

Breast Cancer

Why is genetic testing for Breast Cancer an emerging public health issue?

Other than skin cancer, breast cancer is the most common type of cancer among women in the United States and is the second leading cause of cancer death in women. In 2001, there will be about 40,600 deaths from breast cancer in the United States (40,200 among women, and 400 among men).

In the United States, one out of nine women will develop breast cancer in her lifetime.

About 50-60% of women with inherited BRCA1 or BRCA2 gene changes will develop breast cancer by the age of 70. Women with these inherited gene changes also have an increased risk for developing ovarian cancer.

The prevalence of cancer-predisposing BRCA1 mutations in the general population is estimated to be between 1/500 and 1/1000. The prevalence of BRCA2 mutations in the general population is unknown.

What is Breast Cancer?

Breast cancer occurs mostly in women, but can also occur in men. The American Cancer Society (provide link) estimates that in 2001 about 192,200 new cases of invasive breast cancer will be diagnosed among women in the United States. An estimated 1,500 cases will be diagnosed among men.

Types of breast cancer include: (provide link to all of these?)

- **Adenocarcinoma**, which is a general type of cancer that starts in glandular tissues
- **Ductal carcinoma in situ (DCIS)**, the most common type of noninvasive breast cancer
- **Infiltrating (or invasive) ductal carcinoma (IDC)**, which accounts for 80% of breast cancer
- **Infiltrating (or invasive) lobular carcinoma (ILC)**, accounting for 10-15% of invasive breast cancer
- **Medullary carcinoma**, accounting for 5% of breast cancers

What are the symptoms of Breast Cancer?

Early breast cancer usually does not cause pain and may not produce any symptoms. Changes a woman should watch for include:

- A lump or thickening in or near the breast or underarm area
- A change in size or shape of the breast
- Nipple discharge or tenderness or a nipple pulled back into the breast
- Ridges or pitting of the breast (skin may look like the skin of an orange)
- A change in the way the skin of the breast, areola, or nipple looks or feels (warms, swollen, red, or scaly)
- Pain or discomfort

Fifty percent of breast lumps are found in the upper outer quarter of the breast. Most breast lumps are benign or not cancerous. In the United States, over 80% of biopsied breast abnormalities are proven benign. The most common sign of breast cancer is a new lump or mass. A mass that is painless, hard, and has irregular edges is more likely to be cancerous, but some rare cancers are tender, soft, and rounded.

Most lumps are caused by fibrocystic changes. Cysts are fluid-filled sacs, and fibrosis refers to connective tissue or scar tissue formation. Fibrocystic changes may cause breast swelling and pain. The breasts may feel nodular, or lumpy, and, sometimes, a clear or slightly cloudy nipple discharge is present. Benign breast tumors such as fibroadenomas or papillomas are abnormal growths, but they cannot spread outside of the breast to other organs. They are not life threatening.

If breast cancer cells have grown in the auxiliary lymph nodes, they are more likely to have spread to other organs of the body as well.

What are the causes of Breast Cancer?

Every woman is at risk for breast cancer and the incidence rates increase with age. Most breast cancers occur in women over the age of 50. The risk is especially high for women over age 60. Breast cancer occurs more often in Caucasian women than in African American or Asian women. Factors that increase a woman's chances of developing breast cancer include:

- Personal or family history of breast cancer
- Oral contraceptive use
- Breast density
- Genetic alterations in certain genes such as BRCA1 and BRCA2
- Longer exposure to estrogen either made by the body, taken as a drug, or delivered by a patch
- Late childbearing
- Radiation therapy before age 30
- Alcohol use
- Obesity and high-fat diets

DNA testing for BRCA1 and BRCA2

Genetic testing may indicate whether there is an increased chance of developing breast cancer. Women with a strong family history of breast cancer may choose to have a blood test to see if she has inherited a change in the BRCA1 or BRCA2 gene. Normally, the BRCA1 and BRCA2 genes help to prevent cancer by making proteins that keep cells from growing abnormally. However, if a person has inherited a mutated gene from either parent, chances of developing breast cancer increase. BRCA gene changes also increase the risk for ovarian cancer.

The BRCA1 (located on chromosome 17) and BRCA2 (located on chromosome 13) genes are inherited in an autosomal dominant manner. Children of affected individuals have a 50% chance of inheriting the genetic change themselves.

Recent studies have shown that about 10% of breast cancer cases are directly due to inherited gene changes in breast cancer related genes. Most of these gene changes (mutations) result from a change in

the BRCA1 and BRCA2 genes. About 50% to 60% of women with inherited BRCA1 or BRCA2 mutations will develop breast cancer by the age of 70.

A positive test result from a DNA test indicates that a person has inherited a known BRCA1 or BRCA2 gene alteration. This only provides risk information and cannot tell a person whether or when cancer might develop. Not all women who inherit an altered gene will develop breast cancer. A negative test result is uninformative as other gene alterations may be present that the BRCA1 or BRCA2 test may not detect.

Inherited gene changes of the p53 tumor suppressor gene can also increase a woman's risk of developing breast cancer. Testing the p53 gene is not part of the usual breast cancer genetic testing, but may be done by specialized cancer genetics centers.

Some aspects of a woman's lifestyle may affect her chances of developing breast cancer, such as regular exercise and diet. Scientists are still trying to learn whether having a miscarriage or an abortion increases the risk of breast cancer.

Breast cancer can not yet be prevented, but there are ways to reduce the risk of developing breast cancer, such as limiting alcohol consumption, maintaining a healthy weight, limiting the intake of high-fat foods, and regular exercise.

How is Breast Cancer detected?

Breast cancer can be detected at an early, treatable stage. Annual breast examinations by a medical professional are a required complement to annual screening mammography.

Mammography

A mammogram is a special type of x-ray. Most doctors recommend that women in their 40's and older have mammograms every year. A screening mammogram is the best tool available for finding breast cancer early before symptoms appear. Mammograms also can detect a breast lump before it can be felt. Small deposits of calcium may also be identified. Although most calcium deposits are benign a cluster of very tiny specks of calcium may be an early sign of cancer.

If an area of the breast looks suspicious on the screening mammogram, diagnostic mammograms may be needed. A mammogram cannot prove that an abnormal area is cancer. A biopsy may be advised.

Mammograms may sometimes miss some cancers that are present (false negative) and may find things that are not cancer (false positive).

Breast self-examination (BSE)

BSEs are a recommended component of every woman's breast health program. This exam allows a woman to become familiar with her breasts and what "normal" feels like to her. Although mammography is now the most frequent means of discovering breast cancer, women often find lumps and breast irregularities themselves through BSEs. Despite this, many women do not perform BSE's regularly. Provide link to how to perform a breast self-exam.

Clinical Breast Examination (CBE)

A clinical breast examination is an examination of your breasts by a health professional, such as a physician, nurse practitioner, nurse, or physician assistant.

Biopsy

A biopsy involves the removal of fluid or tissue from the breast in order to make a diagnosis. A biopsy is the only way to tell if cancer is really present. A fine-needle aspiration, needle biopsy, or surgical biopsy may be used.

Ultrasonography

Ultrasonography uses high-frequency sound waves and can often show whether a lump is a fluid-filled cyst or a solid mass. This exam is often used along with mammography.

The American Cancer Society recommends that women ages 40 and older have a screening mammogram every year. Between the ages of 20 and 39, women should have a clinical breast examination by a health professional every 3 years. After age 40, women should have a breast exam by a health professional every year. Women aged 20 or older should perform a BSE every month.

Following the American Cancer Society's guidelines for early detection improves the chances that breast cancer can be diagnosed at an early stage and treated successfully.

How is Breast Cancer treated?

Women with breast cancer now have many treatment options. A woman's treatment options depend on a number of factors. These factors include:

- Age
- Menopausal status
- General health
- Size and location of the tumor
- Stage of breast cancer
- Results of lab tests
- Size of the breast

Breast cancer may be treated with local and/or systemic therapy.

Local treatment

Local therapy is used to remove or destroy breast cancer in a specific area. Surgery and radiation therapy are local treatments. When breast cancer has spread to other parts of the body, local therapy may be used to control cancer in those specific areas.

Surgery is the most common treatment for breast cancer. Surgery may involve a lumpectomy, mastectomy, or reconstructive surgery and breast implant surgery.

Radiation therapy uses high-energy rays to kill cancer cells or shrink the size of a tumor in the breast or surrounding tissue. The radiation may be directed at the breast by a machine, called external radiation, or come from a radioactive material placed directly in the breast (implant radiation). External radiation is done in a hospital or clinic generally 5 days a week for several weeks. Implant radiation requires a longer hospital stay.

Systemic treatment

Systemic treatments are used to destroy or control cancer throughout the body. Chemotherapy, hormonal therapy, and biological therapy are types of systemic treatments. Some patients have systemic therapy to shrink the tumor before local therapy. Others have systemic therapy to prevent the cancer from coming back, or to treat cancer that has spread.

Chemotherapy involves the use of anticancer drugs to kill cancer cells. The drugs may be given in a pill or by injection. The drugs then enter the bloodstream and travel throughout the body.

Hormonal therapy keeps cancer cells from getting the hormones they need to grow. This treatment may involve the use of drugs that change the way hormones work, or surgery to remove the ovaries, which make female hormones.

Biological therapy is a treatment designed to enhance the body's natural defenses against cancer.

Some women at very high risk of developing breast cancer may choose to have a prophylactic mastectomy in order to remove the breasts before cancer develops.

Clinical trial

A woman may want to talk with her doctor about taking part in a clinical trial. Clinical trials are an important option for women with all stages of breast cancer. A clinical trial is a research study of new treatment methods that may help doctors find ways to improve health and cancer care. Studies are done with cancer patients to find out whether promising approaches to cancer prevention, diagnosis, and treatment are safe and effective. Each study tries to answer scientific questions and to find better ways to prevent, diagnose, or treat cancer.

Follow-up care

Regular follow-up exams are important after breast cancer treatment. Regular check-ups ensure that changes in health are noticed. Follow-up exams usually include examination of the breasts, chest, neck, and underarm areas, as well as periodic mammograms.

A woman who has had cancer in one breast should report any changes in the treated area or in the other breast to her doctor right away. Also, a woman who has had breast cancer should tell her doctor about other physical problems, such as pain, loss of appetite or weight, changes in menstrual cycles, unusual vaginal bleeding, or blurred vision. She should also report headaches, dizziness, shortness of breath, coughing or hoarseness, backaches, or digestive problems.

Information and Support Resources

- [American Cancer Society](#)
- [Oncolink](#)
- [GeneClinics](#)
- [National Cancer Institute](#)