Sickle cell trait (SCT)

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). Sickle cell trait results from a genetic mutation/alteration affecting the β globin gene. The variant haemoglobin (haemoglobinopathy) is referred to as Haemoglobin S.

Sickle cell trait is passed from parent to child in genes. Genes carry information about human characteristics such as hair colour. It 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 S mutation in one of their two β globin genes. This person is referred to as a sickle cell carrier/trait and is generally healthy. Doctors may use the terms heterozygous haemoglobin S or sickle cell trait, but they mean the same thing.
- Sickle cell trait is **NOT** the same as sickle cell disease (SCD), however,
    - Carriers may be at risk of having a child with sickle cell disease if their partner is also a sickle cell carrier.
- When both β globin genes have the sickle cell mutation, this person has sickle cell disease. Instead of the patient's red blood cells being round, they become sickle or crescent shaped. These sickle cells are fragile and break down more easily resulting in anaemia and other serious side effects.
- Haemoglobin S 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:
    - β thalassaemia (beta thalassaemia)
    - Haemoglobin C
    - Haemoglobin D^{Punjab}
    - Haemoglobin O^{Arab}
    - δβ thalassaemia (delta-beta thalassaemia)
    - Haemoglobin Lepore
- HPFH (Hereditary Persistence of Foetal Haemoglobin)

Treatment for sickle cell trait

Patients with sickle cell trait generally require no medical treatment and live normal lives.

Sickle cell trait and family planning

The genes for sickle cell disease are common in people of African, Middle eastern, Southern European, Indian, Pakistani, and Caribbean descent.

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

- They or their partner are carriers for haemoglobin S or have sickle cell 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 are able to determine whether there is a risk of having a child with sickle cell disease.

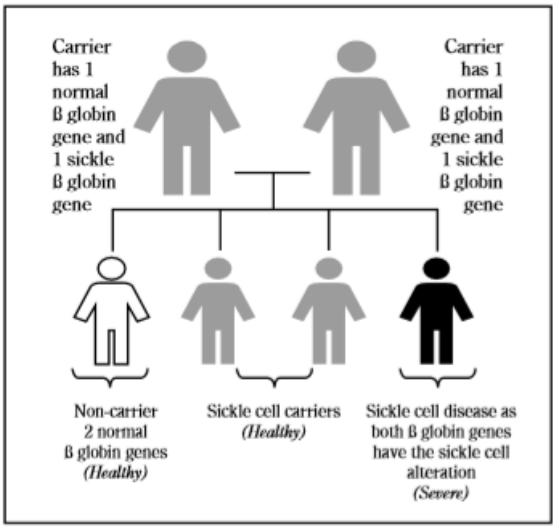
Where there is a risk of having an affected child, several options are available. This condition can be diagnosed as early as the 12th week of pregnancy so termination of pregnancy can be considered, if appropriate. Couples can adopt or can consider assisted reproductive techniques (such as pre-implantation 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 sickle cell carrier, other members of your family should be tested as they may also be carriers and at risk of having children with sickle cell disease. It is recommended that other family members and their partners are screened before having children of their own.

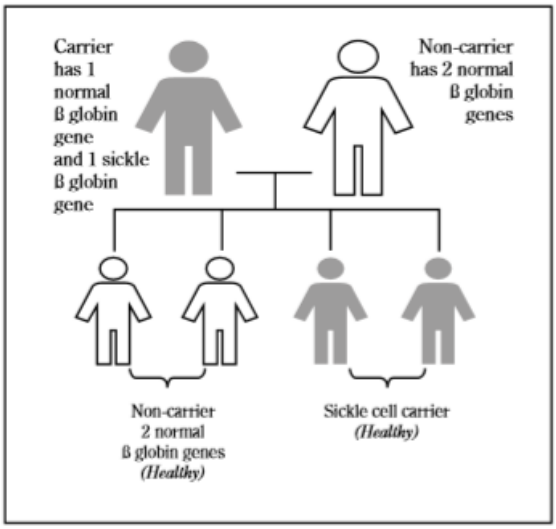
Chances of having child with Sickle Cell Trait or Sickle Cell Disease

Figure 1:
Both parents are sickle cell carriers



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 sickle cell carrier.
- 1 in 4 chance of sickle cell disease.

Figure 2:
Only one parent is a sickle cell carrier



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 sickle cell carrier.

References:

https://www.tasca.org.au

https://hematology.org/education/patients/anemia/sickle-cell-disease

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