Acute Lymphoblastic Leukemia FISH Panels

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 (eg, anemia, thrombocytopenia, leukopenia) and constitutional symptoms (eg, fever, lethargy, weight loss) are common.² In children, joint or extremity pain may be the only presenting symptom.³,⁴

Genetics

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

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
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


Related Information

[Acute Lymphoblastic Leukemia - ALL](#)

Related Tests

**Leukemia/Lymphoma Phenotyping Evaluation by Flow Cytometry 3001780**

*Method: Flow Cytometry*

**Chromosome Analysis, Bone Marrow 2002292**

*Method: Giemsa Band*

**Chromosome Analysis, Bone Marrow with Reflex to Genomic Microarray 2007130**

*Method: Giemsa Band/Genomic Microarray (Oligo-SNP array)*

**Cytogenomic SNP Microarray - Oncology 2006325**

*Method: Genomic Microarray (Oligo-SNP Array)*

**Cytogenomic Molecular Inversion Probe Array, FFPE Tissue - Oncology 2010229**

*Method: Molecular Inversion Probe Array*

**Chromosome FISH, Interphase 2002298**

*Method: Fluorescence in situ Hybridization (FISH)*

**BCR-ABL1, Qualitative with Reflex to BCR-ABL1 Quantitative 2005010**

*Method: Reverse Transcription Polymerase Chain Reaction*
TPMT and NUDT15 3001535
Method: Polymerase Chain Reaction/Fluorescence Monitoring

Thiopurine Methyltransferase, RBC 0092066
Method: Enzymatic/Quantitative Liquid Chromatography-Tandem Mass Spectrometry

Thiopurine Metabolites by LC-MS/MS 2014484
Method: Quantitative Liquid Chromatography/Tandem Mass Spectrometry