Test Definition: CEBPA

CEBPA Mutations, Sequencing

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 originaltube.

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 originaltube.

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

Gross hemolysis	Reject
Other	Paraffin embedded bone marrow aspirate clot Bone marrow biopsies, slides or paraffin shavings Moderately to severely clotted

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Varies	7 days	

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

1. Swerdlow S, Campo E, Harris N, et al: WHO classification of tumours of haematopoietic and lymphoid tissues. WHO Press 2008; pp 109-145

2. Wouters B, Lowenberg B, Erpelinck-Verschueren C, et al: Double *CEBPA* mutations, but not single *CEBPA* mutations, define a subset of acute myeloid leukemia with a distinctive expression profile that is uniquely associated with a favorable outcome. Blood 2009;113:3088-3091

3. Pabst T, Eyholzer M, Fos J, et al: Heterogeneity within AML with CEBPA mutations; only CEBPA double mutations, but not single CEBPA mutations are associated with favourable prognosis. Br J Cancer 2009;100:1343-1346

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).

PDF Report

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

Test ID	Test Order Name	Order LOINC Value
CEBPA	CEBPA Mutations, Sequencing	75009-1

Result ID	Test Result Name	Result LOINC Value
MP019	Specimen:	31208-2
34634	Final Diagnosis:	34574-4