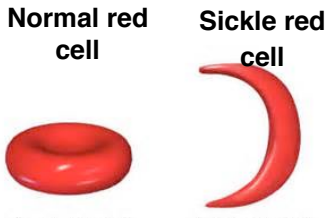
SCU
SICKLE CELL UNIT

What is Sickle Cell Disease?

Sickle cell disease (SCD) is an hereditary disease of the red blood cells. The main job of red blood cells in the body is to carry oxygen. The protein in the red cell that carries oxygen is called haemoglobin. Persons with SCD inherit abnormal haemoglobin genes from each parent which results in the "sickle change", the production of abnormal red blood cells (**sickle cells**). Normal red cells are soft and round and can travel through the circulation without any problems. **Sickle cells** are hard, sticky and curved in shape (like a banana). They do not live long and are destroyed rapidly in the circulation. Because they are hard and sticky, they may also get stuck and cause blockage of blood vessels. When this happens, some parts of the body will not get enough oxygen. This is what causes many of the problems such as anaemia (weak blood), jaundice (yellow eyes), gallstones, bone pain, pneumonia, leg ulcers (sores), increased susceptibility to infection and strokes that can occur in persons with SCD.

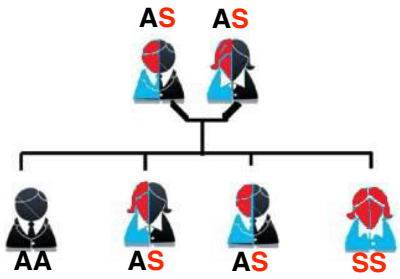


ARE THERE DIFFERENT TYPES OF SCD?

There are many forms of the disease (genotypes). If one inherits the abnormal sickle haemoglobin (Hb S) gene from both parents, the affected person has the most common, as well as one of the most severe forms of the disease (Hb SS disease). If one inherits the sickle gene as well as the gene for another abnormal haemoglobin (eg Hb C) or the gene that does not allow the body to make enough haemoglobin (thalassemia), other forms of SCD may occur such as Hb SC disease and sickle thalassemia (Hb ST). Hb SS disease is often referred to as "full blown" which can lead to the misconception that other forms of the disease are not important. On the contrary, whereas Hb SS disease is a severe form, it is NOT the only severe form of the disease, and even so called MILD forms of the disease can have many problems. In Jamaica, 1 in 150 births are affected by some form of the disease each year.

WHAT IS SICKLE CELL TRAIT?

Persons who have one normal haemglobin (Hb A) gene and one sickle haemoglobin (Hb S) gene are said to be carriers of the disease. They have the sickle cell trait (Hb AS). Other carriers include haemoglobin C trait (Hb AC) and thalassemia trait (Hb AT). Carriers usually have no signs or symptoms and so are often unaware of their status. 1 in 10 persons in Jamaica are carriers of the sickle gene and another 1 in 15 persons carries another gene which puts them at risk of having a child with SCD.



If both parents are carriers of the sickle trait they have a 25% chance of having a child with SCD for EACH pregnancy. *See the diagram above.*

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COMMON PROBLEMS SEEN IN SICKLE CELL DISEASE

Sickle Cell Disease can affect most if not all of the organs in the body. Some of the common problems include:

- **ANAEMIA** or "weak blood" is due to a lower number of red blood cells in the body. This results in a low blood count.
- **INFECTION** may occur due to a weak immune system and the spleen not working properly. The spleen normally helps to filter germs out of the blood to help prevent infection. Sickle red cells can get trapped in the spleen causing damage and preventing it from working normally. Children are at highest risk of getting serious infections. Special vaccines and antibiotics are given to help prevent infection in children with SCD.
- **PAIN** occurs when sickle red cells get trapped and stuck in the small blood vessels. This affects the circulation of blood and prevents oxygen from flowing to the area where blood cells are trapped. Pain can occur in any part of the body in persons with SCD.
- **ACUTE CHEST SYNDROME** is similar to pneumonia in unaffected persons. However, because it occurs not only from infection, but from other causes such as trapping of sickle red cells in the lung (sequestration) and blockage of small blood vessels in the lung preventing oxygen from getting to the lung, it is called acute chest syndrome.
- **GALLSTONES** occur frequently in persons with SCD because the red blood cells break down much faster than normal red blood cells. The red cells release bilirubin, a substance, which collects in the gall bladder and forms a sludge or gallstones.
- **JAUNDICE** or "yellow eyes" also occurs as a result of increased bilirubin from rapid break down of sickle red cells.
- **STROKE** occurs when the blood does not circulate properly to the brain because of sickle red cells blocking blood vessels in the brain. Children with SCD can get strokes.
- **DECREASED URINE CONCENTRATION** occurs because of small amounts of damage to the kidney. Patients need to drink more fluids and often require more frequent trips to the bathroom as they pass urine more frequntly.
- **PRIAPISM** is a painful, purposeless erection due to trapped sickle cells in the penis. If untreated and persisting for long periods it can lead to erectile dysfunction.
- **LEG ULCERS** or sores often occur around the ankles in persons with SCD. The skin around the ankles gets weak because of poor circulation and can break down easily on its own or from minor trauma such as insect bites.

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COMMON PROBLEMS SEEN IN SICKLE CELL DISEASE continued

- **SPLENIC SEQUESTRATION** is a sudden enlargement of the spleen due to trapped sickle red cells, with a drop in the blood count. It is most common in young children and can cause death. Parents / guardians can be taught how to check for an enlarged spleen so that they can seek medical attention for their child quickly.
- **SICKLE RETINOPATHY** (eye disease) occurs from damage to the back of the eyes (retina). It can lead to blindness. Persons with SCD should have their eyes checked each year, from age 12 years, to look for early signs of eye disease.

IMPORTANT FACTS ABOUT SCD

1. Sickle cell disease (SCD) is a genetic disease.
   - Persons with disease inherit the genes from BOTH parents.
   - It is NOT an infection. Problems of the disease such as leg ulcers (sores) and jaundice cannot be “caught”.
   - The risk is the same (1 in 4) for EACH pregnancy when both parents have a trait.
2. 15 % of Jamaicans carry genes that put them at risk for having a child with SCD.
   - 1 in 10 persons are carriers of the sickle gene.
   - A simple blood test can tell you whether you are at risk.
3. Babies can be tested for SCD from immediately after birth.
   - 1 in 150 babies born in Jamaica will have a form of SCD.
   - Children are at risk of dying early from complications of the disease.
   - Early diagnosis of disease allows for simple treatments to be started which can help prevent death in early childhood.
4. SCD is NOT a “black person” disease.
   - Whereas it is more common in persons of African descent, the disease also occurs in other races – Caucasians, Asians and people from the Mediterranean regions.
5. Persons with SCD do not all die young.
   - The lifespan of a person living with a severe form of SCD is, on average, ten years less than persons without disease. Simply put, the average age at which people in Jamaica with a severe form of SCD die is approximately 55 years.
   - Early diagnosis, being informed about their disease and good health care will help patients and their family to learn how to deal with their disease so that they can prevent complications or respond to them quickly when they occur.

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6. Children with SCD can learn like other children and SHOULD be encouraged to go to school as much as possible.
   - They may have to miss school often because of sickness. Parents and teachers should work together to ensure that work is sent home and where possible extra help should be given to these children to allow them to catch up with their school work. If you notice a change in your child's ability at school - speak to your doctor as it may be a sign of a mild stroke.
   - They should be encouraged to participate in all school activities (including sports) to their **PERSONAL LIMIT**.
7. Adults with SCD often have difficulty maintaining a job, especially if they get sick often.
   - Employers should be informed about the disease, problems their employees may have and how best that they can support them.
   - Patients and employers need to work together to ensure that they remain productive members of the workforce.
8. Children with SCD can get strokes.
   - A simple ultrasound test is now available at the Sickle Cell Unit, which can help identify children at higher risk of having stroke.
   - There are treatments that can be started in children to lower their risk of having a stroke.
9. Women with SCD are NOT infertile.
   - They do, however, have an increased risk of complications during pregnancy and delivery.

IMPORTANT HEALTH TIPS FOR PERSONS WITH SCD

Here are some tips to help you / your relative deal with sickle cell disease to prevent complications and know how to respond to them when they occur:

1. Get informed about sickle cell disease.
2. Keep appointments with your doctor for regular check-ups.
3. Ensure that you receive all available immunizations to prevent serious infection.
4. Eat a balanced diet, with lots of fruits and vegetables.
5. Drink plenty of fluids, especially water.
6. Avoid extreme cold, wet or heat.
7. Keep pain medication at home. Pain often starts during the night or early morning.
8. Avoid overexertion.
9. Exercise to your PERSONAL LIMIT.
10. Avoid stressful situations.