

## [Leukemia - Acute Lymphocytic - ALL - Diagnosis](#) [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 01/2016

**ON THIS PAGE:** You will find a list of common tests, procedures, and scans that doctors use to find the cause of a medical problem. To see other pages, use the menu.

Doctors use many tests to find, or diagnose, cancer. They also do tests to learn if cancer has spread to another part of the body from where it started. If this happens, it is called metastasis. For example, imaging tests can show if the cancer has spread. Imaging tests show pictures of the inside of the body. Doctors may also do tests to learn which treatments could work best.

For most types of cancer, a biopsy is the only sure way for the doctor to know whether an area of the body has cancer. In a biopsy, the doctor takes a small sample of tissue for testing in a laboratory. If a biopsy is not possible, the doctor may suggest other tests that will help make a diagnosis.

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:

- The type of cancer suspected
- Your signs and symptoms
- Your age and medical condition
- The results of earlier medical tests

The following tests may be used to diagnose ALL:

- **Blood tests.** If the doctor believes a person has ALL based on the symptoms, he or she will examine the levels of different types of cells in the patient's blood through a test called a [complete blood count \(CBC\)](#) [3]. Low levels of red blood cells and platelets and high levels of white blood cells are common in people with ALL but can also be a sign of other medical problems. In addition, the blood may be examined under a microscope to find out if there are lymphoblasts or other abnormal cells in the blood.
- **Bone marrow aspiration and biopsy.** If the blood test shows abnormalities in the number or appearance of the white blood cells, a [bone marrow aspiration and biopsy](#) [4] will be done. These 2 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. If blood tests (see above) indicate AML, it is better to have the bone marrow aspiration and biopsy at the hospital where treatment will be given so the test does not need to be done again.

A pathologist analyzes the sample(s) removed during a bone marrow aspiration and biopsy. A pathologist is a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease. The sample is used to look for leukemia, and molecular testing and immunophenotyping (see below) may also be done. A common site for a bone marrow aspiration and biopsy is the pelvic bone, 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 the awareness of pain) may be used.

- **Other testing.** Your doctor may recommend running laboratory tests on the leukemia cells to identify specific genes, proteins, and other factors unique to the leukemia. Results of these tests will help decide whether your treatment options include a type of treatment called targeted therapy (see [Treatment Options](#) [5]).
  - Flow cytometry, also called immunophenotyping, and cytochemistry are tests that use chemicals or dyes on the cancer cells to provide information about the leukemia and its subtype. ALL cells have distinctive markers on their surface called cell surface proteins. The pattern of these markers is called the immunophenotype. These tests are used to distinguish ALL from healthy blood cells and from other types of leukemia, which can also involve lymphocytes. Both tests can be done from a blood sample.

- Karyotyping, or cytogenetics, is a test that is used to examine a cell's chromosomes, long pieces of DNA that contain genes. People with ALL may have specific chromosomal changes, including the addition or loss of certain chromosomes, as well as translocations, which means that parts of 1 chromosome have been moved to another chromosome. These changes can be seen under a microscope using special methods. Approximately 25% of patients have a normal karyotype when diagnosed with ALL.

About 20% to 30% of adults with ALL have a particular change in their chromosomes called the Philadelphia chromosome. The Philadelphia chromosome is an example of a small chromosome caused by a translocation. In this instance, it means that genetic material from chromosome 9 breaks off and attaches to chromosome 22 [t(9;22)]. Then, 2 specific genes called *BCR* and *ABL* are brought together to make 1 gene called *BCR-ABL*. Some people may have other types of translocations. For example, many children with ALL have a translocation between chromosomes 12 and 21 [t(12;22)]. These genes are called *TEL* and *AML1*.

- Mutations in genes that are too small to be seen with a microscope and cannot be found with cytogenetic tests have been found using tests called molecular assays. For example, patients with changes in the *IKZF1* gene have a poorer long-term prognosis, which is the chance of recovery. Therefore, testing for these changes during diagnosis helps determine a patient's treatment options.
- **Lumbar puncture (spinal tap).** A lumbar puncture is a procedure in which a doctor uses a needle to take a sample of cerebral spinal fluid (CSF) to look at the make up of the fluid and to find out if it contains cancer cells or blood. Because ALL tends to spread to the CSF, the fluid that flows around the brain and spinal cord, lumbar punctures are done regularly during ALL treatment, and chemotherapy (see [Treatment Options](#) [5]) may be given in the CSF. Doctors generally give an anesthetic to numb the lower back before the procedure.
- **Imaging tests.** The following imaging tests may be used to learn more about the cause of symptoms or to help diagnose infections in patients with ALL.
  - A [computed tomography \(CT or CAT\) scan](#) [6]. A test that creates a 3-dimensional picture of the inside of the body using x-rays taken from different angles.
  - A [magnetic resonance imaging](#) [7] (MRI). A test that uses magnetic fields, not x-rays, to produce detailed images of the body.

Sometimes, a special dye called a contrast medium is given before these scans to provide

better detail on the image. This dye can be injected into a patient's vein or given as a pill to swallow.

After diagnostic tests are done, your doctor will review all of the results with you. If the diagnosis is ALL, results can also help the doctor describe the disease; this is called [classification](#) [8].

*The [next section in this guide is Subtypes and Classification](#) [8]. It explains the system doctors use to describe the extent of the disease. Or, use the menu to choose another section to continue reading this guide.*

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### **Links**

[1] <http://www.cancer.net/cancer-types/leukemia-acute-lymphocytic-all/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/19044>

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

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

[8] <http://www.cancer.net/node/19043>