

Sickle cell anaemia

What is sickle cell anaemia?

Sickle cell anaemia (also known as sickle cell disease) is a blood disorder affecting haemoglobin production. Haemoglobin is a protein in the blood that carries oxygen around our bodies.

Sickle cell anaemia is passed from parent to child in genes. Genes carry information about human characteristics such as eye colour, hair colour and haemoglobin.

Sickle cell anaemia is inherited.

Sickle cell anaemia is not contagious.

Sickle cell anaemia is not transmitted by germs.

Sometimes changes occur to genes, resulting in medical conditions. Such changes occur to beta (β) globin genes in sickle cell anaemia:

- A person normally inherits two β globin genes for the production of the beta globin protein in haemoglobin.
- A person may have the sickle alteration (mutation) in one of their two β globin genes. This person is called a **sickle cell carrier** and is **healthy**.
- Carriers may be at risk of having a child affected with sickle cell anaemia if their partner is also a sickle cell carrier.
- When a person has the sickle alteration (mutation) in **one** of their β globin genes, and they have a certain alteration in their other β globin gene they may have a condition called sickle cell anaemia.

Treatment

Those with sickle cell anaemia require regular medical management to correct the anaemia and to prevent and manage painful crises.

The health of sickle cell carriers

A carrier can expect to be **healthy** and should discuss their carrier state with their doctor.

Sickle cell anaemia and family planning

The genes for sickle cell anaemia are common in people of African, Middle Eastern, Southern European, Indian, Pakistani and Caribbean 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. This condition 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 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.

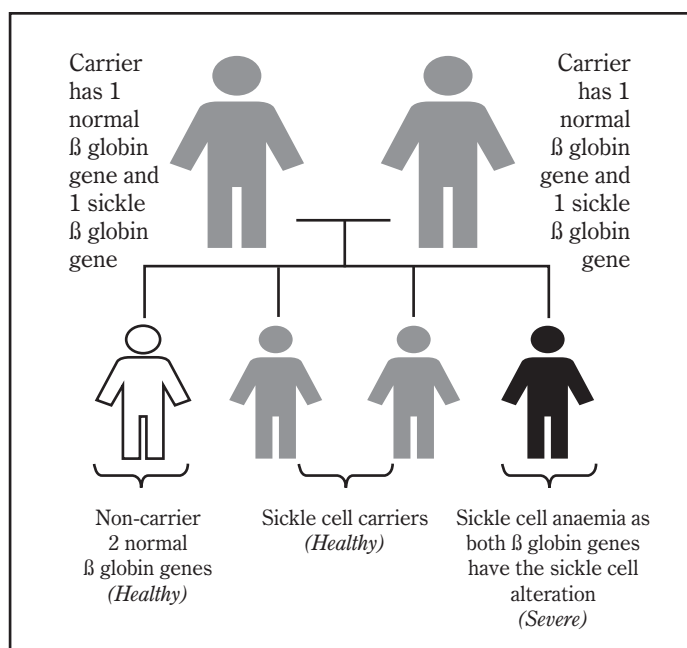
Important information for your family

If you are a carrier of the HbS gene (sickle alteration) other members of your family may also be carriers and at risk of having children with a severe blood condition. It is recommended that other family members **and** their partners be tested for their carrier status **prior** to having children of their own.

Chances of having a child affected by sickle cell anaemia

Figure 1:

Both parents are sickle cell carriers

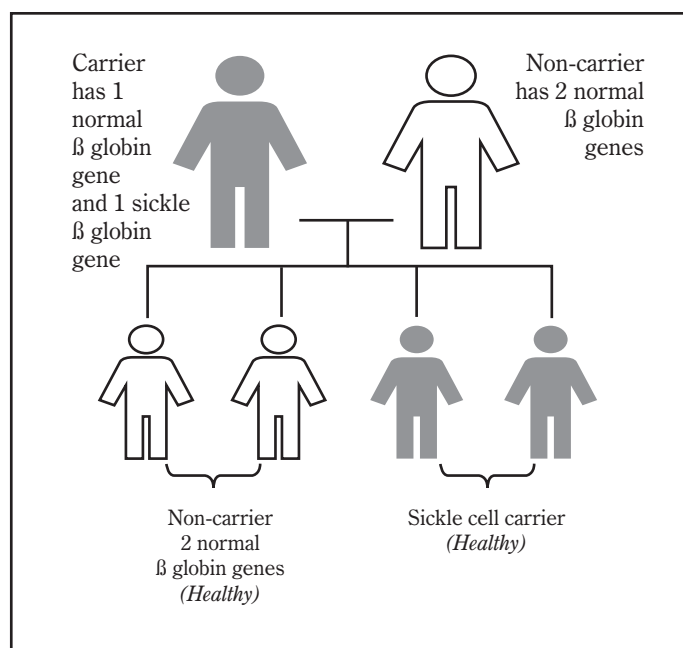


With **each** pregnancy, this couple has a:

- 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 anaemia.

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.

Useful contacts

Mercy Hospital for Women
Genetics
163 Studley Road,
Heidelberg VIC
Australia 3084
Phone: +61 3 8458 4250

Monash Medical Centre
Medical Therapy Unit
246 Clayton Road,
Clayton VIC
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Royal Women's Hospital
Thalassaemia Clinic
Cnr Grattan St & Flemington Rd,
Parkville VIC
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