

# Detection of *ETV6-RUNX1* in Acute Lymphoblastic Leukemia

*DETECTION OF T(12;21) BY RT-PCR IS APPROPRIATE FOR THE DETERMINATION OF ETV6-RUNX1 FUSION TRANSCRIPT SEEN AS A RESULT OF THE T(12;21) TRANSLOCATION*

## Clinical Background

- The *ETV6-RUNX1* (*TEL-AML1*) fusion gene stems from the t(12;21)(p13;q22) chromosomal translocation and is undetectable by conventional cytogenetic analysis.
- *ETV6-RUNX1* is found in 25–30 percent of newly diagnosed precursor B-lineage childhood acute lymphoblastic leukemias (ALL)1–2 and 3 percent of adult B-ALL cases.
- *ETV6-RUNX1*, unlike other ALL-associated chromosomal translocations, is associated with a favorable prognosis.<sup>3</sup>
- Evaluation of *ETV6-RUNX1* expression, coupled with other prognostic markers, provides a framework for the selection of chemotherapy.
- Successful treatment occurs in >90 percent of pediatric cases and results in eradication of *ETV6-RUNX1* fusion transcripts.
- Relapse is associated with reemergence of *ETV6-RUNX1* fusion transcripts and is clinically useful in monitoring patients for disease status.

## Indications for Ordering

- The principal use for this test is to monitor for *ETV6-RUNX1* fusion transcripts in pediatric ALL.
- This test is not designed to detect minimal residual disease.

## Interpretation

- Not detected: There is no evidence of the t(12;21) chromosomal translocation by RT-PCR analysis. A negative result does not entirely exclude the presence of the t(12;21) translocation.
- Detected: There is evidence of the t(12;21) chromosomal translocation by RT-PCR analysis.

## Limitations

- Results of this test must always be interpreted in the context of morphologic and other relevant data, and should not be used alone for a diagnosis of malignancy.
- Samples that do not express *ETV6-RUNX1* fusion transcripts by this test may still express *ETV6-RUNX1* but in quantities below the limit of detection for this test.

## Methodology

Total cellular RNA is isolated from whole blood or bone marrow and subjected to cDNA synthesis, followed by amplification across the *ETV6-RUNX1* fusion gene. Fluorescence is detected and monitored using FRET probes.

## References

1. De Braekeleer E, et al. Cytogenetics in pre-B and B-cell acute lymphoblastic leukemia: a study of 208 patients diagnosed between 1981 and 2008. *Cancer Genet Cytogenet* 2010;200(1):8–15.
2. Stams WA, et al. Expression levels of TEL, AML1, and the fusion products TEL-AML1 and AML1-TEL versus drug sensitivity and clinical outcome in t(12;21)-positive pediatric acute lymphoblastic leukemia. *Clin Cancer Res* 2005;11(8):2974–80.
3. Rubnitz JE, et al. Prospective analysis of TEL gene rearrangements in childhood acute lymphoblastic leukemia: a Children's Oncology Group study. *J Clin Oncol* 2008 1;26(13):2186–91.

## Test Information

**0056008**

***ETV6-RUNX1* (*TEL-AML1*) Translocation, t(12;21) by RT-PCR**

For specific collection, transport, and testing information, refer to the ARUP website at [www.aruplab.com](http://www.aruplab.com).

For information on test selection, ordering, and interpretation, refer to ARUP Consult<sup>®</sup> at [www.arupconsult.com](http://www.arupconsult.com).

## AUTHOR

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