Genetics Uncoded:

NxGen MDx

Facts about

Glucose-6-Phosphate Dehydrogenase Deficiency

What Your Test Results Mean

Carriers of Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD deficiency) have a common enzyme deficiency that is managed by avoiding a specific group of medications and beans that induce anemia. With proper management, individuals with G6PD deficiency may never show symptoms of disease and can lead a normal life.

G6PD Deficiency Explained

G6PD deficiency is the most common of all clinically significant enzyme defects. The enzyme deficiency causes red blood cells to undergo hemolysis faster than the body can replace them. Management of the disease is generally focused on avoiding infection, certain drugs, and fava beans that induce anemia. In severe episodes of hemolytic anemia, individuals may require blood transfusions. Infants with G6PD deficiency are at risk for neonatal jaundice. With proper management, individuals with G6PD dificiency may never show symptoms of disease and can lead a normal life.

How the Genetics Work

G6PD deficiency is caused by pathogenic variations in the *G6PD* gene. Because G6PD deficiency is located on the X chromosome, generally females have two copies of the *G6PD* gene while males only have one copy of the *G6PD* gene. Females inherit one copy from each parent while males inherit the *G6PD* gene from their mother. Females with a variant in one copy of the *G6PD* gene are classified as carriers and have a 50% risk to pass on G6PD deficiency to sons and a 50% risk to pass carrier status onto daughters. Females with a variant in both copies of the *G6PD* gene (homozygotes) and males with a variant in the *G6PD* gene are classified as having G6PD deficiency. Homozygous females will pass G6PD deficiency onto all of their male sons while males with G6PD deficiency will pass on carrier status to each of their female children. Males with G6PD deficiency are not at risk to have a male with G6PD deficiency unless their partners are carriers and/or affected.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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