FACT SHEET
Thalassaemia

A serious inherited blood disorder affecting oxygen transport in the body

novo nordisk haemophilia & haemoglobinopathies foundation

What is thalassaemia?

Thalassaemia is a group of inherited blood disorders that affect the body's ability to produce haemoglobin – the protein in red blood cells that carries oxygen. The condition causes chronic anaemia, which can be mild or severe, depending on the type inherited.
It is passed from parents to children in an autosomal recessive pattern, meaning both parents must carry and pass on the affected gene.

Quick facts

- Thalassaemia is a genetic blood disorder that affects how the body produces haemoglobin.
- It may result in chronic anaemia, which can lead to fatigue, delayed growth, and organ complications.
- There are two major types: Alpha thalassaemia and Beta thalassaemia, each with varying severity. Compound thalassaemia (and other variants) can also occur.
- Thalassaemia trait means a person carries one affected gene but usually does not have symptoms.
- Around 68,000 children are born with various thalassaemia syndromes each year, mainly in Asia, the Mediterranean, and the Middle East, with 1.3 million cases worldwide.

Types of thalassaemia

Thalassaemia is classified into alpha and beta types, depending on which part of the haemoglobin molecule is affected. Alpha thalassaemia involves mutations in the alpha globin genes, while beta thalassaemia affects the beta globin genes. People with thalassaemia trait carry one affected gene and usually do not have symptoms, but they can pass the gene to their children.

Alpha thalassaemia

Silent carrier
1 gene affected, no symptoms.

Alpha thalassaemia trait
2 genes affected, mild anaemia, usually without symptoms.

Haemoglobin H disease
3 genes affected, moderate to severe anaemia, sometimes requiring treatment.

Alpha thalassaemia major
4 genes affected, also called hydrops fetalis – often fatal before or shortly after birth.

Beta thalassaemia

Beta thalassaemia trait
Also called Minor, one gene affected, usually no symptoms or mild anaemia.

Beta thalassaemia intermedia
Both genes affected, moderate anaemia. May require occasional treatment.

Beta thalassaemia major
Cooley's Anaemia, severe anaemia from early infancy; requires lifelong treatment.

Thalassaemia trait

Thalassaemia trait is a carrier state in which a person inherits one normal and one mutated gene. These individuals do not have thalassaemia disease but can pass the gene to their children. While usually symptom-free, some carriers may experience mild anaemia, especially during illness or pregnancy. Many people are unaware they carry the trait until they are tested.

Symptoms and complications

General symptoms
Chronic fatigue, weakness, pale/yellowish skin (jaundice), delayed growth and puberty, bone deformities (face/skull), enlarged spleen, dark urine.

Severe complications
Heart complications, liver damage from iron overload, increased infection risk, shortened lifespan without treatment, lung complications, frequent infections.

Who is affected?

Thalassaemia is most common in regions where malaria was or is historically prevalent, as carrying the gene for thalassaemia offered a survival advantage against severe malaria. High-prevalence areas include:

- South and Southeast Asia
- The Middle East
- The Mediterranean region
- North and East Africa
- Migrant populations in Europe, North America, and Australia

Around 68,000 children are born with various thalassaemia syndromes each year[1] and more than 1.3 million people currently live with the condition[2].

Outcomes vary: Survival is improving in high-income countries, while early complications and limited access to care remain challenges in low-resource settings.

Diagnosis

Thalassaemia is diagnosed through:

- Complete blood count (CBC) to detect anaemia
- Haemoglobin electrophoresis to identify abnormal haemoglobin types
- DNA testing for alpha or beta gene mutations
- Prenatal screening and newborn testing in high-risk populations

Carrier screening is essential for family planning in affected regions.

Treatment options

Treatment availability varies widely between countries and health systems.

Main treatment:

- **Blood transfusions:** Regular transfusions are required for thalassaemia major and intermedia.
- **Iron chelation therapy:** Removes excess iron from the body after repeated transfusions, although it has its own limitations.
- **Folic acid supplements:** Help support red blood cell production.
- **Bone marrow or stem cell transplant:** The only potential cure, most effective in early childhood with a matched donor.
- **Gene therapy:** An emerging option, currently under study in clinical trials.
- **Hydroxyurea:** The first-line treatment in patients with complications but has its own challenges.

Ongoing care may also include:

- **Vaccinations to prevent infections**
- **Monitoring of heart and liver health**
- **Psychological support**
- **Genetic counselling for affected families**

Living well with thalassaemia

Thalassaemia can be managed effectively with lifelong care.

- Regular blood transfusions and iron chelation prevent complications.
- Healthy nutrition and infection prevention support overall well-being.
- Genetic counselling helps families plan and manage risks.
- Advances like gene therapy offer new hope in select cases.
- Holistic care should improve quality of life.

References:

1. Thalassaemia International Federation (TIF), 2023, https://thalassaemia.org.cy/publications/tif-publications/guidelines-for-the-management-of-non-transfusion-dependent-%ce%b2-thalassaemia-3rd-edition-2023/, Last accessed: November 2025

2. Tuo, Y., et al., Global, Regional, and National Burden of Thalassemia, 1990–2021: A Systematic Analysis for the Global Burden of Disease Study 2021, 2024. Available at: pmc.ncbi.nlm.nih.gov/articles/PMC11090906/ Last accessed: November 2025.

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