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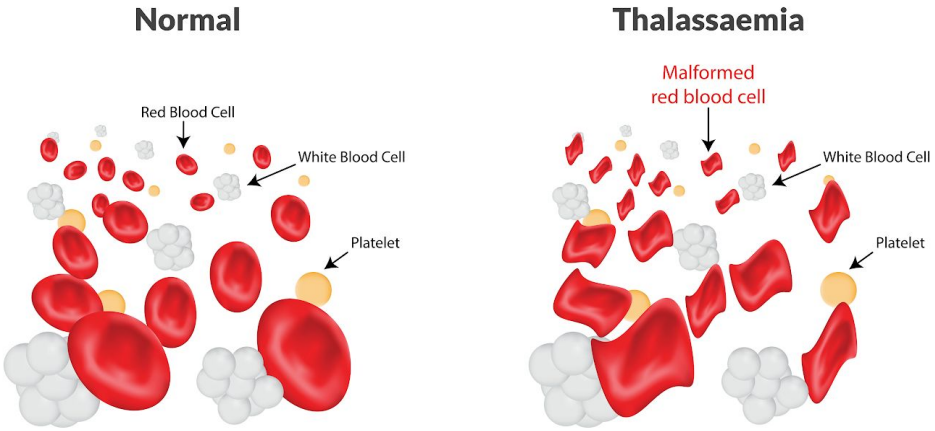
What is Thalassaemia?

Thalassaemia is a genetic blood disorder affecting the production of the red blood cells. Genetic means that this disorder is inherited and can be passed down from one generation to the next.

Abnormal production of blood means that affected individuals do not make adequate amounts of functional red blood cells.



There are several types of thalassaemia, and the most common forms are alpha and beta thalassaemia. Clinically, patients with thalassaemia can present with thalassaemia minor or thalassaemia major.

Thalassaemia Minor

Thalassaemia minor, or Thalassaemia trait, occurs when the person is a carrier of the thalassaemia gene. These people have usually inherited the thalassaemia gene from only one parent, and hence still have a normal functioning haemoglobin gene from the other parent. While thalassaemia minor patients often have a slightly lower red cell count than normal, the majority of them are entirely healthy and lead a normal life, as they are still able to produce sufficient haemoglobin for the body to function. However, the affected person can pass on the abnormal gene to their children.

Thalassaemia Major

Thalassaemia major is a severe form of anaemia. It occurs when the person has inherited two thalassaemia genes, one from each parent. They may suffer from severe anaemia from an early age. These patients often require life-long blood transfusion support, and they suffer from poor growth and development as children. Over time, they may experience iron overload due to repeated blood transfusions, and this can cause damage to their organs, including the heart and liver. As a result, many thalassaemia major sufferers have a shorter lifespan.

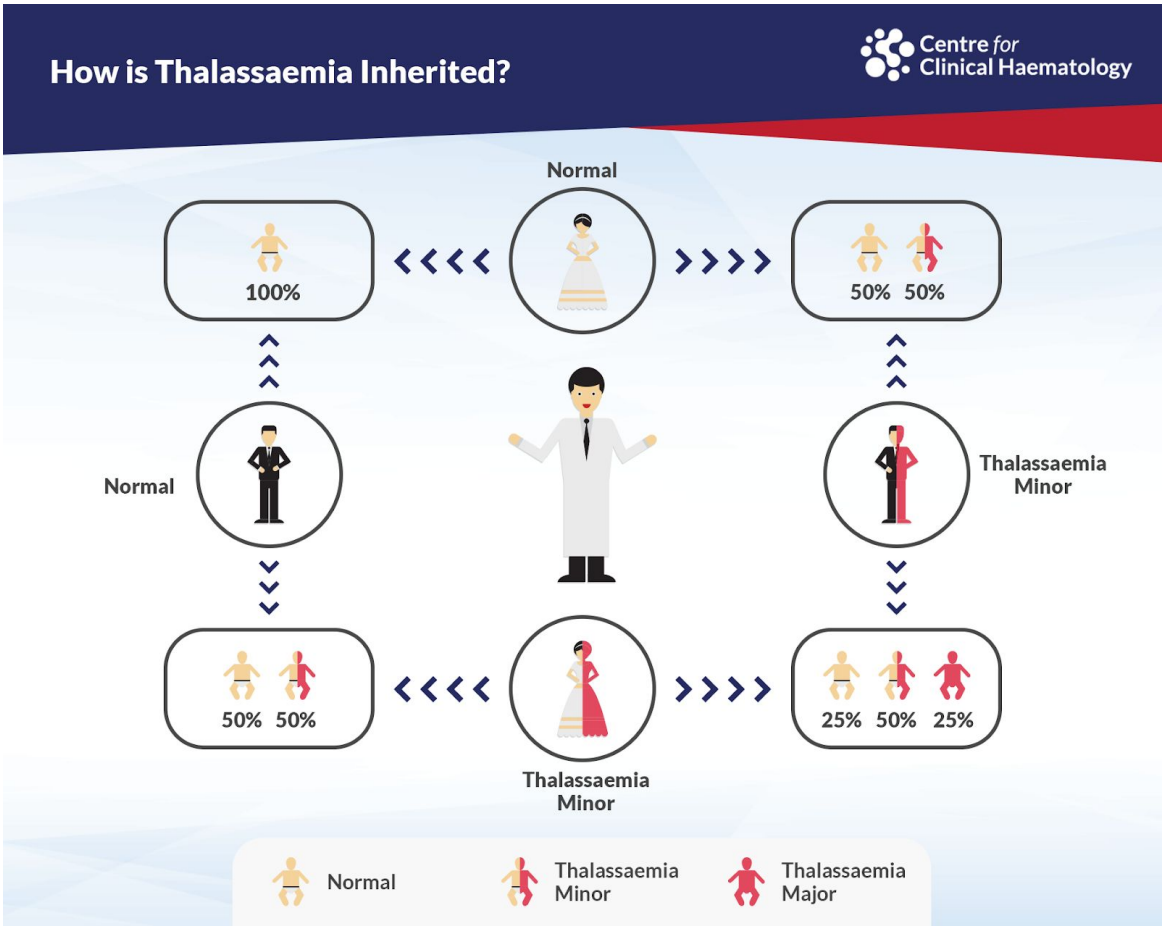
Signs & Symptoms of Thalassaemia

The symptoms of thalassaemia can vary, and some people have no visible symptoms, while others develop symptoms later in adolescence. Some of the most common symptoms include:

- Fatigue
- Weakness
- Pale or yellowish skin
- Bone deformities, especially facial features
- Delayed growth and development
- Abdominal swelling
- Dark urine

How is Thalassaemia inherited?

Thalassemia is an inherited condition, passed to children by parents who carry the mutated thalassemia gene. If both parents carry thalassemia minor, their children may have thalassemia minor, or they may have completely normal blood, or they may have thalassemia major. The chances are the same with each pregnancy, no matter how many children the couple may have.



Five commonly asked questions about Thalassaemia:

1. Should I go for Thalassaemia Screening?

As thalassaemia can be passed on from one generation to another, you and your partner should go for a **thalassaemia screening** if you are:

- Planning to start a family
- Have a family history of thalassaemia
- One of you is already diagnosed with thalassaemia

Thalassaemia screening involves a simple blood test which is readily available.

2. What are the dietary restrictions for Thalassaemia?

Thalassaemia Minor: Unless you also have iron deficiency in addition to thalassaemia, there is no need to avoid a particular food. If you have iron deficiency, avoid food that makes it harder for your body to absorb iron such as coffee, tea, milk, egg whites and soy protein.

Thalassaemia Major: People with thalassaemia may have iron overload, either from the disease or from frequent blood transfusions. Unless your doctor recommends it, do not take vitamins or other supplements that contain iron. Iron-rich foods such as oysters, red meat, peanut butter, grain cereal, leafy green vegetables, dates and watermelons should also be avoided.

3. Are there any activities to avoid if one has Thalassaemia?

Thalassaemia Minor: In most cases, people with thalassaemia minor have no fitness or health issues.

Thalassaemia Major: Some people may have trouble participating in vigorous forms of exercise. However, many others with thalassaemia can participate in moderate physical activities, including biking, running, and walking. If a person with thalassaemia has problems with their joints, there are many types of low-impact exercises to choose from, including yoga, swimming, or water aerobics.

4. What is the life expectancy of someone with Thalassaemia?

Thalassaemia Minor: A person with the thalassaemia trait has a normal life expectancy.

Thalassaemia Major: Heart complications arising from thalassaemia major can make this condition fatal before the age of 30 years.

5. What are the treatments for Thalassaemia?

Thalassaemia Minor: People with thalassaemia minor may have no anaemia or only mild anaemia; hence it is not considered an illness and does not require any medical treatment.

Thalassaemia Major:

- **Blood Transfusion**: Severe forms of thalassaemia may require frequent blood transfusions.
- **Bone Marrow / Stem Cell Transplant**: A bone marrow/stem cell transplant replaces the damaged stem cells with the healthy stem cells from a compatible donor. At present, this is the only treatment that can cure Thalassaemia and eliminate the need for lifelong blood transfusions.
- **Gene Therapy**: Thalassaemia gene therapy is currently undergoing clinical trials. Gene therapy aims to achieve a stable introduction of normal haemoglobin gene into the stem cells in the bone marrow. This will allow people who have thalassaemia to make their own healthy red blood cells and haemoglobin.