

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 9/28/2000
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Beta Globin (HBB) Sequencing, Fetal

ARUP test code 3004550

Maternal Contamination Study Fetal Spec	Fetal Cells	Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	whole Blood	
BG FE Specimen	Cultured Amnio	
BG FE Interp	Negative	

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

RESULT

No pathogenic variants were detected in the HBB gene.

INTERPRETATION

No pathogenic variants, including the familial Hbs (p.Glu7Val) variant, were detected in this fetal sample in the coding regions, exon-intron boundaries, proximal promoter, 5' untranslated region, or 3' polyadenylation signal of the beta globin (HBB) gene. None of the additional targeted HBB pathogenic intronic variants, IVS-I-110, IVS-II-654, IVS-II-705, and IVS-II-745, were detected. This result decreases the likelihood of, but does not exclude, the possibility of beta thalassemia in this fetus as not all pathogenic HBB gene variants are detected by this assay. The clinical presentation may vary due to other genetic modifiers or co-existing conditions.

Please refer to the background information included in this report for the methodology and limitations of this test.

RECOMMENDATIONS

Genetic consultation is recommended.

COMMENTS

Likely benign and benign variants are not reported. Variants in the following region(s) may not be detected by NGS with sufficient confidence in this sample due to technical limitations: None

This result has been reviewed and approved by [REDACTED]

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BACKGROUND INFORMATION: Beta Globin (HBB) Sequencing, Fetal

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. Structural hemoglobinopathies may result in sickling disorders, microcytic or hemolytic anemia, cyanosis, or erythrocytosis.

EPIDEMIOLOGY: Incidence varies by ethnicity.

CAUSE: Pathogenic germline variants within the HBB gene.

INHERITANCE: Usually autosomal recessive, infrequently autosomal dominant.

CLINICAL SENSITIVITY: Up to 99 percent, depending upon ethnicity, for beta thalassemia and hemoglobinopathies associated with the HBB gene.

GENE TESTED: HBB (NM_000518)
Deletion/duplication detection is not available for this gene.

METHODOLOGY: Probe hybridization-based capture of all coding exons, exon-intron junctions, 5' proximal promoter and untranslated region, 3' polyadenylation signal, and intronic variants c.93-21G>A (IVS-I-110), c.316-197C>T (IVS-II-654), c.316-146T>G (IVS-II-705), and c.316-106C>G (IVS-II-745) of the HBB gene, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants that do not meet acceptable quality metrics.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.

LIMITATIONS: A negative result does not exclude a diagnosis of beta thalassemia. This test detects variants within the coding regions and intron-exon boundaries of the HBB gene. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants upstream of c.-250, deep intronic variants (other than those described in methodology section above), and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) variants, or repeat expansions. Noncoding transcripts were not analyzed.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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.

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VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	24-051-160986	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	24-051-160986	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
BG FE Specimen	24-051-160986	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
BG FE Interp	24-051-160986	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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 24-051-160986
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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