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CLINICAL AND LABORATORY CHARACTERISTICS OF ACUTE LEUKEMIA

Zaynutdinova D.L., Sabitkhodjaeva S.O., Yusupova M.I.
Tashkent State Medical University

Abstract: Acute leukemia (AL) is a hematologic neoplasm characterized by excessive proliferation of undifferentiated blast cells in the bone marrow and peripheral blood. Understanding the relationship between laboratory and clinical manifestations of the disease is crucial for assessing prognosis, selecting treatment strategies, and evaluating therapeutic response. This article analyzes the clinical significance of hematologic parameters, immunophenotypic markers, cytogenetic, and molecular alterations in patients with AL, based on scientific studies published between 2020 and 2025.

Keywords: acute leukemia, blasts, leukemia, thrombocytopenia, anemia

Relevance of the problem: Acute leukemia is a clonal (oncologic) disease of hematopoietic cells originating in the bone marrow due to mutations in hematopoietic stem cells. As a result of these mutations, the affected cell lineage loses its ability to differentiate into mature blood cells. It is one of the most rapidly progressing and life-threatening malignancies in hematologic oncology, often resulting in severe clinical outcomes.

Globally, leukemia ranks 13th among newly diagnosed cancers and constitutes a significant portion of hematologic malignancies. Although the incidence varies by region, acute forms of leukemia are widely recognized as one of the leading causes of cancer-related mortality. The World Health Organization (WHO) reports hundreds of thousands of new cases annually, the majority being acute myeloid leukemia (AML) and acute lymphoblastic leukemia (ALL).

In developed countries, the rate of diagnosis is higher, likely due to greater access to diagnostic modalities such as bone marrow biopsy and imaging performed under sedation. However, underreporting in some regions suggests that the global incidence may be underestimated. In countries with stable economies, especially among elderly patients, early bone marrow aspiration under sedation is routinely performed, improving diagnostic accuracy. Worldwide, approximately 2.43 million people live with leukemia, with around 300,000 new cases annually, accounting for 3.2% of new cancer cases and 3.9% of cancer deaths.

Leukemia is the most common oncologic disease in children, comprising more than 9% of all pediatric cancers. Incidence rates vary markedly by region, with the highest rates reported in Australia, New Zealand, North America, and Western Europe, and the lowest in West Africa.

In Uzbekistan, acute leukemias remain a major challenge in oncohematology. Approximately 2,500 oncohematologic diseases are diagnosed annually. Although specific epidemiological data on acute leukemia prevalence are limited, the estimated incidence is about 5 cases per 100,000 population per year.

Diagnosis of leukemia relies on detecting abnormal cells through CBC, bone marrow aspiration for morphological, cytochemical, immunophenotypic, cytogenetic, and molecular analyses, along with instrumental evaluation of internal organs. Laboratory testing, including CBC, peripheral blood smear, biochemical tests, immunophenotyping, and genetic analyses, is essential for correlating clinical and laboratory findings(9).

1. Complete blood count and peripheral blood smear

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CBC determines the percentage of blasts, as well as anemia, thrombocytopenia, and neutropenia associated with leukemia. For instance, anemia is observed in 96% of patients and thrombocytopenia in over 80% (4). PBS provides morphological analysis of blasts, differentiating lymphoid and myeloid forms, thus assisting in leukemia subtype classification (9). Studies have revealed a negative correlation between hemoglobin levels and disease severity, and a positive correlation between leukocyte/blast counts and disease progression (7).

2. Clinical relevance of biochemical indicators

Serum lactate dehydrogenase (LDH) serves as an index of cellular turnover and proliferation. A Chinese cohort study demonstrated that LDH ≥ 570 U/L was associated with increased 60-day mortality (10). Additionally, oxidative stress biomarkers have been investigated - leukemia patients exhibit decreased antioxidant levels and elevated MDA concentrations (3). Inflammatory markers such as CRP and ESR are related to infectious complications in leukemia (7).

3. Immunophenotypic markers

In acute myeloid leukemia (AML), leukemic stem cells (LSCs) play a key role in disease relapse. LSCs typically exhibit a $CD34^{+}/CD38^{-}$ phenotype and express several specific markers, including CD33, CD96, CD123, CD90, and DAPK.

A 2024 clinical study found significantly elevated CD96 expression in AML patients compared to controls ($p = 0.008$) and a strong correlation with chemotherapy response ($p < 0.001$) (Soliman et al., 2024). This marker indicates LSC resistance to therapy and serves as a poor prognostic indicator (8).

4. Cytogenetic and molecular alterations

Cytogenetic and molecular testing are essential for prognosis assessment, risk stratification, and treatment strategy selection.

- FLT3-ITD mutations: associated with a high relapse rate and poor prognosis. FLT3 inhibitors (midostaurin, quizartinib, gilteritinib) are used in combination with chemotherapy.
- NPM1 mutations: generally predict favorable outcomes, particularly in the absence of FLT3-ITD. Patients achieving complete remission may receive reduced post-remission therapy; those with high-risk features are candidates for hematopoietic stem cell transplantation (HSCT).
- BCR-ABL1 (Philadelphia chromosome): in ALL, tyrosine kinase inhibitors (imatinib, dasatinib, ponatinib) combined with chemotherapy improve remission and survival rates.
- IDH1/IDH2 mutations (in AML): targeted therapies such as ivosidenib, olutasidenib (IDH1), and enasidenib (IDH2) are effective in relapsed or refractory cases.
- TP53 mutations: linked to poor prognosis and chemoresistance. Hypomethylating agents (azacitidine, decitabine) or investigational therapies may be used (5).

5. Bone marrow examination

Bone marrow aspiration (BMA) and biopsy (BMB) remain the gold standard for leukemia diagnosis. These methods assess the proportion of blasts, cellular origin, and maturation stage. Flow cytometry and immunohistochemistry (IHC) aid in precise classification based on surface marker expression (9).

6. Clinical–laboratory correlation

Clinically, advanced-stage manifestations - organomegaly, lymphadenopathy, mucocutaneous or gastrointestinal bleeding, and leukostasis - often correlate with laboratory abnormalities. Elevated leukocyte and blast counts increase leukostasis risk and disease severity (7).

- Patients with leukocyte count $<50\times10^{9}$/L often show low MRD-33 - indicating good prognosis.

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- 9p21 deletion correlates with thrombocytopenia (<50×10⁹/L) and high MRD-33 - a poor prognostic marker.
- Decreased LDH levels are associated with unfavorable mutations.
- Normal cytogenetics correlate with low MRD-33, low LDH, and platelet counts >50×10⁹/L - favorable prognosis (5).

In ALL, risk assessment incorporates the following factors:

- Clinical indicators: age, WBC count;
- Cytogenetic/genomic factors: favorable or adverse translocations;
- Therapeutic response: morphological or MRD status during induction.

These factors collectively stratify patients into low-, intermediate-, and high-risk groups, guiding individualized treatment approaches (1).

Conclusion. Acute leukemias are among the most complex and aggressive malignancies of the hematopoietic system. Recent research demonstrates that correlations between clinical and laboratory findings are critical for evaluating disease course, prognosis, and therapeutic response.Parameters such as CBC, LDH, CRP, immunophenotypic markers ($CD34^+/CD38^-$, CD33, CD96, CD123), and molecular mutations (FLT3, NPM1, TP53, IDH1/2, BCR-ABL1) assist in risk stratification and personalized treatment planning.Markers like CD96 and MRD are emerging as novel prognostic biomarkers for therapy response - decreased MRD levels predict favorable outcomes and prolonged remission, whereas elevated CD96 expression is linked to poor prognosis.Integration of laboratory, immunophenotypic, cytogenetic, and molecular data in clinical practice enhances early diagnosis, optimizes treatment efficacy, and improves patient quality of life.

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