ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

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

Glycogen Storage Disease, Type 1A (G6PC), 9 Variants

ARUP test code 2013740

Glycogen Storage Disease, Specimen	Whole Blood
Glycogen Storage Disease, Allele 1	c.248G>A *
Glycogen Storage Disease, Allele 2	Negative
Glycogen Storage Disease, Interp	See Note

Indication for testing: Carrier screening or diagnostic testing for glycogen storage disease type 1A.

Positive: One pathogenic variant, p.R83H (c.248G>A), was detected in the G6PC gene; therefore this individual is at least a carrier of glycogen storage disease type 1A. At-risk family members should be offered testing to determine carrier status for the identified variant. This individual's reproductive partner should be offered screening for the disorder.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Glycogen Storage Disease, Type 1A (G6PC), 9 Variants
CHARACTERISTICS: Infants typically present at 3 to 4 months of age with hepatomegaly, lactic acidosis, hyperuricemia, hyperlipidemia, hypertriglyceridemia and/or hypoglycemic seizures. Other characteristics include growth delay leading to short stature, osteoporosis, delayed puberty, renal disease, and hepatic adenomas with potential for malignancy. With treatment, affected individuals often live into adulthood.
INCIDENCE: 1 in 20,000 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: G6PC pathogenic variants.
VARIANTS TESTED: p.Q27Rfs (c.79delC), Y128Tfs (c.379_380dupTA), p.R83H (c.248G>A), p.R83C (c.247C>T), p.G188R (c.562G>C), p.Q242X (c.724C>T), p.Q347X (c.1039C>T), p.G270V (c.809G>T), p.F327del (c.979_981delTTC).
CLINICAL SENSITIVITY: 99 percent in Ashkenazi Jewish individuals; varies by ethnicity in non-Ashkenazi Jewish individuals.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than those tested will not be detected. Diagnostic errors can occur due to rare sequence variations.

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
Glycogen Storage Disease, Specimen	23-062-104354	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Allele 1	23-062-104354	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Allele 2	23-062-104354	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Interp	23-062-104354	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