Sickle cell disease: 'The silent life wrecker'

Sickle cell disease (SCD) is a 'silent' problem with devastating effect in many parts of Kenya. There is often very low awareness of this condition amongst people, and not much attention is given to the problem by the Ministry of Health (for example, SCD is not included in routine health statistics, and there are no national treatment guidelines). YET SCD causes high rates of serious health problems and excessive early deaths in Kenya (many children die undiagnosed and more than 90% die before 5 years of age!) because of the lack of recognition and comprehensive care. High school dropout rates have been observed among those diagnosed with the disease often due to prolonged ill health. In countries where comprehensive care is available, many people with SCD live normal lives into their 40s and 50s, or longer than this.

SCD is an inherited condition that affects the red blood cells in people's bodies. Normally, red blood cells (RBCs) transport oxygen throughout the body, which is essential to the way our bodies work. This is facilitated by a structural component called haemoglobin inside the red blood cells, which has the ability to bind with oxygen. However, a genetic variation affecting the structure of the haemoglobin may give rise to what is referred as a sickle cell disorder (SCD). The type of abnormal haemoglobin found in many cells of people with SCD is called haemoglobin S (HbS). The problem is that RBCs containing HbS are less flexible and more fragile than usual.

In people with SCD, most of the time their red blood cells have the normal 'doughnut' shape typical of people without this condition. But from time to time, particularly on exposure to low oxygen levels, the shape of the red blood cells with HbS, changes to a sickle shape (like the sickle used for cutting grass), giving this condition its name.. This transient 'sickling' of RBCs, makes them unable to carry oxygen efficiently. These structurally abnormal cells are frequently prematurely destroyed from the body leading to lack of blood (anaemia), and often get stuck in small blood vessels causing bone pains (the hallmark of the disease), stroke, and widespread organ damage. Collectively, these abnormal changes are termed as sickle cell crises. In addition, people with SCD are less resistant to many different types of infection, such as pneumonia and infections of the blood.

But SCD is a very variable condition, with different people experiencing very different severity of symptoms. Some people with the illness in Kenya have very few symptoms, even without much medical care. In more severely affected people, symptoms often start around the age of 6 months, usually with temporary but very painful swelling of the fingers or toes. As they get older, children often develop swelling of an organ called the spleen in the abdomen, so that their stomachs become noticeably swollen accompanied with yellowness of eyes. Later, this swelling disappears as the spleen shrinks through damage caused by sickling of RBCs. People with SCD generally have some level of anaemia all the time, because abnormal RBCs are more rapidly broken up than normal flexible 'doughnut' shaped cells. At younger ages, the condition is often not recognised by families or even health workers. Instead, they may be treated for other conditions, such as malaria or boils.

The way that SCD is inherited in families is that people with the condition carry a SCD gene from both parents, giving them 2 SCD genes – the situation needed to cause symptoms. In this case, both parents must have passed on one SCD gene to their child, but having only one SCD gene does not cause any symptoms in the parents. People who carry one SCD gene are completely healthy, but they do have the risk of future children being affected by the condition if their partner also carries one gene. It is known that the risk of having an effected child if both parents carry one SCD gene is 1 in 4, and this risk will apply to each child they have.

SCD is known to have originated as part of mankind's fight against malaria many thousands of years ago. Having RBCs with HbS makes people more resistant to infection with malaria. This is particularly an advantage to people with one SCD gene, who have an in-built resistance to malaria and at the same time have no problems associated with carrying one gene. This in-built resistance to malaria is the reason why SCD is particularly common in parts of Kenya where malaria is also common, such as the Coast and Western Kenya. However, migration means that the condition can be found anywhere in the country.

There are many ways in which people with SCD can look after their own health at home, including making sure that they always take plenty of fluids to drink, use painkillers like paracetamol when needed, keep warm in cold weather and manage levels of exercise within each individual's tolerance. However, regular lifelong medical care is essential to maintaining health, including the prevention and treatment of infections (using antibiotics and vaccines against pneumonia); prevention of malaria through regular use of preventive medicines, prescription of blood builders and transfusions when needed. However, the cost of treatment is generally enormous –the crises are unpredictable and frequent. Majority poor families sometimes give up seeking any form of health care.

As important as all these medical treatments is the education and counselling that people with SCD can benefit from, to understand the cause and therefore the lifelong medical care needed, including that the condition cannot be 'cured'. Without this understanding, many affected people spend time, money and emotional energy seeking care from other sources, such as traditional healers. Such forms of care may be very supportive to some people, but should not replace medical care. As a genetic condition, affected families can also experience conflict and blame within the home, and counselling can help families to resolve these problems. In addition, to these forms of care, in other parts of the world another treatment called 'hydroxyurea' is used in some people with SCD to reduce the frequency of sickling attacks, and this may be available in Kenya in the near future, once more research is done to check its safety and effectiveness here. Looking to the future, scientists are trying to develop 'stem cell' transplantation techniques that may one day offer the chance of a 'cure' for people with SCD. Currently, success with these procedures is very limited.

Overall, Kenya requires comprehensive information and care programmes on SCD, to create more public awareness of SCD, while avoiding any risks of stigmatising people with the condition; to ensure at least basic care packages in all facilities in the region like the district hospitals/peripheral facilities around the region; and to have more trained counsellors to help affected families – as has been done in other parts of Africa (especially Ghana and Nigeria). In other parts of Africa, different types of SCD screening programmes have been set up. Some screening programmes target new born babies in hospital, to understand as soon as possible if a child has this condition, since starting medical care early gives the child the best chance of a healthy future. Another form is pre-marital screening, offered to young people so that they can understand any risk they carry of having a child with this condition in future. Pre-marital screening has been successful in reducing the burden of other similar inherited diseases of the blood in the Middle East. The important point is that screening programmes for young adults can equip them to make informed choices about their future.

Raising awareness about the disease is only the *first step* to better health. Although we must take the whole staircase for better outcomes, we *just* have to take this *first step to start off*.