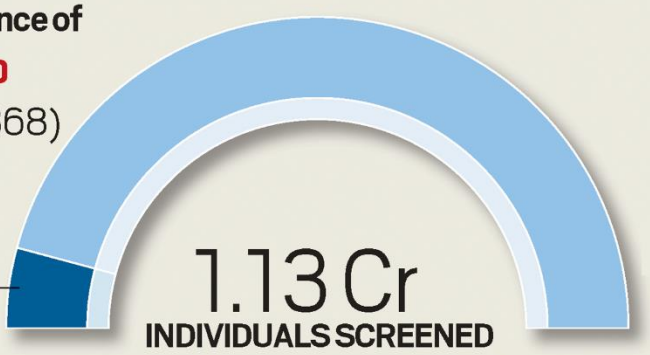
SICKLE CELL

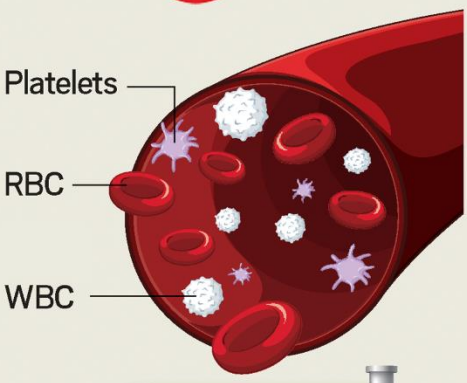
Studied and Presented by Rtn Hassan Mayet. Rotary Club Chikhli River Front

SICKLE CELL ANAEMIA (SCA) IN INDIA

The Indian EXPRESS

Prevalence of **8.75%** (9,96,368)





In 2011, a nationwide screening by the Ministry of Tribal Affairs and Indian Council of Medical Research (ICMR) covered 1,13,83,664 individuals

SCD Support Corner data

Carriers **9,49,057**

SCA **47,311**

An estimated 30,000-40,000 children are born with the disorder in the country each year

15 states in India have the highest incidence of the disease, with **MAHARASHTRA** topping the chart

WHAT IS SCA?

A genetic condition, Sickle Cell Anaemia (SCA) affects haemoglobin, which is responsible for carrying oxygen in red blood cells. SCA can result in severe anaemia and even death.

SCA symptoms usually appear by the age of six

Extreme pain

Fatigue

Swelling of hands and legs

OTHERS SYMPTOMS: Severe anaemia, Pain due to bone and joint damage, Ulcers, Frequent infections

TREATMENT

Bone marrow transplant, which is an expensive option, is currently the only cure for this disease. If diagnosed early, patients can lead a healthy life with pain medication (as needed), drinking 8 to 10 glasses of water each day. Blood transfusion and prescription medication

Source: Sickle Cell Anaemia Support Corner data

Graphic: IE Design

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Sickle Cell Disease is a group of inherited red blood cell disorders that affect hemoglobin, the protein that carries oxygen through the body. Normally, red blood cells are disc-shaped and flexible enough to move easily through the blood vessels. In sickle cell disease, red blood cells become crescent- or "sickle"-shaped due to a genetic mutation. These sickled red blood cells do not bend or move easily and can block blood flow to the rest of the body.

The blocked blood flow through the body can lead to serious problems, including stroke, eye problems, infections, and episodes of pain called pain crises.

Sickle cell disease is a lifelong illness. A bone marrow transplant is currently the only cure for sickle cell disease. Gene therapy is also being explored as another potential cure, but there are other effective treatments that can reduce symptoms and prolong life. If you have sickle cell disease, your healthcare team will work with you on a treatment plan to reduce your symptoms and manage the condition.

The condition affects more than 100,000 people in the United States and 20 million people worldwide. In the United States, most people who have sickle cell disease are of African ancestry or identify themselves as Black.

Sickle cell disease is caused by an abnormality of the β-globin gene and is characterised by sickling of the red blood cells. Globally, sub-Saharan African countries share the highest burden of the disease. Sickle cell disease is an inherited autosomal recessive disorder of the β-globin gene characterised by clinical manifestations such as haemolytic anaemia and recurrent episodes of vascular occlusion. Sickle cell disease refers to a group of inherited red blood cell disorders, with sickle cell anaemia indicated as one of the disorders. Disease presentation and progression in patients with sickle cell anaemia is highly variable. Individual differences in clinical presentations are also believed to be dependent on environment, the extent of sickling, vascular endothelium, platelets, leucocytes, and plasma proteins.

To achieve this, government need to adopt a proactive strategy in addressing gaps that have been identified in this study, as well as instituting other relevant measures, such as continuous media engagement and public health interventions relating to genetic counselling. Reforms in areas that can help reduce the disease burden, include training of practitioners and equipping sickle cell disease treatment centers according to World Health Organization specifications.

Sickle cell is an inherited disease caused by a defect in a gene.

- A person will be born with sickle cell disease only if two genes are inherited—one from the mother and one from the father.

- A person who inherits just one gene is healthy and said to be a "carrier" of the disease. A carrier has an increased chance of having a child with sickle cell disease if he or she has a child with another carrier.

For parents who are each carrier of a sickle cell gene, there is a 1 in 4, or a 25 % chance of having a child with sickle cell disease.

SYMPTOMS

The following is a list of symptoms and complications associated with sickle cell disease. However, each person may experience symptoms differently. Symptoms and complications may include:

- **Anaemia.** Because sickled cells are short-lived or destroyed, there are less red blood cells available in the body. This results in anaemia. Severe anaemia can make you feel dizzy, short of breath, and tired.
- **Pain crisis, or sickle crisis.** This occurs when the flow of blood is blocked to an area because the sickled cells have become stuck in the blood vessel. The pain can occur anywhere, but most often occurs in the chest, arms, and legs. Infants and young children may have painful swelling of the fingers and toes. Interruption in blood flow may also cause tissue death.
- **Acute chest syndrome.** This occurs when sickling occurs in the chest. This can be life-threatening. It often occurs suddenly, when the body is under stress from infection, fever, or dehydration. The sickled cells stick together and block the flow of oxygen in the tiny vessels in the lungs. It resembles pneumonia and can include fever, pain, and a violent cough.
- **Splenic sequestration (pooling).** Crises are a result of sickle cells pooling in the spleen. This can cause a sudden drop in haemoglobin and can be life-threatening if not treated promptly. The spleen can also become enlarged and painful from the increase in blood volume. After repeated episodes, the spleen becomes scarred, and permanently damaged. Most children, by age 8, do not have a working spleen either from surgical removal, or from repeated episodes of splenic sequestration. The risk of infection is a major concern of children without a working spleen. Infection is the major cause of death in children younger than age 5 in this population.
- **Stroke.** This is another sudden and severe complication of people with sickle cell disease. The misshapen cells can block the major blood vessels that supply the brain with oxygen. Any interruption in the flow of blood and oxygen to the brain can result in severe brain damage. If you have one stroke from sickle cell anaemia, you are more likely to have a second and third stroke.

- **Jaundice, or yellowing of the skin, eyes, and mouth.** Jaundice is a common sign and symptom of sickle disease. Sickle cells do not live as long as normal red blood cells and, therefore, they are dying faster than the liver can filter them out. Bilirubin (which causes the yellow colour) from these broken down cells builds up in the system causing jaundice.
- **Priapism.** This is a painful obstruction of the blood vessels in the penis by sickle cells. If not promptly treated, it can result in impotence.

The symptoms of sickle cell disease may look like other blood disorders or medical problems. Always consult your health care provider for a diagnosis.

SICKLE CELL DISEASE IN INDIA:

India has been ranked the country with the second highest numbers of predicted SCD births, with 42,016 [interquartile range (IQR): 35,347-50,919] new-borns estimated to have been born with sickle cell anaemia in 20104. SCD is characterized by considerable variability in clinical severity.

3% of India's tribal population suffers from sickle cell anaemia, another 23% is it carrier.

Sickle cell anaemia is a genetic disorder that cannot be cured but only be treated. Proper interventions, modern treatments and therapy of tribal communities in the Nilgiris, Tamil Nadu, have been found to lessen the disease burden over a period of time.

The normal lifespan of a sickle cell anaemia patient is approximately 40 years. Proper interventions and modern treatments and therapy have been found to lessen the burden of the disease over a period of time.

WORLD SICKLE CELL DAY

The United Nations recognises June 19 as World Sickle Cell Day to promote awareness on sickle cell anaemia and other sickle cell disorders, which are rare diseases affecting people across the world.

Closer home, in India, an estimated three per cent of the tribal population suffers from sickle cell anaemia and another 23 per cent carries and transmits the sickle cell gene to their children. Nearly all tribal communities living in forested terrains across the country, have cases of sickle cell anaemia.

How is sickle cell disease diagnosed?

Along with a complete medical history and physical exam, you may have blood and other tests. Many states routinely screen new-borns for sickle cell so that treatment can begin as soon as possible. Early diagnosis and treatment can reduce the risk of complications.

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Haemoglobin electrophoresis is a blood test that can determine if a person is a carrier of sickle cell, or has any of the diseases associated with the sickle cell gene.

TREATMENT:

Early diagnosis and prevention of complications is critical in sickle cell disease treatment. Treatment aims to prevent organ damage including strokes, prevent infection, and treat symptoms. Treatment may include:

- **Pain medications.** This is for sickle cell crises.
- **Drinking plenty of water daily (8 to 10 glasses).** This is to prevent and treat pain crises. In some situations, intravenous fluids may be required.
- **Blood transfusions.** These may help treat anaemia and prevent stroke. They are also used to dilute the sickled haemoglobin with normal haemoglobin to treat chronic pain, acute chest syndrome, splenic sequestration, and other emergencies.
- **Vaccinations and antibiotics.** These are used to prevent infections.
- **Folic acid.** Folic acid will help prevent severe anaemia.
- **Hydroxyurea.** This medication helps reduce the frequency of pain crises and acute chest syndrome. It may also help decrease the need for blood transfusions. The long-term effects of the medication are unknown.
- **Regular eye exams.** These are done to screen for retinopathy.
- **Bone marrow transplant.** Bone marrow transplants can cure some people with sickle cell disease. The decision to have this procedure is based on the severity of the disease and ability to find a suitable bone marrow donor. These decisions need to be discussed with your doctor and are only done at specialized medical centres.

COMPLICATIONS:

Any and all major organs are affected by sickle cell disease. The liver, heart, kidneys, gallbladder, eyes, bones, and joints can suffer damage from the abnormal function of the sickle cells and their inability to flow through the small blood vessels correctly. Problems may include the following:

- Increased infections
- Leg ulcers
- Bone damage
- Early gallstones
- Kidney damage and loss of body water in the urine
- Eye damage
- Multiple organ failure

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LIVING WITH SICKLE CELL:

- Sickle cell disease is a life-long condition. Although the complications of sickle cell disease may not be able to be prevented entirely, living a healthy life-style can reduce some of the complications.
- It is important to eat a healthy diet with lots of fruits, vegetables, whole grains, and protein, and drink lots of fluids.
- Do not take decongestants because they cause constriction of blood vessels and could trigger a crisis.
- Other factors that may trigger a crisis include high altitudes, cold weather, swimming in cold water, and heavy physical labour.
- Avoid infections by getting an annual flu shot, washing your hands frequently, avoiding those who are sick, and getting regular dental exams.

THE FOLLOWING POINTS NEED TO BE KEPT IN MIND WHILE RESPONDING TO SICKLE CELL ANAEMIA:

- Sickle cell anaemia cannot be cured but can be treated. Regular awareness drives and camps can educate the people about the sickle cell and its effects.
- Marriage counselling will go a long way in arresting the spread of the disease.
- Screening all age groups is essential from new-born to the old, with emphasis on the adolescents, antenatal women and new-born.

- When screening antenatal women for sickle cell diseases, husbands may also be screened to ensure the child will not be at risk. If a foetus is found to have sickle cell disease legal medical termination of pregnancy may be advised.
- Rehabilitation of sickle cell patients' needs attention with compensation and suitable employment opportunities provided.