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

Physician: Doctor, Example

Patient: Patient, Example

DOB	10/17/2002
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Gamma Globin (HBG1 and HBG2) Sequencing

ARUP test code 3001957

Specimen HBG FGS	Whole Blood

HBG FGS Interpretation

Negative

RESULT
No pathogenic variants were detected in the HBG1 and HBG2 genes.

INTERPRETATION
No pathogenic variants were detected in the HBG1 or HBG2 genes using bidirectional sequencing of all coding regions, intron/exon boundaries, and proximal promoters. This result reduces the likelihood of a gamma globinopathy or nondeletional gamma 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. If HPFH is suspected, consideration may be given to HBB deletion/duplication analysis (Deletion/Duplication Analysis by MLPA, ARUP test code 3003144) to assess for deletional HPFH. If suspicion for a hemoglobinopathy remains, consideration may be given to Hemoglobin Reflexive Cascade (ARUP test code 2005792). If unexplained hemolytic anemia or neonatal hyperbilirubinemia is present, consideration may be given to the Hereditary Hemolytic Anemia Panel Sequencing (ARUP test code 2012052) to assess for additional genetic etiologies.

COMMENTS
Reference Sequences: GenBank # NM_000559.2 (HBG1), NM_000184.2 (HBG2)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Gamma Globin (HBG1 and HBG2) Sequencing
CHARACTERISTICS: Variants in the gamma globin genes, HBG1 and HBG2, may occasionally result in either a quantitative defect (gamma thalassemia or nondeletional hereditary persistence of fetal hemoglobin) or a qualitative abnormality (gamma variant). Gamma variants resulting in unstable, high- and low-oxygen affinity or M hemoglobin variants may result in hemolytic anemia/hyperbilirubinemia, erythrocytosis/cyanosis, or methemoglobinemia in neonates, respectively. Clinical symptoms related to gamma globin variants commonly resolve after the first six months of life given the switch from fetal hemoglobin expression to adult hemoglobin expression.
INCIDENCE: Unknown.
INHERITANCE: Autosomal dominant.
CAUSE: Pathogenic germline variants in HBG1 or HBG2.
CLINICAL SENSITIVITY: Unknown. Gamma globin variants are a rare cause of neonatal hemolytic anemia, cyanosis, erythrocytosis, or methemoglobinemia.
METHODOLOGY: Long range PCR followed by nested PCR and bidirectional sequencing of all coding regions, intron-exon boundaries, and 5' proximal promoters of the HBG1 and HBG2 genes.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations or repeat element insertions. Large deletions/duplications, distal regulatory region variants, deep intronic variants, and hybrid gene events 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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Specimen HBG FGS	24-092-400356	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HBG FGS Interpretation	24-092-400356	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