

Acute Lymphoblastic Leukemia FISH Panels

Last Literature Review: July 2020 Last Update: May 2025

Acute lymphoblastic leukemia (ALL) is an aggressive leukemia of B- or T-lineage immature lymphoid cells. B-cell ALL (B-ALL) is primarily a disease of early childhood. Fluorescence in situ hybridization (FISH) testing identifies rearrangements in specific genes used in risk stratification and treatment decisions for children and adults newly diagnosed with B-ALL.

Disease Overview

Incidence

B-ALL occurs in 1.6/100,000 individuals per year, and is the most common leukemia in childhood.¹

Symptoms

Bone marrow failure (e.g., anemia, thrombocytopenia, leukopenia) and constitutional symptoms (e.g., fever, lethargy, weight loss) are common.² In children, joint or extremity pain may be the only presenting symptom.^{3,4}

Genetics

Pediatric ALL	Adult ALL	Ph-Like ALL
<i>BCR-ABL1</i>	<i>BCR-ABL1</i>	<i>CRLF2</i>
<i>KMT2A (MLL)</i>	<i>KMT2A (MLL)</i>	<i>JAK2</i>
<i>ETV6-RUNX1</i>	<i>TCF3 (E2A)</i>	<i>EPOR</i>
<i>CEP4</i>	<i>IGH</i>	<i>CSF1R</i>
<i>CEP10</i>	<i>MYC</i>	<i>ABL1</i>
		<i>ABL2</i>
		<i>PDGFRB</i>

Ph, Philadelphia chromosome

Test Interpretation

Test Results

Pediatric FISH

- Normal: no evidence of *BCR-ABL1* t(9;22), *KMT2A* (MLL) rearrangement, *ETV6-RUNX1* t(12;21), *RUNX1* amplification or copy number gain with *CEP4* and/or *CEP10*
- Abnormal: one of the above rearrangements or translocations detected

Featured ARUP Testing

[Acute Lymphoblastic Leukemia \(ALL\) Panel by FISH, Adult 2002647](#)

Method: Fluorescence in situ Hybridization (FISH)

- Recommended FISH panel for adults with newly diagnosed B-ALL
- Probes include *BCR-ABL1* t(9;22), *KMT2A* (MLL) 11q23 rearrangement (partner not determined), *TCF3* (E2A) rearrangement (partner not determined), *IGH* rearrangement (partner not determined), *MYC* rearrangement

[Acute Lymphoblastic Leukemia \(ALL\) Panel by FISH, Pediatric 2002719](#)

Method: Fluorescence in situ Hybridization (FISH)

- Recommended FISH panel for children with newly diagnosed B-ALL
- Probes include *BCR-ABL1* t(9;22), *KMT2A* (MLL) 11q23 rearrangement (partner not determined), *ETV6-RUNX1*, t(12;21), *CEP4*, *CEP10*

[Ph-Like Acute Lymphoblastic Leukemia \(ALL\) Panel by FISH 3000455](#)

Method: Fluorescence in situ Hybridization (FISH)

- Diagnosis, prognosis, and monitoring of *BCR-ABL1*-like B-ALL
- Probes include *CRLF2* rearrangement, *JAK2* rearrangement, *EPOR* rearrangement, *CSF1R* rearrangement, *ABL1* rearrangement, *ABL2* rearrangement, *PDGFRB* rearrangement
- Order when other major prognostic markers (e.g., *BCR-ABL1*, *ETV6-RUNX1*) are negative

Testing Strategy

At diagnosis, the minimum ALL workup includes bone marrow aspirate for morphology, immunophenotyping, cytogenetics (e.g., karyotyping and fluorescence in situ hybridization [FISH]), and other molecular testing, as indicated.

Adult FISH

- Normal: no evidence of *BCR-ABL1* t(9;22), *KMT2A* (*MLL*) rearrangement, *TCF3* (*E2A*) rearrangement, *IGH* rearrangement, or *MYC* rearrangement
- Abnormal: one of the above rearrangements/translocations or copy number change detected

Ph-Like ALL FISH

- Normal: no evidence of rearrangement involving *CRLF2*, *JAK2*, *EPOR*, *CSF1R*, *ABL1*, *ABL2*, or *PDGFRB*
- Abnormal: one of the described rearrangements detected

Prognostic Issues

More information on the prognostic significance of identified genetic rearrangements can be found in the ARUP Consult [Acute Lymphoblastic Leukemia](#) topic.

Limitations

Panels detect only the specific aberrations targeted by the FISH probes included. Chromosome alterations outside the regions complementary to these probes will not be detected.

References

1. American Cancer Society. [Key statistics for acute lymphocytic leukemia](#). Last revised Jan 2020; accessed Jul 2020.
2. Terwilliger T, Abdul-Hay M. [Acute lymphoblastic leukemia: a comprehensive review and 2017 update](#). *Blood Cancer J*. 2017;7(6):e577.
3. National Comprehensive Cancer Network. [NCCN Clinical Practice Guidelines in Oncology: Acute lymphoblastic leukemia](#). Version 2.2019. Accessed Oct 2019.
4. Paul S, Kantarjian H, Jabbour EJ. [Adult acute lymphoblastic leukemia](#). *Mayo Clin Proc*. 2016;91(11):1645-1666.

Related Information

[Acute Lymphoblastic Leukemia - ALL](#)
[BCR-ABL1 \(BCR::ABL1\) Qualitative and Quantitative Testing](#)
[Cytogenomic Microarray, Oncology](#)
[Leukemia/Lymphoma Phenotyping Evaluation by Flow Cytometry](#)
[TPMT and NUDT15](#)

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