

Beta Thalassemia

Beta thalassemia is a rare, inherited blood disorder characterized by low levels of hemoglobin, a protein that resides in red blood cells and carries oxygen throughout the body

Beta thalassemia is often classified into three types:



Minor

Carriers are typically asymptomatic, but may have mild anemia



Intermedia

Causes mild to moderate anemia and may have variable transfusion needs; some patients may need more transfusions as they age



Major

Leads to severe anemia and may cause enlarged liver and spleen, resulting in a need for regular blood transfusions

Beta thalassemia can lead to:



Production of fewer and less healthy red blood cells



Potential to develop severe anemia



Other serious symptoms, such as abnormal blood clots

Key Statistics



The total annual incidence of symptomatic beta thalassemia is estimated at 1 in 100,000 people globally.



While beta thalassemia remains a rare disease, its prevalence has increased in the United States by approximately 7.5% over the last 50 years.



Beta thalassemia affects men and women equally



Beta thalassemia has been found to occur most frequently in people from Mediterranean countries, the Middle East, and Central and Southeast Asia

Diagnosis and Symptoms

Beta thalassemia is usually inherited in an autosomal recessive manner, which means **both copies of the HBB gene in each cell have mutations**. Healthcare professionals typically look at a person's medical history, symptoms, physical exam and laboratory test results to make a diagnosis.

Symptoms are often dependent on disease severity and treatment and can include:



Anemia



Bone and muscle abnormalities



Abnormalities of the spleen, liver and heart



Growth deficiencies



Hepatic and endocrine complications



Cardiac complications (pulmonary hypertension, arrhythmia, thrombosis)

Disease Management



Stem cell transplant: most common for patients < 16 years of age and/or those with an appropriate match

Individuals with beta thalassemia major and some with intermedia **require regular red blood cell transfusions**

Supplementation with **folic acid**, a B vitamin, boosts the production of red blood cells in certain individuals

Prognosis

In more advanced stages, **heart problems** such as congestive heart failure, abnormal heart rhythms (arrhythmias) and liver fibrosis may be associated with severe beta thalassemia and can impact a patient's survival.

Despite being a chronic illness, **beta thalassemia is still treatable**. With the proper care, patients with beta thalassemia can better manage their disorder.



Advances in red blood cell transfusions have further prolonged survival in recent years. Although there are therapies approved to treat anemia associated with beta thalassemia in multiple countries, it's important that there are further treatment advances for this underserved population.