Genetics Uncoded:

Facts about Sickle Cell Anemia

What Your Test Results Mean

Carriers typically show no symptoms of sickle cell anemia; however, carriers are at an increased risk of having a child with sickle cell anemia or other sickling hemoglobinopathies. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Sickle Cell Anemia Explained

Sickle cell anemia is an inherited disorder that affects red blood cells. Sickle cell anemia accounts for approximately 60-70% of sickle cell disease in the United States. Sickle cell anemia is a lifelong condition characterized by painful episodes, anemia, organ damage, infections, lung problems, leg ulcers, bone damage, and strokes. Individuals with sickle cell disease have rigid, sickle-shaped red blood cells. The abnormal red blood cells can clog blood vessels, preventing delivery of oxygen to tissues and organs, causing the symptoms of the disease.

Management of this disease is generally focused on good hydration, avoidance of extreme temperatures and fatigue, and pain management. Prophylactic penicillin is recommended in childhood to reduce the risk of infection. Hydroxyurea has been shown to decrease painful episodes and increase life span. Individuals with the disease may require joint replacement, hemodialysis, kidney transplantation, splenectomy, cholecystectomy, red blood cell exchange transfusion for strokes, and other treatments for symptoms of the disease. Stem cell transplantation is curative; however, identifying matched donors has proven to be a barrier to transplantation.

How the Genetics Work

The clinical features of sickle cell anemia can be explained by a single pathogenic variant, Glu6Val, in the *HBB* gene. All individuals have two copies of the *HBB* gene. Carriers of sickle cell anemia have the Glu6Val variant in one copy of the *HBB* gene while individuals with sickle cell anemia have the Glu6Val variant in both copies the *HBB* gene, one inherited from each parent.

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.

NxGenMDx, LLC
801 Broadway Ave NW, Suite 203
Grand Rapids, MI, 49504
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