Hereditary Breast Cancer

Most people develop breast cancer as a result of multiple exposures that occur over the course of a lifetime. However, some people, about 5 to 7 percent (%), who are diagnosed have an inherited gene mutation. Inherited conditions are passed to an individual through their blood relatives. People who have a hereditary breast cancer (HBC) gene mutation are at an increased risk to develop breast and possibly other cancers compared to the general population.

What Causes Hereditary Breast Cancer?
HBC is caused by an inherited change called a gene mutation. Genes are the set of instructions that tell all of the cells in the body what to do. Genes determine physical characteristics, such as hair and eye color, nose shape and blood type. A mutation is a change in a gene's DNA that causes it to stop working. In the case of HBC, a gene that normally helps to prevent breast and some other cancers has stopped working. Therefore, these types of cancer are more likely to develop and occur at a younger age.

The majority of HBC is due to a mutation in either the BRCA1 or BRCA2 genes. A person who has a BRCA1 or BRCA2 mutation has "Hereditary Breast and Ovarian Cancer" syndrome and has an increased risk for breast, ovarian, prostate, male breast, pancreatic, skin (melanoma) and other cancers. There are also a number of other genes associated with HBC that greatly increase breast cancer risk, some that only moderately increase breast cancer risk and others that are not studied so the breast cancer risk is not yet known.

Hereditary breast cancer genes		
High risk	**Moderate risk**	**Increased risk**
BRCA1	***ATM***	***BARD1***
BRCA2	***CHEK2***	***BRIP1***
CDH1	***PALB2***	***NBN***
PTEN		***RAD50***
STK11		***RAD51C* and *RAD51D***
TP53		***NF1***
		Other genes…

How is Hereditary Breast Cancer Passed on Through a Family?
Each person inherits two copies of most genes; one from the mother and one from the father. Inheriting just one copy of a gene mutation increases the risks for breast and possibly also other types of cancer. There is a 50 percent chance that a person with an HBC gene mutation will pass it to each of his or her children. However, if a person does not inherit an HBC gene mutation that is present in his or her family member(s), that person is still more likely than the general population to develop breast cancer.

Why is it Important to Diagnose Hereditary Breast Cancer in a Family?
People who have an HBC syndrome are at higher risk to develop breast and other cancers than the general population. For someone who already has been diagnosed with breast cancer, having an inherited gene mutation may indicate a higher risk of developing another cancer, including another breast cancer.

Because HBC is inherited, the diagnosis also affects family members. If genetic testing identifies the specific mutation causing the increased risk for breast cancer in a family, then other family members can be tested for that same genetic mutation.

It is very important for people with HBC to consider specialized cancer risk-reduction and early detection strategies as directed by their health care providers. Management plans include specific cancer screening exams and/or preventive surgery and can be tailored to each patient and his or her family members. Cancer screening exams are medical tests performed to ensure that any existing cancers are identified at their earliest, most treatable stages.

How is Hereditary Breast Cancer Diagnosed?
A medical and family history review is used to screen for the possibility of an HBC syndrome. A genetic counselor usually conducts this screening process and assesses the family's risk. During the screening, the patient and counselor will create a multi-generation family tree. Some signs that suggest breast cancer may run in a family include:

- Close relatives with breast, ovarian or other related cancers.
- Pre-menopausal breast cancer diagnoses.
- Multiple related cancers in an individual, such as breast and ovarian cancer in a single person.
- Male breast cancer.
- Ashkenazi Jewish ancestry.

Further tests may be needed if the medical and family history review suggests the possibility of a HBC syndrome.

Genetic Testing
A person's blood or saliva can be tested for a genetic mutation that causes HBC. This testing may be limited to the BRCA1 and BRCA2 genes or may also include other genes. In general, there are three types of results:

- Positive for a mutation. If a mutation is found, then the diagnosis of HBC is confirmed. Next, other family members may have a genetic test to learn whether or not they carry the same mutation and have the same HBC syndrome.
- Negative for a mutation. The cancer in the family may not be hereditary. However, a negative genetic test result does not eliminate the possibility of HBC in the family, as current genetic testing technology is not able to identify all mutations that cause HBC or the gene that caused it was not included in the test.
- Variant of unknown significance. A variant is a gene change that does not provide clear information regarding cancer risks. A variant may represent benign genetic differences from one person to the next or may actually represent a true genetic mutation. Until more information regarding the variant is collected by researchers, it remains unknown whether a genetic variant increases cancer risks. Thus, a variant result should not be used to make medical decisions.

If you are concerned about the possibility of an HBC syndrome in your family, you are encouraged to discuss your personal and/or family history with your health care provider. Your physician may refer you to the Cancer Genetics Program of the MD Anderson Cancer Center at Cooper for a genetic evaluation and discussion of testing options. If genetic testing is warranted and you choose to proceed, a blood or saliva sample will be taken during your visit to start the process. Please note that health insurance companies may cover most, if not all, of the cost of genetic testing on a case-by-case basis.

Resources

FORCE
www.facingourrisk.org
FORCE is a nonprofit organization for women who are at high risk of developing breast and/or ovarian cancer due to their family history and genetic status and for members of families in which a BRCA mutation may be present. Check out the “Resource Guide”, “Message Board” and “Chat” sections.

Be Bright Pink
www.bebrightpink.com
Bright Pink is a national non-profit organization that provides education and support to young women who are at high risk for breast and ovarian cancer.

National Ovarian Cancer Coalition (NOCC)
www.ovarian.org
The NOCC’s mission is to raise awareness and to promote education about ovarian cancer. The coalition strives to improve the quality of life for ovarian cancer survivors. The site includes sections on detection, treatment, coping, surviving and more.

Susan G. Komen Breast Cancer Foundation
www.breastcancerinfo.com
The “Komen Facts for Life” features a section on genetics and breast cancer. Check out “Komen Connection, ABCs of Breast Cancer and Resources and References.”

Y-Me National Organization for Breast Cancer Information
www.y-me.org
Y-Me provides information and support to those touched by breast cancer, their families, medical professionals and the public. Unique resources include “ShareRing” and “Male Breast Cancer Support.” The “ShareRing” network is a free, monthly one-hour teleconference featuring a breast cancer related presentation by a health care professional followed by a question and answer session and moderated small group discussion. Information is available in Spanish.

Young Survival Coalition
www.youngsurvival.org
The Young Survival Coalition (YSC) is dedicated to the critical issues unique to young women and breast cancer. YSC works with survivors, caregivers and the medical, research, advocacy and legislative communities to increase the quality of life for women 40 and younger diagnosed with breast cancer.

National Cancer Institute
www.cancer.gov
This site has valuable cancer-related health information on more than 200 cancer types, clinical trials, cancer statistics, prevention, screening, risk factors, genetics and support resources. Information is available in Spanish.

American Cancer Society (ACS)
www.cancer.org
The ACS is a voluntary national health organization that supports research, provides information about cancer and offers many programs and services to patients and their families. Information is available in Spanish.

Genetic Alliance, Inc.
www.geneticalliance.org
This organization supports individuals with genetic conditions and their families, educates the public and advocates for consumer-informed public policies. This site provides information on genetic policy, research and a helpline for people with genetic questions.