PML-RARA t(15;17) Quantitative by RT-PCR

Tests Description:

Acute Promyelocytic Leukemia is a rare acute myeloid leukemia characterized by a proliferation of promyelocytes with a balanced chromosomal translocation t(15;17) in the majority of cases. The translocation results in the fusion of the retinoic acid receptor alpha gene (RARA) on chromosome 17 with, most commonly, the promoter region of the promyelocytic gene (PML) on chromosome 15. This results in a fusion gene, PML-RARA, that produces mRNA that can be detected by reverse-transcription PCR-based assays. These assays have a lower limit of detection that is required for monitoring and early detection of patients that may fail initial treatment.

Clinical Sensitivity:

PML-RARA fusion occurs in 90% of cases with Acute Promyelocytic Leukemia (APL). This test is designed to detect the most common PML-RARA fusion forms; type A (short, S-form, BCR-3), and type B (Long, L-form, BCR-1). These forms occur as the result of PML-RARA translocation with different breakpoints, and occur in ~40% and 55% of PML-RARA positive cases, respectively.

Analytical Sensitivity:

- The assay is able to detect ~5 and 10 copies of PML/RARA BCR1 fusion within a pool of normal cells with a sensitivity of 90% and 100%, respectively.
- The assay is able to detect 5 and 10 copies of PML/RARA BCR3 fusion within a pool of normal cells with a sensitivity of 83% and 100%, respectively.
- Translocations involving other genes or gene partners will not be detected by this assay. Negative results cannot rule out the presence of PML/RARA fusions that are below the assay's limit of detection, the presence of the variable form (v), or the presence of other RARA fusions.

Methodology:

RNA is isolated from patient's sample and reversed transcribed to cDNA. The two PML-RARA tested fusion forms and a control gene (ABL1) are then amplified and quantified by real time PCR using specific primers for both the PML-RARA fusions and ABL1 gene. Results are reported as a normalized ratio of PML-RARA transcripts to ABL1 gene multiplied by 10^2.

Specimen Requirements:

At least 3mL of whole blood or bone marrow in lavender top (EDTA) tube. Specimens must be received within **24 hours** of collection due to lability of RNA.

Turn Around Time:

7 days

CPT Code:

- 81315

Shipping Instructions:

Please enclose **test requisition** with sample. **All information must be completed before sample can be processed.**

Place sample in a styrofoam mailer and ship at room temperature by overnight Federal Express to arrive Monday through Saturday. Specimens must be received within **24 hours of collection** due to lability of RNA.

Ship to:
Genetics and Genomics Diagnostic Laboratory
3333 Burnet Avenue NRB 1042
Cincinnati, Ohio 45229
513-636-4474

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changing the outcome together

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References:

Gallagher RE, et al. (1995) Characterization of acute promyelocytic leukemia cases with PML-RAR alpha break/ fusion sites in PML exon 6: identification of a subgroup with decreased in vitro responsiveness to all-trans retinoic acid. Blood. 86(4):1540-7.

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