

# Difference Between Alpha and Beta Thalassemia

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## Key Difference – Alpha vs Beta Thalassemia

Thalassemia is a heterogeneous group of disorders caused by inherited mutations that decrease the synthesis of either alpha or beta globin chains, leading to anaemia, tissue hypoxia and red cell hemolysis related to the imbalance in globin chain synthesis. There are two major forms of thalassemia as alpha thalassemia and beta thalassemia. **In alpha thalassemia, there is a decrease in the number of alpha globin chains whereas in beta-thalassemia it is the number of beta globin chains that goes down.** This is the key difference between alpha and beta thalassemia.

## What is Alpha Thalassemia?

In alpha thalassemia, some of the genes that are responsible for the coding of the alpha globin chains are deleted. The alpha globin gene generally has four copies. The severity of the disease depends on the number of missing copies.

### Hydrops Fetalis

Synthesis of alpha globin chains is completely suppressed when all four copies of the alpha globin gene are missing. Since alpha globin chains are required for the synthesis of both fetal and adult haemoglobin, this condition is not compatible with life; therefore in utero termination of the pregnancy happens if the fetus is affected by this condition.

### HbH Disease

This condition is caused by the absence of three copies of the alpha globin gene. This results in moderate to severe hypochromic microcytic anaemia with associated splenomegaly.

### Alpha Thalassemia Traits

This is caused due to the absence or the inactivity of one or two copies of the alpha globin gene. Although the alpha thalassemia traits do not cause anaemia, they can

decrease mean corpuscular volume and mean corpuscular haemoglobin levels while increasing the red blood cell count over  $5.5 \times 10^{12}/L$ .

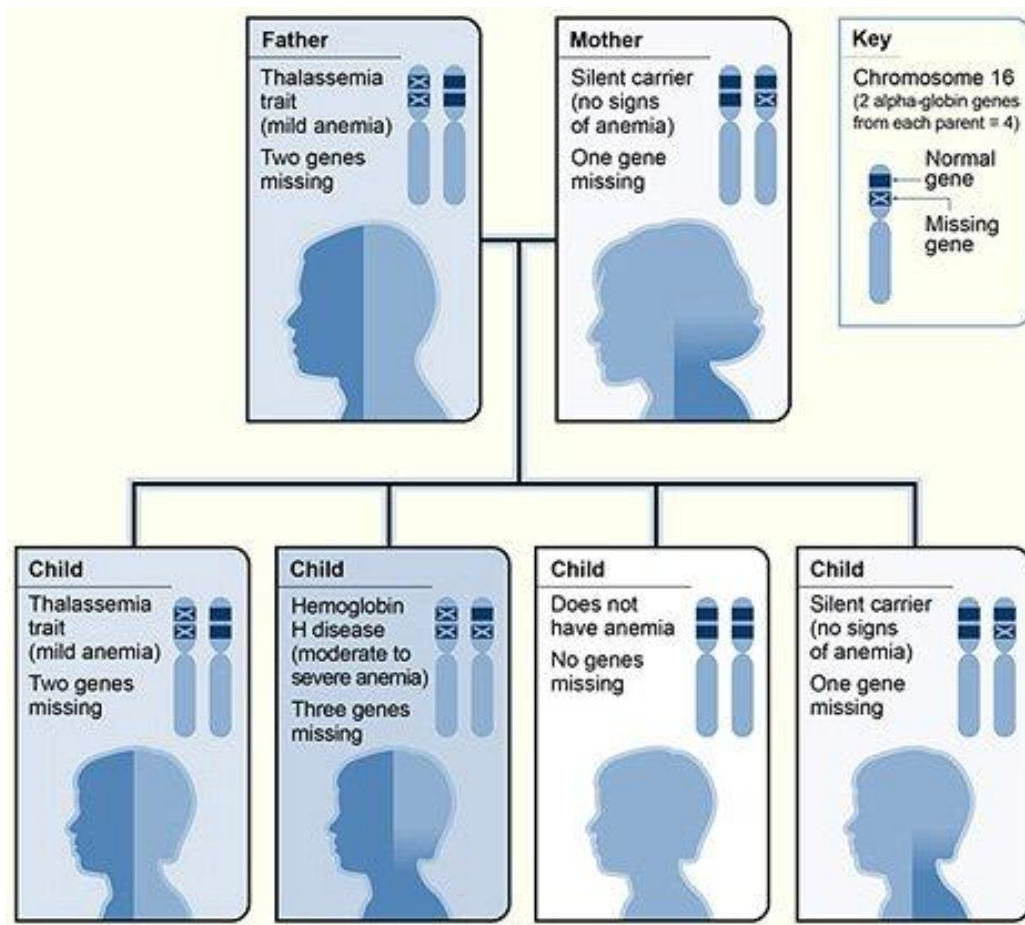


Figure 01: Inheritance of Alpha Thalassemia

Diagnosis of alpha thalassemia is through the globin chain synthesis studies.

## Management

Patients with a mild form of anaemia usually do not require any treatments. Administration of iron and folic acid is advocated only in some patients. Those with the severe forms of alpha thalassemia require a lifelong blood transfusion.

## What is Beta Thalassemia?

In beta thalassemia, the amount of beta globin chains goes down.

## **Beta Thalassemia Major**

If both parents are carriers of the beta thalassemia trait, the possibility of an offspring inheriting beta thalassemia major is 25%. In beta thalassemia major, the production of beta globin chains is either completely suppressed or drastically reduced. Since there aren't enough beta globin chains for them to combine with, the excess alpha globin chains get deposited in both mature and immature red cells. This leads to premature hemolysis of red cells and ineffective [erythropoiesis](#).

## **Clinical Features**

1. Severe anaemia, which becomes apparent at 3- 6 months after birth.
2. Splenomegaly and hepatomegaly
3. Thalassemic facies

The changes in facial features are due to the expansion of bones because of bone marrow [hyperplasia](#). The x-ray radiograph shows the hair-on-end appearance of the skull which is typically seen in beta thalassemia.

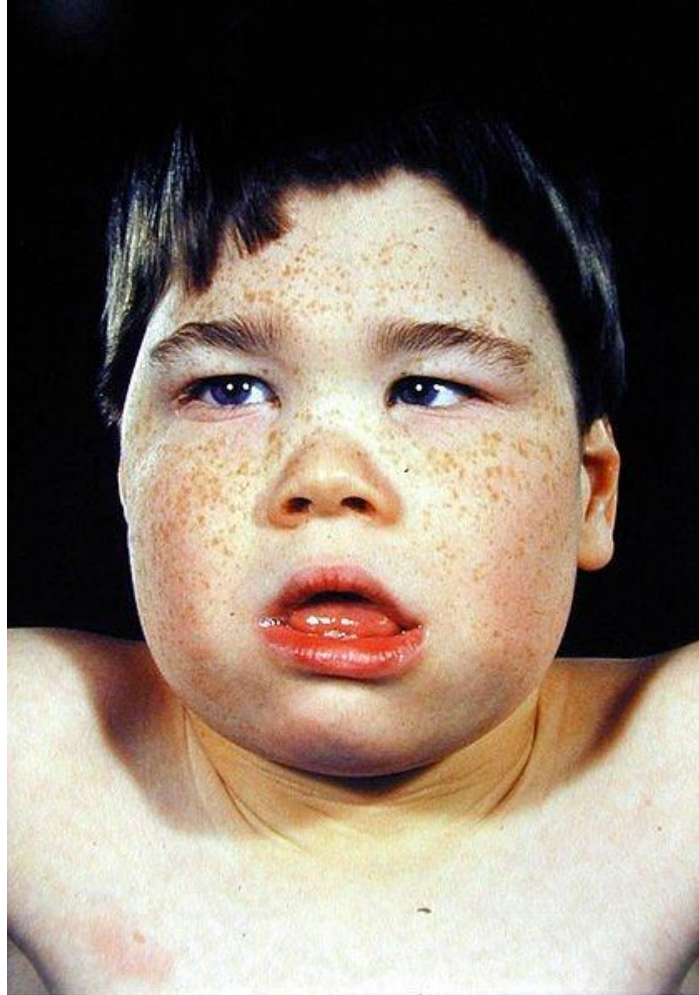


Figure 02: Thalassemic facies

## Laboratory Diagnosis

High-performance liquid chromatography (HPLC) is the major method used in the diagnosis of haematological diseases nowadays. Beta thalassemia major HPLC shows the presence of reduced levels of HbA with unusually high levels of HbF. A full blood count will reveal the existence of hypochromic microcytic anaemia, and the examination of a blood film will indicate the presence of increased amount of reticulocytes along with basophilic stippling and target cells.

## Treatment

- Regular blood transfusions
- Iron chelation therapy
- [Folic acid](#) (if the dietary intake of folic acid is not satisfactory)
- Splenectomy (sometimes used to reduce the blood requirement)
- Bone marrow transplantation

- Gene therapy for screening and therapeutic purposes

## Beta Thalassemia Trait/Minor

Beta thalassemia minor is a common condition which is often symptomless. Although the signs and symptoms are similar to those of alpha thalassemia, beta thalassemia is more severe than its counterpart. The diagnosis of beta thalassemia minor is made if the HbA<sub>2</sub> level is more than 3.5%.

## Thalassemia Intermedia

Thalassemia intermedia refers to cases of thalassemia of moderate severity that do not need regular transfusions.

## What is the Similarity Between Alpha and Beta Thalassemia?

- In both conditions, there is a decrease in the haemoglobin level of blood.

## What is the Difference Between Alpha and Beta Thalassemia?

Alpha vs Beta Thalassemia	
There is a decrease in the number of alpha globin chains.	There is a decrease in the number of beta globin chains.
Deletion of Genes	
Some of the genes responsible for the coding of the alpha globin chains are deleted.	Genes that are responsible for the synthesis of beta globin chains are either partially or completely deleted.
Types	

Hydrops fetalis, HbH disease and alpha thalassemia trait are the major forms of alpha thalassemia.

There are two main forms of beta thalassemia as beta thalassemia major and beta thalassemia minor.

### Diagnosis

Diagnosis of alpha thalassemia is through the globin chain synthesis studies.

High-performance liquid chromatography (HPLC) is the investigation used for the diagnosis of beta thalassemia.

### Clinical Features

In HbH disease, the absence of three copies of the alpha globin gene results in moderate to severe hypochromic microcytic anaemia with associated splenomegaly.

Alpha thalassemia traits do not cause anaemia, they can decrease the mean corpuscular volume and mean corpuscular haemoglobin levels while increasing the red blood cell count over  $5.5 \times 10^{12}/L$ .

- Severe anaemia which becomes apparent at 3- 6 months after birth.
- Splenomegaly and hepatomegaly
- Thalassemic facies

### Treatment and Management

- Patients with a mild form of anaemia usually do not require any treatments.
- Administration of iron and folic acid is advocated only in some patients.
- Those with the severe forms of alpha thalassemia require a lifelong blood transfusion.

- Regular blood transfusions
- Iron chelation therapy
- Folic acid (if the dietary intake of folic acid is not satisfactory)
- Splenectomy (sometimes used to reduce the blood requirement)
- Bone marrow transplantation
- Gene therapy for screening and therapeutic purposes

## Summary – Alpha vs Beta Thalassemia

Thalassemia is a heterogeneous group of disorders caused by inherited mutations that decrease the synthesis of either alpha or beta globin chains that compose the adult haemoglobin HbA. Thalassemia can be categorized broadly into two main categories as alpha thalassemia and beta thalassemia. In alpha thalassemia, the amount of alpha chains is decreased, and in beta-thalassemia, the number of beta chains is decreased. This is the main difference between alpha and beta thalassemia.

### Reference:

1. Kumar, Parveen J., and Michael L. Clark. Kumar & Clark clinical medicine. Edinburgh: W.B. Saunders, 2009.

### Image Courtesy:

1. “Thalassemia alpha” By National Heart Lung and Blood Institute (NIH) – National Heart Lung and Blood Institute (NIH) (Public Domain) via [Commons Wikimedia](#)
2. “Atr-x” By Gibbons R. – Gibbons R. Alpha thalassaemia-mental retardation, X linked. Orphanet J Rare Dis. 1, 15. 2006. doi:10.1186/1750-1172-1-15. PMID 16722615 ([CC BY 2.0](#)) via [Commons Wikimedia](#)

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