

Client: Example Client ABC123  
123 Test Drive  
Salt Lake City, UT 84108  
UNITED STATES

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 12/31/1752  
**Sex:** Female  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 01/01/2017 12:34

**Alpha Globin (HBA1 and HBA2) Sequencing and Deletion/Duplication, Fetal**

ARUP test code 3019566

Maternal Contamination Study Fetal Spec	Fetal Cells
	Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood
HBA FGA FE Int	<p>Negative</p> <p>RESULT No pathogenic variants were detected in the alpha globin gene cluster.</p> <p>INTERPRETATION According to information provided to ARUP, this fetus is reported to have ascites and pleural effusions. No pathogenic variants were detected in the alpha globin genes, HBA1 and HBA2, by sequencing in this prenatal sample. No large deletions or duplications were detected in the alpha globin gene cluster (HBZ, HBM, HBA2, HBA1, HBQ1) or its HS-40 regulatory region in this prenatal sample. This significantly reduces the probability of, but does not exclude, alpha-thalassemia disease or trait in this fetus. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.</p> <p>RECOMMENDATIONS Genetic consultation is recommended.</p> <p>COMMENTS Reference Sequences: GenBank # NM_000558.5 (HBA1), NM_000517.6 (HBA2), NG_000006.1 (Alpha globin gene cluster) Nucleotide numbering begins at the "A" of the ATG initiation codon. Likely benign and benign variants are not reported.</p> <p>This result has been reviewed and approved by [REDACTED]</p> <p>BACKGROUND INFORMATION: Alpha Globin (HBA1 and HBA2) Sequencing and Deletion/Duplication, Fetal</p> <p>CHARACTERISTICS: Alpha thalassemia is caused by decreased or absent synthesis of the hemoglobin alpha chain resulting in variable clinical presentations. Alpha (+) thalassemia results from variants of a single HBA2 globin gene (-a/aa) and is</p>

H=High, L=Low, \*=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example  
ARUP Accession: 25-107-113653  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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clinically asymptomatic (silent carrier). Alpha (0) thalassemia (trait) is caused by variants of both HBA2 globin genes (-a/-a) or variants in the HBA1 and HBA2 globin genes on the same chromosome (--/aa) and results in mild microcytic anemia. Hemoglobin H disease occurs due to variants of three alpha globin genes (--/-a) and results in hemolysis with Heinz bodies, moderate anemia, and splenomegaly. Hb Bart Hydrops Fetalis Syndrome results when variants occur in all four alpha globin genes (---/---) and is lethal in the fetal or early neonatal period. Alpha globin gene triplications result in three active alpha globin genes on a single chromosome. Nondeletional alpha globin variants may be pathogenic or benign; both may result in an abnormal protein detectable by hemoglobin evaluation. Pathogenic nondeletional variants often have a more severe effect than single gene deletions.

INCIDENCE: Carrier frequency in Mediterranean (1:30-50), Middle Eastern, Southeast Asian (1:20), African, African American (1:3).

INHERITANCE: Autosomal recessive.

CAUSE: Pathogenic variants in the alpha globin gene cluster.

CLINICAL SENSITIVITY: 99 percent.

METHODOLOGY: Bidirectional sequencing of the HBA1 and HBA2 coding regions, intron-exon boundaries and 3' polyadenylation signal. Multiplex ligation-dependent probe amplification (MLPA) of the alpha globin gene cluster (HBZ, HBM, HBA1, HBA2, HBQ1) and the regulatory region multispecies conserved sequence 2 (MCS-R2, also known as HS-40 regulatory region).

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Sequence analysis will not detect all regulatory region variants or variants in alpha globin cluster genes other than HBA1 and HBA2. Sequencing of both HBA1 and HBA2 may not be possible in individuals harboring large alpha globin deletions on both alleles. This assay is unable to sequence HBA2-HBA1 fusion genes; thus, HBA1 or HBA2 sequence variants occurring in cis with a 3.7 kb deletion or other HBA2-HBA1 hybrid gene will not be detected (e.g., HbG Philadelphia will not be detected when in cis with the 3.7 kb deletion). It may not be possible to determine phase of identified sequence variants. Specific breakpoints of large deletions/duplications will not be determined; therefore, it may not be possible to distinguish variants of similar size. Individuals carrying both a deletion and duplication within the alpha globin gene cluster may appear to have a normal number of alpha globin gene copies. Rare syndromic or acquired forms of alpha thalassemia associated with ATRX variants will not be detected. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	25-107-113653	4/17/2025 1:18:00 PM	4/17/2025 1:18:49 PM	4/18/2025 11:01:00 AM
Maternal Contam Study, Maternal Spec	25-107-113653	4/17/2025 1:18:00 PM	4/17/2025 1:18:49 PM	4/18/2025 11:01:00 AM
HBA FGA FE Int	25-107-113653	4/17/2025 1:18:00 PM	4/17/2025 1:18:49 PM	4/18/2025 11:01:00 AM

END OF CHART

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