

Information for people who have thalassaemia or are parents of a child with thalassaemia

Introduction

You are receiving this information because you or your child has thalassaemia. It describes what the condition is, what symptoms may occur and what treatments may be considered.

What is thalassaemia and what causes it?

Thalassaemia is a condition you are born with. It is hereditary, meaning that you have inherited the trait for it from both your biological parents.

Thalassaemia is caused by a hereditary mutation in genes that control how the body produces haemoglobin. Haemoglobin is a protein that gives blood its red colour and is needed for the blood to transport oxygen to the body's tissues and organs.

The hereditary mutation leads to haemoglobin being produced incorrectly or in insufficient quantity, which causes a shortage of healthy haemoglobin and leads to anaemia. The abnormally formed red blood cells are broken down faster than normal red blood cells, which can cause jaundice and, later in life, gallstones.

Common symptoms

The first symptoms of thalassaemia may occur during the first year of life, but they may also develop gradually during childhood. The condition can sometimes cause significant problems.

How parents can discover that their child may have thalassaemia:

The child may:

- become pale, feel tired and lack the energy to move around much
- find it difficult to eat
- develop a yellowish skin tone
- develop an enlarged liver or spleen
- grow slowly

Adults can develop the same symptoms as children, but may also suffer from:

- fragile bones
- leg ulcers
- liver problems
- heart failure

These symptoms may be due to a combination of the condition itself and the side effects of treatment.

Treatment

If you or your child has thalassaemia but is feeling fine, treatment may sometimes not be necessary. In the event of long-term or recurrent anaemia, treatment is required to reduce the symptoms of anaemia. Treatment is administered in hospital.

- The most common treatment is a blood transfusion. Each treatment takes about two hours
- Blood transfusions are given every two to six weeks depending on individual needs. Treatments may be given periodically or continuously throughout childhood. A doctor monitors how children with thalassaemia are doing and how they are growing
- You or your child may need medication to manage the extra iron supplied via blood transfusions. Excess iron is harmful to several of the body's organs, which is why medication is needed to counter the iron build-up
- The only curative treatment currently available in Sweden is a stem cell transplant. In a stem cell transplant you receive new stem cells to help the body produce healthy, well-functioning red blood cells with normally structured haemoglobin. Not everyone can receive this treatment; it is a high-risk and complex procedure requiring an extended follow-up. Your doctor will explain more about how the procedure works and the details of the treatment.

To learn more about your situation and any possible treatment, contact your doctor or the healthcare centre you belong to.

If you have any questions or if your child shows symptoms of anaemia between scheduled appointments, contact the medical care unit providing the treatment.

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