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

Physician: Doctor, Example

Patient: Patient, Example

DOB	7/22/1993
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Glycogen Storage Disorders Panel, Sequencing

ARUP test code 3001627

Glycogen Storage Disease Specimen	Whole Blood

GSD NGS Interp: Negative

RESULT
No pathogenic variants were detected in any of the genes tested.

INTERPRETATION
No pathogenic variants were detected in any of the genes tested. This result decreases the likelihood of, but does not exclude, a heritable form of a glycogen storage disease or related disorder. Please refer to the background information included in this report for a list of the genes analyzed, methodology, and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and biochemical/functional assays. If this individual has a family history, determination of a causative familial variant in an affected family member is necessary for optimal interpretation of this negative result. Further testing may be warranted if there is a familial variant that is not detectable by this assay. 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]

BACKGROUND INFORMATION: Glycogen Storage Disorders Panel, Sequencing

CHARACTERISTICS: Glycogen storage diseases (GSD) are a group of inborn errors of metabolism, typically caused by enzyme defects, resulting in a buildup of glycogen in the liver, muscles, and other organs. Common clinical features of these disorders include hepatomegaly, hypoglycemia, slow growth, cardiomyopathy, and muscle weakness. Other disorders with a similar clinical presentation to GSD are included on this panel.

EPIDEMIOLOGY: Incidence of GSD ranges from 1 in 10,000 to 1 in one million, depending on specific types and ethnic backgrounds.

CAUSE: Pathogenic germline variants in the GYS1, G6PC, SLC37A4, GAA, AGL, GBE1, PYGM, PYGL, PFKM, PHKA2, PHKB, PHKG2, PHKA1,

PGAM2, SLC2A2, ALDOA, ENO3, and GYG1 genes are associated with glycogen storage diseases.
Pathogenic germline variants in the ACAT1, ALDOB, CPT2, FBP1, GYS2, LAMP2, LDHA, NHLRC1, OXCT1, PGK1, PGM1, PRKAG2, RBCK1, and SLC16A1 genes are associated with disorders that have phenotypes similar to GSD.

INHERITANCE: Autosomal recessive; X-linked recessive for PHKA1 and PHKA2 genes.

PENETRANCE: Variable

CLINICAL SENSITIVITY: Variable, depending on GSD type and subtype.

GENES TESTED: ACAT1, AGL, ALDOA, ALDOB, CPT2, ENO3*, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, OXCT1*, PFKM*, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4

*One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

METHODOLOGY: Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and confirm reported variants.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants 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 glycogen storage disorder. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants and deep intronic variants will not be identified, including GBE1 (NM_000158.4) intron 15. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

The following regions are not sequenced due to technical limitations of the assay:
ENO3(NM_001374524) exon(s) 1
OXCT1(NM_001364299) exon(s) 5
OXCT1(NM_001364300) exon(s) 1
OXCT1(NM_001364303) exon(s) 1
PFKM(NM_001354735) exon(s) 4
PFKM(NM_001354736) exon(s) 4
PFKM(NM_001354740) exon(s) 1
PFKM(NM_001354741) exon(s) 2
The following may not be detected:
An Ashkenazi Jewish founder mutation in GBE1 (HGMD ID: CX153579)

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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Glycogen Storage Disease Specimen	23-332-131265	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
GSD NGS Interp	23-332-131265	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