

## **Leukemia - Chronic Myeloid - CML - Diagnosis** [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 08/2014

**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 more about the disease. Some tests may also determine 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. If a biopsy is not possible, the doctor may suggest other tests that will help make a diagnosis. Imaging tests may also be used. This list describes options for diagnosing CML, 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 following tests may be used to diagnose or monitor CML:

**Blood tests** [3]. Many people are diagnosed with CML through a blood test called a complete blood count (CBC) before they have any symptoms. A CBC counts the number of different kinds of cells in the blood. A CBC is often done as part of a regular office visit. People with CML have high levels of white blood cells. When the CML is more advanced, there may also be low levels of red blood cells, a condition called anemia, and either elevated or decreased numbers of platelets.

**Bone marrow aspiration and biopsy** [4]. These two procedures are similar and often done at the same time to examine the bone marrow. Bone marrow has both a solid and a liquid part. A bone marrow aspiration removes a sample of the fluid with a needle. A bone marrow biopsy is the removal of a small amount of solid tissue using a needle. The sample(s) are then analyzed by a pathologist or a hematologist. A pathologist is a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease. A hematologist is a doctor who specializes in treating blood disorders. A cytogenetic analysis (see below) may also be done on the marrow samples. A common site for a bone marrow aspiration and biopsy is the iliac crest of the pelvic bone (see the [Medical Illustrations](#) [5] section), which is located in the

lower back by the hip. The skin in that area is usually numbed with medication beforehand, and other types of anesthesia (medication to block awareness of pain) may be used.

**Molecular testing.** Your doctor may recommend testing the leukemia cells for specific genes, proteins, and other factors unique to the leukemia. Results of these tests will help determine whether your treatment options include a type of treatment called targeted therapy (see the [Treatment Options \[6\]](#) section).

Cytogenetics is a type of genetic testing that is used to analyze a cell's chromosomes, including the number, size, shape, and arrangement of the chromosomes. Occasionally, this test can be done on the peripheral or circulating blood when the CML is first diagnosed, but immature blood cells that are actively dividing need to be used. Because of this, a bone marrow sample (see above) is usually the best way to get a sample for testing.

After treatment begins, cytogenetic and/or molecular testing is repeated on another bone marrow sample to find out if there are fewer cells with the Philadelphia chromosome. All people with CML have the Philadelphia (Ph+) chromosome and the *BCR-ABL* fusion gene (described in the [Overview \[7\]](#) section), so the presence of these changes confirms the diagnosis. For a small number of patients, increased blood cell counts may suggest CML, but the patients do not have the Philadelphia chromosome or the *BCR-ABL* fusion gene. In this situation, they do not have CML but instead have a different type of chronic myeloproliferative disease, a disease in which there are too many red blood cells, white blood cells, or platelets. Treatment of these diseases is different from that of CML.

Cytogenetic testing for CML is used to monitor how well treatment is working and if it is reducing the number of cells with the Philadelphia chromosome. The following tests are sometimes used with cytogenetic testing:

- Fluorescence in situ hybridization (FISH) is a test used to detect the *BCR-ABL* gene and to monitor the disease during treatment. This test does not require dividing cells and can be done using a blood sample or bone marrow cells. This test is a more sensitive way to find CML than the standard cytogenetic tests that identify the Philadelphia chromosome.
- Polymerase chain reaction (PCR) is a DNA test that can find the *BCR-ABL* fusion gene and other molecular abnormalities. PCR tests may also be used to monitor how well treatment is working. This test is quite sensitive and, depending on the technique used, can find one abnormal cell mixed in with approximately 1 million normal cells. This test typically uses a blood sample rather than bone marrow cells.

**Imaging tests.** Doctors may use imaging tests to determine if the cancer is affecting other parts of the body. For example, a [computed tomography \(CT or CAT\) scan \[8\]](#) or [ultrasound \[9\]](#) examination is sometimes used to look at and measure the size of the spleen in patients with CML.

- A CT scan creates a three-dimensional picture of the inside of the body with an x-ray machine. A computer then combines these images into a detailed, cross-sectional view that shows any abnormalities. Sometimes, a special dye called a contrast medium is given before the scan to provide better detail on the image. This dye can be injected into a patient's vein or given as a pill to swallow.

- An ultrasound uses high-frequency sound waves to create a picture of the inside of the body.

After diagnostic tests are done, your doctor will review all of the results with you. If the diagnosis is CML, these results also help the doctor describe the phase of the disease.

*The next section helps explain the different phases of CML. Use the menu on the side of your screen to select Stages, or you can select another section, to continue reading this guide.*

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**Links:**

[1] <http://www.cancer.net/cancer-types/leukemia-chronic-myeloid-cml/diagnosis>

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

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

[4] <http://www.cancer.net/node/24409>

[5] <http://www.cancer.net/node/19108>

[6] <http://www.cancer.net/node/19113>

[7] <http://www.cancer.net/node/19106>

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

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