

Pathophysiology, clinical manifestations, and carrier detection in Thalassemia

Abstract

Thalassemias are genetic disorders inherited from a person's parents. Thalassemias are prevalent worldwide with 25,000 deaths in 2013. Highest rates are in the Mediterranean, Italy, Greece, Turkey, West Asia, North Africa, South Asian, and Southeast Asia. Highest carriers (30% of the population) in the Maldives. Thalassemias are classified into alpha-thalassemia, Beta-thalassemia, thalassemia intermedia, and beta thalassemia minor. The severity of alpha and beta thalassemia depends on how many of four genes for alpha or two genes for beta globin are missing. Hemoglobinopathies imply abnormalities in the globin proteins themselves. Health complications are mostly found in thalassemia major and intermediate patients. Signs and symptoms include severe anemia, poor growth and skeletal abnormalities during infancy. Untreated thalassemia major eventually leads to death, usually by heart failure. Diagnosis by hematologic tests, hemoglobin electrophoresis, and DNA analysis. Individuals with severe thalassemia require blood transfusion, drug therapy i.e., deferoxamine, deferasirox, deferiprone, and bone marrow transplant. Most drugs have side effects. Prevention includes premarital screening, carrier detection, and genetic counselling.