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 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 β thalassaemia?

Genetic mutations/ alterations in the beta globin genes can lead to β thalassaemia:

- A person normally inherits two β globin genes for the production of the beta globin protein in haemoglobin.
- β thalassaemia occurs when a person does not produce enough β globin chains.
- A person may have a mutation in one of their two β globin genes. This person is called a carrier of β thalassaemia and may have a mild anaemia but overall is healthy. Doctors may use the term β thalassaemia minor, heterozygous β thalassaemia or β thalassaemia trait instead, but they mean the same thing. It is important that doctors are aware that their patient is a carrier:
    - Carriers may be at risk of having a child with β thalassaemia major if their partner is also a carrier of β thalassaemia.
    - To help distinguish anaemia from thalassaemia from anaemia caused by low iron levels.
- 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.
- B thalassaemia can be inherited with other types of thalassaemia or variant haemoglobins which can also produce severe conditions.

Treatment for β thalassaemia

β thalassaemia minor requires no treatment, but it is still important that the doctor is aware of their patient having this to prevent confusion with iron deficiency. This could result in unnecessary treatment with iron tablets.

Those with β thalassaemia major will require blood transfusions every 3-4 weeks. Complications of this include accumulation of iron which is prevented and managed with medication.

β thalassaemia and family planning

The genes for β thalassaemia are common in some ethnic groups including people from the Middle East, Mediterranean, India, and South-East Asia.

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

- They or their partner are carriers of β thalassaemia or have β thalassaemia major, or
- There is a family history of thalassaemia or variant haemoglobin, or
- If their family origin is an area listed above.

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

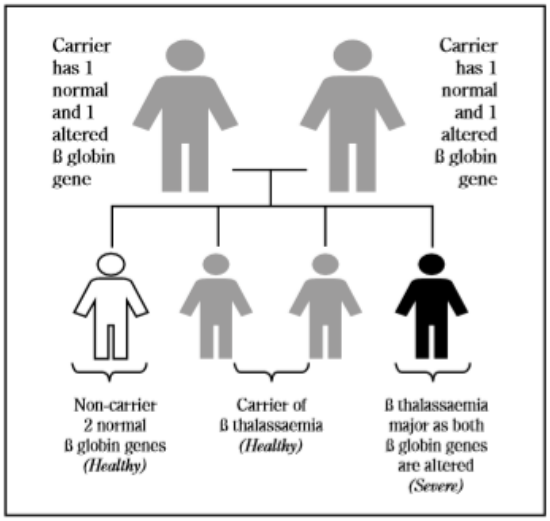
Where there is a risk of having an affected child, several options are available. Thalassaemia can be diagnosed as early as the 12th week of pregnancy so termination of pregnancy can be considered, if appropriate. Couples can adopt or can consider assisted reproductive techniques (such as pre-implantation genetic diagnosis, the use of donor eggs or donor sperm). Others may choose to take the chance of having an affected child. All these options should be discussed with a Genetic Counsellor.

Important information for your family

If you are a carrier of β 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.

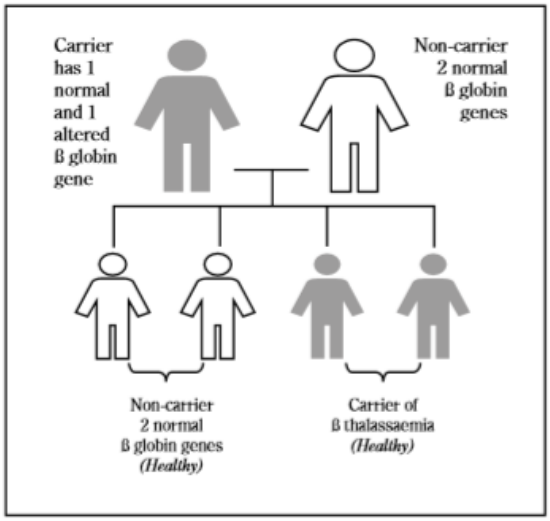
Chances of having a child affected with β thalassaemia major

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.

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.**