Genetics Uncoded:

Facts about Beta Thalassemia

What Your Test Results Mean

Carriers typically show no symptoms of beta thalassemia. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Beta Thalassemia Explained

Beta thalassemia is an inherited disorder that affects red blood cells. The decrease in beta-globin leads to a relative excess of alpha-globin chains, causing the disease. Beta thalassemia major is a lifelong condition characterized by severe microcytic anemia, mild jaundice, bone deformities, and hepatosplenomegaly. Beta thalassemia minor is less severe and carriers of beta thalassemia are generally clinically normal.

Management of beta thalassemia major includes regular blood transfusions as well as chelation therapy to prevent iron overload. Without transfusion and proper treatment, beta thalassemia disease can be fatal within the first two years of life. 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 beta thalassemia can be explained by variants in the *HBB* gene. In general, individuals have two copies of the *HBB* gene. Carriers of beta thalassemia (beta thalassemia minor) have a variant in one copy of the *HBB* gene while individuals with beta thalassemia have variants in both copies of the *HBB* gene, one inherited from each parent. The severity of disease is dependent on the particular variants in the *HBB* gene. Individuals with one mild variant and one severe variant are classified as having beta thalassemia intermedia while individuals with two severe variants are classified as having beta thalassemia major.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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Revised 12/15/16