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

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]/2014
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations

ARUP test code 0051684

G6PD Africa Specimen	Whole Blood
G6PD Allele 1	A376G/G202A *
G6PD Allele 2	Not Applicable
G6PD Mutations Interpretation	See Note

Indication for testing: Diagnostic or risk assessment for G6PD deficiency
Result: ONE COPY OF THE A- COMPLEX VARIANT WAS DETECTED IN THE G6PD GENE
Nucleic Acid Changes: c.[202G>A;376A>G]; Hemizygous
Amino Acid Alterations: p.[Val68Met;Asn126Asp]
Also Known As: G6PD-A(-)

Interpretation:
One copy each of the c.202G>A and c.376A>G variants were detected in the glucose-6-phosphate dehydrogenase (G6PD) gene by targeted analysis, representing the common A- complex allele. The A- allele is a Class III pathogenic variant associated with a mild to moderate decrease in enzyme activity. Thus, this individual is predicted to have G6PD deficiency. All female offspring, but no male offspring, will inherit the A- allele and may be at risk for symptoms.

Recommendations:
Hematologic and genetic consultations are recommended. Family members should be offered testing for the A- complex variant (Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations, ARUP test code 0051684). Medical screening and management should rely on clinical findings and family history.

Reference Sequence: GenBank # NM_001042351.2
Nucleotide numbering begins at the "A" of the ATG initiation codon.

This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations
CHARACTERISTICS: Although G6PD deficiency is usually asymptomatic, it can result in episodic hemolytic anemia triggered by infections, specific foods, and drugs. In newborns, it may be causal for life-threatening acute hemolytic anemia with jaundice. Variants are classified as follows: Class I - severe enzyme deficiency associated with chronic nonspherocytic hemolytic anemia; Class II - severe enzyme deficiency (<10 percent of normal activity); Class III - mild to moderate enzyme deficiency (10-60 percent of normal activity); and Class IV - normal range (>60 percent of normal enzyme activity). G6PD deficiency is best managed by avoiding known environmental triggers. For a list of drugs that may cause adverse reactions in individuals with G6PD deficiency refer to the Clinical Pharmacogenetics Implementation Consortium: https://cpicpgx.org/genes-drugs/.
INCIDENCE: Highly variable but ranges between 5-30 percent in males of African, Asian, Mediterranean, and Middle Eastern descent
INHERITANCE: X-linked.
CAUSE: Hemizygosity for a pathogenic G6PD germline variant in men, and homozygosity or compound heterozygosity in women. Some heterozygous women may be affected due to skewed X-chromosome inactivation.
VARIANTS TESTED: c.376A>G and c.202G>A (A- allele: both variants present in cis; A+ allele: c.376A>G alone; c.202G>A is rarely if ever seen alone).
CLINICAL SENSITIVITY: Variable; dependent on the country of origin.
METHODOLOGY: Polymerase Chain Reaction/Fluorescence Monitoring
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Only the two G6PD gene variants targeted (c.376A>G and c.202G>A) will be detected. This assay cannot determine phase; thus, concurrent detection of c.376A>G and c.202G>A is presumed to reflect the complex A- allele. Diagnostic errors can occur due to rare sequence variations. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation.

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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
G6PD Africa Specimen	25-087-400386	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
G6PD Allele 1	25-087-400386	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
G6PD Allele 2	25-087-400386	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
G6PD Mutations Interpretation	25-087-400386	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