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 hemoglobin protein chain to produce hemoglobin.
- **Beta thalassemia**: occurs when the body doesn’t make enough of the beta hemoglobin protein chain to produce hemoglobin.

**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, 60,000 infants annually are born with thalassemia, and the majority of whom have beta thalassemia.
- Thalassemia affects men and women equally.

**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.