Sickle Cell / Haemoglobin C Disease (Hb S/C):

Information Sheet

Produced by Specialist Haematology, Canterbury Health Laboratories

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What is Haemoglobin?

Haemoglobin (Hb) is a protein found in red blood cells; it carries oxygen from the lungs to all parts of the body and gives blood its red colour. Hb is made up of haem groups, which contain iron and globin chains which are made of protein.

The combination of different globin chains with the haem groups produces the different types of haemoglobin present in people. The normal adult haemoglobin is Hb A.

Where a person has inherited Hb A from both parents, they will have the usual Hb combination, Hb AA.

What is a Variant Haemoglobin?

A variant Hb occurs when the structure of the globin chain is altered. This is the case with sickle haemoglobin (Hb S) and Hb C.

Sickle Haemoglobin (Hb S)

Red blood cells containing only sickle Hb are less flexible than normal and can sometimes pile together and block small blood vessels.

This occurs if a person inherits sickle haemoglobin from both parents, this condition is called **sickle cell anaemia** or homozygous haemoglobin S (Hb SS).

This can also occur if a person inherits sickle Hb from one parent and another variant haemoglobin or thalassaemia from the other parent.

Sickle cell trait (heterozygous haemoglobin S) occurs when a person inherits the normal haemoglobin from one parent and the sickle haemoglobin from the other (Hb AS).

Sickle haemoglobin occurs most frequently in people of African, Caribbean, Indian, Pakistani, Middle Eastern or Eastern Mediterranean ancestry.

Haemoglobin C (Hb C)

Heterozygous haemoglobin C (haemoglobin C trait) occurs when a person inherits the normal haemoglobin from one parent and the haemoglobin C from the other (Hb AC).

When a person inherits the haemoglobin C from both parents, they are said to have homozygous haemoglobin C (Hb CC).

Haemoglobin C occurs most frequently in people from Africa, particularly West Africa. There is also a significant incidence in North Africa, the Caribbean and southern Europe.

What is Sickle Cell /Hb C Disease?

Sickle cell / Hb C (Hb SC) disease occurs when a person inherits a haemoglobin S gene from one parent **and** a Hb C gene from the other parent.

If a person is born with sickle cell/Hb C disease, they will have it all their life. It is **not** contagious or infectious.

How Does it Present?

Because of their rigid shape, sickled red blood cells can't squeeze through small blood vessels as easily as normal red cells. This can cause blockages of small blood vessels, which then stops the oxygen from getting through to where it is needed. This in turn can lead to severe pain and damage to organs.

Symptoms are rarely seen before of the age of six months. The main symptoms of a sickling disorder are episodes of anaemia, pain or infection and are called crises. These can be infrequent in Hb SC disease but when they do occur can be severe.

Anaemia is usually moderate but may worsen during infection, pregnancy or during a crisis.

Splenomegaly (enlarged spleen) occurs in the majority of patients.

Retinal disease and hip problems (aseptic necrosis) are common.

Pregnancy can be associated with serious complications for people with Hb SC disease.

What is the Treatment?

Sickle cell / Hb C disease is a serious condition and does require medical follow-up.

- Patients should try to maintain good general health and nutrition.
- Folic acid supplements may be suggested.
- Special care is needed when having an operation, needing an anaesthetic or at high altitudes.
- Infections must be treated promptly, with severe infections requiring hospitalisation.
- Minor pain crisis can be treated at home with pain killers but if severe may require a patient to be admitted to hospital.
- If the anaemia becomes severe during a crisis a blood transfusion may be needed.

How Will This Affect My Children?

Every child inherits haemoglobin genes from each parent. The type of haemoglobin gene inherited from the parent determines the type of haemoglobin the child will have.

A person with sickle cell / Hb C disease will pass on **either** a Hb S gene or a Hb C gene to each of their children.

When **both** parents carry a gene for a variant haemoglobin or thalassaemia, there is a chance that any child could have a severe form of variant haemoglobin or thalassaemia.

Why is Knowing About Hb SC Disease so Important?

Sickle cell/Hb C disease is a serious condition requiring significant medical intervention.

If your partner has a variant Hb or thalassaemia, future children are at risk of inheriting a serious sickling disorder, thalassaemia or variant Hb.

If you have sickle cell/Hb C disease it is important to have your partner tested for the presence of a variant haemoglobin or thalassaemia before you have children

Is There a Test for Hb SC Disease?

YES ! Blood tests can show whether an individual has sickle haemoglobin or any other variant Hb.

It is important to have your partner tested for a variant Hb or thalassaemia before you have children. Other members of your family who are of reproductive age should also be tested.

The documentation of any variant Hb is important as:

1. It avoids the need for repeat testing in the future.
2. With sickle Hb extra care needs to be taken with the administration of anaesthetics.
3. A variant haemoglobin is an inherited disorder which if passed on to a child by **both** parents could result in a severe condition.

However, if two people who are at risk of having a child with a serious sickling disorder do become parents, there are special prenatal tests that can be done to detect or rule out sickle cell disease in the fetus.

Early diagnosis of a possible severe sickling disorder is important so that treatment can prevent as many complications as possible.

Can Haemoglobin S or Thalassemia Be Prevented?

Sickling disorders cannot be prevented at this time, but a programme of health education, testing for the trait, genetic counselling and prenatal diagnosis can provide families with full medical information to help them have healthy children.

People who think they may have sickle haemoglobin or any other variant Hb should go to their family doctor for advice and testing.

Your family doctor can contact a Haematologist at the Community Laboratory or the Haematology Department at Canterbury Health Laboratories on (03) 364 0300 if further clinical input is required.

The Specialist Haematology Laboratory at Canterbury Health Laboratories provides a comprehensive diagnostic service in conjunction with the family doctor.

Contact for Canterbury Health Laboratories

Laboratory Services: Specialist Haematology Laboratory, Canterbury Health Laboratories.
Address: 524 Hagley Ave, Christchurch 8011.
Phone: (03) 364 0300.

On behalf of the Clinical Haematology Team at Christchurch Hospital