

TriCore Reference Laboratories performs in-house sequencing for CEBPA mutations. The CEBPA gene encodes a transcription factor involved in maturation and differentiation of myeloid cells. CEBPA mutation status is important for diagnosis and risk stratification in acute myeloid leukemia (AML). CEBPA mutations may affect both alleles (biallelic - biCEBPA) or one allele (monoallelic - moCEBPA).

Biallelic CEBPA mutation is associated with increased remission duration and overall survival in most cases. The current 2016 revised WHO classification of hematopoietic tumors includes a category of AML with biallelic CEBPA mutation¹, a favorable prognosis category of AML. In addition, the detection of biallelic CEBPA mutations in younger patients with AML can be associated with germline mutation with predisposition to develop AML. This testing is recommended by the National Comprehensive Cancer Network for all new diagnoses of AML² and can be ordered as a stand-alone test or as part of the Myeloid Disorder Mutation Panel.

REFERENCES

- ¹ Swerdlow SH, et al. WHO Classification of Tumors of Hematopoietic and Lymphoid Tissues. World Health Organization Classification of Tumours. Lyon, France: IARC. 2016.
- ² National Comprehensive Cancer Network. Acute Myeloid Leukemia (Version 1.2019). https://www.nccn.org/professionals/physician_gls/pdf/aml.pdf

CEBPA MUTATION DETECTION, PCR	
Test Code.....MDCEBP	Testing Performed.....Testing initiated on Thursday
CPT Code.....81218	Methodology.....Polymerase Chain Reaction/Sequencing
	Reported.....within 7-21 days
ORDERING	
Ordering Recommendation: Initial test for diagnosis and risk stratification in CN-AML.	
The CEBPA gene encodes a transcription factor involved in maturation and differentiation of myeloid cells. CEBPA mutation status is important for diagnosis and risk stratification in acute myeloid leukemia (AML). Biallelic CEBPA mutation is associated with increased remission duration and overall survival in most cases. The current 2016 revised WHO classification of hematopoietic tumors includes a category of AML with biallelic CEBPA mutation, a favorable prognosis category of AML. In addition, the detection of biallelic CEBPA mutations in younger patients with AML can be associated with germline mutation with predisposition to develop AML. This testing is recommended by the National Comprehensive Cancer Network for all new diagnoses of AML and can be ordered as a stand-alone test or as part of the Myeloid Disorder Mutation Panel.	
Order may be submitted via electronic request. If you do not have electronic ordering capability, a Hematopathology requisition (see Related Documents above) must be completed by ordering provider including clinical indications. This manual requisition must accompany specimen. For specific ordering instructions or questions regarding specimens please call (505) 938-8684.	
COLLECTION	
Specimen	Whole Blood Bone Marrow Specimens should only be collected Monday through Wednesday. Do NOT collect on weekends. Collect whole blood in or add bone marrow aspirate to an appropriately anticoagulated collection tube, invert several times to mix specimen.
Collection Containers	Lavender top tube (EDTA)
Collection Amount	Preferred volume: 5.0mL Minimum volume: 1.0mL
PROCESSING	
Specimen Processing Instructions	Do not centrifuge. Specimen must be kept in original container.
SHIPPING/TRANSPORT AND STORAGE	
Stabilities/Storage: (Collection to initiation of testing)	Temperature Stability Ambient: 24 hours Refrigerated: 5 days Frozen: Unacceptable
Shipping Instructions	Ship refrigerated.
Rejection Criteria	Specimens collected in anticoagulants other than EDTA, serum or plasma, frozen or clotted specimens and severely hemolyzed specimens are unacceptable.