



CAP-ASH Guideline for the Initial Diagnosis of Acute Leukemia

A RESOURCE FOR PATIENTS

Patients and caregivers facing a diagnosis of acute leukemia need to navigate many sources of information.

The College of American Pathologists (CAP) and the American Society of Hematology (ASH) provide practical guidance for patients and caregivers coping with an initial diagnosis of acute leukemia. Based on a clinical guideline the CAP and ASH recently developed, this checklist will help ensure that you receive the best treatment for your unique situation.

DIAGNOSTIC CHECKLIST: Seven Steps to Support Diagnosis and Treatment

1. Make sure your doctors collect and incorporate all critical data into your medical record. This helps guide an integrated, comprehensive approach to treatment. The following is a list of data that should be included in your record:

- Clinical data, such as age, gender, family history of blood disorders, etc.
- Physical exam findings or findings from any imaging tests or scans.
- Results of blood tests.
- Evaluation of the material obtained from a bone marrow test, such as a biopsy or aspiration—ideally, the same doctor should interpret bone marrow aspirates and core biopsies.

2. Expect doctors to perform specialized tests to accurately diagnose and categorize acute leukemia.

Blood and bone marrow tests should include these evaluations:

- Cytogenetic analysis—The pathologist on your care team uses this test to examine chromosomes inside the cells, including inside leukemia cells, under a microscope.
- Molecular genetic analysis—Various laboratory techniques can help the pathologist pinpoint the precise genetic abnormalities occurring in patients with acute leukemia. These techniques include polymerase chain reaction (PCR), next-generation sequencing (NGS), and fluorescence in situ hybridization (FISH). The pathologist selects and performs the appropriate technique to help your care team understand your disease.
- Flow cytometry—This analysis is used to distinguish different types of cells and helps to confirm the type of leukemia. Your clinical team may use this analysis for later comparisons to track treatment progress.
- Cytochemical studies—The pathologist can visualize chemical abnormalities to assist in diagnosis.

Blood and bone marrow tests may also include:

- Cryopreserved cells or nucleic acid—These samples may be obtained and prepared for additional molecular or genetic analyses to be used later in your treatment.

Other evaluations should include:

- Cerebrospinal fluid (CSF)—Evaluation of this fluid is important for patients with confirmed acute lymphoblastic leukemia (ALL) receiving intrathecal therapy (therapy through the spine and spinal fluid).

3. Make sure all laboratory testing will be performed in a laboratory that complies with regulatory and/or accreditation requirements. [Click here to find a CAP-accredited laboratory near you.](#)

4. Support the doctors' ability to preserve cells or specimens from your tests. From these preserved samples, the pathologist on your care team can sometimes perform future evaluations to predict the next step in treatment. Also, the samples may be beneficial should new treatment options become available.

5. If your doctors refer you to another hospital or treatment center with specialized expertise in managing acute leukemia, whenever possible, defer invasive procedures, such as bone marrow biopsies, to the hospital or treatment center to which you've been referred.

- Be sure the referring doctors send the new treatment center all of your laboratory results, data, and information, including pathology slides. Any pending test results should also be forwarded as soon as available.

6. Your pathologist will perform genetic testing to help other members of your care team determine which treatment options are best for you. Genetic markers (genes or parts of DNA used to help identify genetic diseases) should be evaluated according to the table below.* These tests are performed in the laboratory and do not require a procedure for each genetic marker.

ACUTE LEUKEMIA TYPE	PATIENT AGE	TEST FOR THESE GENETIC MARKERS
B-cell precursor acute lymphoblastic leukemia (B-ALL)	Pediatric	<input type="checkbox"/> <i>BCR-ABL1</i> <input type="checkbox"/> <i>ETV6-RUNX1</i> <input type="checkbox"/> <i>KMT2A (MLL)</i> translocations <input type="checkbox"/> <i>iAMP21</i> <input type="checkbox"/> Trisomy 4 and 10 May also test for <input type="checkbox"/> Mutational analysis that includes (but does not limit to) <i>PAX5, JAK1, JAK2, and/or IKZF1</i> <input type="checkbox"/> <i>CRLF2</i> overexpression
B-ALL	Adult	<input type="checkbox"/> <i>BCR-ABL1</i> May also test for <input type="checkbox"/> <i>KMT2A (MLL)</i> rearrangement <input type="checkbox"/> Mutational analysis that includes (but does not limit to) <i>PAX5, JAK1, JAK2, and/or IKZF1</i> <input type="checkbox"/> <i>CRLF2</i> overexpression
T-cell acute lymphoblastic leukemia (T-ALL)	Adult and Pediatric	May also test for <input type="checkbox"/> Mutational analysis for <i>NOTCH1</i> and/or <i>FBXW7</i>
AML—any type	Adult and Pediatric	<input type="checkbox"/> <i>FLT3-ITD</i> May also test for <input type="checkbox"/> Mutational analysis that includes (but does not limit to) <i>IDH1, IDH2, TET2, WT1, DNMT3A, and/or TP53</i>
AML with confirmed core binding factor (AML-CBF)	Adult	Also test for <input type="checkbox"/> <i>KIT</i> mutation
AML-CBF	Pediatric	May also test for <input type="checkbox"/> <i>KIT</i> mutation
AML with suspicion of acute promyelocytic leukemia (APL)	Adult and Pediatric	Also perform <input type="checkbox"/> Rapid detection of <i>PML-RARA</i> <input type="checkbox"/> Coagulation studies for DIC
Patients other than those with confirmed core binding factor AML, APL, or AML with myelodysplasia-related cytogenetic abnormalities	Adult and Pediatric	Also test for <input type="checkbox"/> Mutational analysis for <i>NPM1, CEBPA, and RUNX1</i>

7. Keep your medical records up to date.

- All performed tests, test results, and related commentary from doctors should be entered into your medical records. Hospital medical records are accessible to patients upon request.

* Tests displayed in the table were derived from the “Initial Diagnostic Workup of Acute Leukemia: Guideline from the CAP and ASH,” which is intended to assist physicians and patients in decision making. It is the responsibility of the treating physician or other health care provider, relying on independent experience and knowledge, to determine the best course of treatment for the patient. Because of the rapid advances in genetics, changes may occur to this list. Please consult with your physician to determine which tests are most appropriate for you.

References: Arber DA, Borowitz MJ, Cessna M, et al. Initial diagnostic workup of acute leukemia: guideline from the College of American Pathologists and the American Society of Hematology. *Arch Pathol Lab Med.* 2017; [141\(10\):1342-1393](https://doi.org/10.5858/141.10.1342-1393).