Sickle cell anaemia: causes, signs, symptoms and treatment

SICKLE cell disorders are a group of inherited conditions that affect the red blood cells (erythrocytes). They include sickle cell anaemia, haemoglobin sickle cell disease and beta thalassaemia. Of these the most common and severe is sickle cell anaemia. It is a global health problem, affecting many races and ethnic groups. The World Health Organization estimates that more than 250,000 babies are born each year with the disorder (WHO, 1989).

Anatomy and physiology

In general, anaemia occurs when red blood cells cannot carry as much oxygen as normal – either because there are not enough of them or because there is a problem with the cells themselves. A person with anaemia can feel tired, dizzy and breathless after only slight exertion. Treatment varies because the condition has many causes.

Each red blood cell contains haemoglobin (Hb), a protein used to transport oxygen. Red blood cells take up oxygen in the lungs and deliver it to the body tissues. These cells are round and flexible, and if they contain normal haemoglobin (HbA) they remain so after giving up their oxygen. This roundness and flexibility enables them to flow easily through the tissues (Bloom, 1996).

The red blood cells of people with sickle cell disorder, however, contain unusual haemoglobin – termed sickle haemoglobin or haemoglobin S (HbS). This undergoes changes after oxygen delivery, and can cause the red blood cells to alter shape – particularly when the level of oxygen in the blood is reduced, such as after exertion. The cells become sickle shaped. Sickle cells are rigid and do not flow easily through the tissues (Bloom, 1996).

Sickle cell trait

There is often confusion between sickle cell trait (SCT) and sickle cell anaemia. SCT is not a disease and people with the trait are usually healthy and live normal lives. They do not have side-effects and will not develop sickle cell anaemia. They do, however, carry the gene for HbS and could pass it on to their children (Franklyn, 1990).

Background

There is evidence that sickle cell disease was recognised by black Africans long before its earliest description in 20th century medical literature (Konotey-Ahulu, 1991). West African people were familiar with the disease and gave it specific tribal names. This was long before Herrick (1910) discovered an unusual type of anaemia in a 20-year-old black West Indian student. When Herrick examined the student’s erythrocytes under a microscope, he observed oddly shaped cells. What specifically attracted his attention, however, was the high incidence of sickle and crescent shaped blood cells in this patient.

Sickle cell anaemia occurs mainly in people of African origin, but it is also found in Mediterranean, Middle Eastern and Asian people. In the UK sickle cell anaemia is most common in people of African and Afro-Caribbean descent (approximately 12,500 people) and about 170,000 people are carriers of the sickle cell gene. These figures are estimates based on the most recent data available for population sizes and gene frequencies (Maxwell-Atungo, 2002).

Genetic inheritance

Sickle cell anaemia is a genetic disorder inherited from both parents. It is an example of a disorder in which two people who do not have the condition can produce a child with a genetic disease.

The process of inheritance for sickle cell anaemia is the same as for other characteristics such as height and eye colour: it is determined by pairs of genes, one from each parent. A person who is not born with sickle cell anaemia will never develop it. It is not infectious.

Haemoglobin genes are inherited (Serjeant and Serjeant, 2001). A person with sickle cell trait has inherited sickle haemoglobin (HbS) from one parent and normal haemoglobin (HbA) from the other (Figs 1–4). People with the trait need to take extra care in specific circumstances such as when undergoing general anaesthesia. They should also avoid activities where the amount of oxygen available is or may become compromised, for example climbing mountains.

Children can only be born with sickle cell anaemia if both parents have the gene for sickle haemoglobin (Figs 1–2). When both parents have sickle cell trait, each of their children has a one in four chance of being born with sickle cell anaemia and a one in two chance of being born with sickle cell trait (Fig 2). If one parent has sickle cell anaemia and the other has normal haemoglobin, none of their children will have sickle cell anaemia, but all will have sickle cell trait (Fig 3).

If one parent has sickle cell anaemia and the other has sickle cell trait, at each pregnancy there is a one in two chance of the child being born with sickle cell anaemia and a one in two chance of it having sickle cell trait (Fig 1). None of the children will have normal haemoglobin.

Diagnosis

Sickle cell disease can be diagnosed before or after birth by routine screening methods. A blood test known as haemoglobin electrophoresis is used to identify haemoglobin characteristics in the blood after birth. Prenatal diagnosis involves carrying out DNA studies on

ABSTRACT


Sickle cell anaemia, often called sickle cell disease or sickle cell disorder, is a term covering a number of similar conditions that affect haemoglobin. It has been described as the most common genetic disorder and affects around 12,500 people in the UK, predominantly of African and Afro-Caribbean descent. The severity of the physical, psychological and social consequences of this cluster of disorders can be greatly reduced if families who are affected receive early diagnosis, appropriate support, advice and care.
Anaemia
In general, people with sickle cell disease experience the most severe form of anaemia. Not only do they have fewer red blood cells than people with normal blood counts, their cells also do not live the usual 120 days. The resulting chronic state of anaemia causes breathlessness and fatigue.

Signs
A doctor should be consulted, with a view to obtaining a blood count, if any of these signs are noted:
■ More tired than usual;
■ Pale colour;
■ Loss of appetite;
■ Yellow eyes or skin;
■ Dark urine.

Childhood symptoms
Symptoms rarely start until babies reach the age of four to six months. Symptoms include dactylitis (a painful swelling of the hands and feet), which usually occurs when the baby is between six months and two years old.

The child may experience a painful crisis. Pains can occur, sometimes with swelling and tenderness in various parts of the body. Arms and legs are most often affected, but pain and swelling can also occur in the knees, elbows, back, chest and abdomen. These symptoms may last for a few hours, a week, or longer.

Some children experience splenic sequestration. This occurs when the blood in the spleen sickles and the spleen enlarges quickly. The symptoms are a high temperature accompanied by stomach ache. Splenic sequestration is potentially life-threatening and the child should receive immediate medical attention.

Many children experience jaundice, which results in a slight yellowing of the whites of the eyes. This is caused...
by the quick destruction of red blood cells. Yellow whites of the eyes are not unusual for people with sickle cell anaemia, but extreme yellow could indicate gallstones or sickling in the liver (Anionwu and Jibril, 1986).

Children and adults with sickle cell anaemia pass urine many times a day because the disease has a minor effect on the kidneys. Fluid intake should be increased to keep the kidneys as healthy as possible, and to prevent the blood from thickening and sickling. Some children may have problems with enuresis, or bedwetting. A small number of children with sickle cell anaemia experience strokes. This can occur when cells block the blood vessels in the brain (Oni et al, 1997).

Potential symptoms in teenagers and adults

Potential symptoms include:

- Slow growth and late puberty;
- Sores and ulcers around the ankles;
- Gallstones;
- Blurred vision and even loss of sight;
- Weak bones, often in the shoulder and hip;
- Women with sickle cell anaemia conceive normally and, with proper care, usually have a normal pregnancy. However, pregnancy puts extra stress on the body and increases the woman’s susceptibility to infections or painful crisis;
- Teenage boys and men may suffer from persistent penile erections (priapism). This is extremely distressing and can lead to sterility and/or impotence (Midence and Elander, 1994).

Treatment

If the body stops making new red blood cells or the cells are destroyed more quickly than usual, a low blood count may be the result. A blood transfusion may be required if the blood count falls to a very low level. An extremely low blood count can lead to heart failure and death unless treated in time.

There is, at present, no cure for sickle cell anaemia. However, there are various actions that can be taken at home, school, or work to prevent or relieve symptoms. These include:

- Drinking plenty of fluids throughout the day is vital to prevent dehydration;
- A healthy diet should be eaten, including plenty of fresh fruit and vegetables;
- Strenuous exercise or over-exertion should be avoided;
- The immediate surroundings should be kept clean to avoid infection;
- It is important to keep warm and get plenty of rest;
- Hot baths, massage and relaxation are helpful;
- Non-prescription painkilling drugs, for example, paracetamol, aspirin and ibuprofen can be taken;
- Penicillin can be taken daily to prevent infection;
- Folic acid can be taken daily, if lacking, to encourage healthy new cells.

Actions that should be taken in hospital include:

- Give strong pain relief – opioid analgesics;
- Administer fluids – intravenously if necessary;
- Give antibiotics for infection;
- Occasionally, blood transfusion may be required;
- Bone marrow transplants may be considered (Kirkpatrick et al, 1991).

Conclusion

Sickle cell disease is a global health problem. It can result in distressing acute and chronic symptoms and can be life-threatening. There is no cure, therefore nurses should understand the actions that can prevent or relieve symptoms in order to meet the challenges of caring for patients with sickle cell disease and helping them to minimise its effects on their lives.