Lab Dept: Anatomic Pathology

Test Name: ALPHA GLOBIN (HBA1/HBA2) GENE ANALYSIS

General Information

Lab Order Codes: AGPB

Synonyms: Alpha Thalasseium; HBA1; HBA2; Hemoglobin Bart; Hemoglobin-H Disease; Hydrops Fetalis; Thalassemia, Alpha

CPT Codes: 81269 – HBA1/HBA2, gene analysis, for common duplication/deletions variants

Test Includes: Direct mutation analysis. Deletions within the alpha-globin locus are identified by a multiplex ligation-dependent probe amplification assay. In addition, a PCR based assay is used to detect the presence of the alpha-3.7 and alpha-4.2 deletions.

Logistics

Indications: Diagnosis of alpha-thalassemia. Carrier screening for individuals from high-risk populations.

Lab Testing Section: Anatomic Pathology – Sendouts

Referred to: Mayo Medical Laboratories (MML Test: ATHAL)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 8 – 12 days

Special Instructions: Please submit a Molecular Genetics-Congenital Inherited Diseases Patient Information Sheet (Supply T521)

Specimen must arrive at Mayo within 96 hours of collection.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume:	3 mL (Minimum: 1 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do Not Centrifuge. Specimen should remain in the original collection container. Store and ship at room temperature. Forward promptly. Specimen must arrive at Mayo within 96 hours of collection.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	An interpretive report will be provided
Limitation:	In addition to disease-related probes, the multiplex ligation dependant 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. Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered. A previous bone marrow transplant from an allogenic donor ill interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in our interpretation of results may occur if information given is inaccurate or incomplete. This assay cannot be performed on chorionic villus specimens. Non-deletion types of alpha-thalassemia will not be detected by this assay. This test is not useful for diagnosis or confirmation of beta-thalassemia or hemoglobinopathies. Hemoglobin electrophoresis should usually be done prior to this test to exclude other diagnoses or to identify non deletion types of alpha-thalassemia.
Methodology:	Dosage Analysis by Polymerase Chain Reaction (PCR), Multiplex Ligation-Dependent Probe Amplicfication (MPLA)/Luminex Technology

References:

Mayo Medical Laboratories July 2018
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