SHALBY®
Sanar INTERNATIONAL HOSPITALS

+91-124-3528444

Golf Course Road, DLF Phase-5
Sector - 53, Haryana - 122002

www.sanarhospitals.com
info@sanarhospitals.com

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Understanding Thalassaemia

Thalassaemia is an inherited blood disorder that affects the body's ability to produce healthy red blood cells leading to lower haemoglobin levels. Haemoglobin is a protein present in red blood cells responsible for transporting oxygen throughout the body.

In Thalassaemia, the body produces fewer healthy haemoglobin proteins and the bone marrow produces very less healthy red blood cells. The condition of having fewer red blood cells is called Anaemia. As red blood cells serve the vital role of delivering oxygen to tissues in the body, lack of healthy red blood cells can deprive the body's cells of the oxygen they need to make energy and thrive.

Risk Factors

- **Family history:** Thalassaemia is passed from parents to children through mutated haemoglobin genes
- **Ancestral factor:** Thalassaemia occurs most often in African Americans and in people of Mediterranean and Southeast Asian descent. It is quite common in India as well.

Types

There are two types of Thalassaemia named after defects in the following chains:

- **Alpha Thalassaemia:** Inheritance of four genes, two from each parent, that make alpha globin protein chains. When one or more genes are defective, Alpha Thalassaemia develops. The number of defective genes inherited will determine whether the patient will experience anaemia symptoms.
- **Beta Thalassaemia:** Inheritance of two beta-globin genes, one from each parent. Symptoms of Anaemia and severity of the condition depends on how many genes are defective and which part of the beta globin protein chain contains the defect.

Symptoms

The symptoms for Thalassaemia depend on the type and severity of the condition and may include:

- Fatigue
- Weakness
- Pale or yellowish skin
- Facial bone deformities
- Slow growth
- Abdominal swelling
- Dark urine
- Growth retardation

Diagnosis

Moderate and Severe Thalassaemia are often diagnosed during childhood because symptoms usually appear within the first two years of a child's life. A healthcare provider may perform various blood tests to diagnose Thalassaemia such as Complete Blood Count, Reticulocyte Count, Iron Studies to understand the cause behind Anaemia.

It is confirmed by a simple test of Haemoglobin – Electrophoresis or HPLC and genetic testing.

Treatment

There are various treatment options for moderate to severe Thalassaemia such as:

- **Blood Transfusion:** Involves injections of red blood cells administered through a vein to restore normal levels of healthy red blood cells and haemoglobin
- **Iron Chelation:** A risk with blood transfusion is iron overload as excessive iron may damage internal organs. Iron Chelation is a process to remove excess iron from the body
- **Folic Acid Supplements:** Helps the body produce healthy red blood cells
- **Bone Marrow and Stem Cell Transplant Procedure:** Injecting healthy bone marrow stem cells from the donor into the recipient's bloodstream. The transplanted cells start to make new, healthy blood cells within one month of the transplant

Complications of Thalassaemia

- Iron overload
- Infection
- Bone deformities
- Delayed growth
- Heart problems
- Enlarged spleen

When to seek medical attention

If the parents detect delayed growth milestones in their child accompanied by symptoms of Thalassaemia like pale skin, lethargy and failure to thrive, a medical expert should be immediately consulted for investigative tests, proper diagnosis and treatment.