THALASSEMIA

"Treatable disease with the right knowledge and decisions"

What is Thalassemia?

Thalassemia is an inherited blood disorder that causes the formation of abnormally shaped hemoglobin (a protein that allows red blood cells to carry oxygen). This disorder is caused by the mutation or deletion of certain gene fragments in the DNA that are involved in the production of hemoglobin. These mutations are then passed down from parent to child.

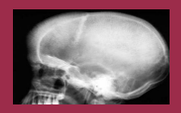
Two types of Thalassemia

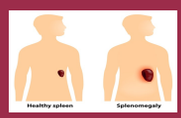
Thalassemia can occur in two forms: α-Thalssemia and β-Thalassemia. This is due to different mutations on the numerous genes involved in hemoglobin production. α-Thalssemia is caused by having at least one mutated α-globin gene. On the other hand, β-Thalassemia is triggered when the β-globin genes show some type of abnormality.

Furthermore, thalssemia is classified into two degrees of severity. This is determined by the number of genes that are mutated in the patient. Out of the four genes that are involved in the production of hemoglobin, 1or 2 mutations are classified as Thalassemia-minor. Consequently, 3 or 4 mutations result in Thalassemia-major.

Symptoms

- Anemia - Fatigue
- Weakness - Dark urine - Bone deformities
- Yellow or pale skin - Abnormal swelling of spleen
- Delayed growth and development





Treatments

Treatments for this condition differ depending on the severity of the patient's condition. Common treatments include regular blood transfusion and the prescription of medication. In more severe cases, bone marrow transplants, splenectonomy (surgical removal of the spleen), and cholecystectomy (surgical removal of the gallbladder) are performed to relieve the symptoms.

However, since the disease is rare, there are cases where the patients are misdiagnosed as iron deficiency. Therefore, it is crucial for patients to visit a hematologist to have clinical testing of their blood.

How to get tested

Blood testing is the most commonly used method to test an individual with Thalassemia. These blood tests are conducted to analyze various characteristics of the blood. Characteristics that can be revealed using blood tests include:

- Red blood cell levels in the blood
- Size of red blood cells
- Shape of red blood cells
- Color of red blood cells
- Hemoglobin distribution in red blood cells
- Iron level in the blood

In addition, pre-natal testing can be done before a baby is born in order to determine the severity of thalassemia beforehand.

Chorionic Villi Sampling is one form of pre-natal testing that can be done around the 11th week of pregnancy. This test involves the removal of a small portion of the placenta and analyzing it.

Amniocentesis is the second type of pre-natal testing that is conducted during the 16th week of pregnancy. For this test, a small sample is taken of the fluid that surrounds the fetus.

More Information

Thalassemia.com

https://medlineplus.gov/ency/article/000587.htm

https://www.healthline.com/health/thalassemia