Members of Uganda Sickle Cell Association marching in Kampala during an awareness week

20,000 babies born with sickle cell disease yearly – study

June 19 is World Sickle Cell Day. It was created by the United Nations in 2008 to increase awareness about sickle cell disease. According to the World Health Organisation, sickle cell disease is the commonest genetic disease worldwide.

The World Health Organisation urges member countries where sickle cell disease is a public health problem to establish health programmes at the national level and operate specialised centres for sickle cell disease and facilitate access to treatment

Prevalence of sickle cell disease/ trait in Uganda

According to results of a study published in 2016 by Prof. Grace Ndeezi, in Uganda, about 20,000 babies are born with sickle cell disease every year.

Ndeezi, a professor of paediatrics and child health at Makerere University, noted that sickle cell disease contributes substantially to death in children younger than five years.

Charles Kiyaga, the national programme co-ordinator for sickle cell disease in the health ministry, notes that about 80% of children born with sickle cell disease die before they reach five years. He attributes this to the fact that they are not diagnosed early and, therefore, not treated in order to prevent complications.

Kiyaga added that between February 2014 and March 2015, the ministry carried out a survey to establish the prevalence and distribution of sickle cell disease and trait in Uganda. The results, which were launched in April 2015, showed that about 13% (one out of seven) of Ugandans carry the sickle cell trait.

Proceeds from Kabaka's birthday run in April were dedicated towards sickle cell disease

Prevention: Only way to stop sickle cell disease

Sickle cell disease is the commonest gene disease in the world, but as Lillian N. Magezi writes it can be stopped in its tracks

Jessica Namuyimbwa, 37, a resident of Maganjo in Wakiso district, had never heard of sickle cell disease until her third child was about seven months old in September 2014. Suddenly, the boy who had been healthy became pale; his eyes and skin became yellow.

"The boy looked unhealthy. When we went for immunisation, I told the nurse about his appearance who said it could be anaemia, which is due to poor feeding. She advised me to continue breastfeeding, in addition to giving the child nutritious foods such as millet porridge, fish and beans," explains Namuyimbwa.

When the boy was about nine months old, his fingers swelled.

"Although he did not seem to be in a lot of pain, I took him to the clinic where a doctor suggested that I have him tested for sickle cell disease. I was reluctant and the doctor suggested that we do a complete blood count. The results showed that my son was anaemic. The doctor said that could be a sign of sickle cell disease and I agreed to do the test," says Namuyimbwa.

She was told to pick the results after a few days. The doctor then prescribed folic acid tablets for the boy.

The doctor explained that if the results were positive for sickle cell disease, the boy would have to take folic acid for the rest of his life.

He explained to Namuyimbwa that sickle cell disease is a genetic blood disorder where one produces faulty red blood cells. As a result, the individual is predisposed to anaemia, therefore, folic acid supplementation is meant to help one produce more blood.

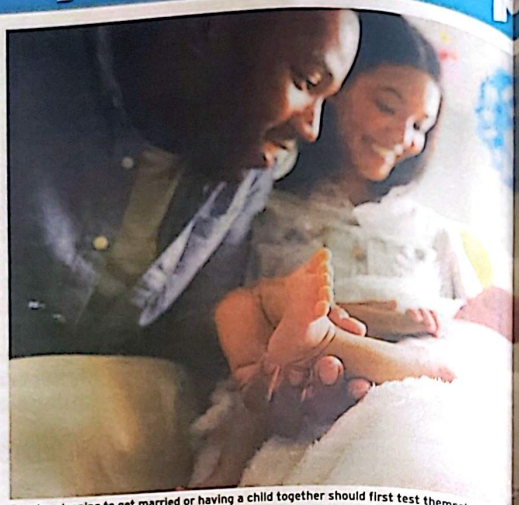
And as the doctor had suspected, the results confirmed that the boy had sickle cell disease.

"I was told that there was no cure for the condition and that the boy had to be given medical treatment and constant reviews throughout his life.

As per the doctor's guidelines, we give him folic acid daily. We also give him fansidar every month to prevent malaria. In addition, he is dewormed regularly," explains Namuyimbwa.

She adds that the doctor recently asked her to take the boy for several tests to determine whether to start him on hydroxurea, a more effective drug that reduces the incidents of pain crises in people with sickle cell disease.

Namuyimbwa discloses that she later went for testing with her husband and it turns out that both of them have the sickle cell trait although they did not have the condition. In addition, blood tests showed that their first two children also carried the sickle cell trait.



Couples planning to get married or having a child together should first test themselves for sickle cell trait

What is sickle cell disease?

Sickle cell disease is a hereditary blood disorder in which one's red blood cells produce abnormal haemoglobin, explains Prof. Christopher Ndugwa, a paediatrician with over 40 years' experience in management of sickle cell disease.

Ndugwa explains that red blood cells contain a component called haemoglobin, which gives them the red colour and enables them to carry oxygen around the body.

Red blood cells are normally disc-shaped (shaped like a doughnut), elastic and flexible, which makes it possible for them to squeeze through small vessels and capillaries. On the other hand, sickled red blood cells are rigid and thus, cannot pass through small vessels and capillaries. Therefore, they end up clogging the vessels, which hinders blood flow, cutting off blood and oxygen supply from some parts of the body.

Clogging of blood vessels and frequent rapturing (breaking down) of sickled red blood cells is the cause of many symptoms and complications brought on by sickle cell disease.

Ndugwa explains that red blood cells take on the shape of a sickle (a farm instrument), which leads to many complications.

A person is born with sickle cell disease when they inherit a gene with a sickle cell trait from each of the parents during conception, explains Charles Kiyaga, the national programme co-ordinator for sickle cell disease in the health ministry and Dr Nicholus Nanyeenya of Rubaga Hospital.

Patients need to take folic acid tablets daily

This means for one to have sickle cell disease, both parents have to be carriers of the sickle cell trait.

Chances of parents who carry the trait of getting a child with sickle cell disease are 25% per pregnancy. Such parents also have a 25% chance of having an unaffected child and a 50% chance of having a child who also is a carrier for every pregnancy.

Link between malaria and sickle cell disease

Prof. Grace Ndeezi, a paediatrician, notes that there are many people with the sickle cell trait in areas with a high prevalence of malaria. This is because having a sickle cell trait protects one from severe malaria.

Kiyaga explains that since malaria is the leading killer for children under five years in Uganda, it means those who die are mainly those without the trait, leaving those with the trait. This means the percentage of people with the sickle cell trait of the general population keeps on increasing.

Complications

Experts note that symptoms of sickle cell disease among children usually start showing at about six months. This is when children start producing the red blood cells with defective haemoglobin. Among infants, the most common symptoms are recurrent fever, pain and swelling of fingers and feet, in addition to jaundice (yellowing of the eyes) and anaemia.

Swelling of fingers, toes

Experts note that swollen digits among children below two years is usually the first sign of sickle cell disease. Men with sickle cell disease can also get priapism. This is an unwanted prolonged painful erection that lasts more than four hours.

Experts note that this is a medical emergency that should be immediately attended to because it can cause death of penile tissue, leading to impotency.

Organ failure

A patient will experience a lot of pain in the parts where blood and oxygen supply are cut off such as arms, legs or back.

Subsequently, cells die off when parts of the body are denied blood and oxygen for some time. For example, patients get unhealing ulcers on the legs or the retina (the part in the eye responsible for sight) can peel off leading to blindness.

If the brain is affected, the patient will get a stroke. This presents with one-sided arm or leg weakness, facial droop, slurred speech, seizures, aphasia or coma.

Clogging of blood vessels in organs can also lead to them getting damaged such as the spleen and lungs.

Pain crises are caused by extremes in temperature (for example extreme coldness), strenuous exercises, dehydration, bacterial infections and other diseases such as malaria

Anaemia

Sickled red blood cells have a shorter lifespan; they break down after 20 days as opposed to the normal cells that breakdown after 120 days.

Continuous breaking down of the red blood cells predisposes people living with sickle cell disease to anaemia, jaundice (yellowing of the eyes) and gallstones.

Other complications suffered by people living with sickle cell disease are crushing of hip joints to cause lameness, kidney malfunctioning, stunting in addition to mental retardation.

Prevention

Test before marriage

Kiyaga advises people planning to get married or having a child together to first test themselves for sickle cell trait. A carrier is advised against marrying or having children with a fellow carrier.

Screening for sickle cell disease or trait costs between sh30,000 and sh60,000 which can be carried out at any reliable health facility.

Sickle cell makes babies anaemic and susceptible to malaria

Managing sickle cell at home

It is advisable that parents, caregivers and people living with sickle cells learn to manage the condition at home. Prof. Christopher Ndugwa, a paediatrician, explains that some patients will frequently get sick, which makes it impractical to always be rushed to hospital. Home management should involve:

- Administering folic acid: it is given daily for life to prevent anaemia
- Hydration: People with sickle cell disease should drink between three to five litres of fluids a day and prevent dehydration.
- Ensure a balanced diet, high in protein
- Proper hygiene to prevent bacterial infections
- Monitor temperature and seek medical advice if it is high, fever could be a sign of bacterial infection or malaria.
- Monitor growth
- Prevent malaria, sicklers have a higher risk of dying from malaria due to anaemia. So malaria should be prevented by sleeping under an insecticide-treated bed net.
- Check spleen size regularly by looking for a mass on the left side of the abdomen and seek medical attention if there is any swelling.

People with sickle cell disease should eat a balanced diet and drink of fluids to prevent dehydration

Treatment of sickle cell

The only cure for sickle cell disease is a bone marrow transplant, but it cannot be done in Uganda. Charles Kiyaga, the national programme co-ordinator for sickle cell disease in the health ministry, explains that with proper care and management, people living with sickle cell disease can lead full and productive lives. In addition to treating complications as they present, people living with sickle cell disease are given the following treatment:

- Folic acid daily to improve blood production
- Monthly prophylaxis for malaria
- Antibiotic prophylaxis every month to protect against bacterial infections
- Routine deworming
- Routine vaccinations as advised by the health ministry. Vaccinations should include the pneumococcal vaccine, which protects one against pneumonia.

Routine treatments include:

- Malaria prophilaxis (monthly). The best medicine to use is Fansidar because it lasts longer in the body than other anti-malarials.
- Antibiotics (monthly)
- Deworming (monthly)
- Routine immunisations as advised by the health ministry
- Regular reviews

People living with sickle cell disease should be registered at a sickle cell clinic where they can be reviewed regularly.

One of the things that need to be reviewed regularly is blood level because a patient might appear considerably fine, but when their blood level is reducing.

Doctors also look for any signs of malaria, jaundice, check spleen size, hydration status, infections or any other complications of the disease.