MAYO CLINIC LABORATORIES

TEST OBSOLETE

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

Beta-Globin Cluster Locus, Deletion/Duplication, Varies

Test ID: WBDDR

Explanation:
On the effective date, WBDDR will be replaced with WBGDR, a blood-only profile-specific code for (THEV1, REVE2, MEV1, HAEV1, HBEL1, HBELP).

Recommended Alternative Test:

Beta-Globin Gene Cluster Deletion/Duplication, Blood

Test ID: WBGDR

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 alterations result in elevated hemoglobin (Hb) A2 and can have slightly elevated Hb F levels

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

Diagnosing complex thalassemias where the beta-globin gene and 1 or more of the other genes in the beta-globin cluster have been deleted

Evaluating and classifying unexplained increased Hb F 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 Hb P-Nilotic

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

Investigating newborns with Hb A levels greater than Hb F 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:
Only orderable as a reflex. For more information see:
-HAEV1 / Hemolytic Anemia Evaluation, Blood
-HBEL1 / Hemoglobin Electrophoresis Evaluation, Blood
-MEV1 / Methemoglobinemia Evaluation, Blood
-REVE2 / Erythrocytosis Evaluation, Blood
-THEV1 / Thalassemia and Hemoglobinopathy Evaluation, Blood and Serum

Cautions:
Non-deletional subtypes of beta thalassemia or hereditary persistence of fetal hemoglobin are not detected by this assay.

Hemoglobin electrophoresis and sequencing analysis of the beta-globin gene will be performed prior to this test to exclude other diagnoses or to indicate the diagnostic utility of this testing platform.

In addition to disease-related probes, the multiplex ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as 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.