Genetics Uncoded:

Facts about

Alpha Thalassemia

What Your Test Results Mean

There are two types of alpha thalassemia carriers, alpha thalassemia silent carriers or alpha thalassemia trait. Silent carriers have no symptoms, while individuals with trait may be identified to have mild anemia in the presence of normal iron levels. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Alpha Thalassemia Explained

Alpha thalassemia is an inherited disorder caused by decreased synthesis of the hemoglobin alpha chain, a protein in the red blood cells required for oxygen delivery. There are two types of alpha thalassemia—Hb Bart syndrome (hydrops fetalis) and Hb H disease. In the most severe type, Hb Bart syndrome, pregnancies may result in stillbirth or early infant death. Anemia, enlargement of the spleen and liver, mild jaundice, and skeletal changes (particularly in the face) are general features of Hb H disease.

Management of Hb Bart syndrome is focused on care of the mother during pregnancy as well as supportive care in infancy, as it is typically a fatal disease. Individuals with Hb H disease require annual follow up with a hematologist and may require red blood cell transfusions. Other complications such as splenomegaly, gallstones, and leg ulcers may require appropriate medical or surgical treatment.

How the Genetics Work

The clinical features of alpha thalassemia can be explained by deletions or variants in the *HBA1* or *HBA2* alpha-globin genes. Typically, individuals have two copies of the *HBA1* and *HBA2* genes, for a total of four alleles encoding for the alpha chain component of hemoglobin in red blood cells. Individuals who are silent carriers have lost one of the alpha-globin allele (∂-/∂∂) and are clinically normal, whereas individuals with trait have lost two copies of the alpha-globin gene allele, either in *cis* or in *trans*, (--/∂∂.) or (∂-/∂-) and may have mild anemia. Individuals with Hb H disease have lost three of the alpha-globin gene alleles (∂-/--). In the most severe type of alpha thalassemia, Hb Bart syndrome, all four alpha-globin alleles are lost (--/--).

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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