Family planning...?

Please consider having a carrier test for genetic blood disorders

Genetic Blood Disorders

Around *1 in 20 people worldwide is thought to have inherited an altered globin gene which they could pass on to their children. This may potentially result in a genetic blood disorder, affecting haemoglobin production. Haemoglobin is a protein in the blood that carries oxygen around our bodies.

These inherited blood disorders are passed from parent to child in genes. Genes carry information about human characteristics such as eye colour, hair colour and haemoglobin.

These genetic blood disorders are inherited.
These genetic blood disorders are not contagious.
These genetic blood disorders are not transmitted by germs.

Sometimes changes occur to genes, resulting in medical conditions. The globin genes carry information for individuals to make haemoglobin, however changes in the globin genes can result in the following:

- **Beta thalassaemia major** - a severe anaemia requiring life long treatment, including monthly blood transfusions.
- **Alpha thalassaemia** - one type can be fatal for baby and dangerous for mother during pregnancy.
- **Sickle cell disease** - a condition which may require regular blood transfusions. Extremely painful blockages of blood vessels occur throughout life due to changes in the shape of the red blood cells.
- **Haemoglobin E** - usually a benign condition, but if inherited with beta thalassaemia or sickle cell, can result in a severe condition requiring life long treatment.

The health of people who carry an altered globin gene

A person who carries an altered globin gene is termed a 'carrier'. A carrier is **healthy**. It is important that their doctor knows they are a carrier.

What does it mean to be a carrier of an altered globin gene?

If you are a carrier of an altered globin gene it is important that your partner has a blood test to determine his or her carrier status. It is only if you are **both** carriers of certain globin gene alterations that you have a risk of having a child affected with a severe genetic blood disorder.

Reasons for family planning

The genes for these genetic blood disorders are common in people of Mediterranean, Middle Eastern, African (including Caribbean or African American) origin, as well as any Asian countries: India, Pakistan, Sri Lanka, Bangladesh, Indonesia, Philippines, Thailand, Vietnam and China etc. Other origins include Pacific Islands and New Zealand Maoris.

Couples planning to have a family, or where the woman is already pregnant, should consider having a blood test to see if they are a carrier if they were born in or have family (even distant relatives) from any of the areas listed above. This test is needed to determine what chance carriers have of having a child affected by a genetic blood disorder.

Those at risk of having an affected child have options. These conditions 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 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.

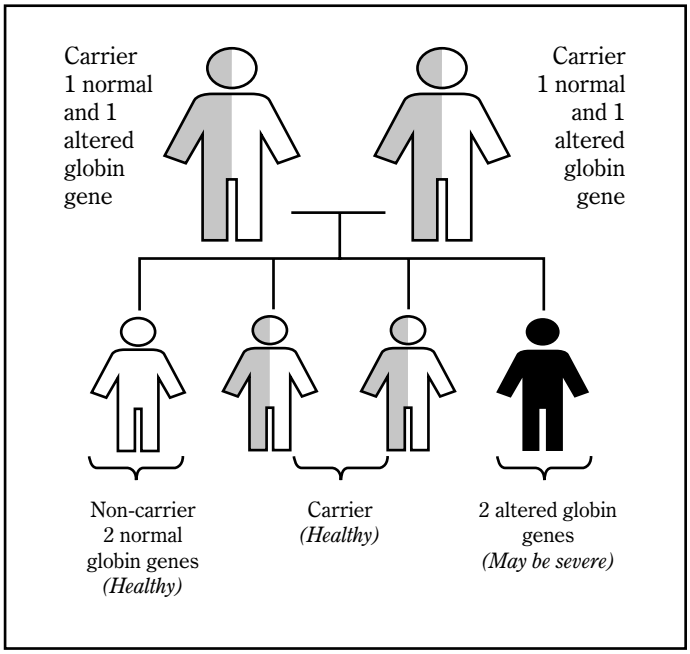
Important information for your family

If you or your partner are carriers of an altered globin gene, other members of your families may also be carriers and at risk of having children with a severe blood disorder. It is recommended that all 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 by a genetic blood disorder

Altered genes are passed on from parents to children in the following fashion:

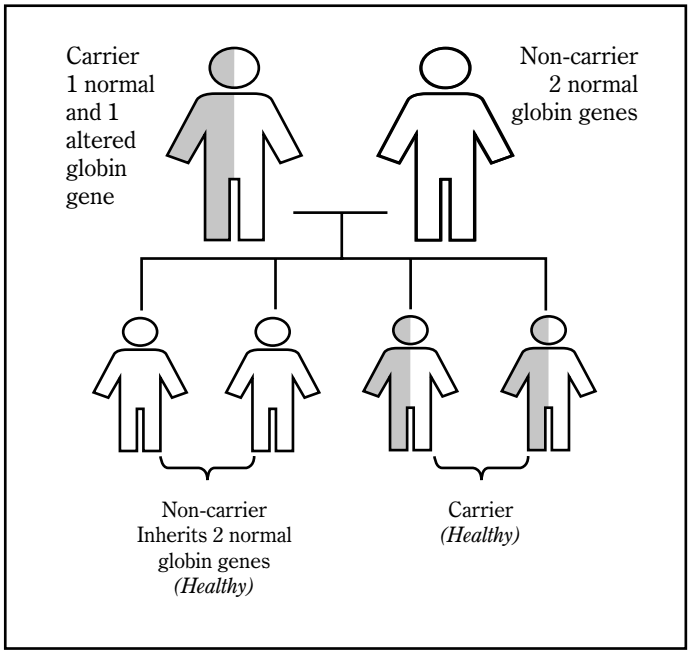
Figure 1:
Both parents are carriers



With **each** pregnancy, this couple has a:

- 1 in 4 chance of having a child who inherits 2 normal copies of the globin gene and is a non-carrier.
- 2 in 4 chance of having a healthy carrier, like its parents. This child has one normal copy of the globin gene and one altered copy of the globin gene.
- 1 in 4 chance of having a child which may have a severe genetic blood disorder. This child has inherited 2 altered copies of the globin gene.

Figure 2:
Only one parent is a carrier



With **each** pregnancy, this couple has a:

- 2 in 4 chance of having a child who inherits 2 normal copies of the globin gene and is a non-carrier.
- 2 in 4 chance of having a child who is a healthy carrier - like its parent. This child has one normal copy of the globin gene and one altered copy of the globin gene.

**Note: (ref: Modell B, Darlison (2008) Global epidemiology of haemoglobin disorders and derived service indicators - Bulletin of the World Health Organisation Vol 86, number 6, June 2008: 417-496)*

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