

I NEED TO KNOW ABOUT THALASSAEMIA

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What is thalassaemia?

Thalassaemia is one of the most common genetic blood disorders in the world. It causes low haemoglobin levels, also known as anaemia. The anaemia is the result of abnormal haemoglobin production. The red cells in people with thalassaemia are small and pale due to a lack of haemoglobin.

What is haemoglobin?

Haemoglobin has two components:

1. Haem – the part with iron. This carries oxygen, and
2. Proteins – there are actually two pairs of proteins (four in total) which mesh together to hold the haem in place. One pair are called ‘alpha chains’ and the others are ‘beta chains’.

What causes thalassaemia?

All proteins are controlled by genes. Thalassaemia is caused when there is a defect in one or more alpha or beta gene. Four genes (two from each parent) are needed to make the alpha proteins and two genes required to make beta proteins.

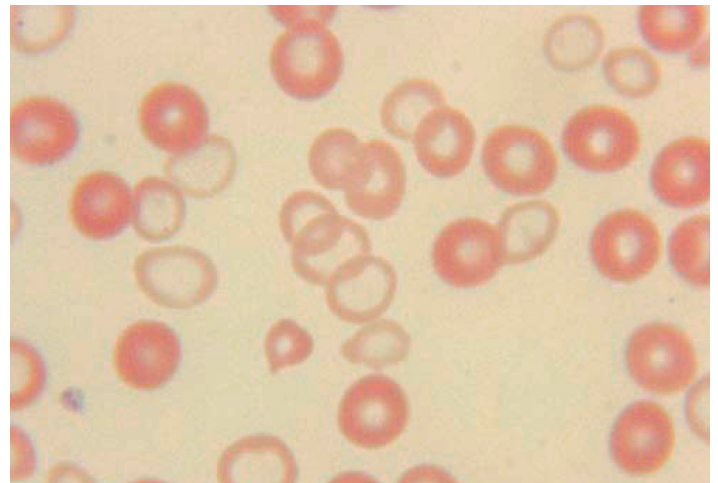
What are the different types of thalassaemia?

There are two main types:

- **Alpha thalassaemia** occurs if one or two of the alpha genes are missing. There are usually no obvious health problems except for mild anemia, which can cause slight fatigue. This ailment can be mistaken for an iron deficiency because the red blood cells will appear small when viewed under a microscope. If three genes are affected, patients will require occasional blood transfusions during times of physical stress, fevers or other illnesses.

The most severe form of this condition is due to missing four genes and is known as alpha thalassaemia major or hydrops fetalis. This condition is extremely rare and babies who have this disorder usually die before or shortly after birth.

- **Beta thalassaemia** occurs if one beta gene is changed and usually requires no treatment. If both genes are affected, it can result in moderate to severe anaemia. A severe form of beta thalassaemia is known as beta thalassaemia major or Cooley’s anaemia.



Above: Thalassaemia red cells are small and pale. The larger, darker ones are from a recent transfusion.

How is thalassaemia treated?

Treatment for thalassaemia usually involves regular blood transfusions. These transfusions give the patient a temporary supply of healthy red blood cells with normal haemoglobin levels that carry oxygen to the body. Bone marrow transplant can be an option with some patients, especially children.

Are there problems from regular transfusions?

Regular transfusions can cause a build up of iron which may become toxic to the body. The only way to remove this excess is by taking a drug that allows the body to excrete it safely.

BLOOD FACT

In Greek, ‘Thalassa’ means ‘of the sea’ and ‘Haema’ means ‘blood’. Thalassaemia literally means ‘watery blood’.