TEST CHANGE

Glucose-6-Phosphate Dehydrogenase (G6PD) 2 Mutations

0051684, G6PD AFRIC

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavender (EDTA), pink (K2EDTA), or yellow (ACD sSolution A or B).
Specimen Preparation:	Transport 3 mL whole blood. (Min: 1 mL)
Transport Temperature:	Refrigerated.
Unacceptable Conditions:	Frozen specimens in glass collection tubes.
Remarks:	
Stability:	Ambient: 72 hours; Refrigerated: 1 week; Frozen: 1 month
Methodology:	Polymerase Chain Reaction (PCR) /)/Fluorescence Monitoring
Performed:	VariesMon, Thu
Reported:	4-10 days
Note:	This assay detects the following variants: c.376A>G and c.202G>A in the G6PD gene.
CPT Codes:	81247
New York DOH Approval Status:	This test is New York DOH approved.

Interpretive Data:

Refer to report. Background Information for 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.

Reference Interval: