

# Screening Information on Sickle Cell & Thalassaemia



## **What are Sickle Cell Disorders & Thalassaemia Major?**

They are both inherited conditions of the blood, which affect haemoglobin, the oxygen-carrying substance which gives blood its red colour.

Children with Sickle Cell Disorder can have attacks of severe pain (crisis) or sudden life-threatening infections.

Children with Thalassaemia Major are severely anaemic and need blood transfusions every four weeks and regular injections for life.

## **What are Sickle Cell & Thalassaemia Carriers (trait)?**

Both Sickle Cell and Thalassaemia disorders exist in two forms; one is the disorder form and the other is the carrier form (trait). People who are carriers are usually healthy people.

## **Does it affect me?**

People who are African, Caribbean, Asian, Middle-Eastern and Mediterranean origins are most 'at risk' of Sickle Cell and Thalassaemia.

About 1 in 4 Africans and 1 in 8 African-Caribbean's carry Sickle Cell trait. The incidence of Thalassaemia trait also varies from 1 in 7 to 1 in 30 people who originate from Asian, Mediterranean and Middle Eastern countries. Caucasian people can also be affected, but this is less common.

## **Nobody in my family has Sickle Cell or Thalassaemia, why should I have a blood test?**

Your parents or relatives may be healthy carriers, as this does not affect their health and well being they may be unaware of their carrier status.

## **How do I find out if I am a carrier?**

Your GP can arrange a simple blood test for free of charge.

## **Does my doctor recommend testing?**

Yes, especially if you are thinking of having a baby or if you are already pregnant. Please ask your midwife to arrange this test.

## **Do you suggest partner and family testing?**

Yes, if you are planning to have a baby it is advisable to find out whether your partner is a carrier or not.

If you are a carrier there is a chance that your brothers and sisters may be carriers as well.

Please contact your G.P Surgery to arrange a blood test.

## **What will happen once I have had my blood test?**

The professional who requested the blood test will inform you of your result. You will receive a letter, information and haemoglobinopathy card from the Sickle Cell and Thalassaemia Service. You can then book a counselling session at your convenience.

Bristol OSCAR provides genetic counselling at the Sickle Cell and Thalassaemia Centre which enables individuals the opportunity to discuss their results and any implications it may have on the family.

## **If I am pregnant will my baby be tested?**

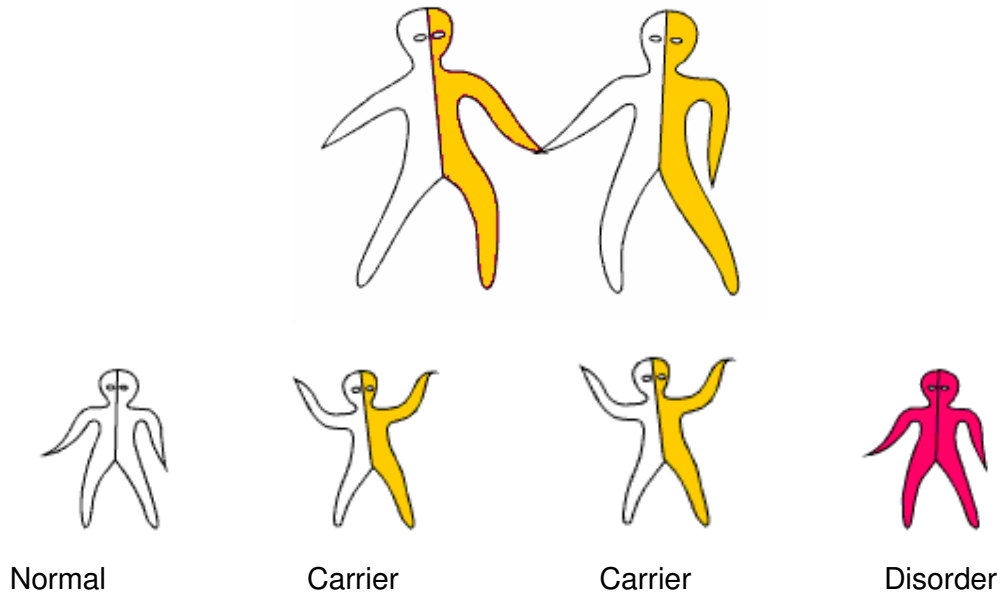
You can be offered prenatal testing if your baby is at risk of developing a disorder.

You will be offered testing for your baby when the midwife carries out the dried blood spot sample 5 – 8 days after birth.

## How do you get Sickle Cell Disorders & Thalassaemia Major?

If both parents are healthy carriers (trait), there is a 1 in 4 chance (25%) that in each pregnancy the child will have Sickle Cell Disorder or Thalassaemia Major.

The diagram below shows how a child could inherit a **Disorder** if both parents carry a **Trait**.



**Normal** means that this individual has inherited normal genes in the **Haemoglobin** and this is written as **Hb AA**

**Carrier** means that this individual has inherited an abnormal gene in the **Haemoglobin** and this is written as **HbA?** (? Is referring to the abnormal gene e.g. S, D, E, Beta Thalassaemia and many others)

**Carrier or Trait** means the same. **HbAS** is an individual with Sickle Cell Trait.

**Disorder** means that the individual has inherited two abnormal genes in the **Haemoglobin** and this is written as **Hb??** (? Is referring to the abnormal genes e.g. S, D, E, Beta Thalassaemia and many others)

**HbSS (Sickle Cell Anaemia)** is an individual with one of the Sickle Cell Disorders.

### Who can I contact for further information?

#### **Bristol O.S.C.A.R**

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