Alpha Thalassemia (HBA1 / HBA2)

Alpha Thalassemia is caused by defects in the HBA1 or HBA2 genes. These genes encode for the protein alpha-globin, a component of hemoglobin. There are two forms of the disease: Hb Bart syndrome and HbH disease. The former is more severe, affecting unborn babies. They suffer from general edema (swelling from fluid buildup), anemia, heart defects, and enlargement of the liver and spleen. Most are stillborn, or die within a few days of birth. Carrying an Hb Bart fetus may be harmful to the mother. HbH disease involves moderate anemia, jaundice, and enlargement of the liver and spleen. Abnormal skeletal changes are sometimes seen. Symptoms may begin in either childhood or adulthood. Generally, those with HbH can live a near-normal lifespan, although some may need blood transfusions if anemia becomes severe.

The inheritance of the faulty genes is complex, but involves a number of categories of both carriers and those with symptoms. The disease is relatively common, particularly in South-East Asia. Other regions badly affected include India, the Middle East, Africa, and Mediterranean countries. Worldwide, about 1 in 48 people are carriers for the condition.

Sources

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