Overview

Useful For
Initial evaluation of acute myeloid leukemia, both for assigning an appropriate diagnostic subclassification and as an aid for determining prognosis

Special Instructions
- Hematopathology Patient Information

Method Name
Mutation Detection in DNA Using Sanger Sequencing

NY State Available
Yes

Specimen

Specimen Type
Varies

Advisory Information
This test is intended for use at the time of diagnosis and not for disease monitoring.

Shipping Instructions
Specimen must arrive within 7 days (168 hours) of collection.

Necessary Information
The following information is required:

1. Pertinent clinical history
2. Clinical or morphologic suspicion
3. Date of collection
4. Specimen source

Specimen Required
Submit only 1 of the following specimens:

Specimen Type: Peripheral Blood

Container/Tube: EDTA (lavender top or ACD (yellow top)

Specimen Volume: 3 mL

Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
3. Label specimen as blood.

**Specimen Stability:** Ambient (preferred)/Refrigerate

**Specimen Type:** Bone marrow

**Container/Tube:** EDTA (lavender top) or ACD (yellow top)

**Specimen Volume:** 2 mL

**Collection Instructions:**
1. Invert several times to mix bone marrow.
2. Send specimen in original tube.
3. Label specimen as bone marrow.

**Specimen Stability:** Ambient (preferred)/Refrigerate

**Specimen Type:** Extracted DNA from blood or bone marrow

**Container/Tube:** 1.5- to 2-mL tube

**Specimen Volume:** Entire specimen

**Collection Instructions:** Label specimen as extracted DNA from blood or bone marrow and provide indication of volume and concentration of the DNA.

**Specimen Stability:** Frozen (preferred)/Refrigerate/Ambient

**Forms**
1. [Hematopathology Patient Information](#) (T676) in Special Instructions
2. If not ordering electronically, complete, print, and send a [Hematopathology/Cytogenetics Test Request](#) (T726) with the specimen.

**Specimen Minimum Volume**
1 mL

**Reject Due To**

<table>
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<th>Gross hemolysis</th>
<th>Reject</th>
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<tbody>
<tr>
<td>Other</td>
<td>Paraffin embedded bone marrow aspirate clot Bone marrow biopsies, slides or paraffin shavings Moderately to severely clotted</td>
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Clinical and Interpretive

Clinical Information
Acute myeloid leukemia (AML) with mutated CCAAT/enhancer-binding protein alpha (CEBPA) gene is a diagnostic category in the current WHO classification of hematopoietic neoplasms.(1) In addition, CEBPA mutation on both alleles (so-called double mutation status) is considered a good prognostic feature in adults with newly diagnosed AML who have a normal karyotype or do not contain an alternate diagnostic genetic abnormality.(2,3) Thus, evaluation for CEBPA mutations is necessary for accurate diagnosis in the current classification system and contributes prognostic information for a large group of AML patients.

Reference Values
An interpretive report will be provided

Interpretation
The results will be given as positive or negative for CEBPA mutation and, if positive, the mutation will be described and single or double mutation status will be indicated.

Cautions
The assay is performed using Sanger sequencing, which has a sensitivity of 20%. This means that 20% or more of the DNA in the sample must be mutated to be detected. Consequently, this test is intended for use at the time of diagnosis, and not for disease monitoring.

Clinical Reference

Performance
Method Description
Total DNA is extracted from the sample and the entire, single exon of CEBPA amplified by PCR followed by Sanger sequencing with evaluation by capillary electrophoresis. Review of the sequence data is performed using a combination of automated calls and manual inspection.(Unpublished Mayo method).
Test Definition: CEBPA
CEBPA Mutations, Sequencing

No

Day(s) and Time(s) Test Performed
Monday through Friday

Analytic Time
5 days

Maximum Laboratory Time
8 days

Specimen Retention Time
DNA 3 months

Performing Laboratory Location
Rochester

Fees and Codes

Fees
- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

Test Classification
This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

CPT Code Information
81218-CEBPA (CCAAT/enhancer binding protein [C/EBP], alpha) (eg, acute myeloid leukemia), gene analysis, full gene sequence

LOINC® Information

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