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**

- Globally, 1.5% of the population have the genetic mutation that causes one of three types of beta thalassemia.
- Beta thalassemia affects men and women equally.
- Beta thalassemia has been found to occur most frequently in people from Mediterranean countries, the Middle East, North Africa, India, 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 and liver 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.
- With proper disease management patients can enjoy a near-normal lifestyle and can experience normal physical and emotional development from childhood to adulthood, including parenthood.
- Advances in red blood cell transfusions have further prolonged survival in recent years. However, there continue to be very limited options for patients living with anaemia due to beta thalassemia who are dependent on long-term red blood cell transfusions.