Summary

- Thalassaemia major is a genetic condition that affects the production of haemoglobin, resulting in severe anaemia.
- This condition is usually diagnosed within the first six months of life.
- Treatment options include regular blood transfusions and bone marrow transplants.
- It is suggested that all couples be tested for their thalassaemia carrier status before starting a family.

Thalassaemia is the most common inherited blood condition 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 (a) and beta (b). Alpha-thalassaemia involves genetic changes in two genes (HBA1 and HBA2). Beta-thalassaemia involves changes in one gene (HBB). Alpha-thalassaemia is more common in countries in Africa, Asia and the Middle East. Beta-thalassaemia is more common in Mediterranean countries.

**Thalassaemia minor**

Thalassaemia minor refers to people who have genetic changes in one copy of the HBB gene (for beta-thalassaemia) or in one copy of each of the HBA1 and HBA2 genes (for alpha-thalassaemia), but still have second copies of these genes that don’t have genetic changes. They are known as ‘carriers’ of the condition.

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.

For example, a person with beta-thalassaemia minor has one changed copy of the HBB gene, and one unchanged copy of the gene. Because they have one functioning copy of the gene, their health is not affected by beta-thalassaemia.

The only way to detect if someone is a carrier is by a specific blood test for thalassaemia.

The World Health Organisation estimates that five to seven per cent of the world’s population has thalassaemia minor.

**Thalassaemia major**

Thalassaemia major refers to when a person has changes in both copies of the affected gene or genes. This means that they have no fully functioning copies of the genes.

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**

If two people who are carriers of the same type of thalassaemia have children, their children have a: 

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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 regular red blood cells that survive for around four months. Lack of haemoglobin results in reduced oxygen to every cell in the body.

Symptoms appear in early childhood. They include:
- severe anaemia – red blood cells are produced without enough 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 blood 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 taken orally (by mouth) or injected.

**Diagnosis of thalassaemia**
Thalassaemia can be diagnosed through:
- blood tests – a full blood test is used to measure the amount of haemoglobin and the different kinds of blood cells in a blood sample, and a haemoglobin test measures the types of haemoglobin present
- genetic testing.

**Pre-pregnancy testing for thalassaemia**
It is suggested that all couples thought to be at risk 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 Health
- Mercy Hospital for Women.

If you are already pregnant, consult with your family doctor regarding options for testing during pregnancy.

**Options for couples where both partners are 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**
At this time, a bone marrow transplant is the only chance of a cure for thalassaemia major, 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. Remember 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.

**Genetic counselling and thalassaemia**

If your child or another family member has been diagnosed with thalassaemia, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand thalassaemia and what causes it, how it is inherited, and what a diagnosis means for your child’s health and development. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If thalassaemia runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

**Thalassaemia and Sickle Cell Australia** is a support network for people affected by thalassaemia and other related genetic blood conditions in Australia.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by thalassaemia.

**Where to get help**

- **Your GP (doctor)**
- **Thalassaemia and Sickle Cell 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
- **Monash Health – Genetics Clinic** Tel. (03) 9594 2026
- **Victorian Clinical Genetics Services** Tel. 1300 118 247