MAYO CLINIC LABORATORIES

1-800-533-1710

G6PDB

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

Patient ID	Patient Name		Birth Date	Gender	Age
SA00008280	TESTINGRNV, G6PD NEG		1981-09-04	F	34
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00008280	SA00008280	CLIENT, CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		03 May 2016 00:00			

G6PD Full Gene Sequencing

Phenotype

MCR

Normal G6PD activity expected

No variants predicted to cause reduced G6PD activity were identified.

Interpretation

MCR

The absence of a G6PD deficient allele suggests that this individual is not at increased risk for developing acute hemolytic anemia when taking rasburicase or other drugs contraindicated in individuals with G6PD deficiency.

Additionally, this result reduces the likelihood of, but does not rule out, the diagnosis or carrier status of 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).

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing. Consultation with a genetics professional may be of benefit. 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.mayomedicallaboratories.com about the test G-6-PD, QN, RBC (test code G6PD).

ADDITIONAL INFORMATION

VARIANT EVALUATION

Evaluation and categorization of variants is performed using the most recent published ACMG recommendations as a guideline. Variants are classified based on known, predicted, or possible pathogenicity and reported with interpretive comments detailing their potential or known significance.

In addition, the World Health Organization (WHO) classification system for G6PD variants was referenced to assign identified variants to one of the four following G6PD variant classes based on published literature (Relling et al. 2014. Clin Pharmacol Ther. 96(2):169–174):

WHO Class I variant: severe G6PD enzyme deficiency with <10% of normal G6PD enzyme activity, associated with chronic nonspherocytic hemolytic anemia. WHO Class II variant: G6PD enzyme deficiency with <10% of normal G6PD enzyme activity, NOT associated with chronic nonspherocytic hemolytic anemia. WHO Class III variant: G6PD enzyme deficiency with 10–60% of normal G6PD enzyme activity, not associated with chronic nonspherocytic hemolytic anemia. WHO Class IV variant: Normal G6PD enzyme activity.

This result should be interpreted in the context of clinical findings, family history, and other laboratory testing. Consultation with a genetics professional may be of benefit. 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. Please note that the information at this link 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 G-6-PD, QN, RBC (test code G6PD).

Multiple in silico evaluation tools may be used to assist in the interpretation of these results. The accuracy of predictions made by in silico evaluation tools is highly dependent upon the data available for a given gene, and predictions made by these tools may change over time. Results from in silico evaluation tools should be interpreted with caution and professional clinical judgment. Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported.

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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RECLASSIFICATION OF VARIANTS POLICY

At this time, it is not standard practice for the laboratory to retrospectively 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.

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

CAUTIONS:

Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. If testing was performed because of a family history of G6PD deficiency, it is often useful to first test an affected family member. Identification of a pathogenic variant in an affected individual would allow for more informative testing of at risk individuals.

Samples may contain donor DNA if obtained from patients who received heterologous blood transfusions or allogeneic blood or marrow 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 blood or marrow transplantation, a pre-transplant DNA specimen is recommended for testing. Fluorescent DNA sequence analysis was used to test for the presence of variants in all 13 exons and intron/exon boundaries of the G6PD gene. Laboratory developed test.

Reviewed by MCR

Mary Karow

Received: 04 May 2016 13:20 **Reported:** 04 May 2016 13:50

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