Alpha (α) thalassaemia

What is alpha (α) 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 alpha globin genes in alpha (α) thalassaemia:

- A person normally inherits four α globin genes for the production of the alpha globin protein in haemoglobin.
- A person may have two or three of the normal four alpha globin genes for haemoglobin production. This person is called a **carrier of α thalassaemia** and is healthy.
- Carriers may be at risk of having a child affected with Haemoglobin H disease or Bart's hydrops fetalis if their partner is also a carrier of certain types of α thalassaemia.
- When a person has only one alpha globin gene, they have **Haemoglobin H disease** and require regular medical care. Individuals with Haemoglobin H disease may experience lifelong anaemia of mild to moderate degree. Occasionally it may be severe.
- When a person has no alpha globin genes, they have a severe condition called **Bart's hydrops fetalis**. Bart's hydrops fetalis affects a foetus long before birth, resulting in death during pregnancy or shortly after birth. This is a fatal condition which is dangerous for both the mother and baby during pregnancy.

Treatment

Those with Haemoglobin H disease may require blood transfusions to correct anaemia. There is no treatment or cure for Bart's hydrops fetalis.

The health of carriers of α thalassaemia

A carrier can expect to be **healthy**. It is important that their doctor knows they are a carrier of α thalassaemia.

Alpha thalassaemia and family planning

The genes for a thalassaemia are common in people of Asian origin, as well as those of African, Middle Eastern and Mediterranean origin.

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. 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 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.

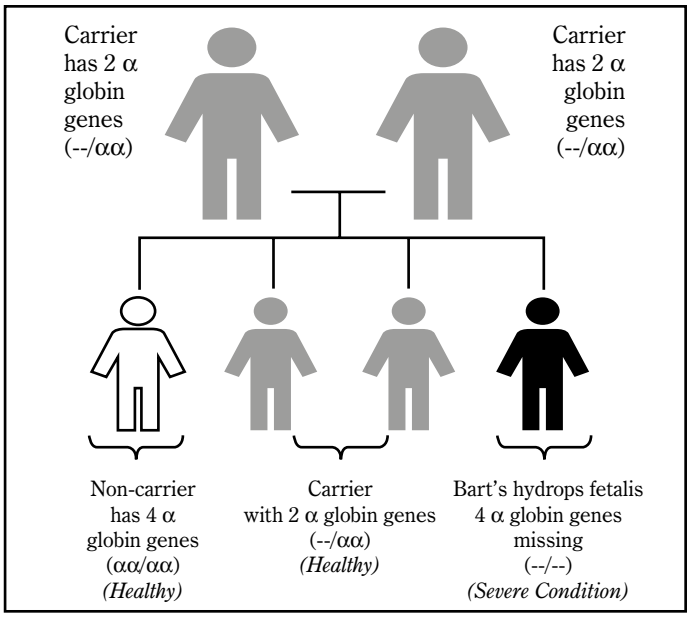
Testing can be arranged by your local doctor or by contacting the hospitals listed at the end of this pamphlet.

Important information for your family

If you are a carrier of alpha thalassaemia, other members of your family may also be carriers and at risk of having children with a severe form of alpha thalassaemia. It is recommended that all other family members **and** their partners be tested for their carrier status **prior** to having children of their own. DNA testing is utilized to detect carriers of alpha thalassaemia.

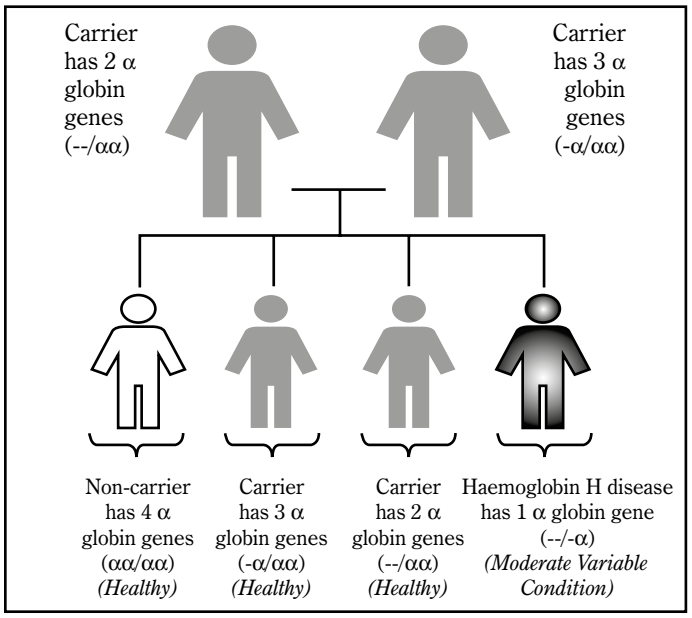
Chances of having a child affected with α thalassaemia

Figure 1:
Carrier parents have 2 alpha (α) globin genes



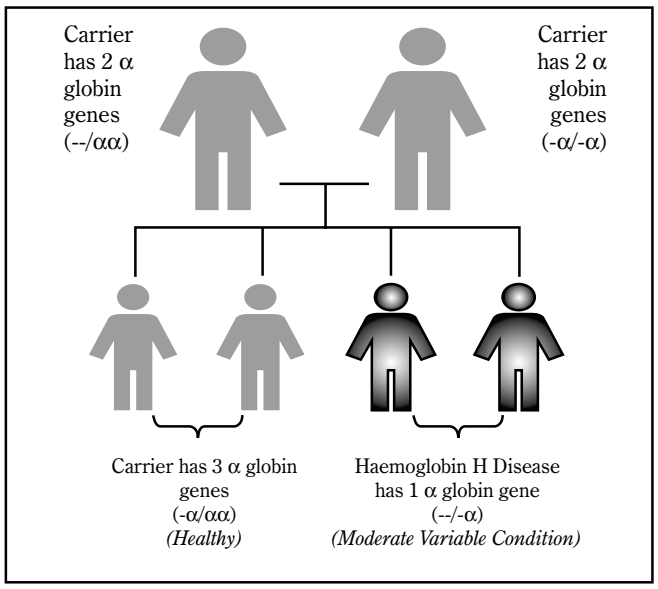
With **each** pregnancy, this couple has a:
- 1 in 4 chance of having a child who is not a carrier of α thalassaemia.
- 2 in 4 chance of having a healthy carrier.
- 1 in 4 chance of Bart's hydrops fetalis.

Figure 2:
Carrier parents have 2 and 3 α globin genes



With **each** pregnancy, this couple has a:
- 1 in 4 chance of having a child who is not a carrier of α thalassaemia.
- 2 in 4 chance of having a healthy carrier.
- 1 in 4 chance of having a child with Haemoglobin H disease.

Figure 3: Carrier parents have 2 α globin genes



With **each** pregnancy, this couple has a:
- 2 in 4 chance of having a healthy carrier.
- 2 in 4 chance of having a child with Haemoglobin H disease.

Useful contacts

Thalassaemia Society of NSW
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Level 7 King George V Building
Missenden Road
CAMPERDOWN NSW 2050
Phone: +61 2 9550 4844
Mobile: 0400 116 393
www.thalnsw.org.au

Children's Hospital at Westmead
Haematology Department
Cnr Hawkesbury Road and
Hainsworth Street,
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 Rd & Darcy Rds
Westmead NSW 2145
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Liverpool Hospital
Department of Haematology
Ground Floor
Cnr Elizabeth & Goulburn Street
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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

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