

CHILDREN'S CANCER & BLOOD FOUNDATION

Thalassemia

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Thalassemia comes from the Greek word "thalassa" for sea. Thalassemia means the ***anemia of the sea***. It is also called Cooley's Anemia for the pediatrician from Detroit, Michigan, Dr. Thomas B. Cooley, who described the disease in 1925.

What is Thalassemia?

The thalassemias are a diverse group of genetic blood diseases. Thalassemia is the most common inherited single gene disorder in the world. Scientists and public health officials predict that thalassemia will become a worldwide issue in the next century. With global improvements in childhood disease prevention and treatment, more focus will be given to diagnosing disorders. It is our hope that by providing education about the disease we can raise awareness, encourage people to get tested for the trait, and spread knowledge about comprehensive treatment.

The thalassemias are a diverse group of genetic blood diseases characterized by absent or decreased production of normal hemoglobin, resulting in a microcytic anemia of varying degree. The thalassemias have a distribution concomitant with areas where malaria is common. People with Thalassemia Minor are able to fight malaria better than those who do not have it and therefore, in parts of the world where malaria existed Thalassemia Minor increased. This was of great value in the past since malaria was rampant and deadly. However, malaria is no longer a problem in many areas of the world, and Thalassemia Minor has no advantage to people in these areas. The alpha thalassemias are concentrated in Southeast Asia, Malaysia and southern China. The beta thalassemias are seen primarily in the Mediterranean Sea area, Africa and

Southeast Asia. Due to global migration patterns, there has been an increase in the incidence of thalassemia in North America in the last ten years.

In the thalassemia patient, a mutation or deletion of the genes that control globin production occurs. This leads to a decreased production of the corresponding globin chains and an abnormal hemoglobin ratio. This abnormal ratio leads to decreased synthesis of hemoglobin and the expression of thalassemia. The globin that is produced in normal amounts winds up in excess and forms red cell aggregates or inclusions. These aggregates become oxidized and damage the cell membrane, leading to hemolysis, ineffective erythropoiesis, or both. The quantity and properties of these globin chain aggregates determine the characteristics and severity of the thalassemia.

Thalassemia Minor: what does it mean?

Your blood count may be a little lower than other people of your age and sex, but this produces no symptoms. You were born with this condition and you will have it all of your lifetime. There is no need for treatment and most people who have inherited this are not sick and probably do not know they have it. A mild form of Thalassemia minor may be mistaken for iron deficiency anemia. Iron medicines are not usually necessary and will not help your anemia. They could even be harmful if taken over a long period of time.

If you marry a person who does not have Thalassemia Minor, your children may have Thalassemia Minor. If you marry a person who does have Thalassemia Minor, some of your children may have Thalassemia Major. You must decide if you want to take this risk in planning your family.

How common is Thalassemia Minor?

About 3-6% of Americans of Italian and Greek descent have Thalassemia Minor. Thalassemia has also been found in people of many ethnic backgrounds, so it cannot be called a Mediterranean disease. Other areas affected are the Mid East, India, Pakistan and Southeast Asia.

Thalassemia Minor does not change into Thalassemia Major.

Thalassemia Major

Thalassemia Major occurs when a person inherits two Thalassemia genes, one from each parent. Both parents must have Thalassemia Minor. When two individuals who have Thalassemia Minor marry, there is a 25% chance that any pregnancy can result in a child with Thalassemia Major. Two of four children will have Thalassemia Minor and 1 of 4 will be normal.

These chances are present with each pregnancy. Some families have had only one ill child, while others have had all of their children affected.

What are the symptoms of Thalassemia Major?

An infant with Thalassemia Major appears normal at birth. If a child is well for the first five years of life, a diagnosis of Thalassemia Major is unlikely.

The double dose of two Thalassemia genes causes an anemia that is so severe that regular blood transfusions must be given throughout life.

- A newborn with Thalassemia Major appears normal at birth.
- As they grow, infants with Thalassemia Major exhibit paleness and fussiness.

- Weakness and slow growth appear in the first or second years of life.
- The abdomen may swell due to an enlarged liver and spleen.
- Changes occur in the appearance of the face and head. The eyes appear slanted and the cheekbones become prominent.

Treatment for Thalassemia Major involves blood transfusions that must be given every 4 to 6 weeks to sustain life.

Complications that may arise from regular blood transfusions include an overload of iron build up in vital organs causing diabetes, liver disease and heart failure. The spleen may become so enlarged or overactive that it has to be removed surgically. In the past, many patients died in their teens due to these complications.

Today, a medication called deferoxamine (Desferal) can remove iron from the body, but must be given by slow infusions under the skin or in a vein over 10-12 hours five to seven times a week using a battery driven pump. Trials are under way with a new oral form of this medication and early results have been encouraging. This promises to dramatically improve the quality of life for sufferers of Thalassemia Major.

With transfusions and with the continuous use of Desferal, the life expectancy of patients with Thalassemia Major has been greatly improved with many patients reaching their thirties and even beyond. Management of thalassemia is not enough. Researchers are investigating two potentially curative treatments: bone marrow transplantation and gene therapy. Both methods have shown promise.

If Thalassemia Minor is harmless, why test for it?

If a person has Thalassemia Minor, the cause of the slight anemia is known and no other blood tests or treatments such as iron are needed. More important, since individuals with Thalassemia Minor can pass the Thalassemia gene to their children, most people would like to know if there is a risk that their children could inherit this severe blood disease.

A safe and reliable prenatal test to diagnose Thalassemia Major in a fetus as early as 10-12 weeks after conception has been developed. Couples who are at risk may want to consider this possibility.

Research is being carried out focusing on

- 1) finding ways to increase hemoglobin production in red blood cells;
- 2) finding new medicines to remove iron, especially ones that can be taken orally
- 3) transplantation of normal bone marrow or stem cells
- 4) gene therapy to replace the Thalassemia genes with normal genes

Children's Cancer and Blood Foundation is the oldest and largest thalassemia clinic in the United States. We are a NIH designated center and at the forefront of research and treatment for this disease.