before age 50
- Ovarian cancer at any age
- Two primary breast cancers in the same individual at any age
- Both breast and ovarian cancer in same individual at any age
- Male breast cancer at any age
- Two or more breast cancers in a family, one under 50
- Three or more relatives with breast cancer at any age
- Ashkenazi Jewish ancestry, breast cancer at any age
- Triple negative breast cancer at any age
- Identified BRCA mutation in the family
- Pancreatic cancer with breast or ovarian cancer in the same person or on the same side of the family

Do not hesitate to remind your treatment team of any characteristics you see in your history that may qualify you for genetic testing. If there are any questions that cause you or your treatment team doubt, seek the advice of a genetic counselor or professional. A genetics professional uses 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.

Potential Outcomes of Genetic Testing:

- A negative test result will allow you to move forward with breast conservation, if recommended. Other treatments may be recommended by your treatment team.
- A positive test for either a BRCA1 or BRCA2 gene may limit your surgical options. At the present time, breast conservation followed by radiation therapy may not be a recommended option. Instead, bilateral mastectomy and high risk ovarian surveillance (transvaginal ultrasound exam and CA-125 testing) or oophorectomy (ovary removal) are often recommended treatments.

Positive Gene Impact on Your Children:

If you are diagnosed with a positive mutation in either BRCA gene, you can pass the mutated gene equally to either your sons or daughters. This places your children at increased risk for developing breast, ovarian and, to a much lesser extent, prostate, colon and pancreatic cancer. Each child has a 50 percent risk of inheriting the mutation from a carrier parent. Knowing if you are a carrier allows you to tell your children so that they can pursue genetic testing (after age 18) to see if they inherited the gene from you. If they are identified as carriers, your daughters can take steps of "high risk" surveillance.

High Risk Surveillance Recommendations:

- Perform monthly breast self-exams and have an annual clinical exam beginning at age 25
- Start having screening mammograms or breast MRIs between ages 25 – 35
- Start transvaginal ultrasound exams and CA-125 testing between ages 25 – 35

Chemoprevention Recommendations:

- Drugs such as tamoxifen or Evista® be taken
- Oral contraceptives be taken for ovarian protection

Prophylactic Surgery Considerations:

- Prophylactic mastectomy (after breast-feeding desired children) to prevent occurrence
- Prophylactic ovary removal (after age 35 or having desired children) to prevent occurrence

Deciding Whether to Test

The decision to have genetic testing should be made in partnership with a healthcare professional skilled in genetic counseling. Your counselor can also provide additional resources on BRCA genes/ mutations and genetic testing.

Genetic Counselor Will:

- Evaluate your risk
- Determine whether you meet criteria for testing
- Answer your questions about the process
- Assist you with financial questions
- Obtain informed consent for testing
- Protect your confidentiality
- Refer you for psychological counseling as needed

Testing Assessment:

- Have you or any family member had breast cancer before age 50? Yes ☐ No ☐
- Have you or any family member had ovarian cancer at any age? Yes ☐ No ☐
- Has any male in your family had breast cancer? Yes ☐ No ☐

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