

Breast Cancer - Diagnosis [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board \[2\]](#), 05/2014

ON THIS PAGE: You will find a list of the common tests, procedures, and scans that doctors can use to find out what's wrong and identify the cause of the problem. To see other pages, use the menu on the side of your screen.

Doctors use many tests to diagnose cancer and find out if the cancer has spread or metastasized to other parts of the body beyond the breast and the lymph nodes under the arm. Some tests may also help the doctor decide which treatments may be the most effective. For most types of cancer, a biopsy is the only way to make a definitive diagnosis of cancer. A biopsy is the removal of a small amount of tissue for examination under a microscope. See below for more information about the types of biopsies that can be performed. If a biopsy is not possible, the doctor may suggest other tests that will help make a diagnosis. Imaging tests may be used to find out whether the cancer has spread. This list describes options for diagnosing this type of cancer, and not all tests listed will be used for every person. Your doctor may consider these factors when choosing a diagnostic test:

- Age and medical condition
- Type of cancer suspected
- Signs and symptoms
- Previous test results

The series of tests needed to evaluate a possible breast cancer usually begins when a woman or her doctor discover a mass or abnormal calcifications on a screening mammogram, or a lump or nodule in the woman's breast during a clinical or self-examination. Less commonly, a woman might notice a red or swollen breast or a mass or nodule under the arm.

The following tests may be used to diagnose breast cancer or for follow-up testing after the cancer has been diagnosed. Not every person will need all of these tests.

Imaging tests

Diagnostic mammography [3]. Diagnostic mammography is similar to screening mammography except that more pictures of the breast are taken, and it is often used when a woman is experiencing signs, such as nipple discharge or a new lump. Diagnostic mammography may also be used if something suspicious is found on a screening mammogram.

Ultrasound [4]. An ultrasound uses high-frequency sound waves to create an image of the breast tissue. An ultrasound can distinguish between a solid mass, which may be cancer, and a fluid-filled cyst, which is usually not cancer. Ultrasounds are not used for screening.

MRI [5]. An MRI uses magnetic fields, not x-rays, to produce detailed images of the body. An MRI can also be used to measure the tumor's size. A special dye called a contrast medium is given into the patient's vein before the scan to help create a clear picture of the possible cancer. A breast MRI may be used after a woman has been diagnosed with cancer to check the other breast for cancer or to find out how much the disease has grown throughout the breast. It may also be used for screening, particularly along with mammography for some women with a high risk of developing breast cancer.

Surgical tests

Biopsy [6]. A biopsy is the removal of a small amount of tissue for examination under a microscope. Other tests can suggest that cancer is present, but only a biopsy can make a definite diagnosis. The sample removed during the biopsy is analyzed by a pathologist. There are different types of biopsies, classified by the technique and/or size of needle used to collect the tissue sample.

- A fine needle aspiration biopsy uses a thin needle to remove a small sample of cells.
- A core needle biopsy uses a wider needle to remove a larger sample of tissue. This is usually the preferred biopsy technique for finding out whether an abnormality on a physical examination or an imaging test is cancer. A vacuum-assisted biopsy removes several large cores of tissue. Local anesthesia, medication to block pain, is used to lessen a patient's discomfort during the procedure.
- Image-guided biopsy is used when a distinct lump cannot be felt, but an abnormality is seen with an imaging test, such as a mammogram. During this procedure, a needle is guided to the location with the help of an imaging technique, such as mammography, ultrasound, or MRI. A stereotactic biopsy is done using mammography to help guide the needle. A small metal clip may be put into the breast to mark where the biopsy sample was taken, in case the tissue is cancerous and more surgery is needed. This clip is usually titanium so it will not cause problems with future imaging tests, but check with your doctor before you have additional imaging tests. An image-guided biopsy can be done using a fine needle, core, or vacuum-assisted biopsy (see above), depending on the amount of tissue being removed. Imaging tests may also be used to help do a biopsy on a lump that can be felt, in order to help find the best location.
- A surgical biopsy removes the largest amount of tissue. This biopsy may be incisional, which is the removal of part of the lump, or excisional, which is the removal of the entire lump. Because surgery is best done after a cancer diagnosis has been made, a surgical biopsy is usually not the recommended way to diagnose breast cancer. Most often, non-surgical core needle biopsies are recommended to diagnose breast cancer. This means that only one

surgical procedure is needed to remove the tumor and to take samples of the lymph nodes.

If cancer is diagnosed, surgery is needed to remove the cancer in the breast. It is also needed to evaluate the lymph nodes for cancer in a procedure called a sentinel lymph node biopsy. Sometimes, treatment may be given before surgery, called neoadjuvant therapy, to shrink the cancer; see [Treatment Options](#) [7]. The goal of surgery is to achieve clear surgical margins, which means that there are no cancer cells at the edge of the tissue removed during surgery. If there is cancer in the lymph nodes, the cancer is called lymph node-positive breast cancer or node-positive; if there is no cancer in the lymph nodes, the cancer is called lymph node-negative breast cancer or node-negative. More information about lymph node evaluation can be found in [Stages](#) [8].

Tumor features. Examination of the tumor under the microscope is used to determine if it is invasive or in situ; ductal or lobular; how different the cancer cells look from healthy cells, called the grade; and whether the cancer has spread to the lymph nodes. The margins or edges of the tumor are also examined and their distance from the tumor is measured, which is called margin width.

Molecular testing of the tumor

The standard tests to further evaluate an invasive breast cancer include estrogen receptor (ER), progesterone receptor (PR), HER2, and Ki67 tests. ER is often measured for DCIS as well. These tests are used to determine the appropriate treatment plan to lower the chance that the cancer will return. Your doctor may recommend additional laboratory tests on your tumor sample to identify specific genes, proteins, and other factors unique to the tumor.

ER and PR. Breast cancer cells with these receptors depend on the hormones estrogen and/or progesterone to grow. The presence of these receptors helps determine both the patient's risk of recurrence and the type of treatment that is most likely to lower the risk of recurrence. Generally, hormonal therapy (see [Treatment Options](#) [7]) works well for ER-positive and/or PR-positive cancers, also called hormone receptor-positive cancers. About 75% to 80% of breast cancers have estrogen and/or progesterone receptors. Learn about [ER and PR testing recommendations from ASCO and the College of American Pathologists \(CAP\)](#) [9].

HER2. About 20% to 25% of breast cancers have an increase in the number of copies of a gene called the *human epidermal growth factor receptor 2 (HER2)*. This is called HER2-positive cancer. The gene makes a protein that is found on the cancer cell and is important for tumor cell growth; these types of cancers usually grow more quickly. The HER2 status helps determine whether a certain type of drug, such as trastuzumab (Herceptin), lapatinib (Tykerb), pertuzumab (Perjeta), or ado-trastuzumab emtansine (Kadcyla, also known as T-DM1), might help treat the cancer. In addition, about 50% of HER2-positive tumors also have hormone receptors and can benefit from both types of therapy. Several tests are available to test for HER2. [Read ASCO's and CAP's recommendations for HER2 testing for breast cancer](#) [10].

If a person's tumor does not have ER, PR, and/or HER2, the tumor is called triple-negative. Triple-negative breast cancers make up about 15% of invasive breast cancers and are the most common type diagnosed in women with *BRCA1* mutations. This type of breast cancer usually grows and spreads more quickly than hormone receptor-positive disease. Triple-negative breast

cancer seems to be more common among younger women, particularly younger black women.

Ki67. How quickly a cell divides into two cells, called tumor proliferation, can be measured in a tumor sample and is referred to as Ki67 or MIB1. How well chemotherapy works to treat a tumor has been linked with how quickly tumor cells grow and divide. Hormone receptor-positive cancers are most commonly slow growing with a low risk of recurrence if they are treated with hormonal therapy, but some are more rapidly growing with a higher risk of recurrence. In these cancers, chemotherapy may play an important role in reducing the risk of recurrence. In contrast, most HER2-positive and triple-negative cancers are fast growing and are treated with chemotherapy, with HER2 targeted therapy for a cancer that is also HER2-positive. In some situations, Ki67 may be used to help to plan treatment or to help estimate a patient's chance of recovery, but it is not used in many hospitals because the results are highly variable. Ki67 results depend on the laboratory doing the testing, the method of testing, and what part of the tumor is tested. Standardization of the testing methods and training appear to improve the results, so there is increasing interest in measuring tumor proliferation more routinely. In addition, molecular testing may be a more accurate measure of proliferation (see below).

Genetic testing of the tumor. Tests that look at the biology of the tumor are commonly used to understand more about a woman's breast cancer, particularly for cancers that have not spread to other organs. The tests below look at the genes in the tumor cells, not the genes a person inherits, to help predict the risk of cancer recurrence and to help choose the type of treatment, such as hormonal therapy or hormonal therapy plus chemotherapy. They can also help determine if radiation therapy would be helpful. Tests are used to look at several genetic measures, such as ER, PR, HER2, and tumor proliferation. These tests are usually performed after surgery, on a sample of the tumor that was removed (see [Treatment Options](#) [7]). A person with a higher risk of recurrence will likely need chemotherapy, while a person with a lower risk of recurrence can possibly avoid chemotherapy and its potential side effects. For more information about genetic tests, what they mean, and how the results might affect your treatment plan, talk with your doctor.

- Oncotype Dx[®] is a test that evaluates 16 cancer-related genes and five reference genes to estimate the risk of the cancer coming back in a place other than the breast and nearby lymph nodes, within 10 years after diagnosis for women with stage I or stage II (see [Stages](#) [8]), node-negative, ER-positive breast cancer treated with hormonal therapy alone. Results are mainly used to help make decisions about whether chemotherapy should be added to a person's treatment with hormonal therapy. Recent research suggests that this test may be useful to decide about use of chemotherapy in addition to hormonal therapy in node-positive disease in some situations.
- MammaPrint[®] is a similar test that uses information about 70 genes to predict the risk of the cancer coming back for early-stage, low-risk breast cancer. It is approved by the FDA for estimating the risk of recurrence in early-stage breast cancer, but it is not yet known if this test can predict whether chemotherapy will work.
- Additional tests are widely available with unknown impact on treatment choice and are being studied. Talk with your doctor about any test you are considering.

Blood tests

The doctor may also need to do several types of [blood tests](#) [11] to learn more about the cancer:

Complete blood count. A complete blood count (CBC) is used to measure the number of different types of cells, such as red blood cells and white blood cells, in a sample of a person's blood. It is done to make sure that your bone marrow is functioning normally.

Serum chemistry. These tests are often done to look at minerals in your blood, such as potassium and calcium, called electrolytes and specialized proteins called enzymes that can be abnormal if cancer has spread. However, many noncancerous conditions can cause changes in these tests, and they are not specific to cancer.

- Alkaline phosphatase is an enzyme that can be associated with disease that has spread to the liver, bone, or bile ducts.
- Blood calcium levels can be high if cancer has spread to the bone.
- Total bilirubin count and the enzymes alanine aminotransferase (ALT) and aspartate aminotransferase (AST) evaluate liver function. High levels of any of these substances can indicate liver damage, a sign that the cancer may have spread to that organ.

Hepatitis tests. These may be used to check for evidence of prior exposure to hepatitis B and/or hepatitis C. If you have evidence of an active hepatitis B infection, you may need to take a special medication to suppress the virus before you receive chemotherapy. Without this medication, the chemotherapy can help the virus to grow and cause damage to the liver.

Blood tumor marker tests. Serum tumor markers are proteins found in a person's blood that can be associated with cancer. High levels of a serum tumor marker may be due to cancer or a noncancerous condition. Tumor marker testing is not recommended for early-stage breast cancer because the markers are not usually high, but they may be useful to monitor the growth of recurrent or metastatic disease along with symptoms and imaging tests. Tumor markers should not be used to monitor for a recurrence, as it does not appear to improve a patient's chance of recovery. Learn more about [tumor markers for breast cancer](#) [12].

Additional tests

The tests your doctor recommends to evaluate whether the cancer has spread and its stage depends on your medical history, symptoms, how much the disease has grown in the breast and lymph nodes, and the results of your physical examination. Read [Stages](#) [8] for more information. Many of these tests may not be done until after surgery. These tests are generally only recommended for patients with later-stage disease.

- An x-ray is a way to create a picture of the structures inside of the body, using a small amount of radiation. A chest x-ray may be used to look for cancer that has spread from the breast to the lungs.
- A [bone scan](#) [13] may be used to look for spread of cancer to the bones. A radioactive dye or tracer is injected into a patient's vein, and then the scan is performed several hours later using a special camera. The tracer collects in areas of the bone that are healing, which occurs in response to damage from the cancer cells. The areas where the tracer collects appear dark, compared to healthy bone, which appears gray. Some cancers do not cause the

same healing response and will not show up on the bone scan. Areas of advanced arthritis or healing after a fracture will also appear dark.

- A computed tomography (CT or CAT) scan [14] may be used to look for tumors in organs outside of the breast, such as the lung, liver, bone, and lymph nodes. A CT scan creates a three-dimensional picture of the inside of the body with a special x-ray machine. A computer combines these images into a detailed, cross-sectional view that shows abnormalities including most tumors. A CT scan can also be used to measure the tumor's size and if it is shrinking with treatment. A contrast dye may be injected into a patient's vein before the scan to provide better detail.
- A positron emission tomography (PET) scan [15] may also be used to find out whether the cancer has spread to organs outside of the breast. Similar to a CT scan, a PET scan is a way to create pictures of organs and tissues inside the body. A small amount of a radioactive sugar substance is injected into a patient's vein. This sugar substance is then taken up by cells that use the most energy because they are actively dividing. Because cancer cells tend to use energy actively, they absorb more of the radioactive substance. A scanner then detects this substance to produce images of the inside of the body. Areas that are most active appear as bright spots, and the intensity of the brightness can be measured to better describe these areas. A combination PET/CT scan [16] may also be used to measure the size of tumors and to more accurately determine the location of the bright spots. A PET/CT scan will also show any abnormalities in the bone, similar to the bone scan.

After diagnostic tests are completed, your doctor will review all of the results with you. If the diagnosis is cancer, these results also help the doctor describe the cancer; this is called staging. If there are suspicious areas found outside of the breast, at least one area will be biopsied if possible to confirm the diagnosis of cancer.

The next section helps explain the different stages for this type of cancer. Use the menu on the side of your screen to select Stages, or you can select another section, to continue reading this guide.

Links:

[1] <http://www.cancer.net/cancer-types/breast-cancer/diagnosis>

[2] <http://www.cancer.net/about-us>

[3] <http://www.cancer.net/node/24584>

[4] <http://www.cancer.net/node/24714>

[5] <http://www.cancer.net/node/24578>

[6] <http://www.cancer.net/node/24406>

[7] <http://www.cancer.net/node/18626>

[8] <http://www.cancer.net/node/18625>

[9] <http://www.cancer.net/node/29856>

[10] <http://www.cancer.net/node/29831>

[11] <http://www.cancer.net/node/24716>

[12] <http://www.cancer.net/node/29851>

[13] <http://www.cancer.net/node/24410>

[14] <http://www.cancer.net/node/24486>

[15] <http://www.cancer.net/node/24648>

[16] <http://www.cancer.net/node/24565>