

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 2/21/2022
Sex: Male
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 3004457

Spcm G6PD

Whole Blood

G6PD Interp

Negative

RESULT

No pathogenic variants were detected in the G6PD gene.

INTERPRETATION

No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the G6PD gene. This result decreases the likelihood of, but does not exclude, a diagnosis of glucose-6-phosphate dehydrogenase (G6PD) deficiency. Please refer to the background information included in this report for limitations of this test.

RECOMMENDATIONS

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

COMMENTS

Likely benign and benign variants are not included in this report.

This result has been reviewed and approved by [REDACTED]

BACKGROUND INFORMATION: Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD) Sequencing

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:

<https://cpicpgx.org/genes-drugs/>.

EPIDEMIOLOGY: Highly variable but ranges between 5-30 percent in males of African, Asian, Mediterranean, and Middle Eastern descent.

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

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 22-054-101197
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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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.

INHERITANCE: X-linked.

PENETRANCE: Low.

CLINICAL SENSITIVITY: 98 percent.

GENE TESTED: G6PD (NM_001042351)

METHODOLOGY: Probe hybridization-based capture of all coding exons and exon-intron junctions of the G6PD gene, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and to confirm reported variants that do not meet acceptable quality metrics. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes.

LIMITATIONS: A negative result does not exclude a diagnosis of G6PD deficiency. This test only detects variants within the coding regions and intron-exon boundaries of the G6PD gene. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants, deep intronic variants, and large deletions/duplications will not be identified. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) variants, or repeat expansions. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Specm G6PD	22-054-101197	2/23/2022 8:49:00 AM	2/23/2022 8:49:19 AM	2/23/2022 8:50:00 AM
G6PD Interp	22-054-101197	2/23/2022 8:49:00 AM	2/23/2022 8:49:19 AM	2/23/2022 8:50:00 AM

END OF CHART

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

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