CEBPA mutation analysis, *CEBPA* sequencing

**CPT Code(s):** 81403

**Service Code (IU Health):** 53025516

**Ordering Recommendation:** *CEBPA* mutation analysis is recommended to determine prognosis in patients with newly diagnosed cytogenetically normal acute myelogenous leukemia (CN-AML).

**Synonyms:** *CEBPA* mutation analysis, *CEBPA* sequencing, Mutation test for CN-AML

**Methodology:** Sanger sequence analysis. The *CEBPA* cDNA reference sequence used isNM_004364.3.

**Performed:** Mon-Fri

**Reported:** 6-9 days

**Specimen Requirements**

**Patient Preparation:** None required for whole blood

**Collect:** Lavender (EDTA) tubes; buccal swab; DNA

**Specimen Volume:** Blood: 2-6 mL whole blood; Buccal swab (contact Lab for collection tube)

**Storage/Transport:** Refrigerated/Room temperature

**Unacceptable Conditions:** Grossly hemolyzed or clotted

**Remarks:**

**Stability:** 2 weeks refrigerated; 1 month frozen

**Reference Interval:** by report

**Interpretive Data**
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Characteristics: The CCAAT/enhancer binding protein alpha, CEBPA, is an important transcription factor for granulocytic differentiation and has received attention as a favorable prognostic indicator in acute myeloid leukemia patients with a normal karyotype (AML-NK). CEBPA is located on chromosome 19q13, and has a GC-rich (more than 70%) coding region in a single exon. On average, CEBPA mutations are identified in 15% of AML-NK patients.

Analytical sensitivity and specificity: >99%

Limitations: Only the coding and immediate flanking regions of the CEBPA gene are analyzed by DNA sequencing. Changes in the promoter and other non-coding regions are not detected by this assay. In addition, the presence of a large intragenic deletion of the CEBPA gene will not be detected by sequence analysis. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

Reference: