CEBPA Mutation Analysis

Background

Mutations in the CEBPA gene are identified in 15-18% of acute myeloid leukemia (AML) with normal cytogenetics, and AML with mutated CEBPA represents a provisional diagnostic entity in the 2008 WHO classification.\(^1\)

AML with mutated CEBPA displays distinct clinicopathologic features including a favorable clinical course, and the identification of CEBPA mutations may assist in treatment selection.\(^2-6\) CEBPA mutation analysis is recommended for cases of AML with normal cytogenetics in current National Comprehensive Cancer Network (NCCN) and European LeukemiaNet guidelines.

Clinical Indications

Cleveland Clinic Laboratories offers CEBPA mutation analysis for classification and prognostic assessment of new acute myeloid leukemias, especially those with normal cytogenetics. Concurrent NPM1 and FLT3 studies are also recommended (see Acute Myeloid Leukemia Mutation Profile technical brief).

Interpretation

Mutations in CEBPA include single and dual (usually biallelic) mutations. Initial studies reported that the presence of any CEBPA mutation was associated with a favorable clinical course, while more recent studies have suggested that the favorable clinical course and distinctive clinicopathologic features are limited to AML with dual CEBPA mutations.\(^2-6\) All identified mutations are reported, and cases are classified as wild type (no mutations detected), single mutated or dual mutated.

Limitations of the Assay

Sanger sequencing is expected to identify >99% of mutations, provided that mutations represent at least 15-20% of total CEBPA alleles. This test is not intended for detection of minimal residual disease.

Methodology

DNA is extracted from peripheral blood or bone marrow. The entire CEBPA coding region is amplified by PCR and analyzed by Sanger sequencing.

References


# Test Overview

<table>
<thead>
<tr>
<th>Test Name</th>
<th>CEBPA Mutation Analysis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ordering Mnemonic</td>
<td>CEBPA</td>
</tr>
<tr>
<td>Specimen Requirements</td>
<td>Volume/Size: 5 mL; Type, blood; Container, EDT (Lavender); Transport temperature, ambient.</td>
</tr>
<tr>
<td>Minimum Specimen Requirements</td>
<td>Volume/Size: 3mL</td>
</tr>
<tr>
<td>Alternate Specimen Requirements</td>
<td>Volume/Size, 2ug; Type, blood; Container, EDTA (lavender); Transport temperature, ambient.</td>
</tr>
<tr>
<td>Reference Range</td>
<td>CEBPA mutations are not detected.</td>
</tr>
<tr>
<td>CPT Code</td>
<td>81218</td>
</tr>
</tbody>
</table>

---

**Technical Information Contact:**
Wendy Nedlik, MT(ASCP)
216.444.8410
tenlikw@ccf.org

**Scientific Information Contact:**
James Cook, MD, PhD
216.444.4435
cookj2@ccf.org