

## Thalassaemia

Thalassaemia is passed from parent to child and is the most common inherited blood disorder in the world. This condition is caused by changes to the genes that control the production of 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 anyone of any nationality, but it is particularly prevalent in people with ancestry from Mediterranean countries and across a broad region extending through India, the Middle East and South East Asia. 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 ( $\alpha$ ) and beta ( $\beta$ ), with the most common being  $\beta$ -thalassaemia.

### Thalassaemia minor

$\beta$ -thalassaemia minor is common. People who have  $\beta$ -thalassaemia minor have one copy of the  $\beta$ -thalassaemia gene. It is estimated that 100,000 people in Victoria have  $\beta$ -thalassaemia minor and carry this gene. These individuals are healthy carriers. The only way to detect the carrier state is by a specific blood test for thalassaemia.

### Thalassaemia major

The concern arises when two carriers of  $\beta$ -thalassaemia wish to start a family. If two carriers conceive a child, the child has a:

- 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.

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. The following signs appear in early childhood:

- Severe anaemia, because red blood cells are produced without sufficient oxygen-carrying haemoglobin
- Paleness
- Sleep difficulties
- Poor appetite
- Failure to grow and thrive
- Enlargement of organs, such as the spleen and liver.

### Treatment

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 excreted by the kidneys. The most commonly used medication is injected using a slow infusion pump that runs over several hours.

Oral iron chelators (tablets) may be prescribed where the infusion treatment is unsuitable or ineffective; however, these medications may have severe side effects and must be carefully monitored. One of these oral iron chelators has only recently been approved for use in Australia and is not listed on the Pharmaceutical Benefits Scheme.

## Testing

Couples from communities with a higher than average risk of being carriers should be tested for thalassaemia, especially before starting a family. Testing can be arranged by your family doctor or is available free of charge at:

- Royal Women's Hospital Tel. (03) 8345 2180
- Monash Medical Centre Tel. (03) 9594 2756

## Options for a carrier couple

Couples who both have -thalassaemia minor may elect not to have children and may 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 prior to being transferred to the uterus of the mother.

## Bone marrow transplants

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 ongoing in the hope of finding a cure.

## Where to get help

- Your doctor
- Thalassaemia Australia Inc. Tel. (03) 9888 2211
- Monash Medical Centre Tel. (03) 9594 2756
- Royal Women's Hospital Tel. (03) 8345 2180

## Things to remember

- 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 from communities with a higher than average risk of being carriers should be tested for thalassaemia.

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

Thalassaemia Australia Inc

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