## WHAT HAPPENS IF THALASSAEMIA IS LEFT UNTREATED?

Patients that receive proper treatment enjoy a very good quality of life and their life expectancy is not very different from that of the general population.

However, when not treated properly, thalassaemia can have life-threatening complications on health and even lead to premature death. The function of several organs can be influenced significantly, leading for example to cardiac failure, liver cirrhosis, diabetes, and others serious health conditions.

## FAST FACTS ABOUT THALASSAEMIA

**80% OF PATIENTS WITH THALASSAEMIA LIVE IN LOW AND MIDDLE-INCOME COUNTRIES.**

**8 OUT OF 10 PEOPLE ARE NOT AWARE THAT THEY ARE CARRIERS OF THALASSAEMIA.**

**IN MOST COUNTRIES WITH MEDIUM AND HIGH-DISEASE PREVALENCE, PATIENTS WITH B-THALASSAEMIA MAJOR DO NOT REACH THE AGE OF 20 YEARS, AS A RESULT OF SUBOPTIMAL CARE.**

## WHAT DOES TIF DO?

The Thalassaemia International Federation (TIF) is a non-profit, non-governmental organization that represents over 230 National Thalassaemia Associations from 62 countries across the world and is dedicated to supporting the equal access of every patient with thalassaemia to health, social and other care within patient-centred healthcare settings.

The Federation works in official relations with the World Health Organization (WHO) since 1996, in special consultative status with the United Nations Economic and Social Council (ECOSOC) since 2017, and in official partnership with the European Commission since 2018.

Its extensive educational programme includes a vast series of internationally acclaimed publications as well as events, conferences, workshops, fellowships, etc. with the objective to provide lifelong educational opportunities for health professionals, patients and their families, raise awareness on thalassaemia amongst policy makers and the community at large, and promote effective, disease-specific programmes for the prevention, control and clinical management of thalassaemia within national healthcare systems based on universal coverage.

**For more information you can visit**
**www.thalassaemia.org.cy**

THALASSAEMIA INTERNATIONAL FEDERATION



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# HAVE YOU HEARD ABOUT THALASSAEMIA?

## WHAT IS THALASSAEMIA?

Thalassaemia is a blood disorder caused by a group of genetic defects that affect the production of haemoglobin, a protein molecule in red blood cells enabling them to carry oxygen from the lungs throughout the whole body. This causes a shortage of functional red blood cells and low levels of oxygen in the bloodstream, which lead to a variety of health problems depending on the type and severity of the condition.

There are two main types of thalassaemia:

- **α-thalassaemia**, occurring when a gene or genes related to the α-globin protein are missing or changed (mutated), and
- **β-thalassaemia**, occurring when similar genetic defects affect the production of the β-globin protein

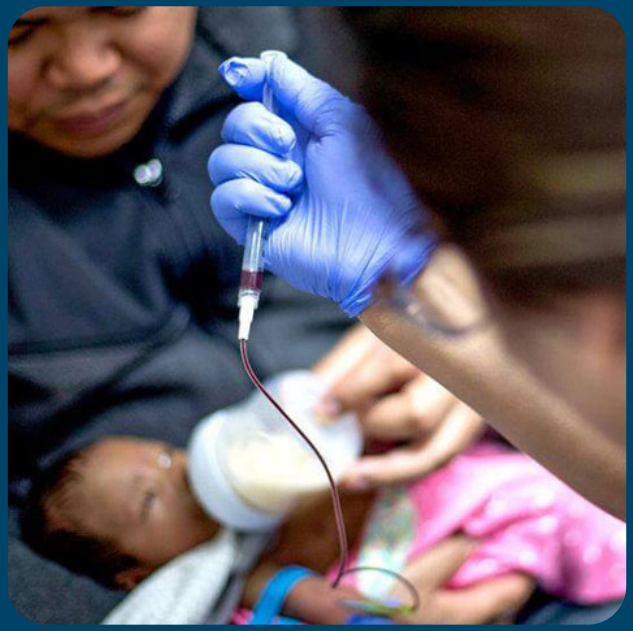
This leaflet will focus on β-thalassaemia, classified into 2 main forms; ***thalassaemia major*** and ***thalassaemia minor***. Inheriting the gene from both parents causes thalassaemia major. Inheriting it from one parent causes thalassaemia minor. People with thalassaemia minor, also known as having the thalassaemia trait or as thalassaemia carriers, have the gene but do not have the disorder, and are as a result generally healthy and symptom-free.

On the contrary, individuals with β-thalassaemia major suffer from severe, life-threatening anaemia, usually resulting in poor growth, a number of serious health problems and a shortened lifespan, if left untreated.

## WHAT CAUSES THALASSAEMIA AND WHO IS MOST AT RISK?

Thalassaemia is an inherited disease, meaning that it is caused by problems within our genetic make-up, and not because of nutrition or other environmental factors. This is why the percentage of patients and carriers of the disease within the total population differs in various areas of the world.

Thalassaemia can affect individuals of any nationality and ethnicity. Nonetheless, it is particularly common in **people of Mediterranean ancestry** and across a broad region extending through **India, the Middle East, Southeast Asia and Africa,** whereas its prevalence is constantly increasing all across the world, due to global population movements and absence of effective prevention programmes.



## WHAT ARE THE SYMPTOMS OF THALASSAEMIA?

Most people born with thalassaemia experience health problems that can manifest from a few months after birth up until the first 2 years of life. Less severe cases may not be noticeable until later in childhood or even until adulthood.

The symptoms of thalassaemia can vary. They may include severe anaemia and other health problems, such as:

- **delayed growth and development**
- **excessive tiredness and fatigue**
- **yellow or pale skin (jaundice)**
- **dark urine**
- **bone deformities, especially in the face**
- **enlarged spleen, liver, or heart**

## WHAT DOES THALASSAEMIA TREATMENT INCLUDE?

Patients with severe forms of thalassaemia must receive regular blood transfusions throughout their life at regular intervals, usually every 2–5 weeks. In this case, the condition is called ***transfusion-dependent thalassaemia (TDT)***. When patients need to have transfusions occasionally, in certain circumstances and/or for defined periods of time, their condition is called ***non-transfusion-dependent thalassaemia (NTDT)***.

Because of these blood transfusions, a substance called iron gradually accumulates inside the body and becomes toxic for many organs. As a consequence, patients must also receive regularly, usually on a daily basis, drugs that remove iron from the body, called chelation agents. These drugs may be pills that can be taken orally, or liquid solutions that must be injected in the body over a period of several hours through a needle in the abdomen or other parts of the body.

In addition, the provision of multidisciplinary care, the treatment involving the collaboration of many different medical and scientific disciplines, is essential in monitoring and treating common complications associated with the disorder and in ensuring, as such, the highest standard of patients' well-being.