Thalassemia
A group of inherited disorders characterized by reduced or absent amounts of hemoglobin, the oxygen-carrying protein inside the red blood cells.
• Types of Thalassemia
• 1) Thalassemia trait
• 2) Thalassemia major
• **Thalassemia trait**

• **People with thalassemia trait carry thalassemia, but they are not ill. They are healthy and normal, however, some may have slight anemia.**

• **People with thalassemia trait also have slightly more hemoglobin called hemoglobin A2 in their blood.**
• Thalassemia trait is present at birth, it remains the same for life, and it can be handed down from parents to children.
• Thalassemia major
• It is further divided into two types.
• 1) Alpha thalassemia
• 2) Beta thalassemia
• **Alpha thalassemia**

• Alpha thalassemia is the result of changes in the genes for the alpha globin component of hemoglobin.
There are two main types of alpha thalassemia disease.

1) hemoglobin H disease and alpha thalassemia major. The two diseases are quite different from beta thalassemia as well as from one another.
Individuals with hemoglobin H disease can experience events of hemolytic anemia—
anemia caused by the rapid breakdown of the red blood cells.

These events are thought to be triggered by various environmental causes, such as infection and/or exposure to certain chemicals.
Hemoglobin H disease is in most cases milder than beta thalassemia.
• **Alpha thalassemia major**

• It is a very serious disease that results in severe anemia that begins even before birth. Most affected babies do not survive to be born or die shortly after birth.
• **Beta thalassemia**

• Beta thalassemia may be the best-known type of thalassemia and is also called Cooley's anemia.

• It is caused by a change in the gene for the beta globin component of hemoglobin.

• Beta thalassemia causes variable anemia that can range from moderate to severe, depending in part on the exact genetic change underlying the disease.
• Beta thalassemia can be classified based on clinical symptoms.

• *Beta thalassemia major* usually causes severe anemia that can occur within months after birth.

• If left untreated, severe anemia can result in insufficient growth and development, as well as other characteristic physical complications that can lead to a dramatically decreased life-expectancy.
• *Beta thalassemia intermedia* describes the disease in individuals who have moderate anemia that only requires blood transfusions intermittently, if at all.
Pathophysiology of thalassemia

Two main features contribute to the pathogenesis of thalassemia.

1) Inadequate globin gene production leading to decreased level of normal hemoglobin.

2) Imbalance in alpha and beta globin gene production.
• In bone marrow there is disruption of the maturation of red blood cell leading to ineffective erythropoiesis.
• The marrow is hyperactive but the patient is relatively few reticulocytes and sever anemia.
Symptoms of Thalassemia

People with thalassemia major may experience the following:

- Paleness
- Headaches
- Fatigue
• Shortness of breath
• Jaundice
• Spleen enlargement
• Diagnosis of Thalassemia

• The diagnosis of thalassemia trait and thalassemia major is made from microscopic examination of the blood, which shows many small, pale red blood cells, and from other blood tests that show reduced levels of adult hemoglobin in the blood.
• **Treatment of Thalassemia**

• **Thalassemia trait**

• Normally, there are no treatments recommended. However, the doctor may suggest taking iron medication if they feel it is necessary.
• **Thalassemia major**

• The primary treatment is regular blood transfusions, usually every four weeks.

• In addition to the blood transfusions, doctors recommend injections of Desferal to help the body flush out the extra iron created by the new blood.

• The injections are given under the skin from a small pump 5 to 7 nights a week.
Additionally, splenectomy (removal of the spleen), bone marrow transplants and chelation therapy are being researched as possible treatments for thalassemia.
• Complication

• Many complication seen are due to increased iron diposition from repeated blood transfusions.

• Endocrine disfunction include hypothyroidism, gonadal failure, hypoparathyridism, and DM.
• CHF, and cardiac arrhythmias.
• Hemosiderosis.
• Risk of v. hepatitis due to repeated transfusion.
• New approaches to the transfusion management of thalassemia

• Recent advances in the treatment of patients with thalassemia major have centered around the removal of iron from individuals already overloaded due to repeated transfusions.

• In this report we present therapeutic maneuvers designed to decrease the rate of iron accumulation.
we describe a method for obtaining units of blood from normal donors that contain primarily young red cells ("neocytes").

These cells have prolonged in vivo survival as measured by the interval between transfusions (30 +/- 2.5 days to 43 +/- 4.5 days) and 51Cr red cell survival (43.8 days versus 27.8 days).

Supertransfusion with neocytes is effective in decreasing the rate of iron accumulation in thalassemia.
New Advances in Iron Chelation Therapy

Deferoxamine,

Administered regularly by subcutaneous infusion in appropriate doses, is a safe and effective approach to the treatment of transfusional iron overload.
- Deferiprone
- At a dose of 75 mg/kg/day, reduces or maintains iron stores in the majority of patients receiving regular red cell transfusions.
- Deferiprone alone readily achieves reductions in iron stores in patients with lower rates of transfusional iron loading than are found in thalassemia major.