Lab Dept: Anatomic Pathology

Test Name: ALPHA GLOBIN CLUSTER LOCUS DEL/DUP

General Information

Lab Order Codes: AGDD

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: Presence or absence of deletions or duplications within the alpha globin gene cluster, with interpretive report.

Logistics

Indications: Useful for the diagnosis of alpha-thalassemia.

This test is not useful for diagnosis or confirmation of beta-thalassemia or hemoglobinopathies. Sequence variants, other than the alpha T-Saudi and hemoglobin constant spring alterations, are not detected by this assay. For detection of single point and other nondeletion variants, see Mayo's catalog for guidance.

Lab Testing Section: Anatomic Pathology – Sendouts

Referred to: Mayo Clinic Laboratories (MML Test: AGDD)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: 9 – 13 days

Special Instructions: Specimens are preferred to be received at the testing lab within four (4) days of collection. Extraction will be attempted for specimens received after four days, and DNA yield will be evaluated to determine if testing may proceed.

To ensure minimum volume and concentration of DNA is met, the preferred volume of blood must be submitted. Testing may be canceled by the reference lab if DNA requirements are inadequate.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top (EDTA) tube Alternative: yellow top ACD tube or green top Sodium Heparin (no gel) tube
Draw Volume:	3 mL (Minimum: 1 mL) blood – optimal volume is strongly preferred
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 preferred to arrive at Mayo within 96 hours of collection. Specimen stable ambient (preferred) for 4 days, refrigerated or frozen for 4 days.
Patient Preparation:	A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant.
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	An interpretive report will be provided
Limitation:	Hemoglobin electrophoresis should usually be done prior to this test to exclude other diagnoses. 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. Rare alterations (i.e., polymorphisms) exist that could lead to false-negative or false-positive results. If the results obtained do not match the clinical findings, additional testing should be considered. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given is inaccurate or incomplete.

Methodology:	Polymerase Chain Reaction (PCR)/Quantitative Polymerase Chain Reaction (qPCR)/Multiplex Ligation-Dependent Probe Amplification (MLPA)
References:	Mayo Clinic Laboratories December 2024
Updates:	12/19/2024: Initial entry. Replaces obsolete AGPB. 04/10/2025: Updated methodology, stability, and acceptable specimen type.