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Thalassemia: A genetic journey through red blood cell disorders.

Dewber Sara*

Department of Medicine, Oslo University Oslo, Norway

Correspondence to: Dewber Sara, Department of Medicine,Oslo University Oslo, Norway, E-mail: saradewber8@gmail.com

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Introduction

Thalassemia is more than a medical diagnosis—it's a lifelong genetic journey that affects millions across the globe. As a red blood cell disorder rooted in inherited mutations, thalassemia disrupts the body's ability to produce hemoglobin, the vital protein responsible for transporting oxygen. This condition not only challenges the physical health of those affected but also reshapes their emotional, social, and economic landscapes. Thalassemia is an inherited blood disorder characterized by reduced or absent production of hemoglobin, the oxygen-carrying molecule in red blood cells. Hemoglobin is composed of two alpha and two beta protein chains. Mutations in the genes responsible for these chains lead to two main types of thalassemia: Caused by mutations in the alpha-globin genes [1].

Caused by mutations in the beta-globin genes. Depending on the number and severity of gene mutations, thalassemia can range from asymptomatic carrier states to life-threatening anemia requiring lifelong treatment. Thalassemia is passed down in an autosomal recessive pattern, meaning both parents must carry the defective gene for a child to be affected. Carriers often show mild symptoms or none at all. Interestingly, the genetic mutations responsible for thalassemia evolved as a protective mechanism against malaria. In regions historically plagued by malaria—such as South Asia, the Mediterranean, and parts of Africa—carrying one thalassemia gene offered partial immunity, leading to its prevalence in these populations [2].

Symptoms of thalassemia vary widely depending on the type and severity: Mild anemia, often mistaken for iron deficiency. Moderate anemia, occasional transfusions needed. Severe anemia, requiring regular blood transfusions from early childhood. Detects anemia and abnormal red blood cells. Identifies abnormal hemoglobin types. Confirms mutations in globin genes. Prenatal screening and carrier testing are crucial in high-risk populations to prevent severe forms of the disease [3].

Treatment depends on the severity of the condition: Lifesaving for thalassemia major but can lead to iron overload. Removes excess iron from the body to prevent organ damage. Support red blood cell production. The only potential cure, though limited by donor availability and risk factors. Emerging therapies include gene editing and gene therapy, which aim to correct the underlying genetic defect. Thalassemia affects over 300,000 births annually worldwide, with the highest prevalence in India, Southeast Asia, the Middle East, and the Mediterranean. In India alone, over 10,000 children are born with thalassemia major each year, and an estimated 40 million people are carriers [4].

Access to diagnosis and treatment varies significantly. In high-income countries, patients often receive comprehensive care, while in low-resource settings, many face delayed diagnosis, inadequate treatment, and early mortality. The economic burden of thalassemia is substantial. Regular transfusions, iron chelation, and hospital visits can cost thousands annually. In countries without universal healthcare, families often bear these costs, leading to financial hardship. Socially, thalassemia affects education, employment, and mental health. Children may miss school due to frequent hospitalizations, and adults often face stigma or discrimination. Psychological support and community awareness are essential components of holistic care [5].

Conclusion

Recent breakthroughs in gene therapy offer hope. Techniques like CRISPR and lentiviral vectors aim to correct the faulty genes responsible for thalassemia. Clinical trials have shown promising

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results, with some patients achieving transfusion independence. Additionally, prenatal diagnosis and preimplantation genetic testing allow families to make informed reproductive choices, potentially reducing the incidence of severe thalassemia. Despite its challenges, many individuals with thalassemia lead fulfilling lives. With proper treatment, education, and support, patients can pursue careers, relationships, and personal goals. Support groups and advocacy organizations play a vital role in raising awareness, promoting research, and empowering patients. The journey through thalassemia is not just medical—it's emotional, communal, and deeply human.

References

1. Hallek M, Cheson BD, Catovsky D, et al. Guidelines for the diagnosis and treatment of chronic lymphocytic leukemia: a report from the International Workshop on Chronic Lymphocytic Leukemia updating the National Cancer Institute–Working Group 1996 guidelines.. Am J Hematol. 2008;111(12):5446-56.
2. Shanafelt TD, Ghia P, Lanasa MC, et al. Monoclonal B-cell lymphocytosis (MBL): biology, natural history and clinical management. Leukemia. 2010;24(3):512-20.
3. Zent CS, Kay NE. Autoimmune complications in chronic lymphocytic leukaemia (CLL). Best Pract Res Clin Haematol. 2010;23(1):47-59.
4. Puente XS, Pinyol M, Quesada V, et al. Whole-genome sequencing identifies recurrent mutations in chronic lymphocytic leukaemia. Nature. 2011;475(7354):101-5.
5. Tam CS, O'Brien S, Wierda W, et al. . Long-term results of the fludarabine, cyclophosphamide, and rituximab regimen as initial therapy of chronic lymphocytic leukemia. Am J Hematol. 2008;112(4):975-80.

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