ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	5/24/1952
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) Sequencing

ARUP test code 2007163

G6PD Deficiency (G6PD) Seq Specimen	Whole Blood

G6PD Deficiency (G6PD) Seq Interp

Negative

TEST PERFORMED - 2007163
TEST DESCRIPTION - Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the G6PD gene.

INTERPRETATION
No pathogenic variants were detected in the glucose-6-phosphate dehydrogenase (G6PD) gene using bidirectional sequencing of all coding regions and intron/exon boundaries. This result significantly decreases, but does not exclude, a diagnosis of or carrier status for G6PD deficiency.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_001042351.1 (G6PD)
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: Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) Sequencing

Characteristics: G6PD deficiency can cause chronic hemolytic anemia, food-, drug- and infection-mediated hemolytic anemia, and acute hemolytic anemia with jaundice in the newborn - which can be potentially life-threatening. Ethnic-specific variants are common in individuals of African, Southeast Asian and Mediterranean descent. Most mutations identified to-date have been classified according to the following scheme: Class I - severe enzyme deficiency with chronic non-spherocytic hemolytic anemia (CNSHA); Class II - severe enzyme deficiency with less than 10 percent of the normal activity; Class III - mild to moderate enzyme deficiency (10 to 60 percent of normal activity); and Class IV - very mild to almost normal enzyme activity (greater than 60 percent normal activity with no clinical consequences).
Incidence: Varies by ethnicity; 7 in 10 Kurdish Jewish males; 1 in 6 to 10 African American males; 1 in 7 to 9 Arabic males; 1 in 6 to 16 Southeast Asian males.
Inheritance: X-linked recessive.
Penetrance: Variable depending on mutation and sex.
Cause: Pathogenic mutations in Glucose-6-Phosphate Dehydrogenase (G6PD) gene.
Clinical Sensitivity: Expected greater than 98 percent.
Methodology: Bidirectional sequencing of the entire G6PD coding region and intron-exon boundaries.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications will not be detected. Mutations in genes other than G6PD are not evaluated.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
G6PD Deficiency (G6PD) Seq Specimen	20-084-402434	3/24/2020 9:54:00 AM	3/25/2020 11:03:58 PM	4/10/2020 2:33:00 PM
G6PD Deficiency (G6PD) Seq Interp	20-084-402434	3/24/2020 9:54:00 AM	3/25/2020 11:03:58 PM	4/10/2020 2:33:00 PM

END OF CHART