CEBPA Gene Mutation Analysis

TEST NAME: CEBPA

TEST CODE: CEBPBM; CEBPBL

(Order CEBPBM for Bone marrow samples, CEBPBL for Whole Blood samples)

CPT CODE: 81403

CLINICAL USE:

CEBPA is an intronless gene located on 19q13.11, encoding a transcription factor C/EBPα which binds to and regulates genes involved in cell cycle progression and homeostasis. CEBPA mutations occur in ~10-20% of all cytogenetically normal (CN) AMLs. Many different types of CEBPA mutations have been reported across the entire CEBPA coding sequence and include insertions and/or deletions, and double mutations (usually biallelic) are common. Cytogenetically normal (CN) AMLs with biallelic CEBPA mutations (but without a FLT3 mutation) have been reported to have a relatively good prognosis compared to AMLs with wild type CEBPA.

METHODOLOGY:

PCR amplification of the entire CEBPA coding sequence is performed, followed by Sanger sequencing to detect the presence of absence of mutations.

REFERENCE RANGE: NA

SPECIMEN REQUIREMENTS:

- 5-10 mL of blood or bone marrow-purple (EDTA) tube or yellow (ACD) tube within 24 hours of collection.
- DNA 10ug at a minimum of 100ng/µL.

Note: If sample cannot arrive at laboratory within 24 hours of draw, refrigerate until sample can be transported and then transfer it at room temperature.

SPECIAL HANDLING: NA

TEST PERFORMANCE SCHEDULE: Weekly

TURN AROUND TIME: 7-10 days

STAT AVAILABILITY: NA

REFERENCES:

1. Thomas Pabst et al. Dominant-negative mutations of *CEBPA*, encoding CCAAT/enhancer binding protein-α (C/EBPα), in acute myeloid leukemia. Nature Genetics, Vol 27, March 2001.

2. Claire et al. Prognostic Significance of *CEBPA* Mutations in a Large Cohort of Younger Adult Patients With Acute Myeloid Leukemia: Impact of Double *CEBPA* Mutations and the Interaction With *FLT3* and *NPM1* Mutations. J Clin Oncol 28:2739-2747, 2010.

3. Schlenk et al. Mutations and treatment outcomes in cytogenetically normal acute myeloid leukemia. N Engl J Med 358 :1909 (2008).

4. Baldus et al. Clinical outcome of de novo acute myeloid leukemia patients with normal cytogenetics is affected by molecular genetic alterations: a concise review. Brit J Haematology 137: 387 (2007)