**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.

AML with mutated *CEBPA* displays distinct clinicopathologic features including a favorable clinical course, and the identification of *CEBPA* mutations may assist in treatment selection. 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.

**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. 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.

**References**
### Test Overview

<table>
<thead>
<tr>
<th>Test Name</th>
<th>CEBPA Mutation Analysis</th>
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</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>
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<tr>
<td>Alternate Specimen Requirements</td>
<td>Volume/Size, 2ug; Type, blood; Container, EDTA (lavender); Transport temperature, ambient.</td>
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<tr>
<td>Minimum Specimen Requirements</td>
<td>Volume/Size: 3mL</td>
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<td>Reference Range</td>
<td>CEBPA mutations are not detected.</td>
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<td>Billing Code</td>
<td>89259</td>
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<td>CPT Code</td>
<td>81403</td>
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