Hereditary Breast and Ovarian Cancer Syndrome

Hereditary Breast and Ovarian Cancer (HBOC) syndrome is an inherited, genetic condition that increases the risk for breast, ovarian and other cancers. Inherited conditions are passed down in families. Parents can pass HBOC syndrome to their children.

HBOC syndrome is caused by changes (mutations) in the *BRCA1* and *BRCA2* genes. Normally, the *BRCA1* and *BRCA2* genes help protect the body from developing certain cancers. A mutation in either the *BRCA1* or *BRCA2* genes causes an increased risk for certain cancers.

- 5 to 10 out of every 100 people with breast cancer have HBOC syndrome.
- About 15 out of every 100 people with ovarian cancer have HBOC syndrome.

Cancer Risks

People with HBOC syndrome have an increased risk of developing certain cancers. Your exact risk may depend on your personal and family history. Talk with your doctor or genetic counselor about your risk.

Breast Cancer

Females with HBOC syndrome:

- Have a 6 in 10, or higher, chance of developing breast cancer in their lifetime. This is higher than the average risk for females to develop breast cancer (about 1 in 12).
- Have a higher chance of developing breast cancer before age 50 or before menopause.
- Have a higher chance of developing a second (new) breast cancer after having a first breast cancer diagnosis.

Males with HBOC syndrome have a higher chance of developing breast cancer in their lifetime than males without HBOC syndrome.

Ovarian Cancer

Females with HBOC syndrome a much higher chance (up to 58 in 100) of developing ovarian cancer in their lifetime. The average female's chance of developing ovarian cancer is 2 in 100.

Prostate Cancer

Males with HBOC syndrome have up to a 6 in 10 chance of developing prostate cancer in their lifetime. The average male's chance of developing prostate cancer is about 1 in 10.

Other Cancers

People with HBOC syndrome may have a higher chance of developing other cancers, such as melanoma skin cancer and pancreatic cancer. The risk for pancreatic cancer is higher if you have a family history of pancreatic cancer.

Diagnosis

Diagnosing HBOC syndrome can help plan for additional cancer screening and preventive care. People with HBOC syndrome are at much higher risk for breast, ovarian and other cancers than people in the general population.

A person with HBOC syndrome who already has cancer has a risk that a new cancer may also develop. You may reduce your cancer risk or cancer may be detected earlier by following specific cancer screening and prevention guidelines for people with HBOC syndrome.

Family History

You may be asked to gather information about your family history of cancer. Your genetic counselor will record this information in a family tree to assess you and your family's risk.

Signs that HBOC syndrome runs in a family may include:

- Multiple close relatives with breast, ovarian or other related cancers
- A family member with breast cancer diagnosed before age 50 or before menopause
- A family member with multiple related cancers, such as breast and ovarian cancer
- A family member with ovarian cancer
- A family member with pancreatic cancer
- A male family member with breast cancer
- Ashkenazi Jewish ancestry

Your genetic counselor may recommend genetic testing if your medical and family history show signs of possible HBOC syndrome.

Genetic Testing

Genetic testing for HBOC syndrome involves a blood or saliva test that looks for mutations in the *BRCA1* or *BRCA2* genes. An HBOC syndrome diagnosis is confirmed if a mutation is found.

Genetic tests are not perfect. Even if the test does not find a *BRCA1* or *BRCA2* gene mutation, you may still have a mutation that cannot be found with current technology. More research is still needed for technology to be able to find all gene mutations related to HBOC syndrome. You can continue to ask your doctor about updates to genetic testing over time.

Genetic testing may also find an uncertain result, called a variant of uncertain significance (VUS). This result means the genetic test found a gene change that does not provide clear information about cancer risks. With a VUS result, your doctor may order further testing to help understand what the result means for you.

Family Members

The *BRCA1* and *BRCA2* gene mutations can be passed down in a family from parents to children. Your body has 2 copies of every gene. Each parent gives you one copy of every gene. With HBOC syndrome, only one copy of the *BRCA1* gene or the *BRCA2* gene needs to be

affected by a mutation. One inherited gene mutation in either copy of the *BRCA1* or *BRCA2* genes will cause HBOC.

- A person with a *BRCA1* or *BRCA2* gene mutation has a 50% (1 in 2) chance to pass the gene mutation to each child.
- *BRCA1* and *BRCA2* gene mutations do not typically skip generations.
- Males and females have equal chances to inherit or pass on the mutation.

Family members may benefit from genetic testing when a person in the family has HBOC syndrome. Testing can help determine which family members may have increased cancer risks due to the same gene mutation.

Before having children, people with a *BRCA1* or *BRCA2* gene mutation may want to talk with their partner about genetic testing. If both parents have a *BRCA1* or both parents have a *BRCA2* mutation, their children are at risk for a genetic condition called Fanconi anemia. Fanconi anemia is caused by inheriting either 2 *BRCA1* or 2 *BRCA2* gene mutations. Fanconi anemia is a rare childhood blood disorder that causes bone marrow failure, physical changes, and increased blood cancer risks.

If a family member does not have a *BRCA1* or *BRCA2* gene mutation, screening recommendations are made based on family history.

Screening and Prevention

Screening helps find cancer as early as possible, when it may be easier to treat. It is very important for people with HBOC syndrome to follow specific cancer prevention and early detection guidelines. The recommendations below are general guidelines for people with HBOC syndrome. Your care team will make a specific screening and prevention plan for you and your family.

General Screening Recommendations for Females with HBOC

Breast Cancer Screening

- **At age 18**, start monthly breast self-exams and practice general breast awareness.
- **At age 25,** (or earlier, depending on family history), start breast exams by a health care provider every 6 to 12 months.
- **At age 25,** (or earlier, depending on family history), start yearly breast MRI scans.
- **At age 30**, start yearly mammograms and breast MRI scans. These screenings may be alternated every 6 months. For example, if you have a mammogram in January, you will have a breast MRI in June.
- Talk with your doctor about prescription medicines for prevention, such as tamoxifen.
- Talk with your doctor about preventive breast surgery. Females may consider surgery to remove the breasts to prevent cancer.

Breast Cancer Symptoms

Changes in the breasts can 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
- Swelling, soreness or rash
- Skin dimpling or puckering
- Nipple changes or discharge

Many changes in the breast are not caused by cancer. If you have any of these changes for **more than 2 weeks**, contact your doctor right away.

Ovarian Cancer Screening
Surgery to remove the ovaries and fallopian tubes can significantly decrease the risk of developing ovarian cancer. If you have this surgery, you will not be able to get pregnant naturally. Talk to your doctor about the risks and benefits of this surgery and fertility options if you want to have biological children after this surgery.

If you choose to have this surgery, it is recommended at the following ages:
- **Between ages 35 to 40,** (or earlier, depending on family history) for females with a *BRCA1* mutation
- **Between ages 40 to 45,** (or earlier, depending on family history) for females with a *BRCA2* mutation

Talk to a doctor specializing in gynecology and genetics about the benefits and limitations of other non-surgical risk reducing options such as prescription medicines, imaging, or blood work.

Ovarian Cancer Symptoms
Ovarian cancer symptoms are not very specific. Other common conditions can also cause the same symptoms. Symptoms to watch for include:
- Swelling or bloating of the abdomen
- Feeling of pain or pressure in the pelvic area
- Change in appetite
- Feeling full after eating only a small amount
- Changes in bowel or bladder habits (such as urgency to urinate or frequency of urination)

Ovarian cancer is difficult to find based on symptoms and screening alone. Talk with your doctor if you have symptoms that will not go away or if you have new symptoms.

General Screening Recommendations for Males with HBOC

Breast Cancer Screening
- **At age 35**, start breast self-exams and have a clinical breast exam each year.
- **At age 50,** (or earlier, depending on family history), consider a mammogram yearly.

Prostate Cancer Screening
- **At age 40**, start prostate cancer screening (a PSA blood test and rectal exam each year).
- Talk with your doctor about the benefits, risks and limitations of prostate cancer screening.

Pancreatic Cancer Screening

- Pancreatic cancer screening can be considered for individuals with HBOC and a close family history of pancreatic cancer. Talk with your doctor about screening options based on your family history, as well as the benefits, risks, and limitations of pancreatic cancer screening.

Screening Recommendations for All

- Learn about signs and symptoms of cancer, especially breast, ovarian, prostate, pancreatic and melanoma. Talk with your doctor about any questions you may have.
- There are no specific guidelines for melanoma screening for people with HBOC syndrome. Your doctor may recommend screening based on your family history.

If you are concerned about HBOC in your family, contact Clinical Cancer Genetics at 713-745-7391 to schedule a visit with a genetic counselor. A genetic counselor will assess your risk and talk with you about further testing and screening and prevention options for you and your family.

Resources

Clinical Cancer Genetics Program
713-745-7391
www.MDAnderson.org/Departments/CCG/
MD Anderson's Clinical Cancer Genetics Program provides hereditary cancer risk assessment and consultation services.

FORCE
www.FacingOurRisk.org
FORCE is a nonprofit organization for people who have hereditary cancer risks.

National Ovarian Cancer Coalition (NOCC)
www.Ovarian.org
The NOCC website includes information about ovarian cancer detection, treatment, coping, surviving and more.

Young Survival Coalition
www.YoungSurvival.org
The Young Survival Coalition (YSC) supports young people with breast cancer.

Genetics (National Library of Medicine)
www.MedlinePlus.gov/Genetics/
This website provides patient-friendly information about genetics and genetic related conditions.