

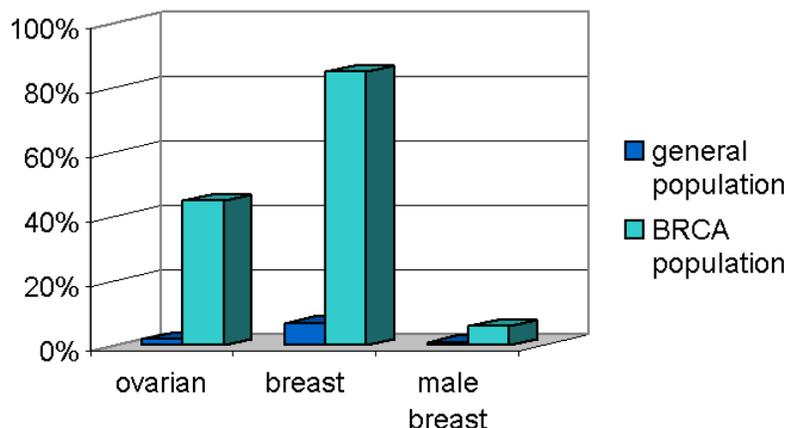
Hereditary Breast and Ovarian Cancer Syndrome

Hereditary Breast and Ovarian Cancer syndrome (HBOC) is an inherited tendency to develop breast, ovarian and other cancers. Inherited conditions are passed to an individual through their blood relatives. Although most cancers are not inherited, about 5 percent (%) of people who have breast cancer and about 10% of women who have ovarian cancer have HBOC.

What are the cancer risks for people with HBOC?

- A higher than usual risk of developing breast cancer (40% to 87% lifetime risk) (See graph below).
- A higher than usual risk of breast cancer developing before age 50, or before women reach menopause.
- An increased risk of developing a second primary breast cancer (40% to 60% risk).
- A higher than usual risk of developing ovarian cancer (10% to 44% lifetime risk).
- In men, the risk of breast cancer is also higher than usual (6% lifetime risk).
- In men, the risk of prostate cancer may also be increased, and tends to occur at younger ages than in the general population. Men should discuss the benefits, risks and limitations of prostate cancer screening with their doctor.
- Risks for some other cancers, including pancreatic cancer and melanoma, are also increased.

HBOC cancer risks compared to the general population



What causes HBOC?

HBOC is caused by an inherited change in a gene, called a mutation. Genes are the set of instructions that tell all of the cells in our bodies what to do. Genes determine our hair and eye color, the shape of our nose, our blood type and the number of fingers and toes we have. A mutation is a change in a gene's DNA that causes the gene to stop working. In the case of HBOC, a gene has stopped working that normally helps to prevent breast, ovarian and some other cancers. Therefore, these types of cancer are more likely to develop and are also more likely to occur at a younger age than usual. The majority of HBOC is due to a mutation in either the BRCA1 or BRCA2 genes.

What are my chances of inheriting HBOC?

Each person has two copies of all genes, including each of the BRCA genes. One copy is inherited from their mother, and one copy is inherited from their father. One inherited gene mutation in either copy of a BRCA gene will cause HBOC. There is a 50% chance that a person with HBOC will pass the change or mutation to each of their children. Inheriting this mutation causes an increased risk for several types of cancers (refer to Page 1). However, if a person does not inherit the mutation, they have the 12% general population lifetime risk to develop breast cancer and the 1% to 2% general population lifetime risk to develop ovarian cancer.

Why is it important to diagnose HBOC?

People who have HBOC are at much higher risk to develop breast, ovarian and other cancers than people in the general population. For someone who has cancer, the diagnosis of HBOC may indicate a higher risk of developing a new cancer in the future. For these reasons, individuals with HBOC should follow different screening and prevention guidelines than people at average risk for breast, ovarian and prostate cancer. Following the early detection and prevention guidelines for people with HBOC can reduce these cancer risks.

Because HBOC is inherited, the diagnosis also affects family members. If genetic testing identifies the specific mutation causing HBOC in a family, then other family members can be tested.

How is HBOC diagnosed?

Family History

A medical and family history review is used to screen for the possibility of HBOC. A genetic counselor usually conducts this screening process and assesses the family's risk for HBOC. During the screening, the patient and counselor will create a multi-generation family tree. Some signs that suggest HBOC 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 individual;
- male breast cancer; and

- Ashkenazi Jewish ancestry.

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

Genetic Testing for HBOC

Genetic testing of a blood or saliva sample can identify a mutation in the BRCA1 or BRCA2 gene. If a mutation is found, then the HBOC diagnosis is confirmed. Next, other family members may have a genetic test to learn whether or not they carry the same mutation and have HBOC.

Sometimes, genetic testing will not find a mutation in the BRCA1 or BRCA2 genes even in persons with a clinical history that suggests HBOC. This does not necessarily mean that they do not have HBOC. A negative genetic test may be due to the fact that the current genetic testing technology is not able to identify all mutations or other genes that may cause HBOC.

A third, but rare result is a variant. A variant is a gene change that does not provide clear information regarding cancer risks. In these cases, further testing may be ordered to help clarify the result.

How is HBOC managed?

It is very important for people with HBOC to reduce their cancer risk by following specialized cancer prevention and early detection guidelines. Management plans include specific cancer screening exams, and/or preventive surgery and are tailored to each patient and their family by a team of specialists. Cancer screening exams are medical tests performed to ensure that any existing cancers are identified at their earliest, most treatable stages. General screening and prevention guidelines for persons with HBOC are outlined below.

Recommendations for Women

Breast Cancer Screening Recommendations

- **Beginning at age 18** - monthly breast self-exams and annual clinical breast exams.
- **Beginning at age 25** (or earlier, depending on family history) - semi-annual clinical breast exams and annual mammogram and breast MRI. The mammogram and breast MRI may be staggered six months apart (for example, you would have a mammogram, then six months later a breast MRI, then six months later another mammogram).
- You may also wish to discuss medicines that are prescribed as a preventive measure, such as tamoxifen, with your doctor. Preventative surgical removal of the breasts, also referred to as prophylactic bilateral mastectomy, is another option for women with HBOC.

Symptoms of Breast Cancer

Although many changes in the breast are not caused by cancer, some changes to look for include:

- a lump or mass in your breast;
- enlarged lymph nodes in the armpit;
- changes in breast size, shape, skin texture or color;
- skin redness;
- skin dimpling or puckering; and

- nipple changes or discharge.

If any of these changes are present for **more than two weeks**, contact your doctor immediately.

Ovarian Cancer Screening Recommendations

- **Beginning at age 30** - pelvic exam, vaginal ultrasound and a blood test for a tumor marker called CA-125. These screening tests are usually done every six months. However, ovarian cancer is difficult to detect by symptoms and screening alone.
- **For women between ages 35-40 or older who have completed childbearing** - surgical removal of the ovaries and fallopian tubes, called prophylactic bilateral salpingo-oophorectomy. This protective surgery will significantly decrease the risk of developing ovarian cancer. You may also wish to discuss medicines that are prescribed as a preventive measure, such as oral contraceptives, with your doctor. Research studies indicate that oral contraceptives can reduce the risk of ovarian cancer by more than 50%.

Symptoms of Ovarian Cancer

The symptoms of ovarian cancer are not very specific; the same symptoms can also be caused by other conditions. This can make it hard to detect ovarian cancer by symptoms alone.

However, you should talk to your doctor if you have new or persistent symptoms of:

- abdominal bloating (swelling of the abdomen or stomach area);
- a feeling of pain or pressure in the pelvic area;
- change in appetite, particularly if you feel full after eating only a little; and
- changes in bowel or bladder habits (such as urgency or frequency of urination).

Recommendations for Men

- **Beginning at age 35** – breast self-exam and education and a clinical breast exam every six-12 months.
- **Beginning at age 40** – consider a baseline mammogram and consider starting prostate cancer screening.

Recommendations for Men and Women

- Learn about signs and symptoms of cancer(s), especially breast and ovarian cancer, prostate cancer, melanoma and pancreatic cancer
- No specific guidelines exist for pancreatic cancer and melanoma screening. Your doctor may recommend screening based on family history

Where can I find more information?

If you are concerned about HBOC in your family, contact the department of Clinical Cancer Genetics at 713-745-7391 to schedule a consultation with a genetic counselor. The genetic counselor will perform a complete cancer risk-analysis and discuss further testing and medical management options for you and your family members. More information about the Clinical Cancer Genetics program is also available at <http://www.mdanderson.org/departments/ccg>.

Resources

Hereditary Breast and Ovarian Cancer

FORCE

<http://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

<http://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.

Breast and Ovarian Cancer

National Ovarian Cancer Coalition (NOCC)

<http://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

<http://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

<http://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 healthcare professional followed by a question and answer session and moderated small group discussion. Information is available in Spanish.

Young Survival Coalition

<http://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 diagnosed with breast cancer ages 40 and under.

General Cancer and Genetics

National Cancer Institute

<http://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)

<http://www.cancer.org>

The ACS is a voluntary national health organization with local offices around the country. The ACS 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.

<http://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.

Understanding Genetic Testing

<http://nci.nih.gov/sciencebehind>

This site has a slide show that provides information about the role of DNA, chromosomes and genes in hereditary cancers.