BCR/ABL t(9;22), p210 kD

(major breakpoint)

Indication: Chronic myeloid leukemia (CML) is a clonal myeloproliferative neoplasm. Most cases of CML are associated with the presence of t(9;22) resulting in a small derivative chromosome 22 known as the Philadelphia (Ph1) chromosome. Cytogenetically, CML is characterized by the presence of a reciprocal translocation between chromosome 9 and 22 [t(9;22)(q34;q11)], and as a consequence, the ABL proto-oncogene on chromosome 9 is fused to the BCR gene on chromosome 22. In 95% of CML patients and approximately 5% of Ph1 positive adult ALL patients, the breakpoint on chromosome 22 is located between exons 12 and 16 of the BCR gene, in the so called major break point cluster region M-bcr. The breakpoint on chromosome 9 is located in most cases between exons 1 and 2 in the ABL gene. The transcription product of this BCR-ABL fusion gene is an 8.5 kb aberrant fusion RNA with two junctional variants: b2a2 and b3a2 that give rise to the BCR-ABL chimeric protein (p210), a tyrosine kinase with deregulated activity.

Indication for Testing and Clinical Relevance:

1. Detection of BCR-ABL, p210, M-bcr is useful for diagnosis of chronic myelogenous leukemia (CML).
2. Quantitative real-time PCR (QRT-PCR) provides an appropriate monitoring strategy for patients with BCR/ABL expressing CML.
3. In patients treated with imatinib (Gleevec) therapy, achievement of a major molecular response (MMR) is associated with a high progression-free survival.

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 BCR-ABL transcripts. Control gene transcripts are amplified in parallel for normalization.

Test Parameters: The sensitivity of the assay is approximately one BCR-ABL bearing cell in 100,000 total cells. Levels detected in peripheral blood and bone marrow samples are generally equivalent.

Clinical Background: The BCR-ABL, M-bcr, p210 kD fusion transcript is found in most cases of chronic myelogenous leukemia. The assay will detect both type b3a2 and type b2a2.

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: 81206

Ship Specimens to:

Henry Ford Center for Precision Diagnostics
Henry Ford Hospital
Clinic Building, K6, Core Lab E-655
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