Beta (β) thalassaemia

What is Beta (β) thalassaemia?

Thalassaemia is a group of blood disorders affecting haemoglobin production. Haemoglobin is a protein in the blood that carries oxygen around our bodies.

Thalassaemia is passed from parent to child in genes. Genes carry information about human characteristics such as eye colour, hair colour and haemoglobin.

Thalassaemia is inherited.
Thalassaemia is not contagious.
Thalassaemia is not transmitted by germs.

Sometimes changes occur to genes, resulting in medical conditions. Such changes occur to beta globin genes in beta (β) thalassaemia:

- A person normally inherits two β globin genes for the production of the beta globin protein in haemoglobin.
- A person may have an alteration (mutation) in one of their two β globin genes. This person is called a **carrier of β thalassaemia** and is healthy. Doctors may use the term β **thalassaemia minor** instead but it means the same thing.
- Carriers may be at risk of having a child affected with beta thalassaemia major if their partner is also a carrier of β thalassaemia.
- When a person has alterations (mutations) in both of their β globin genes, they have a severe condition called β thalassaemia major. β thalassaemia major results in severe anaemia requiring life long treatment.

Treatment for β thalassaemia major

Those with β thalassaemia major require regular blood transfusions every 3 to 4 weeks to correct anaemia. Complications of their treatment include accumulation of excess iron, which can be effectively prevented and managed with medication.

The health of carriers of β thalassaemia

A carrier can expect to be **healthy**. It is important that their doctor knows they are a carrier to distinguish any anaemia from anaemia caused by low iron levels.

Beta thalassaemia and family planning

The genes for β thalassaemia are common in people of Middle Eastern, Mediterranean, Indian sub-continent and South-East Asian backgrounds.

Couples planning a family, or early in pregnancy, should have a blood test to determine whether or not they are carriers, if the origin of either of their families is one of the areas listed above; or if they have a family history of any blood disorder or anaemia. This test is needed to determine if there is any risk of having a child affected by a genetic blood disorder.

Those at risk of having an affected child have options. This condition can be diagnosed as early as the 12th week of pregnancy. Termination of pregnancy can then be considered, if appropriate. People can adopt or can consider assisted reproductive techniques (such as preimplantation genetic diagnosis, the use of donor eggs or donor sperm). Others may choose to take the chance of having an affected child. All of these options can be discussed with a Genetic Counsellor.

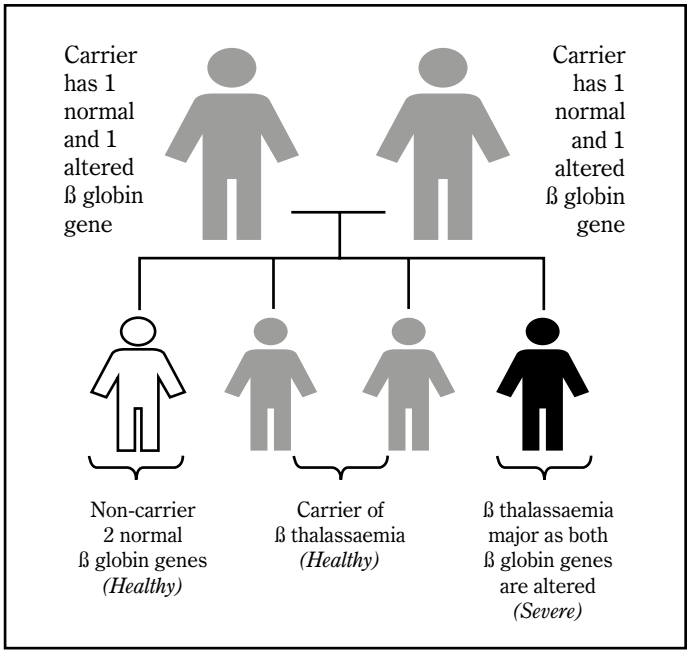
Testing can be arranged by your local doctor or by contacting the hospitals listed at the end of this pamphlet.

Important information for your family

If you are a carrier of beta thalassaemia, other members of your family may also be carriers and at risk of having children with a severe blood condition. It is recommended that other family members **and** their partners be tested for their carrier status **prior** to having children of their own.

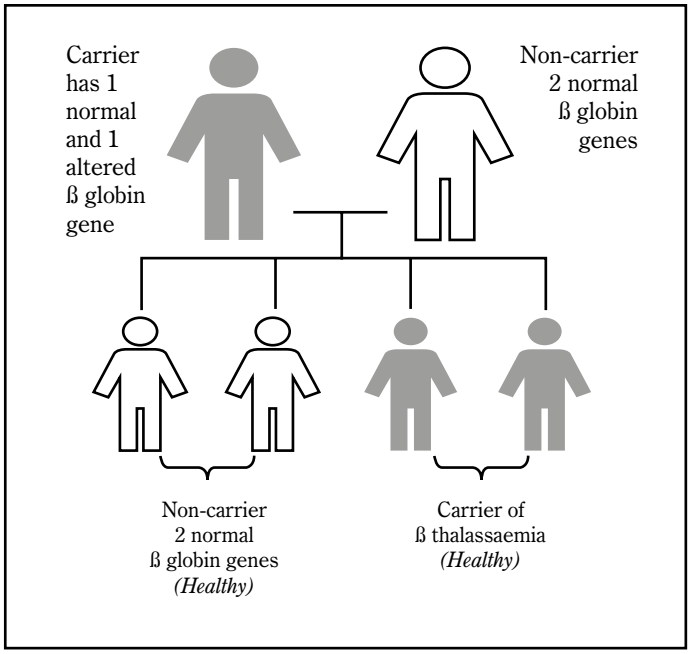
Chances of having a child affected with β thalassaemia major

Figure 1:
Both parents are carriers of β thalassaemia



With **each** pregnancy, this couple has a:
- 1 in 4 chance of having a child with 2 normal β globin genes.
- 2 in 4 chance of having a carrier of β thalassaemia.
- 1 in 4 chance of β thalassaemia.

Figure 2:
Only one parent is a carrier of β thalassaemia



With **each** pregnancy, this couple has a:
- 2 in 4 chance of having a child with 2 normal β globin genes.
- 2 in 4 chance of having a carrier of β thalassaemia.

This fact sheet has been translated into a number of different languages and they are available to download at www.thalassaemia.org.au

Useful contacts

Monash Medical Centre
Medical Therapy Unit
(State Thalassaemia Service, Southern Health)
246 Clayton Road Clayton VIC 3168
Phone: +61 3 9594 2756 or +61 3 9594 3154

Royal Women's Hospital
Thalassaemia Clinic
Cnr Grattan St & Flemington Rd
Parkville VIC 3052
Phone: +61 3 8345 2180

Mercy Hospital for Women
Genetics
163 Studley Road
Heidelberg VIC 3084
Phone: +61 3 8458 4250

Thalassaemia AUSTRALIA
Unifying support and genetics

Thalassaemia Australia Inc.
333 Waverley Road
Mount Waverley VIC
AUSTRALIA 3149

Phone: +61 3 9888 2211
Fax: +61 3 9888 2150
Email: info@thalassaemia.org.au
Website: www.thalassaemia.org.au

Revised: 06/12