Acute Lymphocytic Leukemia (ALL): Diagnosis

How is acute lymphocytic leukemia (ALL) diagnosed?

If your healthcare provider thinks you might have ALL, you will need certain exams and tests to confirm your diagnosis. Your healthcare provider will ask you about your health history, your symptoms, risk factors, and family history of disease. Your healthcare provider will also give you a physical exam.

What tests might I need?

You may have 1 or more of these tests:

Blood tests

These are often the first tests done to diagnose ALL. Blood is taken from your arm or hand with a small needle. The blood is then sent to a lab and tested in many ways (see below).

Bone marrow aspiration and biopsy

This procedure is done by taking out small amounts of bone marrow. This is the thick liquid in the center of your bones. Bone marrow samples are usually taken from the back of the hip (pelvic) bone. First, the aspiration is done. An area over back of your hip is numbed. A long, hollow needle is then put through your skin and into your hip bone. A syringe is used to pull out a small amount of liquid bone marrow. You may have some brief pain when the marrow is removed. A bone marrow biopsy is usually done right after the aspiration. A small piece of bone and marrow is removed with a slightly larger needle that's pushed down into the bone. The biopsy may also cause some brief pain. The bone marrow is then checked for leukemia cells and tested in many ways (see below).

Spinal tap (lumbar puncture)

This procedure is done to look for leukemia cells in your spinal fluid. It's done by putting a thin needle in the space between 2 bones of your spine to take out a small amount of fluid. Numbing medicine is used to make it more comfortable for you.

How blood or bone marrow is tested

The tests done on blood or bone marrow samples may include:

- **Complete blood count (CBC).** This is often the first test done. It measures the numbers of different types of cells in your blood. People with ALL typically have too many early (immature) forms of white blood cells, called lymphoblasts or blasts.
- **Immunophenotyping.** These tests are done on blood, bone marrow, or both. They measure the types and amounts of proteins called antigens on or in the leukemia cells. This can be used to see if you have ALL and which subtype you have. It takes a few days to get the results of these tests.
- **Cytogenetics.** For this test, cells are grown in a lab for a week or more. The chromosomes inside the cells are then stained with special dyes and viewed with a microscope. Major changes in the chromosomes can often be seen with this test. But smaller changes may not be visible.
- **Fluorescence in situ hybridization (FISH).** This test is used to find abnormal changes in the chromosomes and genes of leukemia cells. The cells in the sample are stained with fluorescent dyes that will only attach to certain parts of chromosomes. The cells are then viewed with a microscope using a special light. This test can find some abnormal chromosome and gene changes that can't be seen with standard cytogenetic testing. It's also a quicker test.

- **Polymerase chain reaction (PCR).** This is a very sensitive test that can find and measure some genetic mutations and chromosomal changes that are too small to be seen with a microscope. PCR testing increases or amplifies the amount of genetic material in a sample so that it can be easier to find. This test can find small levels of genetic or chromosomal changes that other tests can't find. This test may be done on bone marrow or a blood sample.

Finding the gene changes for your ALL cells can help decide your treatment. For instance, about 1 out of 4 people with ALL have the **Philadelphia chromosome** in their leukemia cells. This chromosome contains the abnormal gene BCR-ABL1 that helps the leukemia cells grow. Those leukemia cells can be treated with medicines that target cells with this gene change.

Getting your test results

When your healthcare provider has the results of your tests, they will contact you with the results. Your healthcare provider will talk with you about other tests you may need if ALL is found. Make sure you understand the results and what follow-up you need.