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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
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	Negative
Glycogen Storage Disease, Allele 2	Negative
Glycogen Storage Disease, Interp	See Note

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

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

Negative: This sample is negative for the 9 variants tested in the G6PC gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her risk of being a carrier of glycogen storage disease type 1A has been reduced from 1 in 71 to approximately 1 in 7,000.

This result has been reviewed and approved by Pinar Bayrak-Toydemir, M.D., Ph.D.

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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Glycogen Storage Disease, Specimen	19-170-104498	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Allele 1	19-170-104498	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Allele 2	19-170-104498	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Glycogen Storage Disease, Interp	19-170-104498	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