Facts about Glucose-6-Phosphate Dehydrogenase Deficiency

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 deficiency may never show symptoms of disease and can lead a normal life.

How the Genetics Work
G6PD is caused by pathogenic variations in the G6PD gene. Because G6PD is located on the X chromosome, all 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 mothers. Females with a variant in one copy of the G6PD gene are classified as carriers and have a 50% risk to pass on G6PD 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. Homozygous females will pass G6PD onto all of their male sons while males with G6PD will pass on carrier status to each of their female children. Males with G6PD are not at risk to have a male with G6PD 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.