What Your Test Results Mean
Test results indicate that you are a carrier of Glucose-6-Phosphate Dehydrogenase Deficiency (G6PD). Female carriers typically show no symptoms of G6PD; however, carriers are at an increased risk of having a child with G6PD. Risk for the current or future pregnancies is sex dependent. Consultation with a genetic counselor is recommended.

Glucose-6-Phosphate Dehydrogenase Deficiency - Female Carriers Explained
G6PD 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 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 transfusion. Infants with G6PD are at risk for neonatal jaundice. With proper management, individuals with G6PD may never show symptoms of disease and can lead a normal life.

How the Genetics Work
The clinical features of G6PD can be explained by mutations 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. Female with a mutation in one copy of the G6PD gene are classified as carriers while males with a mutation in the G6PD gene are classified as having G6PD. Female carriers have a 50% chance of having an affected male and a 50% chance of having a female carrier.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

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.