

# Family planning...?

Please consider having a carrier test for genetic blood disorders

## Genetic Blood Disorders

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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

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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?

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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

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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

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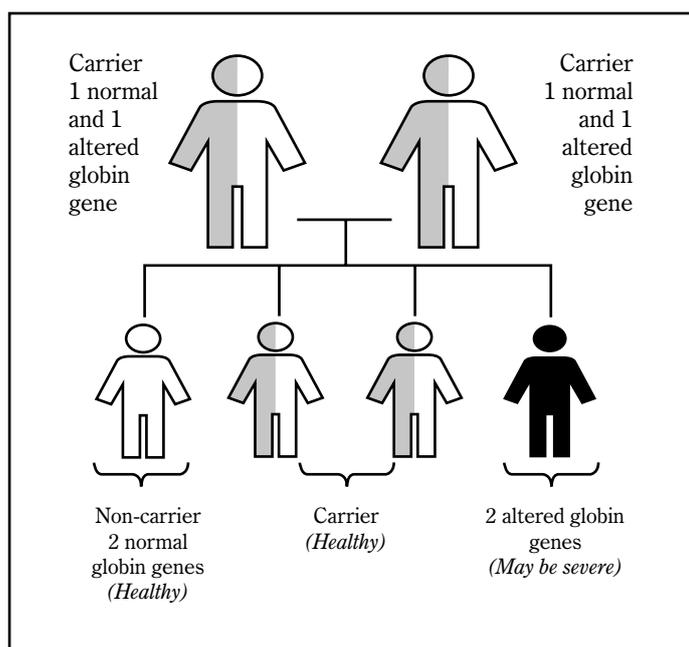
If you or your partner are carriers of an altered globin gene, other members of your families may also be carriers and at risk of having children with a severe blood disorder. It is recommended that all other family members **and** their partners be tested for their carrier status **prior** to having 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:

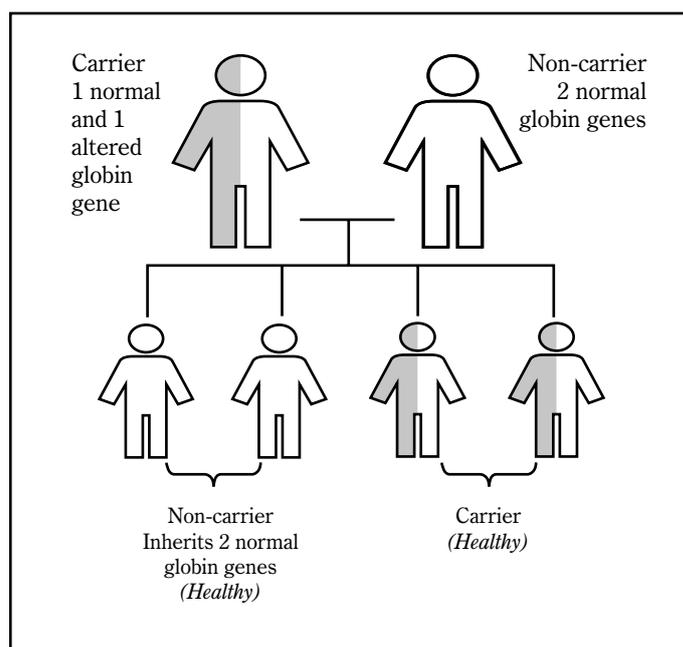
**Figure 1:**

**Both parents are carriers**



**Figure 2:**

**Only one parent is a carrier**



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.

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)

### Useful contacts

#### Thalassaemia Society of NSW

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Missenden Road  
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Phone: +61 2 9550 4844  
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www.thalns.org.au

#### Children's Hospital at Westmead

Haematology Department  
Cnr Hawkesbury Rd and Hainsworth St,  
Westmead NSW 2145  
Ph: +61 2 9845 0000

#### Sydney Children's Hospital

Department of Haematology  
High Street, Randwick NSW 2031  
Ph: +61 2 9382 1111

#### The Prince of Wales Hospital

Department of Haematology  
High Street, Randwick NSW 2031  
Ph: +61 2 9382 4982

#### Royal Prince Alfred Hospital

Department of Haematology  
Level 5, Missenden Road  
Camperdown NSW 2050  
Ph: +61 2 9515 7013

#### Westmead Hospital

Department of Haematology  
Cnr Hawkesbury Road & Darcy Roads  
Westmead NSW 2145  
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#### Liverpool Hospital

Department of Haematology  
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This fact sheet has been translated into a number of different languages and they are available to download at [www.thalassaemia.org.au](http://www.thalassaemia.org.au)