MAYO CLINIC LABORATORIES

NEW TEST

Notification Date: November 19, 2024
Effective Date: December 19, 2024

Beta-Globin Gene Cluster, Deletion/Duplication, Varies

Test ID: WBGDD

Useful for:
Determining the etiology of hereditary persistence of fetal hemoglobin (HPFH), delta-beta thalassemia, or other large deletions involving the beta-globin gene cluster

Diagnosing less common causes of beta thalassemia; these large deletional beta-thalassemia variants result in elevated hemoglobin (Hb) A2 and can have elevated HbF levels

Distinguishing homozygous HbS disease from a compound heterozygous HbS/large beta-globin cluster deletion disorder (ie, HbS/beta zero thalassemia, HbS/delta-beta zero thalassemia, HbS/HPFH, HbS/gamma-delta-beta thalassemia)

Diagnosing complex thalassemias where the beta-globin gene and one or more of the other genes in the beta-globin cluster have been deleted

Evaluating and classifying unexplained increased HbF percentages

Evaluating microcytic neonatal anemia

Evaluating unexplained long standing microcytosis in the setting of normal iron studies and negative alpha-thalassemia testing/normal Hb A2 percentages

Confirming gene fusion hemoglobin variants such as Hb Lepore and HbP-Nilotic

Confirming homozygosity vs hemizygosity of variants in the beta-like genes (*HBB*, *HBD*, *HBG1*, *HBG2*)

Investigating newborns with HbA levels greater than HbF on newborn screen in the absence of transfusion

This test is **not useful for** diagnosis or confirmation of alpha thalassemia, the most common beta thalassemias, or hemoglobin variants. It also does not detect non-deletional HPFH.

Methods:
Polymerase Chain Reaction (PCR) Analysis/Multiplex Ligation-Dependent Probe Amplification (MLPA)

Specimen Requirements:
Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA)
Acceptable: Yellow top (ACD)
Specimen Volume: 4 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in the original tube. **Do not aliquot.**
Specimen Stability Information: Refrigerated (preferred)/Ambient

Cautions:
Non-deletional subtypes of beta thalassemia or hereditary persistence of fetal hemoglobin are not detected by this assay. In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as the internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing.

CPT Code:
81363-HBB (hemoglobin, beta, beta-globin) (eg, beta thalassemia), duplication/deletion analysis

Day(s) Performed: Wednesday, Friday **Report Available:** 25 to 30 days

Questions
Contact Connie Penz, Laboratory Resource Coordinator at 800-533-1710.