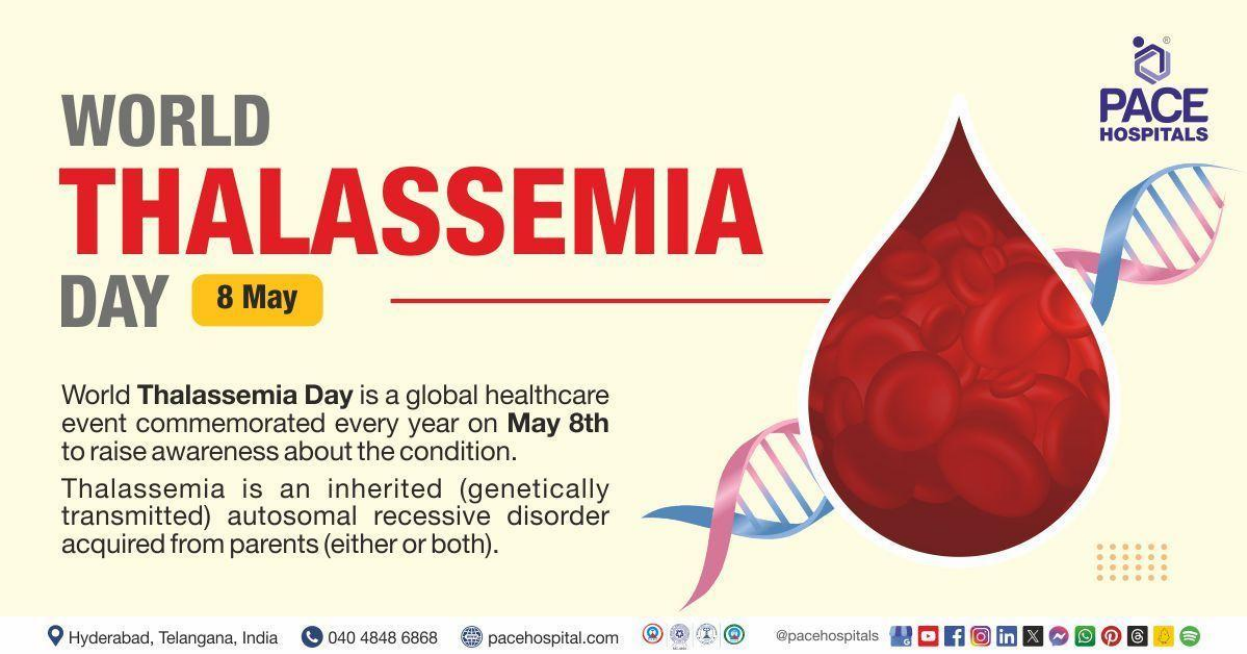
THALASSEMIA



Haemoglobin consists of 2 different pairs of peptide chains (alpha and beta) with the heam molecule attached to each peptide.
The thalassemia are inherited disorders characterized by reduced or absent synthesis of α or β globin polypeptide chain.

Classification:

1. Alpha (α) chain thalassemia – (deficient synthesis of α globulin chains)
2. Beta (β) chain thalassemia– (deficient synthesis of β globulin chains)

In India β thalassemia is more common in communities like Punjabis, Sindhis, Bengalis, Gujaratis, and Jains.

There are three clinical forms of thalassemia

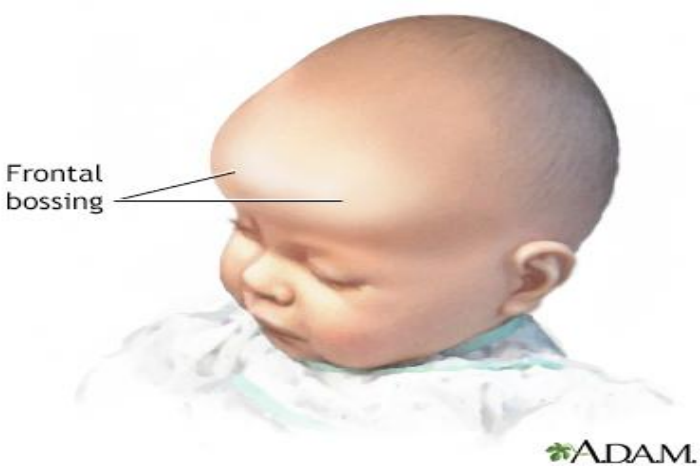
- Thalassemia major
- Thalassemia intermedia
- Thalassemia minor

β Thalassemia major: (homozygous β Thalassemia /Cooley's anemia)
βThalassemia major results when both the β globin genes are defective e.g. genotype β^0/β^0 or β^0/β^+. It is characterized by severe anemia, and requires regular blood transfusion therapy for survival.

Clinical feature of βThalassemia major are:

- Severe hemolytic anemia that develops around 6 months of age
- Severe growth retardation and delayed development
- Skeletal deformities (frontal bossing, malar prominence)
- Susceptibility to infection and to folate deficiency
- Transfusion dependence for survival.

In untreated patients death usually occurs before 5 years of age. In individuals who receive regular blood transfusion therapy, iron overload gradually develops during adolescence unless iron chelation therapy is given, such individuals will die prematurely form damage to organ like heart, pancreas and liver.



Laboratory feature:

- Hb - < 7 gm
- Reticulocytosis (immature RBC)
- Low – MCV, MCH, MCHC
- Hypochromia

β Thalassemia intermedia :-

This results from homozygous inheritance of mild β^{+}Thalassemia (e.g. genotypes β^{+}/β^{+}) it is characterized by moderate degree of anemia. Which does not require regular blood transfusion therapy. Worsening of anemia occurs during infections and pregnancy.

Thalassemia intermedia presents at a later age (2-5 yr.) than thalassemia major. These patients have chronic hemolytic anemia, splenomegaly, and skeletal changes.

Laboratory features :-

- Moderate degree of anemia (hb7-10 gm/dl)
- Blood smear shows less severe abnormalities than Thalassemia major.
- Reticulocytosis (5-10%)
- Variable elevation of hb F on electrophoresis.

β Thalassaemia minor :- (Thalassemia trait, asymptomatic Thalassemia)

Usually, individuals having one normal and one abnormal β globin gene have β Thalassemia minor (e.g. β/β^{+} or β/β^{0}) in this heterozygous carrier state, anemia is either mild or absent. This condition is often detected during the course of routine hematological investigations.

B Thalassemia trait should be distinguished from iron deficiency anemia as iron therapy is not required in the former. **Serum iron and serum ferritin low in iron deficiency anemia, while normal in β thalassemia minor.**

Laboratory features :-

- Hb. Slightly low or normal (>10 gm/dl)
- Low MCV, MCH
- Normal MCHC
- Blood smear shows microcytic hypochromic red cells, target cells and basophilic stippling
- Normal serum ferritin
- Hemoglobin electrophoresis shows elevated hemoglobin A_2 (>3.5%) this is a diagnostic test for β thalassemia minor.

Treatment;

Prevention

Blood transfusion

Gene therapy

Bone merrow transplant