

Thalassemia: Understanding the Inherited Blood Disorder

World
Thalassemia
Day | 8 May

As we observe World Thalassemia Day on May 8th, it is essential to raise awareness about this inherited blood disorder that affects millions of people worldwide. Thalassemia is a genetic condition that impairs the production of haemoglobin, a vital protein in red blood cells responsible for carrying oxygen throughout the body. This deficiency leads to anaemia, fatigue and other complications if left untreated.

Causes of Thalassemia

Thalassemia is caused by mutations in the DNA of cells that produce haemoglobin. These mutations are inherited from parents and affect the production of either the alpha or beta chains of haemoglobin, resulting in alpha-thalassemia or beta-thalassemia, respectively. The severity of thalassemia depends on the number of mutated genes inherited from parents.

♦ Types of Thalassemia

There are two main types of thalassemia: alphathalassemia and beta-thalassemia. Alphathalassemia is caused by mutations in the genes that code for the alpha haemoglobin chain, while beta-thalassemia is caused by mutations in the genes that code for the beta haemoglobin chain. The severity of thalassemia varies depending on the type and number of mutated genes.

Symptoms and Complications

Thalassemia can cause a range of symptoms, including anaemia, fatigue, pale skin and weakness. In severe cases, it can lead to complications such as enlarged spleen, bone deformities and increased risk of infections. If left untreated, thalassemia can be life-threatening.

Prevention and Management

While there is no cure for thalassemia, it can be managed with regular blood transfusions, folic acid supplements and iron chelation therapy. In some cases, bone marrow transplantation may be necessary. Preventing thalassemia is challenging, but genetic counselling and assisted reproductive technology can help identify carriers of the mutated genes and reduce the risk of passing it on to offspring.



Importance of Awareness and Screening

World Thalassemia Day serves as a reminder of the importance of raising awareness about this inherited blood disorder. Early screening and diagnosis are crucial in managing thalassemia effectively. Parents who are carriers of the mutated genes can take steps to prevent passing it on to their children. Moreover, awareness about thalassemia can help reduce the stigma associated with the condition and promote inclusivity and support for those affected.

In Summary

Thalassemia is a complex and debilitating condition that requires attention and awareness. On World Thalassemia Day, we acknowledge the struggles of those affected by this condition and reaffirm our commitment to promoting awareness, prevention, and management of thalassemia. By working together, we can make a difference in the lives of those affected and strive towards a future where thalassemia is no longer a life-threatening condition.

Haematology is the medical specialty that manages thalassemia. Haematologists are doctors who specialize in blood disorders, including thalassemia. They play a crucial role in diagnosing, treating and managing the disease. Regular follow-ups with a haematologist can help monitor the symptoms and complications of thalassemia, ensuring that patients receive appropriate care and maintain a good quality of life.

