Acute Myeloid Leukemia (AML): Diagnosis

How is acute myeloid leukemia (AML) diagnosed?

If your healthcare provider thinks you might have AML, you will need certain exams and tests to make sure. 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 need one or more of these tests:

Blood tests

Blood is taken from your arm or hand with a needle. The blood is then tested in many ways (see below).

Bone marrow aspiration and biopsy

This procedure is done by taking out small amounts (samples) of bone marrow. Bone marrow is the thick, spongy liquid inside bones. Samples are usually taken from the back of the hip (pelvic) bone. For the bone marrow aspiration, the skin over the hip is numbed. A long, hollow needle is then put through the skin and into the hip bone. A syringe is used to pull out a small amount of bone marrow. You may have some brief pain when the marrow is removed. A bone marrow biopsy is usually done just 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 sent to a lab where it's checked for leukemia cells and tested in many ways (see below).

How blood or bone marrow is tested

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

- **Complete blood count (CBC).** This test measures the numbers of different types of cells in your blood. People with AML usually have too many early (immature) white blood cells, called blasts. There might be low numbers of red blood cells and platelets.
- **Immunophenotyping.** This test measures the types and amounts of proteins called antigens on the surface of the leukemia cells. This can be used to see if you have AML, and to find out which subtype you have. Results of this test may take several days.
- **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 another way to look for changes in chromosomes. 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 chromosome 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 even very low levels of leukemia cells in a test sample. It works by increasing the amount of genetic material in the sample so that it can be found. This test can find small levels of chromosome changes that other tests can’t find.
- **Next generation sequencing.** This is a broad term to describe multiple different sequencing methods used to find abnormal cancer cells. Lab tests are used to look at the order (sequence) of DNA and RNA

cells. Comparisons can then be made in the order of cancer cells with those of normal cells to find genetic changes. These tests can help determine prognosis and types of treatment used.

Finding the gene changes in your leukemia cells helps your healthcare provider know if you have AML and the exact type of AML you have.

Getting your test results

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