

## Know what is Thalassemia?

Thalassaemia is the name of a group of genetic inherited disorders of the blood. More specifically, it is a disorder of the haemoglobin molecule inside the red blood cells.

It is an inherited genetic disease, i.e. a disease that is passed from parents to children through the genes. It is not infectious and cannot be passed on from one individual to another by personal or any other contact, through blood transfusion, food or air.

The two main types of thalassaemia are  $\beta$  (beta) and  $\alpha$  (alpha). Both affect the production of normal haemoglobin – a key constituent of human red blood cells. Other abnormal types of adult haemoglobin, also known as structural haemoglobin variants, have been identified. These include haemoglobin S (HbS), haemoglobin E (HbE), haemoglobin C, haemoglobin D and Lepore.

Structural haemoglobin variants can combine with  $\beta$ -thalassaemia to produce other related clinically significant blood disorders.

### The Name Thalassaemia

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The name  $\beta$ -thalassaemia derives from a combination of two Greek words: thalassa meaning sea, i.e. the Mediterranean Sea, and anaemia (“weak blood”).  $\beta$ -thalassaemia is also known as Mediterranean anaemia. Both of these names reflect the fact that the disorder was first described in patients originating from countries around the Mediterranean, and for many years it was believed that thalassaemia occurred exclusively in these countries.

Another term, nowadays less frequently used in literature, for  $\beta$ -thalassaemia is Cooley's anaemia, in recognition of the contribution of Professor Thomas Cooley, a paediatrician in the United States who first described the clinical characteristics of this disorder in patients of Italian origin in 1925.

### Terminology

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Individuals who are carriers of the  $\beta$ -thalassaemia/Mediterranean anaemia/Cooley's anaemia trait (sometimes referred to as stigma), or with  $\beta$ -thalassaemia minor, or the heterozygous form of  $\beta$ -thalassaemia are not patients, but individuals who have inherited a normal haemoglobin (B-globin) gene from one parent and the defective gene from the other parent.

Individuals with  $\beta$ -thalassaemia major or Mediterranean anaemia or Cooley's anaemia, or homozygous  $\beta$ -thalassaemia are those who have inherited both the defective haemoglobin (B-globin) genes, one from each parent. These individuals are going to develop the full-blown disease.

## About carriers of the Thalassaemia trait

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Carriers of the thalassaemia trait do not have a disease. They have no physical or mental symptoms and do not require a special diet or medical treatment. The condition cannot become a serious disease over time – indeed, most carriers will be unaware that they carry the trait unless specifically tested. Some carriers may experience mild anaemia, which may be inaccurately diagnosed as iron deficiency anaemia. However, laboratory tests can differentiate between the two. Pregnant women carriers may experience moderate anaemia which is addressed by prescribing iron supplements during pregnancy.

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