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

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/6/1932
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Beta Globin (HBB) Gene Sequencing (Temporary Referral as of 12/07/20)

ARUP test code 0050578

BGSEQ Specimen	Whole Blood
Beta Globin Full Gene Sequencing	Negative

TEST PERFORMED - 0050578
TEST DESCRIPTION - Beta Globin (HBB) Gene Sequencing
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the HBB gene.

INTERPRETATION
No pathogenic variants were detected in the beta globin (HBB) gene through bidirectional sequencing of the coding region, intron/exon boundaries, proximal promoter, 5' untranslated region, and 3' polyadenylation signal. None of the additional targeted HBB pathogenic intronic variants, IVS-I-110, IVS-II-654, IVS-II-705, IVS-II-745, and IVS-II-765 L1, were detected. This result significantly decreases the probability of, but does not exclude, beta thalassemia or beta thalassemia trait. 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. If clinical suspicion for beta thalassemia remains high, consideration should be given to Beta Globin (HBB) Deletion/Duplication analysis (ARUP test code 2010113). Genetic consultation is recommended.

COMMENTS
Reference Sequences: GenBank # NM_000518.5 (HBB)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Beta Globin (HBB) Sequencing

CHARACTERISTICS: Structural hemoglobinopathies or thalassemias (insufficient or absent beta-chain production).
INCIDENCE: Varies with ethnicity.
INHERITANCE: Usually autosomal recessive, infrequently autosomal dominant.
CAUSE: Pathogenic variants in the HBB gene.
CLINICAL SENSITIVITY: Up to 97 percent, depending upon ethnicity.
METHODOLOGY: Bidirectional sequencing of the HBB coding regions, intron-exon boundaries, 5' proximal promoter and untranslated region, 3'polyadenylation signal, and intronic variants c.93-21 (IVS-I-110), c.316-197 (IVS-II-654), c.316-146 (IVS-II-705), c.316-106 (IVS-II-745), and c.316-86_316-85 (IVS-II-765 L1).
ANALYTICAL SENSITIVITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Large deletions, and variants in distal regulatory elements are not detected.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
BGSEQ Specimen	20-218-107080	8/5/2020 9:15:00 AM	8/6/2020 8:33:00 AM	8/19/2020 11:21:00 AM
Beta Globin Full Gene Sequencing	20-218-107080	8/5/2020 9:15:00 AM	8/6/2020 8:33:00 AM	8/19/2020 11:21:00 AM

END OF CHART

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