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 10μg 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:


