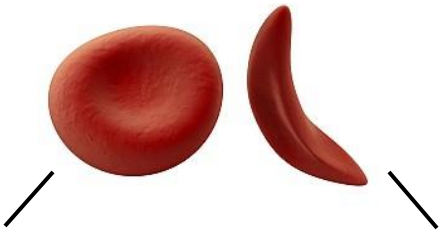
Sickle Cell Disease

Division of Human Genetics
University of Cape Town

What is sickle cell disease?

Sickle cell disease (SCD) is a group of inherited blood conditions which cause the red blood cells in the blood to develop abnormally. Normal red blood cells are flexible and disc-shaped, but in SCD, the red blood cells have a tendency to become sickle-shaped, like a crescent moon (see figure 1). This can cause various problems including pain in the joints, chest pain, shortness of breath, fever, episodes of anaemia and a susceptibility to infection. In between the episodes of illness, people with SCD generally feel well



Normal red blood cell **Sickled red blood cell**

Figure 1: Normal red blood cells are disc-shaped, while sickled cells look like crescent moons.
Image taken from: http://www.transfusionnews.com/wp-content/uploads/2013/02/SickleCellFeb22.jpg

What causes sickle cell disease?

Sickle cell disease is caused by a genetic mistake (mutation) in a specific gene that is responsible for telling the body how to make haemoglobin. Haemoglobin is a protein which is important for the correct formation of red blood cells. When haemoglobin doesn't form properly, the red blood cells become rigid and are likely to get stuck in small blood vessels in the body and block them. This can happen suddenly and can result in a variety of symptoms which depend on the location and severity of the blockage.

How is sickle cell disease inherited?

Sickle cell disease is inherited as a recessive condition (see Autosomal Recessive Inheritance fact sheet **10**). This means that to have sickle cell disease two altered haemoglobin genes must be inherited, one from each parent. Although a person with only one altered haemoglobin gene does not have SCD, they will have what is called a sickle cell trait. This means that although it is very unlikely that they will experience any symptoms of SCD, some of their red blood cells are vulnerable to sickling. Under extreme physical stress such as dehydration and conditions of low oxygen, it is possible that an individual with a sickle cell trait may experience pain in muscles and joints because of sickled cells blocking blood vessels.

What are the symptoms of sickle cell disease?

The symptoms of SCD can be very variable because they are dependent on the proportion of sickled cells in the body, and the location of a blockage. For much of the time, the red blood cells may behave normally and the individual may not suffer from any symptoms. It is only when an environmental factor, such as dehydration or low oxygen, causes many of the red blood cells to sickle, known as a sickle crisis, that an individual will experience symptoms. Most people with SCD experience a few episodes of sickle cell crisis every year.

The most common symptoms of SCD include:

<u>Episodes of pain</u>

Moderate to severe pain can be experienced in the joints, bones or abdomen (tummy) when sickle cells block small blood vessels, reducing oxygen supply to these areas.

<u>Acute chest syndrome</u>

Acute chest syndrome is the sudden loss in the ability of the lungs to take in oxygen and is often the result of a blockage of blood vessels leading to the lungs, or because of a lung infection. Chest pain, shortness of breath and a fever are often symptoms of acute chest syndrome. It occurs most commonly in women who are pregnant or who have just given birth. It is a very serious condition and must be treated rapidly.

<u>Infections</u>

People with SCD are vulnerable to infections. It is important to seek rapid treatment if an infection is suspected.

<u>Episodes of anaemia</u>

Anaemia is caused by a lack of haemoglobin in

Sickle Cell Disease

Division of Human Genetics
University of Cape Town

the blood. Haemoglobin is very important for the transportation of oxygen around the body. Individuals with SCD will generally experience constant but mild anaemia because of the rapid clearance of sickled cells from the blood. However, severe anaemia may be experienced in periods of stress, such as during an infection, and should be treated rapidly. During a severe anaemic episode, a person may feel tired, short of breath, light headed, dizzy, nauseous and may have rapid breathing. A good indicator of anaemia is pale skin which can be easiest to see on the lips, tongue, fingernails or eyelids.

Pulmonary hypertension

Pulmonary hypertension refers to an increase in pressure in the blood vessels between the heart and lungs. The pressure build-up is a result of a blockage in the vessels and can result in shortness of breath, dizziness, fainting and swollen legs.

Stroke

Sickled cells that block a blood vessel leading to or within the brain will cause a stroke. The severity of a stroke depends on which region(s) of the brain have been affected, and for how long. A stroke may cause paralysis in one or more regions of the body, loss of the ability to produce or understand speech, loss of vision in one half of the visual field or even death. Onset can be sudden and medical attention must be sought immediately.

How is sickle cell disease treated?

Although there is currently no cure of SCD, There are a number of management options which can reduce the number of sickling episodes an individual may experience, significantly reducing their risk of severe complications.

Preventing sickling episodes

It is important for a person with SCD to avoid environmental factors which encourage sickling such as being cold, dehydration, lack of oxygen, hard exercise, fever and infection.

Always ensure that a person with SCD is dressed warmly in cold conditions and that they have appropriate bedding to keep them warm at night.

Dehydration can be avoided by drinking a lot of fluid every day and by avoiding excessive alcohol intake.

To ensure that a good oxygen supply is always available for the body, a person with SCD can take folic acid supplements to increase red blood cell production. Smoking should be avoided as this is damaging to the blood vessels and can reduce lung function. People with SCD must also be careful about travelling to places that are very high, such as mountains, as there is less oxygen in the air at high altitudes.

A person with SCD should exercise regularly to remain fit and healthy and to encourage red blood cell production, but they must be careful to keep their exercise gentle such as going for regular walks.

The risk of infection can be reduced by taking antibiotics daily and receiving all of the usual childhood vaccinations as well yearly influenza vaccinations. Maintaining a healthy, balanced diet and taking vitamin supplements will also ensure that the immune system is strong.

If a person with SCD lives in or visits a place with Malaria, they should be careful to protect themselves from mosquito bites and ensure that they take malaria preventing medication.

Treating sickling episodes

An many cases, sickling episodes do not require hospitalisation and can be treated at home with painkillers, fluid, antibiotics and possibly oxygen. However, if the sickling episode is severe then the person should be taken to hospital where they can receive expert care.

Blood transfusions are often helpful in the treatment of severe anaemia and acute lung syndrome because the effect of the sickled cells is reduced by the rapid introduction of healthy red blood cells. However, blood transfusions do have some potential side-effects, so they should only be performed as a treatment option and not routinely.

Contact details:

To make an appointment with a Genetic Counsellor, please contact the Genetic Nurse at:

Tel: (021) 404 6235 or 406 6304