CBFB-MYH11, inv(16)

Indication: For determining the presence or absence of the CBFb-MYH11 fusion transcripts that are associated with the inv (16) chromosomal abnormality seen in AML-M4Eo. This test can be used for diagnosis or for minimal residual disease detection following therapy.

Testing Method: RNA is extracted from white blood cells in bone marrow and/or peripheral blood and reverse transcribed to cDNA. Real-time quantitative PCR is performed to amplify the CBFB-MYH11 transcripts. Control gene transcripts are amplified in parallel for normalization.

Test Parameters: This assay will detect CBFb-MYH11 inv (16) fusion transcripts A, D and E. More than 85% of positive patients have the type A transcript, while types D and E each represent approximately 5% of patients. All other types occur only in sporadic cases. Sensitivity of the test is approximately one CBFb-MYH11 bearing cell in 100,000 total cells. Levels detected in peripheral blood and bone marrow samples are generally equivalent.

Clinical Background: The pericentric inversion of chromosome 16 characterizes a distinct subset of patients with acute myelomonocytic leukemia - M4. This inversion produces fusion transcripts involving the CBFB (core binding factor) gene and the MYH11 (smooth muscle myosin heavy chain) gene. Inv (16) is found in 8-9% of newly diagnosed AML cases.

Turnaround Time: 3-5 business days

Sample requirements:

- 3 ml peripheral blood in lavender top tube (EDTA) if received same day
- Bone marrow aspirate (anticoagulated with either heparin or EDTA and, if possible, placed into tissue culture medium)
- PAXgene tube for peripheral blood or bone marrow (RNA stabilized at room temperature up to 48 hrs or cold for longer periods if shipment delayed)

CPT Codes: 81401

Ship Specimens to:

Henry Ford Center for Precision Diagnostics
Henry Ford Hospital
Clinic Building, K6, Core Lab E-655
2799 W. Grand Blvd.
Detroit, MI 48202