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# MANAGEMENT OF THALASSEMIA

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## Abstract

Thalassemia affects the population of Thalassemia's entire buildings, from the Mediterranean to the Middle East, from sub-Saharan Africa to South and Southeast Asian countries. Today, thalassemia is caused by the continued migration of population from these regions to western countries around the world. Thalassemia affects men and women equally, occurring in about 4.4% of 10,000 births and accounting for about 60,000-70,000 babies born in different types of thalassemia each year.

**Keywords:** Thalassemia, Blood disorder, Birth, Hereditary Disease

## INTRODUCTION

Thalassemia is a group of heterogeneous hereditary diseases inherited by autosomal recessive inheritance, and the rate of hemoglobin production is partial due to the decreased rate of α synthesis. Or it is completely suppressed. -Or β chain, two chains of adult hemoglobin (Hb A). The molecular defects that cause thalassemia are in the human globulin genes that encode the alpha and beta globulin polypeptide chains of hemoglobin on chromosomes # 16 and # 11, respectively.

In addition to pediatricians and hematologists, general practitioners and general practitioners are what is thalassemia, how thalassemia is diagnosed and distinguished from other hypo pigmented microcytic anemia, and of treatment and prevention. You need to know the principles.

## DEFINITION

Thalassemia is a familial (hereditary) blood disorder that causes the body to have an abnormal shape and a lack of hemoglobin.

## CAUSE

Thalassemia occurs when one of the genes involved in hemoglobin production is abnormal or mutated. You have inherited this genetic abnormality from your parents.

If only one of your parents is a carrier of thalassemia, you can develop a form of illness known as minor thalassemia. If this happens, you're probably asymptomatic, but you're a carrier. People with thalassemia develop mild symptoms.

If both parents are carriers of thalassemia, they are more likely to inherit a more serious illness.

## TYPES

Thalassemia can be classified in two ways: depending on which part of hemoglobin is affected and the severity of the condition. 4 Two alpha globins and two beta globin protein chains make up hemoglobin. The two main types of thalassemia are alpha and beta.

***Alpha thalassemia:*** is caused by a deletion of the alpha globin gene, resulting in reduced or absent production of alpha globin chains. There are four alleles in the alpha globin gene, and the severity of the disease ranges from mild to severe, depending on the number of allele deletions. Deletions of the four alleles are the most serious form, no alpha globin is produced, and excess gamma chains (presence during fetal life) form tetramers. It is incompatible with life and causes hydrops fetalis. Allele deletions are the mildest form and are usually clinically silent.

***Beta thalassemia:*** results from a point mutation in the beta globin gene. It is divided into three categories based on the zygosity of beta gene mutations. Heterozygous mutations (beta plus thalassemia) lead to minor beta thalassemias that under produce beta chains. Mild and usually asymptomatic. Beta thalassemia major is caused by a homozygous mutation in the beta globin gene (beta zero thalassemia) and is completely deficient in beta chains.

## SYMPTOMS

***Iron overload:*** People in thalassemia can develop iron overload in their bodies, either by the disease itself or by frequent blood transfusions. Too much iron can damage the heart, liver and endocrine system. The endocrine system contains glands that produce hormones that regulate processes throughout the body. The damage is characterized by excess iron formation. Without proper iron chelating therapy, almost all patients with beta thalassemia can accumulate deadly iron levels.

***Infections:*** People in thalassemia are at increased risk of infections. This is especially true if the spleen has been removed.

***Bone deformities:*** Thalassemia can dilate bone marrow and expand bone. This leads to abnormal bone structure, especially in the face and skull. Bone marrow dilation also makes bones thinner and more brittle, increasing the risk of fractures.

***Spleen enlargement:*** The spleen helps fight infections and filters out unwanted substances such as old and damaged blood cells. Thalassemia often involves the destruction of many red blood cells, and the task of removing these cells causes the spleen to enlarge. Splenomegaly can aggravate anemia and reduces the lifespan of transfused red blood cells. If the spleen is severely enlarged, the spleen should be removed.

***Slow growth:*** Anemia can cause a child to slow down. Even children with thalassemia can delay puberty.

## DIAGNOSIS

***Blood test:*** A blood test can show red blood cell count and size, shape, or color abnormalities. Blood tests can also be used for DNA analysis to look for mutated genes.

***Prenatal testing:*** Before the baby is born, tests can be done to check for the presence and severity of thalassemia. Tests used to diagnose fetal thalassemia include: -

***Chorionic villi sampling***. This test is usually done around the 11th week of pregnancy and requires removal of a small portion of the placenta for evaluation.

***Amniocentesis.*** This test is usually done around the 16th week of pregnancy and examines a sample of fluid around the fetus.

## TREATMENT FOR THALASSEMIA

Treatment depends on the type and severity of thalassemia.

***Blood transfusions:*** These can replenish hemoglobin and red blood cells. The main patients with thalassemia require 8-12 blood transfusions annually. People with less severe thalassemia may need up to eight or more blood transfusions each year during periods of stress, illness, or infection.

***Iron Chelation:*** This removes excess iron from the bloodstream. Blood transfusions can cause iron overload. This can damage the heart and other organs. Doctors may prescribe deferoxamine, a drug given by injection under the skin or into the muscle. Alternatively, you can prescribe deferasirox, which is taken by mouth.

***Folic Acid Supplements:*** People who are transfused or chelated may also need folic acid supplements. These help the development of red blood cells.

***Bone marrow or stem cell transplantation:*** Bone marrow cells produce red blood cells, white blood cells, hemoglobin, and platelets. In severe cases, transplantation from a compatible donor may be an effective treatment.

***Surgery:*** This may be necessary to correct bone abnormalities.

***Gene Therapy:*** Scientists are researching gene therapy techniques to treat thalassemia. Possibility includes inserting a normal beta globin gene into the patient's bone marrow and using drugs to reactivate the gene that produces fetal hemoglobin.

## CONCLUSIONS

In summary, regular assessments and follow-ups with an emphasis on blood transfusion and iron chelating practices are highly recommended to improve the patient's clinical symptoms, life expectancy, and quality of life. In addition, due to the high heterogeneity of β-thalassemia, treatment should be tailored to each patient.

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