

# CEBPA Gene Mutation Analysis

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**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/EBP $\alpha$  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/ $\mu$ L.

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

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## REFERENCES:

1. Thomas Pabst et al. Dominant-negative mutations of *CEBPA*, encoding CCAAT/enhancer binding protein- $\alpha$  (C/EBP $\alpha$ ), 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)