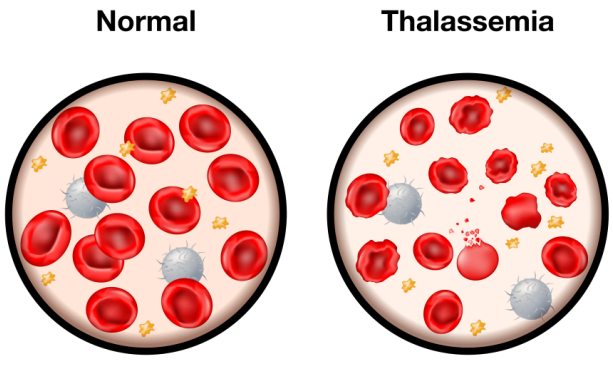
All You Need to Know About Thalassemia Disease



Thalassemia is a hereditary blood disorder transmitted from parents to children through genes. It occurs when the body produces insufficient quantities of haemoglobin, a crucial protein within red blood cells. Inadequate production of haemoglobin leads to impaired functionality and reduced lifespan of red blood cells, resulting in a lower count of healthy cells circulating in the bloodstream.

The role of red blood cells is to transport oxygen to all body cells. It is essential for their proper functioning. Insufficient healthy red blood cells result in inadequate oxygen delivery to the cells of the body, causing symptoms such as fatigue, weakness, and shortness of breath—a condition known as anaemia. Thalassemia can manifest as mild or severe anaemia, with severe cases posing risks of organ damage and death.

Types of Thalassemia

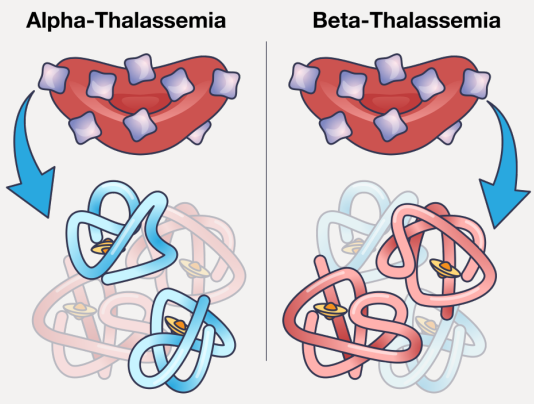
There are many variations of Thalassemia, each with multiple subtypes. Among these are alpha and beta Thalassemia, which encompass the following forms:

- Thalassemia Major
- Thalassemia Minor

To develop Thalassemia Major, one must inherit the gene defect from both parents.

Thalassemia Minor occurs when the faulty gene is inherited from just one parent. Individuals with this type of the disorder are carriers and often do not exhibit symptoms.

Read more:
https://www.cdc.gov/thalassemia/about/?CDC_AAref_Val=
https://www.cdc.gov/ncbddd/Thalassemia/facts.html



Symptoms

Thalassemia is a genetic blood disorder that affects the production of haemoglobin, leading to anaemia. The symptoms of Thalassemia can vary depending on the type and severity of the condition. Here are common symptoms associated with Thalassemia:

- Fatigue
- Weakness, pale skin
- Jaundice (yellowing of the skin and eyes)
- Slow growth in children
- Bone deformities (in severe cases)
- Enlarged Spleen and Liver
- Dark urine
- Abdominal swelling

Causes and risk factors

Thalassemia is caused by genetic mutations that affect the production of haemoglobin, the protein in red blood cells that carries oxygen. These mutations lead to reduced or absent production of normal haemoglobin, resulting in anaemia and other related complications.

Some factors that elevate the risk of Thalassemia are

1. Having a family history of Thalassemia. This condition is inherited from parents who carry mutated haemoglobin genes.
2. Specific ethnic backgrounds. Thalassemia is more prevalent among people with African, Mediterranean and Southeast Asian heritage.

How is thalassemia diagnosed?

Thalassemia diagnosis can occur both prenatally and postnatally:

1. Prenatal diagnosis:

- **Chorionic Villus Sampling (CVS):** This invasive procedure involves taking a sample of chorionic villi from the placenta for genetic testing. It's usually done around 12-10 weeks into the pregnancy.
- **Amniocentesis:** Another invasive test, amniocentesis involves taking a sample of the amniotic fluid for genetic analysis. It's typically done between 20-18 weeks of pregnancy.

Read more: https://pubmed.ncbi.nlm.nih.gov/6275566/
https://pubmed.ncbi.nlm.nih.gov/30141449/

2. Postnatal diagnosis:

- Complete Blood Count (CBC) checks haemoglobin and red blood cell levels. (Thalassemia patients have low levels of both).
- A reticulocyte count shows if your bone marrow makes enough red blood cells.
- Iron studies reveal if anaemia is due to low iron or Thalassemia.
- Hemoglobin electrophoresis diagnoses beta Thalassemia.

Both prenatal and postnatal diagnosis are crucial for early identification and management of Thalassemia. Early diagnosis allows for appropriate medical care and counselling for families.

Treatment

Treatment for Thalassemia depends on the type and severity. It may include:

- Blood Transfusions: Regular transfusions to maintain haemoglobin levels, especially in beta Thalassemia Major.
- Iron Chelation Therapy: To remove excess iron from the body due to frequent transfusions.
- Folic Acid Supplements: Help in red blood cell production.
- Bone Marrow Transplant: Can be curative in some cases, particularly in severe beta Thalassemia.

Things you can do to help

- **Genetic Counselling:** Seek genetic counselling if you have a family history of Thalassemia or are at risk of carrying the gene.
- **Regular Monitoring:** Regularly monitor blood counts, iron levels, and overall health under the guidance of a healthcare professional.
- **Medication Adherence:** Adhere to prescribed medication regimens, such as iron chelation therapy if needed, to manage complications like iron overload.
- **Healthy Lifestyle:** Maintain a healthy lifestyle with a balanced diet rich in iron (for non-transfusion-dependent Thalassemia) and follow recommended exercise routines.

Things to avoid

- **Avoid Iron Supplements:** Unless prescribed by a healthcare provider, avoid taking iron supplements as they can contribute to iron overload in Thalassemia patients.
- **Avoid Unsupervised Treatments:** Do not undergo any treatments or therapies without consulting a healthcare professional specialised in Thalassemia management.
- **Avoid Smoking:** Smoking can exacerbate complications associated with Thalassemia, such as liver and lung problems.
- **Avoid Infections:** Take precautions to avoid infections, as Thalassemia patients may have a weakened immune system.

These guidelines are general in nature and may vary based on individual medical conditions and recommendations from healthcare professionals.

Remember:

Since Thalassemia is a genetic disorder, genetic counselling and testing are essential for couples with a family history to understand their risk of passing the condition to their children. Prenatal testing can also detect Thalassemia in the foetus. Increasing awareness about Thalassemia is the key to empowering individuals and their families. By advocating for genetic screening and early diagnosis, we can pave the way for timely interventions and tailored medical care. Together, let's work towards managing and lessening the impact of this disorder, ensuring healthier lives and brighter futures for all.

References:

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