Beta Thalassemia (HBB)

Beta Thalassemia is caused by mutations in the HBB gene, which encodes for the protein beta-globin, a subunit of hemoglobin. There are two forms of the disease, beta thalassemia major and beta thalassemia intermedia, the latter being less severe. With beta thalassemia major, symptoms develop before the age of two. Severe anemia is common, necessitating frequent blood transfusions. Other symptoms include jaundice, skeletal defects, and enlargement of the heart, liver, and spleen. Delayed adolescence may occur. Over time, excess iron from transfusions builds up in the body, and needs to be removed by chelation drugs. Premature death from cardiac mortality is common, but decreasing as treatments improve. Thalassemia intermedia is associated with mild anemia, some skeletal abnormalities, and in some cases growth inhibition.

The worldwide incidence of beta-thalassemia is 1 in 100,000 new births. Regions with high levels of the disease include Mediterranean countries, the Middle-East, Central Asia, Africa, and the Far East. In the USA, those whose ancestors came from these regions have a higher risk of the disease than other ethnic groups. The mutated gene is inherited in an autosomal recessive manner, typically requiring both parents to be asymptomatic carriers of the faulty gene copy.

Sources

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