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

Physician: Doctor, Example

Patient: Patient, Example

DOB	11/28/1966
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Deletion/Duplication Analysis by MLPA

ARUP test code 3003144

Deletion/Duplication Interpretation

Negative

Deletion/Duplication Analysis by MLPA

TEST PERFORMED - 3003144
TEST DESCRIPTION - Beta Globin (HBB) Deletion/Duplication
INDICATION FOR TESTING - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the beta globin gene cluster.

INTERPRETATION
No pathogenic large deletions/duplications involving the genes of the beta globin gene cluster (HBB, HBD, HBG1, HBG2 and HBE1) or its locus control region were detected. This result decreases the likelihood of, but does not exclude, beta thalassemia or hereditary persistence of fetal hemoglobin (HPFH). Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. This result should be correlated with the results of beta globin gene sequencing (ARUP test code 3004547) and alpha globin sequencing and deletion/duplication analysis (ARUP test code 2011708), which were requested concurrently and reported under separate cover. Genetic consultation is recommended.

COMMENTS
Reference sequences: GenBank # NG_000007.3 (Beta globin gene cluster)

This result has been reviewed and approved by [redacted]

Deletion/Duplication Gene

BG DD

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.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Deletion/Duplication Interpretation	22-229-400949	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Deletion/Duplication Gene	22-229-400949	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