Sickle Cell Disease

An Education Book for Patients and Family

First published 2017

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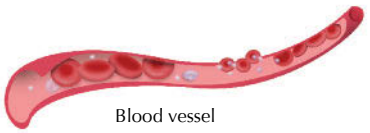
Contents

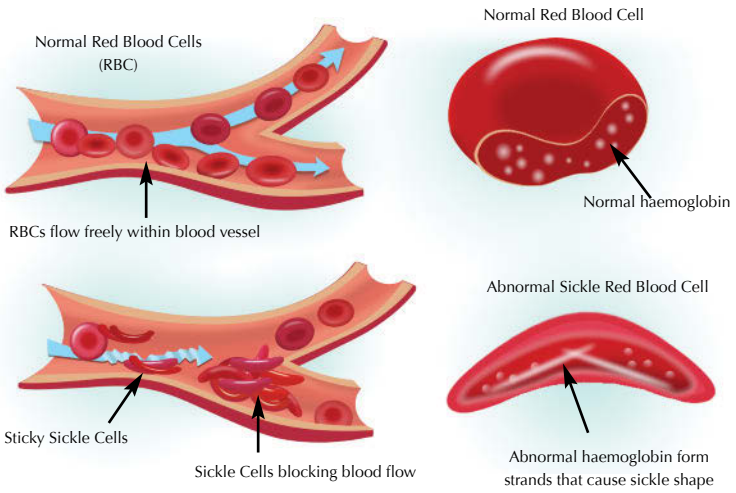
SICKLE CELL DISEASE

Sickle cell disease (SCD) is a disease of the blood. It is a disease of RED BLOOD CELLS. There are different cells in the blood:

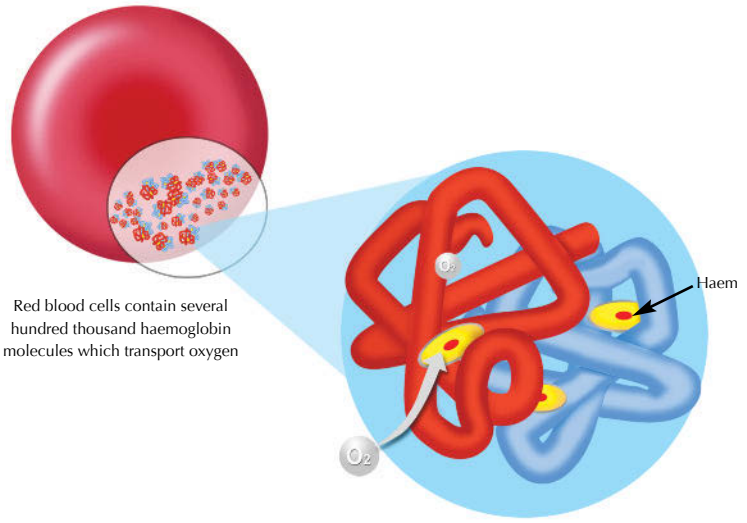
- Red blood cells which carry oxygen
- White blood cells which help fight infection
- Platelets which help stop bleeding if you get a cut



Normal red cells are round and soft; they pass easily through the circulation. Sickle red cells can change shape, to a sickle or banana shape; they are stiff and sticky and can block the circulation causing many problems.



Red blood cells contain a substance **HAEMOGLOBIN** which gives blood its colour and is responsible for carrying oxygen.

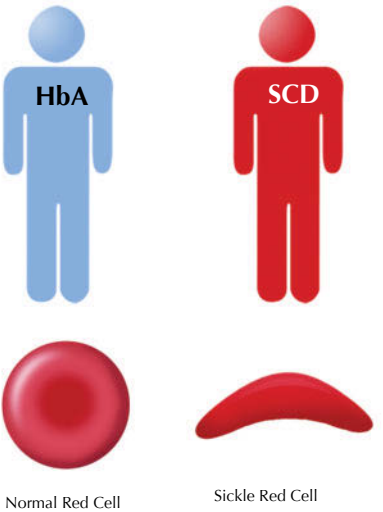


Red blood cells contain several hundred thousand haemoglobin molecules which transport oxygen

Oxygen binds to haem on the haemoglobin molecule

Persons with normal red cells have normal haemoglobin (HbA).

Persons with SCD have sickle haemoglobin (HbS) which is the reason sickle red cells can change their shape.



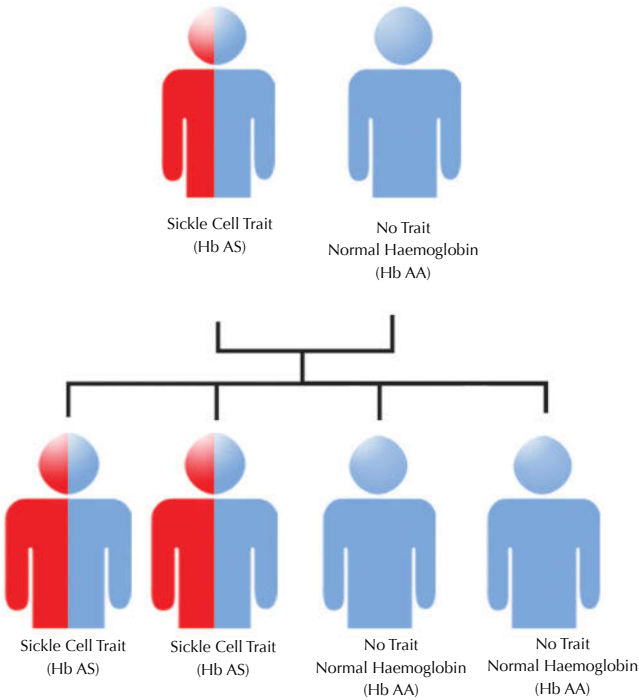
Normal Red Cell

Sickle Red Cell

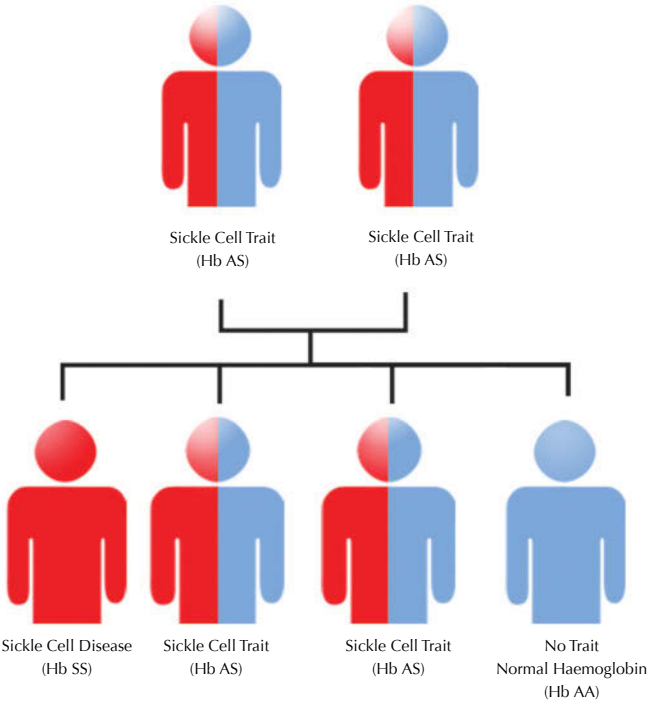
INHERITANCE

Persons inherit genes from their parents. If they inherit haemoglobin A genes from both parents they are Hb AA and have normal red blood cells.

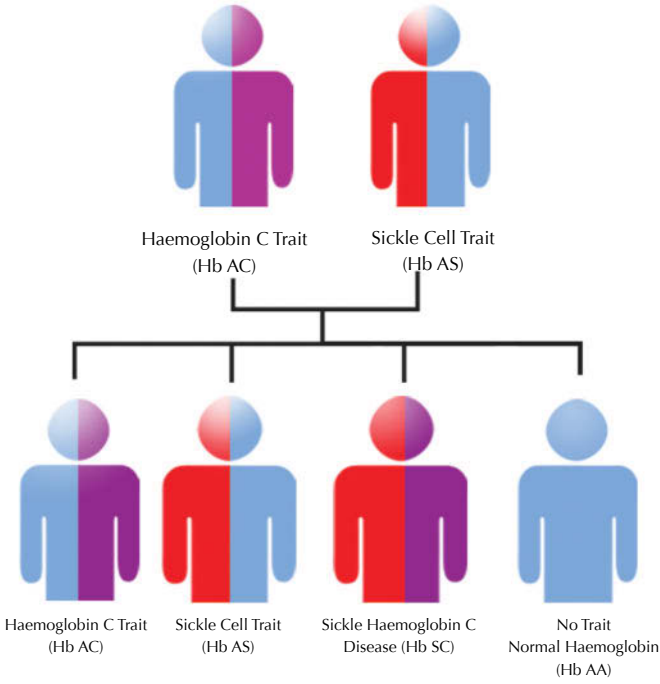
If they inherit a haemoglobin A gene from one parent and the haemoglobin S gene (sickle gene) from the other parent they are ***carriers*** of the disease and have ***sickle cell trait*** (Hb AS). Persons with sickle trait are usually well and unless they get tested do not know that they are carriers of the sickle gene. In Jamaica, 1 in 10 persons (10%) carry the sickle cell gene.

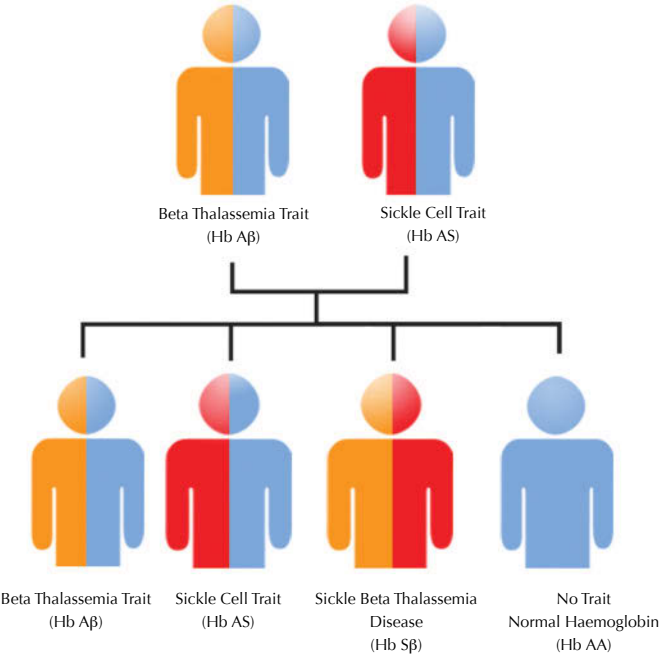


If a person inherits the sickle gene from both parents they will have the most common type of sickle cell disease – Hb SS.



There are also other types of SCD. These will occur if persons inherit the sickle gene from one parent and the gene for another abnormal haemoglobin (eg Hb C) **OR** the gene that prevents them from making normal haemoglobin properly (beta-thalassemia gene). This can cause other types of SCD such has Hb SC disease or Hb Sβ thalassemia.


Haemoglobin C Trait
(Hb AC)
Sickle Cell Trait
(Hb AS)
Haemoglobin C Trait
(Hb AC)
Sickle Cell Trait
(Hb AS)
Sickle Haemoglobin C
Disease (Hb SC)
No Trait
Normal Haemoglobin
(Hb AA)

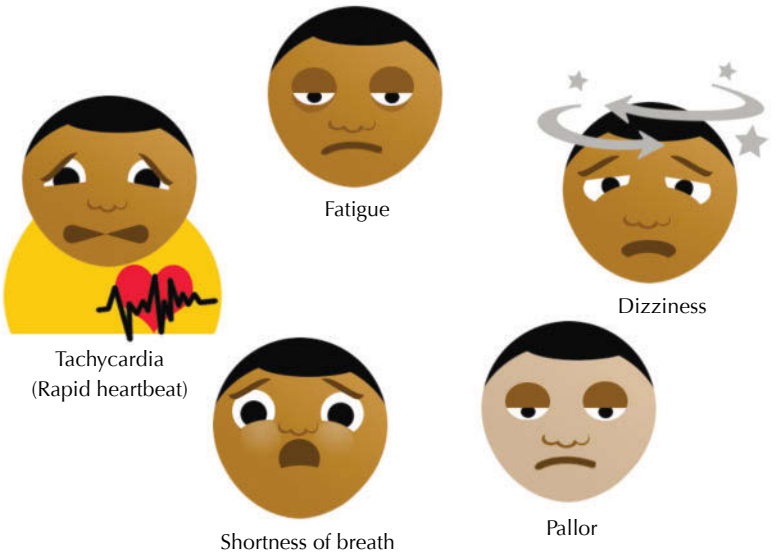

Beta Thalassemia Trait
(Hb Aβ)
Sickle Cell Trait
(Hb AS)
Beta Thalassemia Trait
(Hb Aβ)
Sickle Cell Trait
(Hb AS)
Sickle Beta Thalassemia
Disease
(Hb Sβ)
No Trait
Normal Haemoglobin
(Hb AA)

ANAEMIA

Persons with SCD have **ANAEMIA ("weak blood")**. This occurs because sickle red cells break-down ("die-off") quickly in the circulation **(red cell haemolysis)**; normal red cells live for ~ 120 days while sickle red cells live for only ~ 10 – 21 days. The body cannot make new red cells fast enough to replace them and persons therefore have a *low blood count* (anaemia). Anaemia can cause problems such as pale skin (pallor), becoming tired easily (fatigue) and shortness of breath on exertion.

Anaemia in SCD is not due to lack of iron. Iron tonics will not help "weak blood" in SCD; they can actually trigger problems. Persons with SCD should **NOT** take iron tonics unless advised to do so by a doctor.

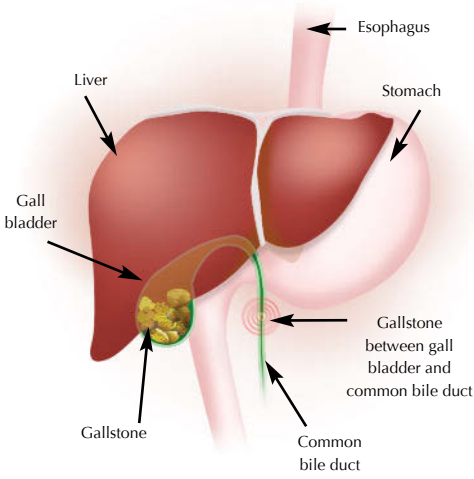
Consequences of Anaemia



JAUNDICE AND GALLSTONES

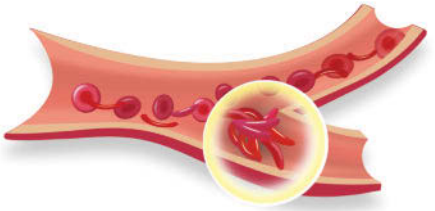
Persons with SCD can have **JAUNDICE ("yellow eyes")**. When sickle red blood cells break-down quickly they release a waste product **(bilirubin)** which can build up in the circulation. Excess bilirubin may cause yellow eyes and dark urine.

Excess bilirubin excretion can also lead to **gallstones**. Over 50 percent of persons with SCD will have gallstones. In most persons gallstones cause no problems. In other persons, it may cause problems such as **chronic upper abdominal pain (belly pains), nausea and fullness after meals**. Eating greasy foods can trigger these symptoms. If symptoms occur frequently or are severe, they may require surgery.



PAINS

Persons with SCD often get **pains** in the bones and around the joints. This occurs because sickle red cells clump / stick together and block the circulation ***(vaso-occlusion)***. This prevents oxygen getting to the tissues and important organs. This can cause pain and sometimes swelling of bones.



- Young children and babies can get **DACTYLITIS (Hand-Foot syndrome**) – pain and swelling of the small bones in the hands and feet due to vaso-occlusion.

Pains can be triggered by

- Changes in temperature – e.g. cold weather, swimming, rain
- Dehydration – e.g. when playing games or sports
- Stress

Persons should try and avoid pain triggers. They should also always keep pain medication at home to use if / when pain starts.

Remember:

- Start pain medication early at the first sign of pains
- Use pain medications appropriate to the level of pain
- Keep warm and drink extra fluids (water or juice)

If pain medications at home are not helping and the pain is persisting or getting worse they should visit a doctor or clinic or go to the nearest hospital for treatment.

INFECTION

Persons with SCD, **especially children under age 4 years**, are at risk of serious infections with certain bacteria (germs) such as *Streptococcus Pneumoniae* (Pneumococcus), *Haemophilus Influenzae* type B (HiB) and Salmonella. These bacteria can cause infections in the blood (sepsis), in the lining of the brain (meningitis), in the lungs (acute chest syndrome) and in bone (osteomyelitis).

High fever (T more than 38º C or 100º F) may be due to a serious infection.

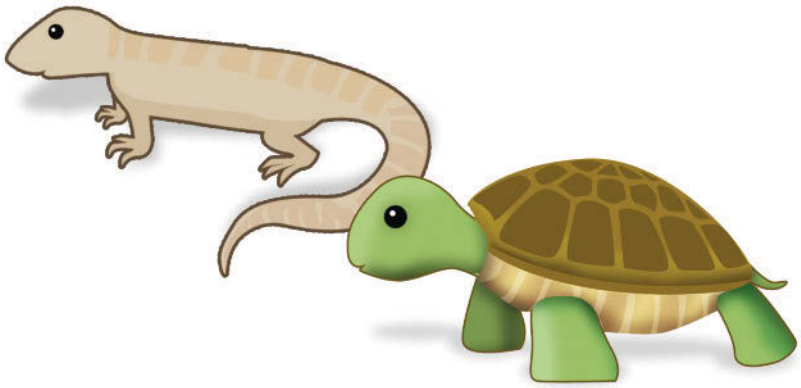
- Persons should learn how to use a thermometer to check for fever.
- Persons with SCD should see a doctor if they have a high fever.

Some children with SCD will need antibiotics such as **Penicillin** during the early years to prevent infections with Pneumococcus.

ALL children and adults with SCD will also need special vaccines to help prevent against infections with Pneumococcus and HiB

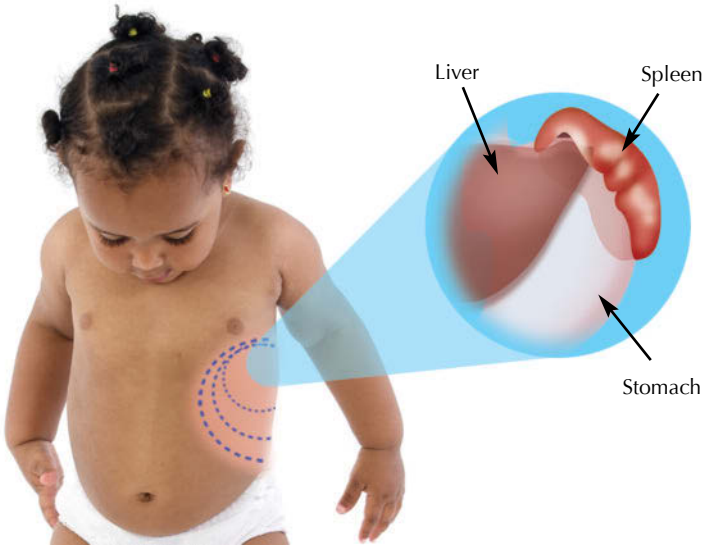
Persons with SCD should practice good food hygiene and ensure that meats are cooked properly. They should NEVER eat foods that contain raw eggs (e.g. egg nog, home-made cake batter) because they can catch germs (such as Salmonella) from these foods.

They should stay away from lizards and turtles which also carry these germs which can cause serious infections.



ACUTE SPLENIC SEQUESTRATION

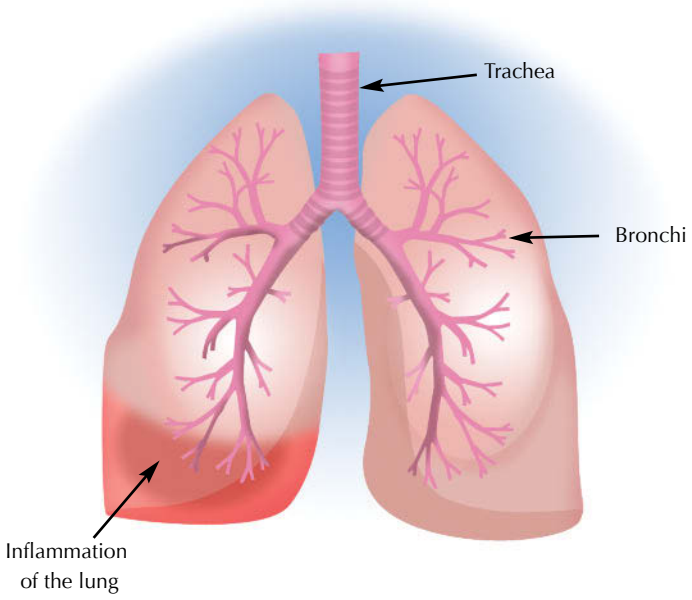
The spleen is an organ in the body. It acts as a filter to help clean the blood; it removes old and damaged blood cells and germs. Sometimes, mostly in young children & babies with SCD, the spleen will trap sickle red blood cells and swell suddenly **(acute splenic sequestration)**. This is a **MEDICAL EMERGENCY** as the blood can become very weak and children can die within hours. Parents should feel their babies / young child's abdomen at least once a day to check for the spleen. If the spleen is swollen, they should take their child to the nearest hospital **IMMEDIATELY.**



ACUTE CHEST SYNDROME

Acute Chest Syndrome (ACS) is similar to pneumonia. It may be due to a lung infection or effects of SCD on lungs. Persons with an ACS may complain of:

- Fever
- Chest Pain
- Shortness of breath
- Cough



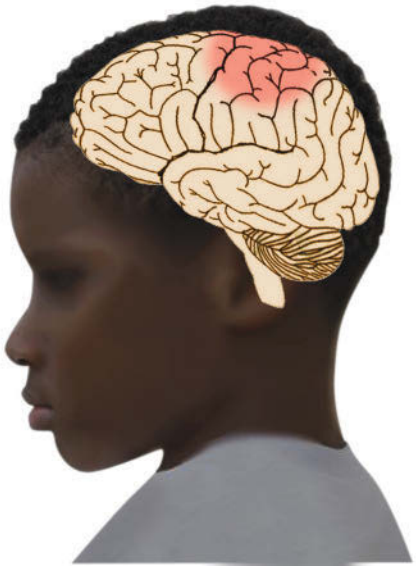
Persons with ACS often need admission to hospital for proper treatment.

STROKE

Stroke is a severe complication of SCD. It can occur in children and is partly due to blockage of circulation in the brain by sickled cells. This prevents oxygen supply to the brain. Signs and symptoms of stroke include:

- Weakness or paralysis
- Slurred speech
- Fits or seizures
- Unsteady walk

If you suspect your child may be having a stroke you should seek medical attention **IMMEDIATELY** so that your child can get **urgent treatment.**



Children who have had a stroke are at high risk for a recurrent (repeat) stroke. There is treatment that can be given to try and prevent recurrent stroke in children.

HOW CAN I KNOW IF MY CHILD IS AT RISK FOR STROKE?

Trans-Cranial Doppler (TCD) Ultrasound is a simple ultrasound screening test which can be used to determine your child's risk of having a stroke. It is recommended that children with severe forms of SCD from age 2 – 16 years have this test done once per year to determine their risk for stroke.

If your child is at risk of having a stroke there is treatment that can be started to try and prevent a first stroke.

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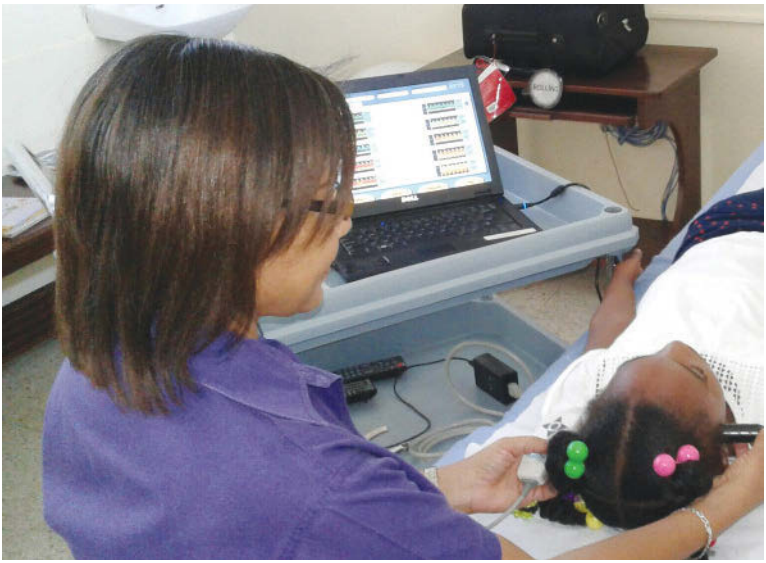
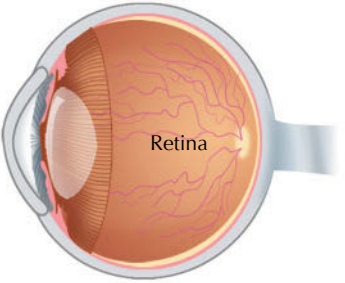


SICKLE RETINOPATHY

Sickle cell disease can also lead to damage to the eyes (retinopathy). This can affect vision and even cause blindness. Sickle retinopathy is more common in persons with Hb SC disease.



ALL persons with SCD, from **age 11 years** should have their eyes checked by an eye specialist once per year to look for signs of retinopathy.

PRIAPISM

Priapism is a persistent, unwanted, painful erection of the penis. It usually occurs in young men but can occur even in childhood. Priapism episodes may be

- Stuttering: these episodes last less than 4 hours
- Prolonged / Major: these episodes last 4 hours or longer

To try and help your priapism to go away, you should:

- Take pain killers
- Drink extra fluids
- Empty the bladder
- Take a warm shower
- Do some light exercise e.g. walking around

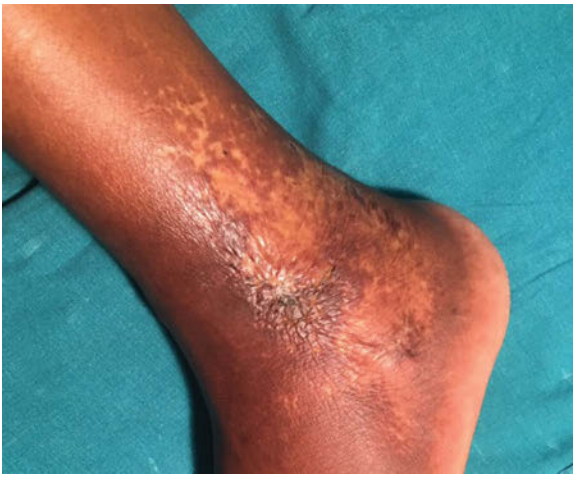
If these measures are not working, you should go to the hospital or doctor. You must try and get to the doctor/ hospital within **4 hours from the start** of the episode for treatment. Episodes of priapism that last longer than 4 hours can lead to problems with sexual function later on.

LEG ULCERS

Leg ulcers (sores) can occur in persons with SCD; often around the ankles. They can occur spontaneously (for no reason), or after minor trauma (such as an insect bite or cut).

Persons with SCD should practice good skin care from an early age. They should:

- Avoid scratches / bites / trauma to the ankles
- Moisturize / lotion their skin daily
- Wear proper fitting shoes / sandals
- Clean and dress cuts / scratches as soon as they occur
- Seek medical attention if any ulcers develop and are not healing properly



KIDNEY PROBLEMS

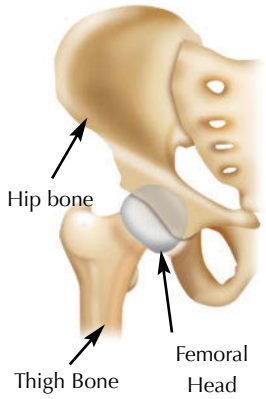
Sickle cell disease can cause damage to the kidneys. One of the problems that can occur from an early age is the inability to concentrate urine properly. As such, persons with SCD make plenty urine. This can lead to problems with bedwetting (enuresis) in older children and even some adults. If you or your child has problems with bedwetting, the following measures may help:

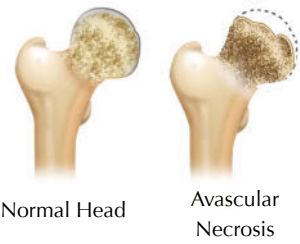
- Limit fluid intake 1 -2 hours before bedtime
- Empty the bladder just before going to bed
- Have someone wake you or set an alarm clock to wake you during the night to empty your bladder (may need to be done more than one time during the night)

Persons with SCD may also pass blood in the urine. If you notice blood in the urine you should go to the doctor immediately.

AVASCULAR NECROSIS OF THE FEMORAL HEAD

The hip joint is an important joint in the body as it bears most of our body weight and allows for activities such as walking, running and jumping. It is a ball and socket joint formed between the thigh bone (femur) and hip bone. The ball is the femoral head. A single artery supplies blood to the femoral head. If blockage occurs in this artery from sickle red cells, it will affect oxygen delivery to the femoral head. This may lead to death of the bone tissues, destruction and collapse of the femoral head. This causes **persistent hip pain accompanied by a limp**.





Avascular necrosis of the femoral head (AVNFH) can occur in children and adults with SCD. Persons with AVNFH will need to use crutches for a very long time. Keeping weight off the femoral head in the early stages will help it heal more quickly. **Using crutches can be extremely challenging for children and teens, but MUST be encouraged.** Surgery is sometimes required if healing does not occur.

PUBERTY

Children with SCD often do not gain plenty weight and may mature later than their friends **(delayed puberty)**. This is partly due to their chronic anaemia. Delay in maturation (sexual development) may cause body image and self-esteem issues in some teens. They should be reassured that they will eventually go into puberty and catch up with their peers. If puberty is significantly delayed they can get treatment to help speed it up.

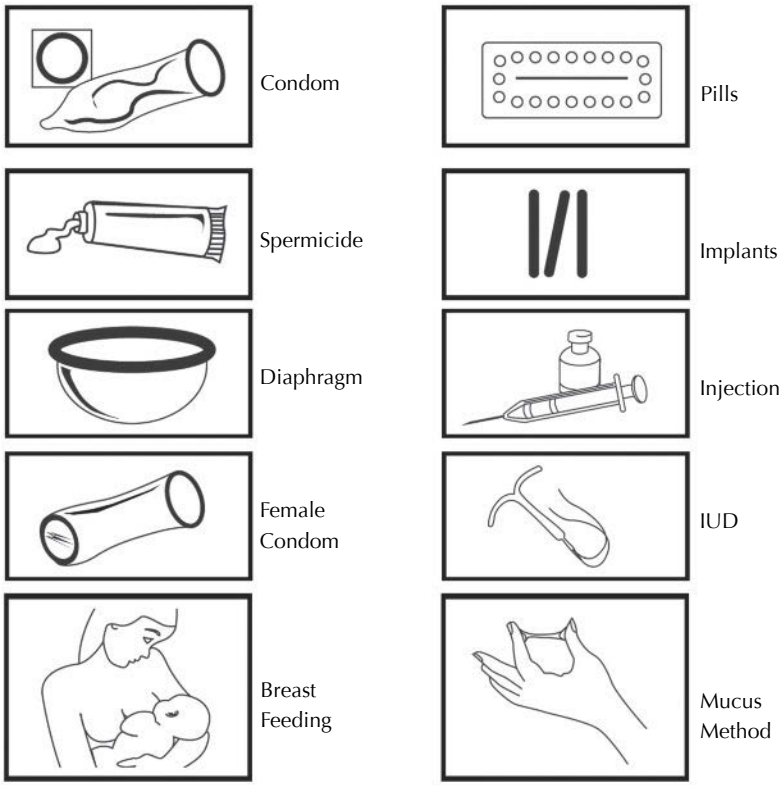
PREGNANCY AND FAMILY PLANNING

Persons with SCD can have children of their own. Pregnancy in women with SCD is however **HIGH-RISK**; women with SCD can have serious problems during and even after pregnancy. These can affect both their and their baby's health. The risk of maternal death around the time of delivery as well as the chance of having a stillbirth is high in women with SCD. They should ensure as much as possible that pregnancies are planned and that during their pregnancy they attend a high-risk clinic.

To prevent pregnancy, persons with SCD should use family planning if they are sexually active. All types of family planning can be used in women and men with SCD.

Persons should also make informed decisions about family planning with their partners. They should ensure that their partners are tested so that they know the risk of having a child themselves with SCD.

Family Planning

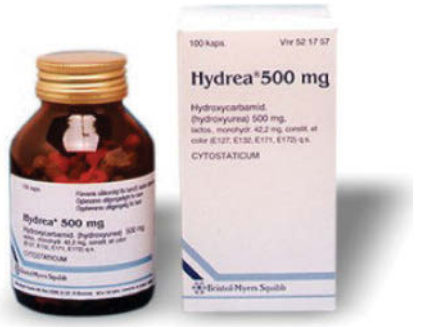


HYDROXYUREA

Hydroxyurea (HU) is a drug approved for the treatment of sickle cell disease. HU has been shown to reduce the frequency of many complications such as:

- Painful crisis
- Acute Chest Syndrome
- The need for transfusion and hospitalizations

HU works by increasing the production of **fetal haemoglobin (Hb F)** in the body. Fetal haemoglobin is the main haemoglobin found in babies at birth. However, soon after birth, the body stops producing Hb F and makes Hb S instead. Hb F helps to protect against the complications of SCD. Persons who take HU regularly have higher Hb F levels in their red blood cells over time. This may protect them from some of the complications of the disease.



Hydroxyurea can be used to treat both adults and children with SCD. If you think that you or your child may benefit from HU, speak with your doctor for further information.

DIET

Persons with SCD do not need a special diet. They should eat a healthy, age appropriate diet at all times which contains lots of fruit and vegetables.

They should **NOT** take supplements that contain iron unless instructed to do so by a doctor. Iron will not improve anaemia (weak blood) in SCD as persons with SCD are not normally iron-deficient (lacking iron in the body).

ORAL HEALTH

Good dental hygiene is important for everyone, even persons living with SCD. They too can have problems such as bad breath, cavities (dental caries), toothache and gum disease.

As part of keeping healthy, persons with SCD should visit their dentist at least once per year for check-ups. Having SCD does not prevent your dentist from doing any dental procedure on you.

Dental hygiene tips:

- Brush teeth at least twice daily
- Use fluoridated toothpaste from age 2 years and older
- Floss your teeth daily
- Clean your tongue
- Limit sugary and acid containing foods & drinks
- Visit your dentist regularly

LIVING WITH SICKLE CELL DISEASE

Persons with SCD can lead fairly normal lives.

School years

Children with SCD must go to school and should be allowed to participate in all school activities to their personal limit. Frequent periods of sickness can affect school attendance and learning. Parents have a responsibility towards their children and must work together with teachers to ensure that children get the most of their education years.

Routine health checks

Trips to the doctor, especially when one feels well, can be time-consuming and bothersome. After all, it means time missed from work or school. These visits however, especially when you have a chronic disease such as SCD, **ARE EXTREMELY IMPORTANT**. They allow for disease education, time to interact with your doctor and nurse and also for SCREENING for disease complications.

As persons get older, SCD can damage organs such as kidneys, lungs and heart. Well health checks will help doctors pick up problems in these organs early so that appropriate treatment can be started to prevent progression (worsening) of damage.

General tips for living with SCD include:

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

THE SICKLE CELL UNIT

The Sickle Cell Unit is a research institution located on the Mona Campus of The University of the West Indies (UWI). The unit seeks to improve the lives of individuals with Sickle Cell Disease through Biomedical Research, Education and Clinical Care.

Opening hours: Monday – Friday, 8.30 a.m. – 4.30 p.m. (Registration until noon)

Exceptions: Public holidays, UWI closure and previously publicized special occasions

Current clinical services:

1. Health maintenance for persons with SCD
2. Disease / genetic counseling
3. Day treatment ward
4. Social work
5. Diagnostic and Laboratory services
    a. Including Trans-Cranial Doppler Ultrasound
6. Specialist clinics
    a. Orthopaedic
    b. Men's Health

SUPPORT SERVICES

1. **NATIONAL HEALTH FUND**

   Sickle Cell Disease is now covered under the National Health Fund in Jamaica. As such, persons with SCD can access some medications at reduced costs. Persons should ensure that they sign up for their NHF card as soon as possible. Without an NHF card they will not be able to access benefits.

2. **SICKLE CELL SUPPORT FOUNDATION OF JAMAICA**

   The Sickle Cell Support Foundation of Jamaica is a non-profit, voluntary organization which seeks to raise public awareness of sickle cell disorders and provide support to patients and their families.
   Contact no: (876) – 927 – 0276
   Website: www.sicklecellfoundationja.org

3. **SOCIAL SERVICES**

   The Sickle Cell Unit has an in-house social worker who is available to assist with various needs. Let your doctor or nurse know if you are interested in speaking with our social worker.

   Social welfare progammes, such as PATH may be available to qualified persons.

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 (sore) 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 decrease the risk of 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, people with SCD will live, on average, to their mid-50s.

   - 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.

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 teacher should work together to ensure that work is sent home and when possible extra help is given to these children to allow them to catch up with their school work.

- 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.

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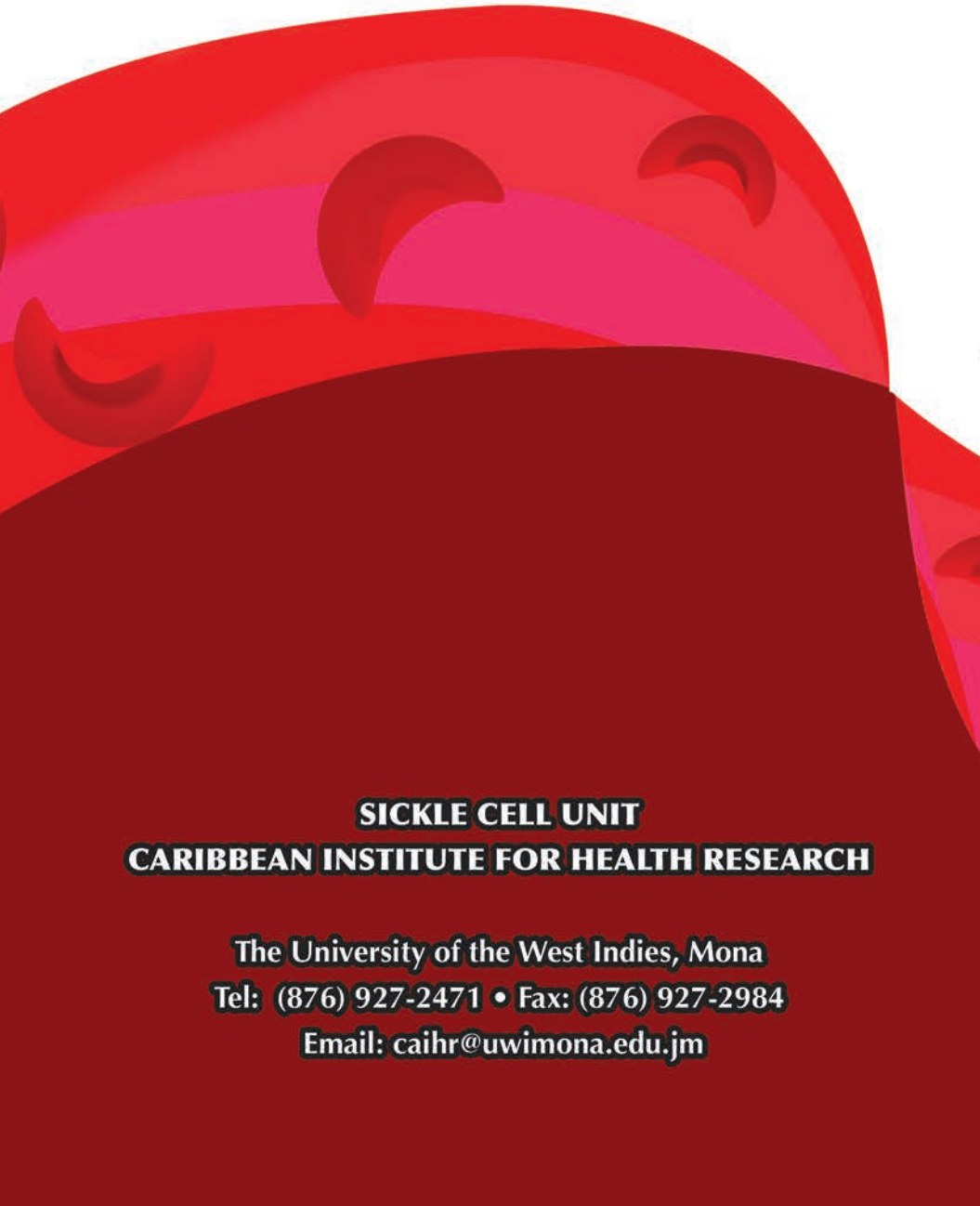

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