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 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.

REFERENCES
