European Commission

ARISE
African Research And Innovative Initiative For Sickle Cell Education

FONDAZIONE PER LA RICERCA FARMACOLOGICA GIANNI BENZI ONLUS

LIVING WELL WITH SICKLE CELL DISEASE

INTRODUCTION

Nigeria with a population of about 180 million has the largest burden of sickle cell disease (SCD) in Africa and worldwide. Nigeria accounts for approximately 190,000 newborns with SCD annually. About 25% of adults in Nigeria have the sickle cell trait, and Northern Nigeria has the largest number of SC trait with a prevalence of 68.2%. In addition, Nigeria has relatively high mortality due to SCD-related infections occurring especially in rural areas. The burden of child mortality from SCD in Nigeria remains disproportionately higher than that of children without SCD. A study found that nearly half of newborns with SCD in Nigeria do not reach their fifth birthday.

While good policies that assist patients with SCD include good management of the disease in the hospitals, prevention campaigns for newborns, parental and patient education, the care available for SCD in Nigeria is still suboptimal. There is a variable and poor utilization of standard-of-care practices for SCD patients. Moreover, an analysis done through a questionnaire survey designed and administered within the ARISE project demonstrated the absence of structured government policies that would make SCD diagnosis and treatments free and available all time within communities, and the inadequacy of manpower (healthcare personnel), drugs and equipment in most parts, especially in the rural areas.

Moreover, poor knowledge and awareness allow different perceptions on SCD among public, especially in the northern part of Nigeria due to ignorance, tradition/culture, and poverty, which also represents a relevant factor preventing the diffusion of good prevention and care practices that may be mitigated by promoting large-scale patient's and parental education initiatives.

The present publication has been tailored to the specific needs of the population of reference, prepared in a friendly and lay language, and will be distributed in collaboration with patients, families and patient organisations. It aims to contribute to reducing the gap affecting SCD patients in Nigeria and in other countries sharing similar problems, concerns and barriers for patients.

The book is developed for patients, parents, families/relatives, healthcare providers, and other health professionals and it is recommended for adult patients and for children older than 10 years. In the case of younger children, it should be read with parental support and guidance for a better understanding.

We hope this book will help you better understand SCD!

° Prof. Adriana Ceci ° Prof. Prof Baba Inusa ° Mr. Fedele (Duccio) Bonifazi

QUESTIONS AND ANSWERS FOR PATIENTS PARENTS/ CAREGIVERS, FAMILIES/ RELATIVES AND PUBLIC

•TOPIC: SCD general features

What is sickle cell disease?

Sickle cell disease (SCD) is an inherited blood disorder that affects haemoglobin. Haemoglobin is contained in the red blood cells. It gives blood its red colour and is responsible for carrying oxygen throughout the body. People with SCD have an atypical form of haemoglobin called haemoglobin S, that causes "sickle" shaped red blood cells that can stick together, blocking blood flow and oxygen from reaching all parts of the body. SCD is a term covering a number of different but similar conditions that affect haemoglobin.

•TOPIC: Causes of SCD, genetics and types of SCD

What causes sickle cell disease?

SCD is a genetic condition that occurs when a child receives two genes, one from each parent, coding for atypical haemoglobin, one of which is haemoglobin S. Genes contain instructions to determine your features (e.g., eyes or hair colour) that are passed on to you from your parents. There are several types of SCD depending on the haemoglobin genes you inherited from your parents. Haemoglobin SS (HbSS) disease is the most common one. It occurs when you receive two genes for haemoglobin S from both parents, thus a person is born with SCD. As the most severe form of SCD, individuals with this form also experience the worst symptoms at a higher rate.
A person has sickle cell trait when the haemoglobin S gene is inherited from only one parent and the normal haemoglobin gene – coding for haemoglobin A- is inherited from the other one.

•TOPIC: Transmission of SCD

Is sickle cell disease contracted, transmitted or acquired?

SCD is not contracted sexually, acquired later in life, or gotten from living with persons with SCD, or sharing objects. SCD is hereditary like hair, or eye colour. You can only get it if both of your parents carry a certain gene trait and pass it on to you. If you are not born with it, you can't get it later in life.

•TOPIC: People affected by SCD

Who are those affected by sickle cell disease?

Most of the people who have SCD are of African ancestry or identified as Blacks. Others are Hispanic Americans from Southern America, and people of Middle Eastern, Asian, Indian and Mediterranean descent. Both males and females are affected by SCD. More than 300,000 babies are born each year with the disease worldwide. Nigeria is the most affected country in Africa with 150,000 children born with SCD each year.

•TOPIC: Genetics test and diagnosis of SCD

How is sickle cell disease diagnosed?

SCD is diagnosed with a simple blood test. The most popular methods for detecting this disease are the full count of blood cells, Hb electrophoresis, and high-performance liquid chromatography (HPLC). These methods have been relied on for a long time in the diagnosis of SCD.

•TOPIC: Genetics test and diagnosis of SCD

Is it important to diagnose SCD early in childhood?

Early new-born screening for SCD is strategic in establishing early comprehensive management which comprises of:

- Scheduled follow-ups to proactively look for crises and complications.
- Prophylactic penicillin treatment to reduce the risk of pneumococcal infections.
- Other prophylaxis e.g. antimalarial in malaria endemic regions.
- Ensuring adequate vaccinations are achieved as per national protocols.
- Counselling for healthy living e.g ensuring optimal nutrition, good hydration etc.

•TOPIC: Complications of SCD

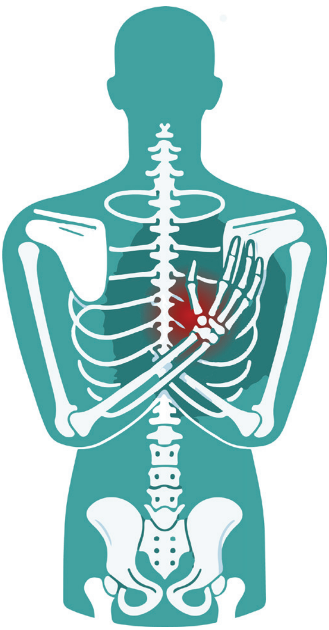
What are the complications of sickle cell disease?

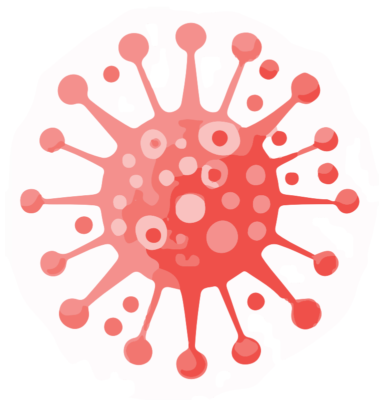
SCD causes many complications. Symptoms and complications are different for each person and range from mild to severe. Complications tend to worsen over time. SCD causes abnormal form of haemoglobin inside red blood cells. This can block blood flow and prevent oxygen from reaching many different organs. Lack of oxygen because of blocked blood flow can lead to: acute chest syndrome, pain and acute crises of pain, joint problems, stroke, and other complications such as retinopathy, kidney damage, etc.

•TOPIC: Infections and prophylaxis

Are infections relevant complications of sickle cell disease?

Yes. SCD increases susceptibility to infections and it is the leading cause of death, particularly in less developed countries. Infections, especially Pneumococcal septicaemia, meningitis, and Salmonella osteomyelitis are the major cause of morbidity and mortality in patients with SCD.





•TOPIC: Various organ complications of SCD

Is it true that sickle cell disease can cause stroke?

Yes. However, it is not the only complication and not necessarily people with SCD have all signs and symptoms. Stroke is one of the major complications and potentially devastating manifestations of SCD that can affect children and adult.

•TOPIC: Various types of pain in SCD and other crises

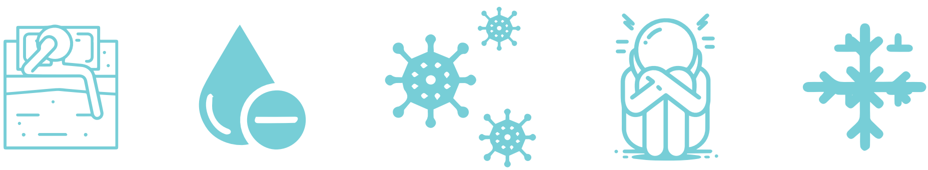
Is pain crisis the most common presentation of sickle cell disease?

Yes. Pain is the most common complication of SCD and the top reason for hospital admission of SCD people. Sickle cells travelling through small blood vessels can get stuck and block blood flow throughout the body causing pain. The pain in SCD can be acute, chronic, or a mixture of the two. Chronic pain in SCD is not simply a continuation of the pain due to a vaso-occlusion; it may be secondary to other complications such as vascular necrosis of bone at various joints.

•TOPIC: Discuss the several factors that could trigger a crisis. Discuss the pathophysiology of vaso-occlusion

What can trigger a sickle cell crisis?

The term “sickle cell crisis” is used to describe several acute conditions such as the vaso-occlusive crisis (acute painful crisis), hepatic crisis, acute chest syndrome and many others. Red blood cells in SCD are not flexible and disc-shaped, but they are curved and stiff. These sickle-shaped blood cells don’t flow through blood vessels easily and can clog the vessels, if this happens blood and oxygen can’t get through, and the part of the body (heart, lungs, kidneys) can’t work the way they should. Several factors can trigger sickle cell crisis, infections, extreme physical exercise, dehydration, cold weather, psychological stress, smoking, pregnancy etc.



POOR SLEEP | DEHYDRATION | INFECTIONS | STRESS | COLD WEATHER

•TOPIC: Pain management and severity assessment. Discuss appropriate tools, acute pain management, modalities of chronic pain management

Is it possible to treat the pain caused by the sickle cell crisis?

Yes. It is possible to treat pain due to SCD crisis, the treatment of pain involves a sequential approach to pain assessment, which includes considering age, developmental stage, functional status, cognitive ability, and emotional state. Assessing pain requires also measuring it: determination of the presence, intensity, duration, and location of pain, as well as efficacy of management. Clinicians ask patients to report their pain levels as a number from 0 to 10. For children who are unable to understand numbers, physicians may use pictures to assess specific signs and positions in the body.

•TOPIC: Pain and other adjuvant drugs for pain

Can morphine be used to treat chronic pain?

Yes. However, the approach to chronic pain caused by SCD is multidisciplinary; chronic pain is often associated with other conditions that enhance its chronicity. These include psychosocial factors such as depression, anxiety, feelings of despair, insomnia, loneliness, helplessness, posttraumatic stress disorder (PTSD), and dependence on pain medications. Management of all types of chronic pain associated with SCD may be enhanced by adding non-pharmacologic approaches (e.g. psychological intervention, occupational therapy, behavioural and cognitive interventions, etc).

•TOPIC: Follow up and monitoring schedule from diagnosis to adulthood

Do I need regular monitoring for complications?

Yes. To reduce the burden of the disease, there is the need for regular monitoring for complications and in some instances even prophylactic treatments are needed. Other disease conditions to be monitored include hepatitis (especially in patients with a history of blood transfusions), renal damage/nephropathy, eye damage/retinopathy, chronic obstructive airway disease, sensorineural deafness, etc.

•TOPIC: Pregnancy and SCD

Does SCD affects pregnancy, and how?

Yes. SCD affects pregnancy, with multiple hospital admissions and, in worse cases, it can even cause miscarriage. Women with SCD should particularly take care of its complications, including, anaemia, urinary tract infection, chest infection, pre-eclampsia, prematurity, blocking of blood vessels etc. During pregnancy, SCD may become more severe and pain episodes can occur more frequently. A pregnant woman with SCD is at higher risk of preterm labour and of having low birth weight baby.

•TOPIC: Discuss fertility, family planning

Is it true that patients with sickle cell disease shouldn't have children?

No. SCD patients are known to have become pregnant and deliver babies. However, recurrent priapism in boys may increase the chances of infertility. Hydroxyurea therapy may also increase the risk for infertility as well as incidence of congenital malformations. It is important therefore to use family planning methods while the woman is on Hydroxyurea and to stop the medications while the couple are trying for pregnancy, during pregnancy and while breastfeeding.

•TOPIC: community, premarital, prenatal, and pregnancy screening programs

Can I avoid giving birth to a child with sickle cell disease?

Yes. This could be avoided through early community diagnosis, premarital screening and counselling, and use of contraceptives. SCD could be diagnosed during the first trimester of pregnancy, if foetus found affected, couples could decide on whether to continue with pregnancy or not.

•TOPIC: Hydroxyurea and risk

Are there pharmacological therapies for sickle cell disease?

Yes. Hydroxyurea is a medicine used to treat both adults and children with SCD. Hydroxyurea is not a cure but can help prevent many complications such as reducing the pain crisis, the acute chest pain crisis and the need for transfusions. This can improve the life expectancy and the quality of patient's life. New promising therapies for SCD are emerging in the recent years.

•TOPIC: Penicillin, vaccination, folic acid, antimalaria

Do I need other drugs routinely apart from analgesics?

Yes. Other medicines such as penicillin prophylaxis, vaccinations, antimalarial and folic acid are recommended to stay healthy. A daily antibiotic is usually recommended like penicillin, or erythromycin (if you are allergic to penicillin). This is especially important to protect against serious infections in children aged under 5 years.

•TOPIC: Diet, nutrition and lifestyle

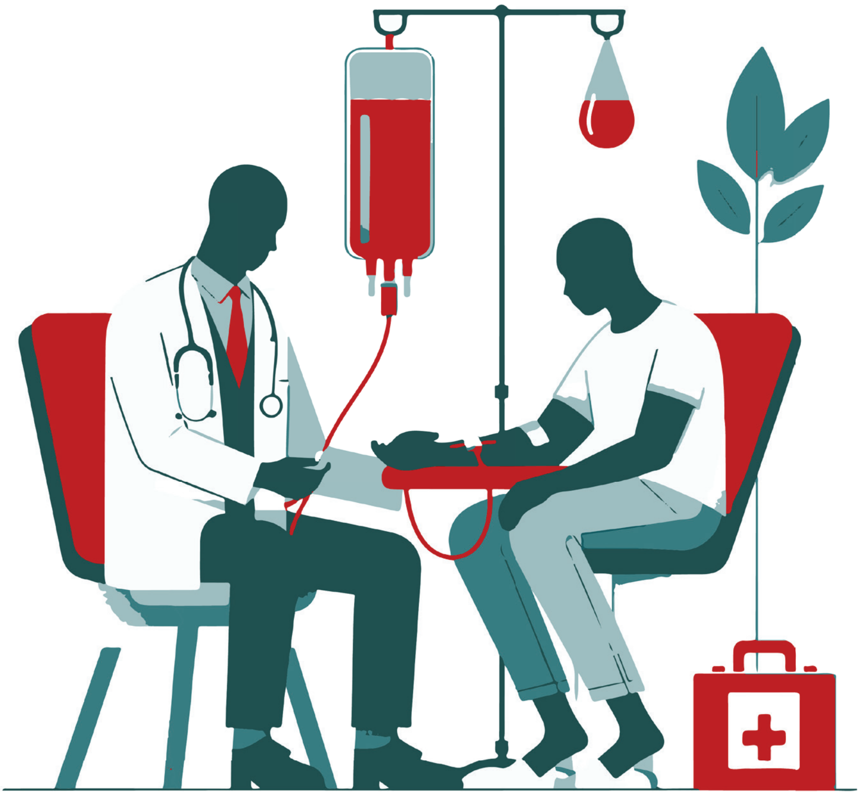
Is it true that healthy lifestyle has synergistic effect on the therapy?

Yes. This is important to ensure optimal health and hygiene at all times, boost psychological well-being and maintain a positive outlook. Living a healthy life can reduce some of the complications, lifestyle modifications are focused on healthy eating and regular check-ups, that are the primary recommendations for patients with SCD.

•TOPIC: Blood transfusion and exchange therapy

Is blood transfusion still a form of remedy for sickle cell disease?

Yes; the aims of transfusion in SCD are both to increase the oxygen-carrying capacity and decrease the proportion of sickle haemoglobin (HbS) relative to haemoglobin A (HbA) to prevent or reverse the complications of vaso-occlusion. Blood transfusion may improve disease complications and plays a prominent role in the management of patients with SCD.



•TOPIC: Infection and blood safety

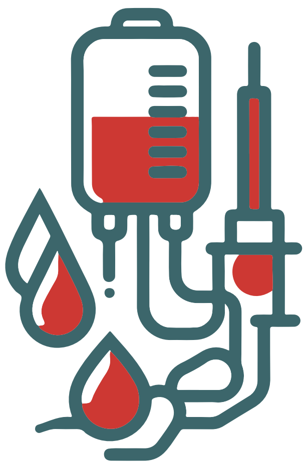
Is blood transfusion safe?

Yes. Blood transfusions are common and safe procedures, but there may be some risks. Infections were once the main risk, but they have become extremely rare with testing and donor screening. Transfusion reactions and other non-infectious problems are now more common than infections. Blood transfusion in SCD can be potentially lifesaving and prevent progressive organ damage but can also be associated with morbidity.

•TOPIC: Indications for exchange transfusion, simple transfusion and chronic transfusion regimes

Is exchange blood transfusion given only for stroke?

No. Exchange blood transfusion is potentially life-saving procedure that is done to counteract the effects of serious jaundice or change in the blood due to SCD. Exchange blood transfusion may be undertaken to rapidly reduce the percentage of sickle cells in the circulation when a patient develops a life-threatening complication of the disease. It is not to be undertaken lightly, as the possibility for complications is considerable.



•TOPIC: Iron overload and chelation

Is there any other possible major complication of transfusion therapy?

A possible complication of transfusions in SCD is iron overload. Iron overload is an undesired consequence of a regular transfusion therapy that can cause the exogenous iron accumulation which may led to organ damage/ failure (e.g., in the liver and the heart).

•TOPIC: Complication in other organs

Is iron overload only found in the liver?

No. Iron overload is not only found in the liver, it is also found in the heart, and pancreas. Excess iron is stored in the organs and too much iron can lead to life-threatening conditions, such as liver disease, heart problems and diabetes.

•TOPIC: Treatment for iron overload

Is there treatment for iron overload?

Yes. There are treatments to reduce iron overload, commonly using iron chelators. Iron chelation therapy involves taking oral or injected medicine to remove excess iron from the body. Medications can include desferroxamine, deferiprone, deferasirox, to be administered throughout the life period after the 10 transfusions.

•TOPIC: Blood and bone marrow transplant and risk

Is there a cure for sickle cell disease?

Cures through stem cell therapy and gene therapy are being explored. Stem cell transplantation is well established, while gene therapy is still being experimented upon. Stem cell therapy requires a relative who is a close enough genetic match to be a donor. Because of the significant risks involved, they are not done often.

•TOPIC: Gene therapy and risk

Can gene therapy create a world without sickle cell disease?

Is possible, gene therapy can cure SCD, but availability, accessibility, and treatment cost make the treatment unavailable in most countries, especially low- and middle-income countries (LMICs). Also, there are some side effects/risks including allergic reactions, cancers, damage to the organs or tissues etc. However, there is the need for Government, NGOs and partners' collaborations, investment, and also provision of the adequate facilities to support treatment.

NOTES

NOTES

10.5281/zenodo.15533631

Creative Commons Attribution 4.0 International
The Creative Commons Attribution license allows re-distribution and re-use of a licensed work on the condition that the creator is appropriately credited.

Author: ALIYU TAMBAYA MANDE

Graphic Design:
ESTHER GANI
GIANFRANCO BONADIES
MARIA CAVALLO

Thanks to ROSA CONTE, FRANCESCA BASILE and LUCIA RUGGIERI for the editorial review.

This document reflects only the authors' view and the EU Research Executive Agency (REA) is not responsible for any use that may be made of the information it contains.

It has been developed within the African Research and Innovative Initiative for Sickle cell Education: Improving Research Capacity for Service Improvement project (ARISE). ARISE has received funding from the European Union's Horizon 2020 research and innovation programme under the Marie Skłodowska-Curie grant agreement No. 824021.