Glucose-6-Phosphate Dehydrogenase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of glucose-6-phosphate dehydrogenase (G6PD) deficiency. Female carriers typically show no symptoms; however, female carriers have an increased chance to have a child with G6PD deficiency. Consultation with a genetic counselor for a more detailed risk assessment is recommended.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

There is a 50% chance for each of your sons to have G6PD deficiency and a 50% chance each of your daughters would be carriers for G6PD deficiency. Female carriers typically show no symptoms of G6PD deficiency. Prenatal diagnostic testing for G6PD deficiency is not recommended. Testing for G6PD deficiency should be considered for your children after birth, particularly if the child is a male. In the absence of symptoms, reflexive testing of your male partner or donor for G6PD deficiency is not indicated due to the X-linked inheritance pattern of the condition.

G6PD Explained

What is G6PD?

G6PD is the most common of all clinically significant enzyme defects. The enzyme (glucose-6-phosphate dehydrogenase) deficiency causes the destruction of red blood cells faster than the body can replace them. Certain triggers can lead to the increased destruction of red blood cells. These triggers include certain medications, infections, foods (ex. fava beans), and stress. When a person with G6PD deficiency is exposed to these triggers, they can experience hemolytic anemia. Symptoms of hemolytic anemia include paleness, yellowing of the skin and whites of the eyes (jaundice), dark urine, enlarged spleen, fatigue, shortness of breath, and rapid heart rate.

Prognosis

Prognosis is generally favorable. Individuals who experience a hemolytic episode usually fully recover with adequate treatment. With proper management, individuals with G6PD deficiency may never show symptoms and lead a normal life.

Treatment

Management of the disease is generally focused on avoiding triggers such as infection, certain drugs, and foods that induce hemolytic anemia. In severe episodes of hemolytic anemia, individuals may require blood transfusions.

Resources

G6PD Association
https://www.g6pd.org/en/Home.aspx

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/glucose-6-phosphate-dehydrogenase-deficiency

National Society of Genetic Counselors
https://www.nsgc.org/

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