MAYO CLINIC LABORATORIES

1-800-533-1710

G6PDZ

Glucose-6-Phosphate Dehydrogenase (G6PD) Full Gene Sequencing, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
321	TESTRNV, TESTING ATLAS		1968-02-19	M	55
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
X100427663	X100427663	Test,Atlas			
Account Information		**Collected**			
C7028846 DLMP Rochester		25 Aug 2023 06:33			

G6PD Full Gene Sequencing, V

G6PD Phenotype

MCR

Normal G6PD activity expected

Result Details

MCR

No reportable variants were detected in the G6PD gene.

Interpretation

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No reportable variants were detected in G6PD. There is no evidence of carrier status or affected status for G6PD deficiency. In addition, this result suggests that this individual is not at increased risk for developing acute hemolytic anemia when taking drugs contraindicated in individuals with G6PD deficiency.

Some individuals with features of G6PD deficiency and involvement of the G6PD gene may have a pathogenic variant that is not detectable by the method utilized. Additionally, if a clinical phenotype has been observed in this individual and/or family, it may be due to a pathogenic variant(s) in another gene(s).

Clinical correlation is recommended. A genetic consultation may be of benefit.

Additional Information

MCR

The following is based on the publication entitled "Technical consultation to review the classification of glucose-6-phosphate dehydrogenase (G6PD), 25 and 27 January 2022, virtual meeting":

Updated WHO Classification of G6PD Variants in Homozygous And Hemizygous Individuals (2022) Note: This system is used for classifying genetic variants, not individual patients

Class	Median G6PD Activity	Hemolysis
A	<20%	Chronic (CNSHA)
B	<45%	Acute, triggered
C	60–150%	No hemolysis
U	Any	Uncertain significance

Global Malaria Programme, Malaria Policy Advisory Group. (2022). Meeting report of the technical consultation to review the classification of glucose-6-phosphate dehydrogenase (G6PD). WHO Reference Number: WHO/UCN/GMP/MPAG/2022.01. Available at www.who.int/publications/m/item/WHO-UCN-GMP-MPAG-2022.01 Accessed July 29, 2022.

Legacy WHO Classification

G6PD Classification	Residual Enzyme Activity (% normal)
Class I	<10% with CNSHA
Class II	<10%
Class III	10–60%
Class IV	60%-150%
Class V	More than twice normal

Method

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Bidirectional fluorescent DNA sequence analysis was used to test for the presence of variants in the G6PD gene (transcript NM_001042351.2; build GRCh37 (hg19)) for all exons and intron-exon boundaries.

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

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Disclaimer

[1] MCR

Evaluation and categorization of variants is performed using available literature and variant classification guidelines. At this time, it is not standard practice for the laboratory to systematically re-review likely pathogenic variants or variants of uncertain significance that are detected and reported. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time. Consultation with genetics professional should be considered for interpretation of this result.

A list of benign and likely benign variants identified for this patient is available from the laboratory upon request.

Multiple in silico evaluation tools may be used to assist in the interpretation of these results and predictions made by these tools may change over time.

Patients with negative G6PD sequencing results may still experience hemolysis either due to genetic variation that is not identified by the methods utilized here or due to other factors.

CAUTIONS :

Rare variants may be present that could impact results. This assay may not detect large deletions or rearrangements that may affect G6PD protein expression. If more than one heterozygous variant is detected, the cis/trans relationship (i.e. whether the variants are inherited on the same or opposite alleles) of these variants typically cannot be definitively determined with the methods used by this assay. In addition, the reported zygosity of variants is based on reported patient sex. Note, due to skewed X-chromosome inactivation, elderly heterozygous carriers may be at increased risk of G6PD deficiency. Therefore, test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Samples may contain donor DNA if obtained from patients who received non-leukocyte reduced blood transfusions or allogeneic hematopoietic stem cell transplantation. Results from samples obtained under these circumstances may not accurately reflect the recipient's genotype. For individuals who have received blood transfusions, the genotype usually reverts to that of the recipient within 6 weeks. For individuals who have received allogeneic hematopoietic stem cell transplantation, a pretransplant DNA specimen is recommended for testing.

For a list of drugs associated with increased risk for hemolysis in G6PD-deficient individuals see the Pharmacogenomics Association Tables on the Mayo Clinic Laboratories webpage, www.mayocliniclabs.com or the list available at the following page: www.g6pd.org/en/G6PDDeficiency/SafeUnsafe/DaEvitare_ISS-it Please note that the information at these links is educational material intended for health care professionals and may not be comprehensive. This educational material is not intended to supersede the care provider's experience and knowledge of her/his patient to establish a diagnosis or a treatment plan. All medications require careful clinical monitoring. If results do not correlate with this individual's clinical presentation, consider G6PD enzymatic studies or other laboratory testing. For information about G6PD enzymatic studies, please contact the laboratory at 1–800–533–1710 or the online test catalog at www.mayocliniclabs.com about the test G6PD Enzyme Activity, B (test code G6PD1).

Reviewed By

MCR

Ann Marie Moyer, M.D., Ph.D.

Received: 25 Aug 2023 11:41 **Reported:** 29 Aug 2023 16:11

Laboratory Notes

[1] This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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