What is Haemoglobin C?

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). Mutations in the globin genes leads to a variant haemoglobin (haemoglobinopathy) being produced. Haemoglobin C results from a genetic mutation/alteration affecting the β globin gene.

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

Inheritance

- A person normally inherits two β globin genes (one from each parent) for the production of the β globin protein in haemoglobin.
- A person may have the haemoglobin C mutation in one of their two β globin genes. This person is referred to as a carrier for haemoglobin C and is generally healthy. Doctors may use the term haemoglobin C trait or heterozygous haemoglobin C, but the mean the same thing.
    - Carriers may be at risk of having a child with haemoglobin C disease if their partner is also a haemoglobin C carrier.
- When both β globin genes have the haemoglobin C gene this person is said to have haemoglobin C disease.
    - These patients may have a mild anaemia but are generally healthy.
- Haemoglobin C can be inherited with other haemoglobin variants or thalassaemia's resulting in severe conditions. Examples of other variant haemoglobins/ thalassaemia's to be aware of include:
    - Haemoglobin S
    - β thalassaemia

Treatment

Treatment is not usually required for haemoglobin C trait or haemoglobin C disease. However, if the haemoglobin C mutation is inherited with any of the other conditions listed above, this can cause more severe disease requiring life-long transfusions and other supportive/preventative treatment.

Haemoglobin C and family planning

The haemoglobin C gene is most common in people from Latin America, the Caribbean region, Africa, Italy and Greece. Couples planning a pregnancy should have a blood test to determine whether they are carriers if:

- They or their partner are carriers for haemoglobin C or have haemoglobin C disease, 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 a severe haemoglobinopathy/thalassaemia.

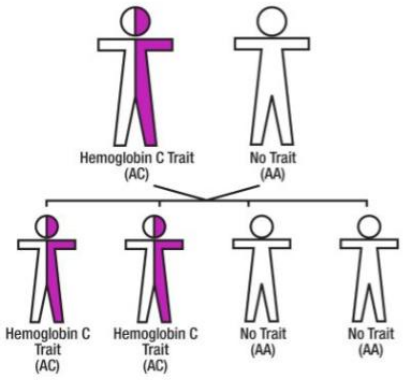
If both parents are found to be carriers of a thalassaemia or variant haemoglobin, this should be discussed with a haematologist to assess the risk for their pregnancy/ future pregnancies. These conditions can be diagnosed as early as the 12th week of pregnancy so termination of pregnancy can be considered, 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 haemoglobin C, other members of your family should be tested as they may also be carriers and at risk of having children with a severe disease. It is recommended that other family members and their partners are screened before having children of their own.

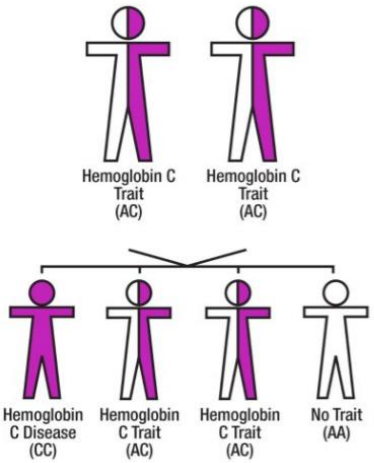
If one parent has haemoglobin C trait and the other parent has normal haemoglobin A, there is a 50 percent (1 in 2) chance **with each pregnancy** of having a child with haemoglobin C **trait**. These are the possible outcomes with each pregnancy. (Please note 'A' refers to the normal haemoglobin gene)

- 50 percent (1 in 2) chance of having a child with Haemoglobin C trait
- 50 percent (1 in 2) chance of having a child without trait



If both parents have haemoglobin C trait there is a 25 percent (1 in 4) chance **with each pregnancy** of having a child with haemoglobin C **disease**. These are the possible outcomes ***with each pregnancy***.

- 25 percent (1 in 4) chance of having a child with Haemoglobin C disease
- 50 percent (1 in 2) chance of having a child with Haemoglobin C trait
- 25 percent (1 in 4) chance of having a child without trait or disease



This is the same inheritance pattern when one parent is a carrier of Haemoglobin C and the other is a carrier of another abnormal haemoglobin. For example Haemoglobin S or β thalassaemia.

References:

Hemoglobin C Trait - St. Jude Children's Research Hospital

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