**Lab Dept:** Anatomic Pathology  
**Test Name:** ALPHA GLOBIN (HBA1/HBA2) GENE ANALYSIS

### General Information

**Lab Order Codes:** AGPB  
**Synonyms:** Alpha Thalassemia; 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
Phone: 507-538-2996 Fax: 507-284-0670