

# THALASSEMIA



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*Shaping Lives...*  
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**By**

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## **INTRODUCTION:**

Thalassemia is a genetic blood disorder that affects the production of haemoglobin. Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. It is caused by mutations in genes that control the production of alpha or beta globin, the two proteins that make up hemoglobin.

## **TYPES:**

There are two main types of thalassemia: **alpha thalassemia and beta thalassemia.**

**Alpha thalassemia** is caused by mutations in the genes that control the production of alpha globin, a protein that is part of hemoglobin. There are four genes that control the production of alpha globin. Alpha thalassemia can be classified as:

**Silent carrier alpha thalassemia:** In this form of alpha thalassemia, one of the four alpha globin genes is missing or mutated. People with this form of the condition usually have no symptoms.

**Alpha thalassemia trait:** In this form of alpha thalassemia, two of the four alpha globin genes are missing or mutated. People with this form of the condition may have mild anemia, but often have no symptoms.

**Hemoglobin H disease:** In this form of alpha thalassemia, three of the four alpha globin genes are missing or mutated. People with this form of the condition may have moderate to severe anemia, as well as other symptoms such as fatigue, jaundice, and an enlarged spleen.

**Alpha thalassemia major or hydrops fetalis:** In this severe form of alpha thalassemia, all four alpha globin genes are missing or mutated. This can be life-threatening before or shortly after birth.

**Beta thalassemia** is caused by mutations in the genes that control the production of beta globin, another protein that is part of hemoglobin. There are two genes that control the production of beta globin, beta thalassemia can be classified as:

**Beta thalassemia minor or beta thalassemia trait:** In this form of beta thalassemia, one of the two beta globin genes is missing or mutated. People with this form of the condition may have mild anemia, but often have no symptoms.

**Beta thalassemia intermedia:** In this form of beta thalassemia, both beta globin genes are affected, but not completely absent. People with this form of the condition may have moderate to severe anemia, as well as other symptoms such as fatigue, jaundice, and an enlarged spleen.

**Beta thalassemia major or Cooley's anemia:** In this severe form of beta thalassemia, both beta globin genes are completely absent or severely mutated. This can be life-threatening in infancy or childhood and requires regular blood transfusions and other medical treatment.

#### **CAUSES/ETIOLOGY:**

- Genetic or Gene mutation

#### **PATHOPHYSIOLOGY:**

- Thalassemia develops in the body due to mutations in the genes that control the production of hemoglobin, it is a protein in red blood cells that carries oxygen throughout the body. The mutations can affect the amount or quality of hemoglobin

produced, leading to a decrease in the ability of the red blood cells to carry oxygen.

- In normal red blood cells, there are two alpha globin chains and two beta globin chains that make up the hemoglobin protein. In thalassemia, mutations in the genes that control the production of either the alpha or beta globin chains leads to a decrease in the amount or quality of hemoglobin production.
- Depending on the specific mutations and the extent of their effect on hemoglobin production, thalassemia can range from mild to severe.

### **CLINICAL MENIFESTATION/SIGN & SYMPTOMS:**

**The clinical symptoms of thalassemia can vary depending on the severity of the condition. This can lead to symptoms such as:**

- Fatigue and weakness
- Pale or yellowish skin
- Shortness of breath
- Dizziness or lightheadedness
- Headaches
- Cold hands and feet

**In more severe cases of thalassemia, other symptoms may be present, such as:**

- Growth and development problems, particularly in children
- Bone deformities, particularly in the face and skull
- Enlarged spleen or liver
- Abdominal swelling and pain
- Dark urine
- Increased risk of infections

## **DIAGNOSIS:**

- **Physical examination**
- **Family history & Medical history.**
- **Complete blood count (CBC):** This test measures the number of red blood cells, white blood cells, and platelets in the blood. People with thalassemia may have a decreased number of red blood cells and hemoglobin.
- **Hemoglobin electrophoresis:** This test measures the different types of hemoglobin in the blood, and can help distinguish between different types of thalassemia.
- **DNA analysis:** This test can detect specific genetic mutations that cause thalassemia, and can be used to diagnose carriers of the condition.
- **Prenatal testing:** If there is a family history of thalassemia or if a parent is a known carrier, prenatal testing may be done to diagnose thalassemia in a developing fetus.
- **Newborn screening:** Many countries have a newborn screening program that tests for certain genetic conditions, including thalassemia.

## **PHARMACOLOGICAL MANAGEMENT:**

- **Blood transfusions:** People with severe thalassemia may require regular blood transfusions to replace the deficient red blood cells. This can help improve symptoms of anemia and increase the overall quality of life.
- **Iron chelation therapy:** Regular blood transfusions can lead to iron overload in the body, which can cause organ damage over time. Iron chelation therapy involves the use of medications such

as deferoxamine, deferasirox, and deferiprone to remove excess iron from the body.

- **Folic acid supplements:** Folic acid is important for the production of red blood cells, and people with thalassemia may require higher doses of folic acid to support red blood cell production.
- **Bone marrow transplantation:** For people with severe thalassemia, bone marrow transplantation may be a curative treatment option. This involves replacing the defective bone marrow with healthy bone marrow from a donor.
- **Gene therapy:** Some experimental treatments for thalassemia involve gene therapy, which aims to correct the underlying genetic mutations that cause the condition.

## **NON PHARMACOLOGICAL MANAGEMENT/PREVENTION:**

- **Genetic counseling:** People who are carriers of thalassemia or who have a family history of the condition may benefit from genetic counseling. This involves meeting with a healthcare provider or genetic counselor who can discuss the risks and options for having a child with thalassemia.
- **Prenatal testing:** If one or both parents are carriers of thalassemia, prenatal testing may be done to determine whether the developing fetus has the condition.
- **Pre-implantation genetic diagnosis:** This is a type of genetic testing that can be done before implantation during in vitro fertilization (IVF). It involves testing the embryos for the

presence of thalassemia mutations before they are implanted in the uterus.

- **Donor sperm or egg:** In some cases, people with thalassemia may choose to use donor sperm or eggs to reduce the risk of passing on the condition to their children.