

[Home](#) > [Types of Cancer](#) > [Leukemia - Acute Myeloid - AML - Childhood](#) > [Leukemia - Acute Myeloid - AML - Childhood - Diagnosis](#)

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## [Leukemia - Acute Myeloid - AML - Childhood - Diagnosis](#)

### [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 07/2015

**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 leukemia. Some tests may also help the doctor choose the treatment that may be the most effective. Not all tests listed will be used for every child. Your child's doctor may consider these factors when choosing a diagnostic test:

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

When a child has [signs and symptoms](#) [3] of leukemia, the doctor will ask about the child's medical history and perform a physical examination. In addition, the following tests may be used

to diagnose AML:

- **Blood tests.** [Complete blood count \(CBC\)](#) [4] and cell type (called differential count) are blood tests done to count the number of each type of blood cell under a microscope and to check if they look abnormal.
- **[Bone marrow aspiration and biopsy](#)** [5]. 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.

A pathologist then analyzes the samples. 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 child's skin in that area will be numbed with medication beforehand, and other types of anesthesia (medication to block the awareness of pain) may be used. From this test, the doctor can find out whether the child has leukemia and, if so, what type of leukemia it is.

- **Molecular and genetic testing.** Your doctor may recommend running laboratory tests to identify specific genes, proteins, and other factors involved in the leukemia. Examining the genes in the leukemia cells is important because AML can be caused by a buildup of mistakes (also called mutations) in the cell's genes. Identifying these mistakes helps diagnose the specific [subtype](#) [6] of AML and choose treatment options. In addition, the results of those tests can also be used to monitor how well treatment is working. Listed below are the more common molecular or genetic tests used for AML.
- Cytochemical and immunohistochemical tests are laboratory tests that are used to find out the exact subtype of AML. In cytochemical tests, a special dye is used that stains the different types of leukemia cells differently based on the chemicals in the cells. For AML, immunohistochemical tests and a test called flow cytometry are used to find markers on the surface of the leukemia cells. The different subtypes of leukemia have different and unique combinations of cell surface markers.
- Cytogenetics is a way to look at a cell's chromosomes (long strands of genes) through a microscope to analyze the number, size, shape, and the arrangement of the chromosomes to find genetic changes in the leukemia cells. Sometimes, a chromosome breaks off and reattaches to another chromosome, which is called a translocation. Other times, part of a chromosome is missing, called a deletion. A chromosome can also be made more than once, most often called a trisomy. Some subtypes of leukemia are caused by chromosome translocations, deletions, or trisomies.

Knowing if there are certain translocations may help doctors determine the AML subtype and plan the best treatment. Fluorescence-in-situ-hybridization (FISH) is a way to detect chromosome changes in cancer cells and is being used more often to help diagnose and determine the subtype of leukemia. This test is done on tissue removed during a biopsy or aspiration (see above).

- The molecular genetics of leukemia cells can also be used to help find out if a person needs more or less chemotherapy and/or stem cell transplantation (see the [Treatment Options](#) [7] section). The goal of this type of testing is to look for very small genetic mutations, called sub-microscopic mutations. People who have the Flt3 (pronounced flit 3) genetic mutation, called an internal tandem duplication (ITD), and specifically those with the mutation on both versions of the gene, have a high risk that the cancer will come back after treatment.

For children with this type of AML, the use of stem cell transplantation may lengthen their lives when used after the first complete remission (see the [Treatment Options](#) [7] section). There are also new drugs being tested that target Flt3-ITD positive cells to find out if the drugs can better treat the leukemia. At the same time, research has shown that children with two leukemia cell gene mutations called nucleophosmin-1 (NPM1) and CEBP $\alpha$  have a better prognosis (chance of recovery) than those without these mutations. If a child has these mutations, the doctor may recommend chemotherapy without stem cell transplantation.

- Recently, highly advanced tools that can look at a person's entire genetic make-up, called whole genome testing and whole exome testing, have been developed. These methods of testing are still emerging but have been used to look for genetic mutations that can lead to cancer. However, these tests are still generally only available in research studies where they are being used to find out if using these tests improves diagnosis, treatment, and cure.

After diagnostic tests are done, your child's doctor will review all of the results with you. If the diagnosis is AML, these results also help the doctor describe the cancer and determine the subtype.

*The [next section in this guide is Subtypes](#) [6] and it helps explain the different subtypes of AML. Or, use the menu on the side of your screen to choose another section to continue reading this guide.*

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## Links

[1] <http://www.cancer.net/cancer-types/leukemia-acute-myeloid-aml-childhood/diagnosis>

- [2] <http://www.cancer.net/about-us>
- [3] <http://www.cancer.net/node/19055>
- [4] <http://www.cancer.net/node/24716>
- [5] <http://www.cancer.net/node/24409>
- [6] <http://www.cancer.net/node/19057>
- [7] <http://www.cancer.net/node/19058>