ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/12/2016
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Beta Globin (HBB) Deletion/Duplication

ARUP test code 2010113

Beta Globin (HBB) DelDup Specimen	Whole Blood
Beta Globin (HBB) DelDup Interp	**Deletion** *

RESULT
One HPFH deletion detected in the beta globin gene cluster.

Nucleic Acid Change: HBBP1 exon 3-OR51V1 downstream; Heterozygous
Commonly Known As: Ghanaian

INTERPRETATION
A deletion of HBBP1 exon 3-OR51V1 downstream was detected by deletion/duplication analysis of the beta globin gene cluster and its locus control region. This large deletion is associated with hereditary persistence of fetal hemoglobin (HPFH). Individuals heterozygous for this deletion typically have normal red blood cell indices but elevated levels of Hb F. A more severe disorder is possible if a second HBB mutation is present on the opposite chromosome that is not detected by this assay.

RECOMMENDATION
Medical management should rely on clinical findings and family history. If suspicion for a clinically significant form of beta thalassemia remains, consideration should be given to beta globin gene sequencing (ARUP test code 0050578) which detects up to 97 percent of all beta globin gene mutations. A hemoglobin evaluation should be offered to this individuals reproductive partner and family members to assess carrier status for hemoglobin variants. Genetic consultation is recommended.

COMMENTS
Reference sequence for beta globin gene cluster: NG_000007.3

This result has been reviewed and approved by Yuan Ji, Ph.D.

H – high L – low * – abnormal C – critical

BACKGROUND INFORMATION: Beta Globin (HBB) Deletion/Duplication
CHARACTERISTICS: Beta thalassemia is caused by decreased or absent synthesis of the hemoglobin beta-chain resulting in variable clinical presentations ranging from mild anemia to transfusion dependence. Hereditary persistence of fetal hemoglobin (HPFH) is a clinically benign condition caused by variants within the beta globin gene cluster that alter normal hemoglobin switching and result in persistent fetal hemoglobin (Hb F) production.
INCIDENCE: Varies by ethnicity.
INHERITANCE: Usually autosomal recessive, infrequently autosomal dominant.
CAUSE: Pathogenic variants within the HBB gene or variants involving the beta globin gene cluster and its regulatory elements.
CLINICAL SENSITIVITY: Varies by ethnicity.
METHODOLOGY: Multiplex ligation-dependent probe amplification (MLPA) of the beta globin gene cluster (HBB, HBD, HBG1, HBG2, HBE1) and its locus control region.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. HBB single base pair substitutions, small deletions/duplications, deep intronic and promoter variants will not be detected. Breakpoints of large deletions/duplications will not be determined; therefore, the precise clinical phenotype associated with a particular deletion (e.g., HPFH vs. delta-beta thalassemia) may not be known. Intragenic deletions in the beta globin cluster genes, other than HBB, may not be detected. This assay does not assess for sequence variants within the coding or regulatory regions of HBB, HBD, HBG1, HBG2 or HBE1. Apparent copy number changes detected solely in the HBG1-HBG2 region will not be reported as they can result from benign sequence variants or gene conversion events.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Beta Globin (HBB) DelDup Specimen	17-130-139152	5/10/2017 10:42:00 AM	5/12/2017 9:47:20 AM	5/17/2017 3:16:52 PM
Beta Globin (HBB) DelDup Interp	17-130-139152	5/10/2017 10:42:00 AM	5/12/2017 9:47:20 AM	5/17/2017 3:16:52 PM

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