Ganga S. Pilli, Gen Med (Los Angel) 2021, Volume 09

Diagnosis of thalassemia

Ganga S. Pilli
KLE University, India

Thalassemia affects approximately 4.4 out of every 10,000 live births throughout the world. This condition causes both males and females to inherit the relevant gene mutations equally because it follows an autosomal pattern of inheritance with no preference for gender. The thalassemias are a group of inherited hematologic disorders caused by defects in the synthesis of hemoglobin chains. There are two main forms – alpha thalassemia and beta thalassemia. The symptoms and severity of beta thalassemia varies greatly from one person to another. A beta thalassemia major diagnosis is usually made during the first two years of life and individuals require regular blood transfusions and lifelong medical care to survive. There are different types of alpha thalassemia. The symptoms vary based on the type of alpha thalassemia that is inherited. Severe anaemia develops and is associated with fatigue, weakness, shortness of breath, dizziness, headaches and jaundice. Affected infants present with failure to thrive. Some infants become progressively develop pallor, feeding problems, diarrhoea, irritability and recurrent fever. Hepatomegaly and splenomegaly may also occur. Complications related specifically to Iron overload results from the blood transfusions required to treat individuals with beta thalassemia major. Thus, early diagnosis, proper treatment for prolongation of life and antenatal screening should be made aware in the families who carry thalassemia gene defect.

Biography

Ganga S. Pilli is working as a Professor, in the Department of Pathology in KLE University, India.