

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB** 9/1/1992  
**Sex:** Male  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Alpha Globin (HBA1 and HBA2) Deletion/Duplication**

ARUP test code 2011622

Alpha Globin (HBA1/2) DelDup Interp

**Deletion** \*

**RESULT**

Two pathogenic deletions, resulting in the deletion of two alpha globin gene copies, were detected in the alpha globin gene cluster.

**DNA VARIANT(S)**

Pathogenic Deletion: -alpha3.7; Homozygous  
Predicted Genotype: -a/-a

**INTERPRETATION**

Two copies of the 3.7 kb alpha globin gene deletion were detected by deletion/duplication analysis of the alpha globin gene cluster and its HS-40 regulatory region. This individual is predicted to have a single functional alpha globin gene on both chromosomes. This result is consistent with alpha (0) thalassemia (trait) often associated with mild anemia and microcytosis.

**RECOMMENDATIONS**

Medical management should rely on clinical findings and family history. This test detects only large deletions/duplications and not pathogenic alpha globin variants. If clinical findings suggestive of alpha-thalassemia disease are present, consider alpha globin gene sequencing. Carrier screening for alpha thalassemia should be offered to this individual's relatives and reproductive partner. Genetic consultation is recommended.

**COMMENTS**

Reference sequence for alpha globin gene cluster: NG\_000006.1

**REFERENCES**

HbVar 3.7kb deletion link: [http://globin.bx.psu.edu/cgi-bin/hbvar/query\\_vars3?mode=output&display\\_format=page&i=1076](http://globin.bx.psu.edu/cgi-bin/hbvar/query_vars3?mode=output&display_format=page&i=1076)

This result has been reviewed and approved by [REDACTED]

**BACKGROUND INFORMATION:** Alpha Globin (HBA1 and HBA2) Deletion/Duplication

**CHARACTERISTICS:** Alpha thalassemia is caused by decreased or absent synthesis of the hemoglobin alpha-chain resulting in variable clinical presentations. Alpha (+) thalassemia results from mutation of a single alpha<sup>2</sup> globin gene (-a/aa) and is clinically asymptomatic (silent carrier). Alpha (0) thalassemia (trait) is caused by mutation of both alpha<sup>2</sup> globin genes (-a/-a), or mutations in the alpha<sup>1</sup> and alpha<sup>2</sup> globin genes on

**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: 26-041-111037  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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the same chromosome, (--)aa) and results in mild microcytic anemia. Hemoglobin H disease occurs due to mutation of three alpha globin genes (--)aa) and results in hemolysis with Heinz bodies, moderate anemia, and splenomegaly. Hb Bart hydrops fetalis syndrome results when mutations occur in all four alpha globin genes (--)aa) 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.

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

INHERITANCE: Autosomal recessive.

CAUSE: Pathogenic mutations in the alpha globin gene cluster.

CLINICAL SENSITIVITY: Varies by ethnicity, up to 95 percent.

METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) of the alpha globin gene cluster (HBZ, HBM, HBA2, HBA1, HBQ1) and its HS-40 regulatory region.

ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.

LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Single exon deletions/duplications may not be detected based on the breakpoints of the rearrangement. Specific breakpoints of large deletions/duplications will not be determined; therefore, it may not be possible to distinguish mutations of similar size. This assay does not assess for nondeletional mutations within the coding or regulatory regions of the alpha globin cluster genes. 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 mutations will not be detected.

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

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

#### Alpha Globin (HBA1/2) DelDup Specimen

whole Blood

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Alpha Globin (HBA1/2) DelDup Interp	26-041-111037	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha Globin (HBA1/2) DelDup Specimen	26-041-111037	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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