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

Physician: Doctor, Example

Patient: Patient, Example

DOB
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Hemoglobin Evaluation Reflexive Cascade

ARUP test code 2005792

Hemoglobin A	97.2 %	(Ref Interval: 95.0-97.9)
Hemoglobin A2	2.3 %	(Ref Interval: 2.0-3.5)
Hemoglobin F	0.5 %	(Ref Interval: 0.0-2.1)
	REFERENCE INTERVAL: Hemoglobin F Access complete set of age- and/or gender-specific reference intervals for this test in the ARUP Laboratory Test Directory (aruplab.com).	
Hemoglobin S	0.0 %	(Ref Interval: 0.0-0.0)
Hemoglobin C	0.0 %	(Ref Interval: 0.0-0.0)
Hemoglobin E	0.0 %	(Ref Interval: 0.0-0.0)
Hemoglobin - Other	0.0 %	(Ref Interval: 0.0-0.0)
Sickle Cell Solubility	Not Performed	
Hemoglobin, Capillary Electrophoresis	Performed	
Hemoglobin Evaluation	See Note	
Beta Globin Full Gene Sequencing	Not Applicable	

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

Beta Globin (HBB) Del/Dup Result	Not Applicable
Alpha Thalassemia HBA1 and HBA2 Seq	Not Applicable
Hemoglobin Lepore (HBD/HBB) 3 Mutations	Not Applicable
Hemoglobin Cascade Interpretation	See Note

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

RESULT
Normal hemoglobin evaluation.
See comments.

COMMENTS
HPLC, and capillary electrophoresis revealed a normal hemoglobin pattern. No large deletions or duplications were detected in the alpha globin gene cluster (HBZ, HBM, HBA2, HBA1 and HBQ1) or its HS-40 regulatory region by alpha thalassemia deletion/duplication testing. These results do not rule out a rare, Greek beta thalassemia variant associated with a normal Hb A2.

Please correlate with clinical and laboratory findings.

Controls were run and performed as expected.
This result has been reviewed and approved by Archana Agarwal, M.D.

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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS.

Alpha Globin (HBA1 and HBA2) Del/Dup Rst	Performed

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Hemoglobin A	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin A2	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin F	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin S	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin C	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin E	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin - Other	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Sickle Cell Solubility	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin, Capillary Electrophoresis	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin Evaluation	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta Globin Full Gene Sequencing	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Beta Globin (HBB) Del/Dup Result	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha Thalassemia HBA1 and HBA2 Seq	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin Lepore (HBD/HBB) 3 Mutations	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Hemoglobin Cascade Interpretation	19-291-140483	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Alpha Globin (HBA1 and HBA2) Del/Dup Rst	19-291-140483	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