Bristol Myers Squibb®

Thalassemia

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

Thalassemia is often classified into two types

Alpha thalassemia: occurs when the body doesn't make enough of the alpha hemaglobin protein chain to produce hemaglobin.

Beta thalassemia: occurs when the body doesn't make enough of the beta hemaglobin protein chain to produce hemaglobin.

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 World Health Organization (WHO) reports that, globally, 40,000 infants annually are born with thalassemia, and the majority of whom have beta thalassemia.

The prevalence of thalassemia has increased by approximately 7.5% in the United States over the last five decades, likely due to an influx of immigration from affected regions.

Thalassemia affects men and women equally.

Thalassemia has been found to occur most frequently in people from tropical and subtropical regions, including Mediterranean countries, Southeast Asia, the Indian subcontinent and Africa.

Diagnosis and symptoms

Both forms of thalassemia are usually inherited. In alpha thalassemia, the severity of the disease depends on the number of gene mutations inherited from your parents, the more mutations, the more severe. In beta thalassemia, the severity is dependent on which part of the hemoglobin molecule is impacted. 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 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 and liver problems** such as congestive heart failure, abnormal heart rhythms (arrhythmias) and liver fibrosis may be associated with severe thalassemia and can impact a patient's survival.

Despite being a chronic illness, **thalassemia is still treatable**. With the proper care, patients with 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 thalassemia in multiple countries, it's important that there are further treatment advances for this underserved population.