

E2A-PBX1 t(1;19) Translocation by RT-PCR and Fluorescence Monitoring

USED TO DETECT THE PRESENCE OF T(1;19) TRANSCRIPTS IN A PATIENT SAMPLE IN WHICH PRECURSOR B-ALL IS SUSPECTED

Clinical Background

- The *E2A-PBX1* fusion transcript is present in some cases of precursor B-cell acute lymphoblastic leukemia (pre B-ALL). This transcript results from a translocation involving the *E2A* gene on chromosome 19 and the *PBX1* gene (also known as PRL) on chromosome 1.
- The t(1;19) translocation is found in 5–6 percent of childhood ALL and in about 3 percent of adult ALL. The t(1;19) translocation identifies a subgroup of precursor B-ALLs with an unfavorable prognosis and is useful in the diagnosis of that category of leukemia (i.e., pre-B ALL).
- Identical t(1;19) breakpoints occur rarely in early B-progenitor ALL, but these breakpoints do not involve *E2A* or *PBX1*, and prognosis is excellent without the need for intensified therapy. It is thus clinically important to assess leukemia cases for the presence of the t(1;19) translocation.

Interpretation

- The *E2A/PBX1* assay will detect the presence of *E2A-PBX1* fusion transcripts by real-time reverse transcription-polymerase chain reaction (RT-PCR).
- A cell line known to contain the t(1;19) translocation is utilized as a positive control. Detection is determined by the presence of an amplification curve. Samples showing no amplification curve will be scored as Not Detected.

Methodology

- Patient RNA is isolated from a whole blood or bone marrow sample and reverse transcribed into cDNA. The cDNA is then subjected to PCR amplification using oligonucleotide primers specific for the *PBX1* gene on chromosome 1 and the *E2A* gene on chromosome 9.

- PCR products are monitored by real-time PCR using a set of fluorescently-labeled probes specific for the *PBX1* gene on chromosome 1.
- The utilization of probes specific for the *PBX1* gene provides additional specificity and diminishes the likelihood of false-positive detection by this assay.
- Each sample is also subjected to amplification for the *ABL* gene to verify cDNA quality and amplifiability.

Indication for Ordering

The principal use for the *E2A/PBX1* assay is to detect the presence of t(1;19) transcripts in a patient sample in which precursor B-ALL is suspected.

Limitation

As currently configured, this assay is intended to serve as a qualitative test and should not be used to detect minimal residual disease.

Reference

1. Van Dongen JJM, et al. Standardized RT-PCR analysis of fusion gene transcripts from chromosome aberrations in acute leukemia for detection of minimal residual disease. *Leukemia* 1999;13:1901–28.

Test Information

0055346

E2A-PBX1, t(1;19) Translocation by RT-PCR

For specific collection, transport, and testing information, refer to the ARUP website at www.aruplab.com.

For information on test selection, ordering, and interpretation, refer to ARUP Consult[®] at www.arupconsult.com.