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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/2/1992
Gender:	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

This result has been reviewed and approved by [redacted]

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

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 alpha2 globin gene (-a/aa) and is clinically asymptomatic (silent carrier). Alpha (0) thalassemia (trait) is caused by mutation of both alpha2 globin genes (-a/-a), or mutations in the alpha1 and alpha2 globin genes on the same chromosome, (--/aa) and results in mild microcytic anemia. Hemoglobin H disease occurs due to mutation of three alpha globin genes (--/-a) 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 (--/--) 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. 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 non-deletional 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

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Alpha Globin (HBA1/2) DelDup Interp	23-123-400250	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha Globin (HBA1/2) DelDup Specimen	23-123-400250	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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