Thalassaemia

Summary

- Thalassaemia major is a genetic disorder that affects the production of haemoglobin, resulting in severe anaemia.
- This disorder is usually diagnosed within the first six months of life.
- Treatment options include regular blood transfusions and bone marrow transplants.
- Couples should be tested for their thalassaemia carrier status.

Thalassaemia is the most common inherited blood disorder in the world. This condition is caused by changes to the genes for haemoglobin. Haemoglobin is a protein in red blood cells that carries oxygen around the body. Changes affecting haemoglobin result in severe anaemia.

Thalassaemia can affect people of any nationality and ethnicity. It is particularly common in people of Mediterranean ancestry and across a broad region extending through India, the Middle East, Asia and Latin America.

Thalassaemia is usually diagnosed within the first six months of life and can be fatal in early childhood without ongoing treatment.

There are two different types of thalassaemia: alpha (α) and beta (β). Alpha-thalassaemia involves genetic changes in copies of two genes, whereas beta-thalassaemia involves changes in copies of one gene. Alpha-thalassaemia is more common in countries in Africa, Asia and the Middle East. Beta-thalassaemia is more common in Mediterranean countries.

In Australia, beta-thalassaemia is the most common form because of the high number of people who have migrated from Mediterranean countries. However, more migration of people from Asia means that the number of people with alpha-thalassaemia has increased in Australia.

Thalassaemia minor

Thalassaemia minor refers to people who have genetic changes in one copy (for beta-thalassaemia) or in one or two copies (for alpha-thalassaemia) of the genes, but still have other copies of thalassaemia genes that don’t have genetic changes. This means that people with thalassaemia minor have mild or no symptoms, but they carry the genetic changes and can pass them on to their children. Thalassaemia minor can refer to people with either alpha- or beta-thalassaemia.

It is conservatively estimated by the World Health Organisation that five to seven per cent of the world’s population has thalassaemia minor. People who have beta-thalassaemia minor, for example, have one copy of the changed beta-thalassaemia gene and are healthy carriers. The only way to detect if someone is a carrier is by a specific blood test for thalassaemia.

Thalassaemia major

Thalassaemia major refers to the condition where people have inherited more than one copy of the genes with genetic changes. Thalassaemia major can refer to people with either alpha- or beta-thalassaemia. People with either alpha- or beta-thalassaemia major will have symptoms associated with the condition. Beta-thalassaemia major is more common than alpha-thalassaemia major.
Carrying the alpha or beta thalassaemia gene

There is a concern when two carriers of alpha or beta thalassaemia wish to start a family. If two carriers conceive a child, the child has:

- 25 per cent risk of developing thalassaemia major because they inherited the thalassaemia gene from both parents
- 25 per cent chance of not inheriting the thalassaemia gene at all
- 50 per cent chance of inheriting the gene from one parent and becoming a carrier.

Symptoms of thalassaemia major

The red blood cells of a person with thalassaemia major can only survive for a few weeks, compared to normal red blood cells that survive for around four months. Lack of haemoglobin results in reduced oxygen to every cell in the body.

Symptoms appearing in early childhood include:

- severe anaemia – red blood cells are produced without sufficient haemoglobin to carry oxygen
- paleness
- sleep difficulties
- poor appetite
- failure to grow and thrive
- enlargement of organs – such as the spleen and liver.

Treatment for thalassaemia major

There is no cure for thalassaemia major and treatment must continue for life. Ongoing treatment includes regular transfusions to boost haemoglobin levels in the blood. However, these transfusions can lead to a build-up of iron and this can cause serious side effects, including diabetes, heart failure and liver disease.

Medications can be used to remove excess iron from the blood. These medications are called iron chelators. They work by binding with the iron and allowing it to be removed by the kidneys. Chelators can be oral or injectable, for example, Exjade is an oral chelator and Desferal is an injectable chelator.

Diagnosis of thalassaemia

It is suggested that all couples be tested for thalassaemia carrier status, especially before starting a family. In Victoria, testing can be arranged by your family doctor or is available free of charge at:

- Royal Women’s Hospital
- Monash Medical Centre
- Mercy Hospital for Women.

Options for a couple who are both carriers of thalassaemia

Carrier couples may elect not to have children, but choose to adopt, foster or consider using donor sperm or eggs. Others may elect to take the risk of having children with thalassaemia major. Couples may also choose prenatal diagnosis with the option of terminating the pregnancy if the foetus is diagnosed with thalassaemia major.

Pre-implantation genetic diagnosis (PGD) may also be used. This is where artificial reproductive technology is used to create an embryo that can be tested for thalassaemia before being transferred to the uterus of the mother.

Bone marrow transplants for thalassaemia major

A bone marrow transplant is the only chance of a cure, but the risks are considerable. The odds of finding a
compatible donor are around 30 per cent and most donors are siblings. This operation is more successful in young children who don’t suffer from iron overload, but graft rejection or even death can still result.

The risks involved mean that bone marrow transplantation is not a viable option for some families. It should be remembered that a person with thalassaemia major can live a normal life with regular transfusions and medication. Genetic research is continuing in the hope of finding a cure.

**Where to get help**

- Your doctor
- **Thalassaemia & Sickle Cell Society of Australia.** Tel. (03) 9888 2211
- **Thalassaemia Services Victoria, Monash Medical Centre.** Medical Therapy Unit Tel. (03) 9594 2756
- Royal Women’s Hospital – Genetic Counselling Service Tel. (03) 8345 2180
- Mercy Hospital for Women – Clinical Genetics Service: Tel. (03) 8458 4250

**This page has been produced in consultation with and approved by:**

Thalassaemia & Sickle Cell Society of Australia