What is Thalassaemia?

Thalassaemia is a group of blood disorders affecting haemoglobin production. Haemoglobin is a protein in red blood cells that carries oxygen around our bodies. Haemoglobin is made up of haem groups containing iron and globin chains. The main globin chains are call alpha (α) and beta (β). People with thalassaemia don't produce enough globin chains. People with thalassaemia don't produce enough globin chains and therefore not enough haemoglobin, resulting in anaemia.

Thalassaemia is an inherited condition meaning it is passed from parent to child in genes. Genes carry information about human characteristics such as hair colour. Thalassaemia is **NOT** contagious and **NOT** transmitted by germs.

What is α Thalassaemia?

Genetic mutations/ alterations in the alpha globin genes can lead to α thalassaemia:

- A person normally inherits four α globin genes for the production of the alpha globin protein in haemoglobin.
- α thalassaemia occurs when a person doesn't produce enough α globin chains.
- A person may have only two or three of the four normal α globin genes. This person is a carrier of α thalassaemia and is normally healthy.
    - α thalassaemia carriers may be at risk of having a child with Haemoglobin H disease or Bart's hydrops fetalis if their partner is also an α thalassaemia carrier.
    - α thalassaemia carriers may be mistaken for having iron deficiency.
- When a person has only one α globin gene, they have a condition known as Haemoglobin H disease. These patients usually have life-long mild-moderate anaemia, it is not usually severe.
- When a person has no α globin genes, this causes a severe condition known as Bart's hydrops fetalis. This results in death of the baby during pregnancy or shortly after birth.

Treatment for α Thalassaemia

α thalassaemia carriers generally require no treatment. Patients with Haemoglobin H disease may require blood transfusions to help manage their anaemia. Only a small number of patients survive with Bart's hydrops fetalis, those who do survive require life-long blood transfusions that begin during the pregnancy. These patients are also susceptible to many other growth and developmental complications.

α Thalassaemia and Family Planning

The genes for α thalassaemia are common in some ethnic groups including people from Asia, Africa, the Middle East and Mediterranean.

Couples planning a pregnancy should have a blood test to determine whether they are carriers if:

- They or their partner are carriers for α thalassaemia or have haemoglobin H disease, or
- There is a family history of thalassaemia, or
- If their family origin is an area listed above.

Testing can be arranged by your local doctor. By testing we are able to determine whether there is a risk of having a child with thalassaemia.

Where there is a risk of having an affected child, several options are available. Thalassaemia can be diagnosed as early as the 12th week of pregnancy so termination of pregnancy can be considered, if

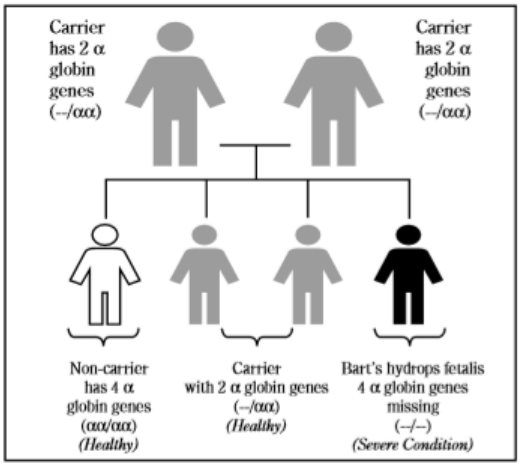
appropriate. Couples can adopt or can consider assisted reproductive techniques (such as pre-implantation genetic diagnosis, the use of donor eggs or donor sperm). Others may choose to take the chance of having an affected child. All of these options should be discussed with a Genetic Counsellor.

Important information for your family

If you are a carrier of α thalassaemia, other members of your family should be tested as they may also be carriers and at risk of having children with a severe thalassaemia. It is recommended that other family members and their partners are screened before having children of their own.

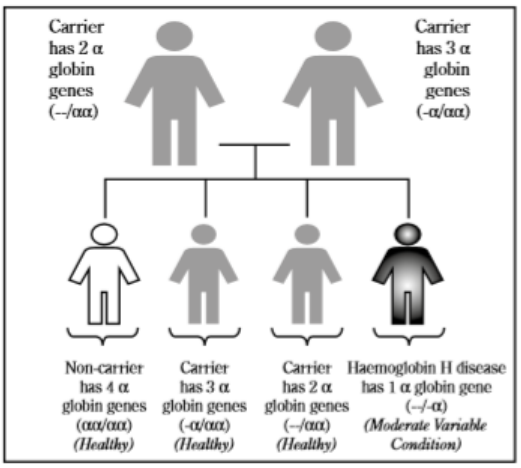
Chances of having a child affected with α thalassaemia

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



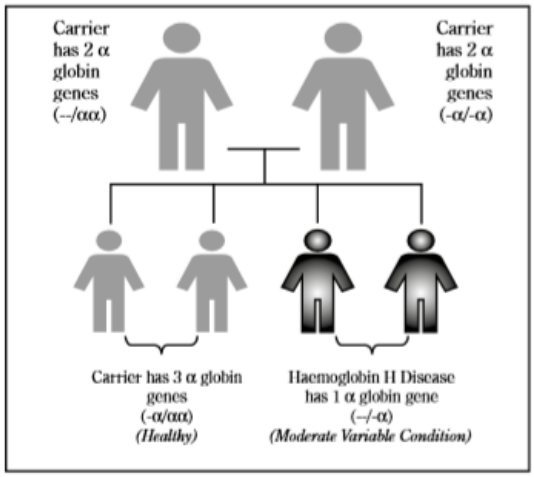
With **each** pregnancy, this couple has a:
- 1 in 4 chance of having a child who is not a carrier of α thalassaemia.
- 2 in 4 chance of having a healthy carrier.
- 1 in 4 chance of Bart's hydrops fetalis.

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.

References:

https://www.tasca.org.au

https://www.thalassemia.org/learn-about-thalassemia/about-thalassemia/#thalassemiabrochures

For Medical Professionals seeking further information or advice, please contact Pathlab to discuss with one of our Haematologists: **07 858 0795**