Living with Sickle Cell Disease: 
an information leaflet for patients 
(and for other members of their family!)

This leaflet has been written by two doctors who have specialised in treating 
patients with blood disorders. It explains Sickle Cell Disease: what causes the 
condition, the common problems that occur in patients and available treatment.

We hope this information will help patients and their families understand and 
therefore, live with Sickle Cell better.

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Living with Sickle Cell Disease

In many parts of the world, sickle cell disease is the most common genetic condition. It results from inheriting two sickle haemoglobin genes or one sickle haemoglobin gene in combination with another abnormal haemoglobin gene. One gene is passed on to you from each of your parents. In other words, you are born with the condition as a result of the genes you have. Sickle cell disease comprises of a number of different genotypes. Haemoglobin SS also known as sickle cell anaemia is the most common genotype. Other forms of sickle cell disease include haemoglobin SC and haemoglobin S beta thalassaemia.

The picture below illustrates how Sickle Cell Anaemia (SS) can be inherited from two parents who themselves have inherited one normal adult haemoglobin (A) and one sickle haemoglobin (S).

In the example shown, there is a 1 in 4 or 25% chance for EACH child born into this family to inherit two sickle haemoglobin genes (SS) resulting in sickle cell anaemia. There is also a 1 in 4 or 25% chance for EACH child to inherit two normal haemoglobin genes (AA) and a 1 in 2 or 50% chance for EACH child to inherit one normal gene (A) and one sickle haemoglobin gene (AS). Children with genotype AS are carriers of the sickle cell gene which means they can pass it on to their children if they marry a partner who is also a carrier. Carriers of the sickle cell gene are healthy individuals with normal blood counts and do not suffer any of the complications of sickle cell disease.

More on Sickle Cell...

Sickle cell is a disorder of haemoglobin. Haemoglobin is a substance in red blood cells that carries oxygen around the body. It gives blood the red colour. In people with sickle cell disease, the haemoglobin is abnormal and behaves differently from that in people without this condition.

With the abnormal haemoglobin, red blood cells change their shape. Normally red blood cells are flexible and doughnut shaped. However, in sickle cell they become curved in the shape of a sickle. Sickle cells are rigid and sticky.
Sickle cells get stuck in blood vessels and block blood flow. This results in reduced oxygen supply to parts of the body, and causes tissue damage and pain. When this happens repeatedly, long-term damage happens.

Sickle shaped cells are also destroyed more quickly than normal red cells. Sickle cells are destroyed in less than 20 days rather than the 120 days for normal red blood cells. This leads to anaemia (low number of red blood cells) and jaundice (yellowness of the white part of the eyes).

**Taking care of yourself...**

Medical treatment continues to improve. Many people live fruitful and productive lives well into adulthood.

It is important for people with sickle cell disease to learn about various aspects of their condition and know how to manage their illness. It is a lifelong disorder and there are a number of practical measures that will help you take care of yourself and enable you cope with your condition. Understanding how to manage your condition will help you take charge of your health and avoid crises. Simple measures go a long way.

Also, it is important to have access to medical care from professionals who are familiar with looking after patients with your condition. You will need regular outpatient follow-up in a clinic to help you stay well. Your doctor will be able to monitor your health, detect any complications and recommend appropriate treatment early. Treatment is available to help reduce the frequency of painful crises and prevent infections, which will hopefully reduce the need for admission to hospital.
What you can do to keep well

Factors that can bring on sickle cell crises or make them worse include dehydration (not having enough water or fluids in your body), stress (including school exams!), infections (like malaria which is common in Nigeria and caused by bites from mosquitoes carrying the malaria parasite or pneumonia which is a chest infection caused by bacteria), change in the weather (like the rainy season or harmattan which make the weather colder).

So, it is VERY important to:

1. Ensure you are drink plenty of fluids regularly throughout the day. The best fluid to drink is water but others such as juices and soft drinks are also acceptable. It is advisable to avoid drinks that may dehydrate you such as tea, coffee and alcohol.
2. Try not to get too cold or too hot. Avoid swimming in cold water.
3. Eat a balanced diet.
4. Have adequate rest and sleep. Don’t overdo it!
5. Avoid unnecessary physical and emotional stress.
6. If you wish to exercise, do so in moderation. Listen to your body.
7. Do not smoke. Smoking damages the lungs and robs the body of oxygen.
8. Have regular check-ups.
9. Ask your doctor about immunizations you require.
10. Know when to seek urgent medical attention. Tell your mum and dad (or whoever is looking after you) when you really don’t feel very well despite your usual painkillers OR if you

And your medication...

1. Take folic acid daily. This vitamin helps make new red blood cells.
2. Take penicillin twice daily at a dose prescribed by your doctor. If you are allergic to penicillin, a different antibiotic is given. This is to help prevent severe infections that can occur.
3. You should take antimalarials regularly (Paludrine or Daraprim).
4. Pain medication - know about simple painkillers that you may use and carry your medication with you at all times so that you can start them early.
5. It is recommended that children and adults with sickle cell disease be given the pneumococcal vaccine to protect against some common severe infections.
Complications sometimes happen

Sickle cell disease can lead to a number of complications. Not everyone with sickle cell disease is affected in the same way. Some people have more problems than others. The way the illness affects you may change over time. Different parts of the body can be affected. Some complications are short-lived and others are long-term.

Sometimes you know you are unwell because of the symptoms you have, such as pain. However, some problems show no obvious symptoms, particularly in the initial stages. Problems can be detected early if you have regular check ups. You should have a check up with your doctor at least every six months even if you feel well.

**Cries**

Sickle cell crises refer to recurrent episodes of pain that come and go. It is the most common complication that occurs. The pain is often in the arms, legs, chest and back. Other parts of the body can be affected as well. Sometimes crises can be severe and you will need hospital admission. When the crisis is not so severe, it may be managed at home. When you are having a crisis, it is important to increase your fluid intake, take painkillers and rest. The crisis will usually resolve within a few days. If you do not get better or you have symptoms such as fever, difficulty in breathing, severe headache, leg or arm weakness, you should go and see your doctor immediately.

People tend to be well in between crises and are described as being in ‘steady state’ during this period.

Chest crisis (acute chest syndrome) is another type of crisis. This is a serious complication and will require emergency hospital admission. Fever, cough, chest pain and shortness of breath are some of the symptoms.

If you suffer with frequent crises, your doctor may recommend medication to help reduce the chances of you getting a crisis.

**Infections**

People with sickle cell disease tend to be more prone to certain infections. Infections in individuals with sickle cell disease can be very serious. This is because the spleen does not work properly. The spleen is an organ in the upper left hand side of the abdomen that helps people fight germs and infection. Infections can happen in the bone (osteomyelitis), lungs (pneumonia), blood (septicaemia), covering of the brain (meningitis), as well as in other areas of the body. People with sickle cell disease can still get malaria even when they take anti-malarial tablet regularly. Malaria can be
much worse if you have sickle cell. Infections can trigger off crises so it is important that you see your doctor if you suspect you have an infection, including malaria.

**Anaemia worsens**

People with sickle cell disease tend to have low numbers of red blood cells. A blood test will show you have a low haemoglobin level. Anaemia may cause you to feel tired easily. It is important for you to know what your usual haemoglobin level is when you are well. During a crisis or when you have another complication, the anaemia may get worse. At these times, a blood transfusion may become necessary.

**Splenic sequestration**

This complication is serious and can pose great danger. It is more common in children. Large amounts of blood get trapped in the spleen and there is rapid swelling of the spleen. It causes severe anaemia and the affected person may be very weak, pale and collapse.

It is important for parents to know how to look out for this problem. Parents should be taught how to feel the tummy for the spleen size by their doctor. Parents should check the spleen size regularly and whenever their child is unwell. They should seek medical attention immediately if they think the spleen has increased in size and their child is unwell. Prompt medical attention can be life saving. Ask your child’s doctor about this.

**Hand-foot syndrome**

This is painful swelling of the hands and feet. It is one of the earliest signs of sickle cell disease and occurs in babies and young children. It can be described as a crisis affecting the bones in the hands and feet.

**Joint damage**

The blood supply to bones around joints may become blocked. This results in damage known as avascular necrosis. It causes chronic pain and difficulty in walking and movement. The pain usually lasts longer than a few weeks and may be present everyday. The hip joint is most commonly affected, but the shoulder joints can be affected as well. You may need to take painkillers regularly. Surgery may be required. Hip replacements have been done successfully by teams of experienced orthopaedic surgeons who work closely with sickle cell specialists (haematologists) and physiotherapists to manage this complication safely. Let your doctor know if you have ongoing joint pain.
Kidney problems

Sickle cell disease can affect the kidneys. The kidneys are not able to help the body retain water very well and as a result many people find they pass a lot of urine, sometimes waking up at night to do so. As plenty of fluid is lost in the urine, it is very easy for people with sickle cell to become dehydrated. This is one of the reasons why you need to drink plenty fluids. Bedwetting (enuresis) sometimes happens as a consequence of the illness. If so, restricting fluid intake an hour before going to bed and parental waking may help.

Another type of problem in the kidney may result in passing blood in the urine. If you notice blood in your urine, you will need to see your doctor. Kidney damage may result in protein leaking from the kidneys. Monitoring your urine for protein and blood tests when you go for regular check ups will help keep an eye on your kidney function. If protein is present, treatment to help slow the progress of kidney damage may be given. Also if you have significant kidney damage, it is advisable for you not to use painkillers such as Ibuprofen, Nurofen, Diclofenac and Voltarol. These painkillers belong to a group of medication called non-steroidal anti-inflammatory drugs that can cause further damage to the kidneys.

Priapism

This happens in males. It is prolonged painful erection caused by sickle cells blocking blood flow out of the penis. Priapism commonly occurs at night or in the early hours of the morning, though it can happen at anytime. It may or may not be related to sexual activity. It tends to be a recurrent problem. People with repeated episodes are more likely to have a prolonged episode lasting for hours. Long lasting episodes may result in permanent damage leading to problems with sexual function and impotence.

Seek URGENT medical attention if you have:

- Severe pain not getting better with your usual painkillers
- Pain not typical of the pain you usually get
- Fever
- Chest pain
- Difficulty in breathing
- Unusual headache
- Abdominal swelling
- Sudden loss of vision
- Weakness or loss of feeling in arm or leg
- Priapism lasting more than two hours
Avoiding dehydration and emptying your bladder before going to bed may help prevent priapism. Exercise such as climbing up and down stairs, a warm shower and painkillers may help during an attack. If an attack lasts more than 2 hours you need to seek urgent medical attention. Speak to your doctor if you suffer with this problem. Try not to feel embarrassed about discussing this with your doctor as there are now a number of treatments available to help.

**Leg ulcers**

Ulcers (open sores) around the ankle may occur. Injury and insect bites play a role in starting the ulcers, so it is important to try and prevent these as much as possible. Leg ulcers often last for many months. Regular change of dressings, a good diet, adequate rest and leg elevation can all help with healing.

**Eye problems**

Blocked blood flow in the small blood vessels at the back of the eye causes the eye to make new blood vessels to bypass the blocked ones. These new blood vessels are thin, weak and tend to burst causing bleeding. The retina which is the seeing part of the eye may detach and lead to problems with vision.

Eye check ups about once a year will help detect any problems. The specialist should look into the back of your eyes to assess if there are any sickle cell related problems. If you develop any problems with your vision or you see things floating, see an eye specialist as soon as possible.

**Gallstones**

As a result of continuous breakdown of red blood cells, gallstones can develop. This may result in abdominal pain and worsening jaundice. A scan of the abdomen is used to look for gallstones. Should you have problems as a result of gallstones, surgery to remove your gallbladder may be recommended.

**Stroke**

This is a known complication of sickle cell disease. It can occur at any age, though more common in children. Weakness and loss of feeling in the arm or leg are common symptoms of strokes. Speech may be affected. Parents of young children need to be vigilant as their child may not be able to complain.

It is possible to identify children at high risk of developing stroke by a special ultrasound of the blood vessels in the brain called a transcranial doppler scan. This scan can identify areas where blood flow in the brain is abnormal. If a child is found to have areas in the brain where the blood supply is affected, treatment can be given to try to prevent strokes before they happen.
Another kind of stroke known as ‘silent stroke’ occurs in sickle cell disease. There are no obvious physical manifestations. This is more common than the obvious strokes and is detected by special brain scans.

**Other Organ damage**

Long-term organ damage may occur. It tends to be more common as people get older. The lungs, heart and liver may be affected.

Also, a condition known as pulmonary hypertension is sometimes a complication of sickle cell. The pressure in the large blood vessel that takes blood from the heart to the lungs (pulmonary artery) is abnormally high. Shortness of breath on an on-going basis is one of the symptoms. However, you may have no symptoms. Your doctor may ask for a heart scan (echocardiogram) to detect this problem.

Liver problems may be detected when you have a blood test. Generalised itching and worsening jaundice may be due to liver problems.

If you have long-term organ damage, your doctor will recommend appropriate treatment, if needed.
Other Treatments

**Blood transfusions** are sometimes needed, often as an emergency. Transfusions may be required when anaemia becomes more severe due to one of the complications. They may also be required on a regular basis to prevent certain complications such as strokes and may be required before surgery.

**Hydroxyurea (or Hydroxycarbamide)** is an oral medication that can reduce the frequency of crises and acute chest syndrome. It can also decrease the need for blood transfusions. It is not suitable for everyone. Your doctor will let you know if this medication is appropriate for you. Hydroxyurea should only be prescribed by a doctor with experience in using this medication and regular monitoring by a specialist is required.

In some parts of the world, a number of people with severe sickle cell disease have been cured with **bone marrow transplants**. Bone marrow transplants are mostly successful when they are done in childhood. They are not recommended for all children because they can cause serious side effects in some patients. You should discuss whether a transplant is suitable for you or your child with a specialist who will be able to explain the process to you and answer any questions you may have.

There is plenty of research going on in sickle cell disease. Work is in progress to develop new and effective treatments. Researchers are seeking to find ways to halt the effects of the disease and ultimately would hope to find a universal cure.

**Contact information**

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