

inv(16) for AML(CBFB-MYH11) by RT-PCR

Clinical Background

CBFB (core binding factor β chain)-MYH11 (myosin heavy chain 11 gene) fusion transcripts are detected in approximately 10 percent of *de novo* acute myelogenous leukemias (AML). One-half of these cases belong to AML subtype M4 with abnormal eosinophils (AML-M4Eo). The fusion results from a pericentric inversion on chromosome 16 inv(16)(p13q22) or, rarely, from the translocation t(16;16)(p13;q22). Based on the breakpoints in the *CBFB* and *MYH11* genes, greater than 95 percent of analyzed inv(16) cases belong to three types: A (88 percent), D (5 percent), and E (5 percent). Recent literature reports that inv(16) is associated with a better prognosis.

Indications For Use

The principal use for this test is to detect the presence of CBFB-MYH11 fusion transcripts in patients with AML-M4Eo or an indication of inv(16). This test is not intended to detect minimal residual disease.

Interpretation

- Negative: inv(16) types A, D, and E are not detected.
- Positive: inv(16) type A, D, or E are detected.

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. A negative result does not preclude the presence of CBFB-MYH11 fusion transcripts below the limit of detection or the presence of a type other than A, D, or E.

Methodology

Total RNA extracted from patient samples is analyzed by RT-PCR using a nested assay. PCR products are detected by electrophoresis and UV-illumination of ethidium bromide stained gels.

Reference

1. Van Dongen JM, et al. Standardized RT-PCR analysis of fusion gene transcripts from chromosome aberrations in acute leukemia for detection of minimal residual disease. *Leukemia* 1999; 13:1901-1928, and references therein.

Test Information

0092209

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