



## Sickle Cell Disease

**For Prelims:** Biotechnology, [Sickle Cell disease](#), Genetic disorder, [Tribal communities](#)

**For Mains:** Challenges and Governmental Initiatives faced by the [tribal communities](#) relating to the treatment and accessibility of sickle cell disease (SCD)

[Source: TH](#)

### Why in News?

Amidst the unavailability of essential drugs to treat [Sickle Cell Disease \(SCD\)](#) at district healthcare institutions, there is growing concern about the challenges faced by people from marginalised Indigenous Tribal communities in managing the treatment of SCD.

### What is Sickle-Cell Disorder?

#### ▪ About:

- [Sickle Cell Disease \(SCD\)](#) is an **inherited haemoglobin disorder** characterised by a **genetic mutation** that **causes red blood cells (RBCs) to assume a sickle or crescent shape** rather than their normal round shape.
- This **abnormality** in RBCs **results in increased rigidity, impairing their ability to circulate effectively** throughout the body. Consequently, individuals with SCD often experience complications such as **anaemia, organ damage, recurrent and severe pain episodes, and a shortened lifespan.**
- As per the Ministry of Health and Family Welfare, marginalised tribal populations are most vulnerable to SCD.

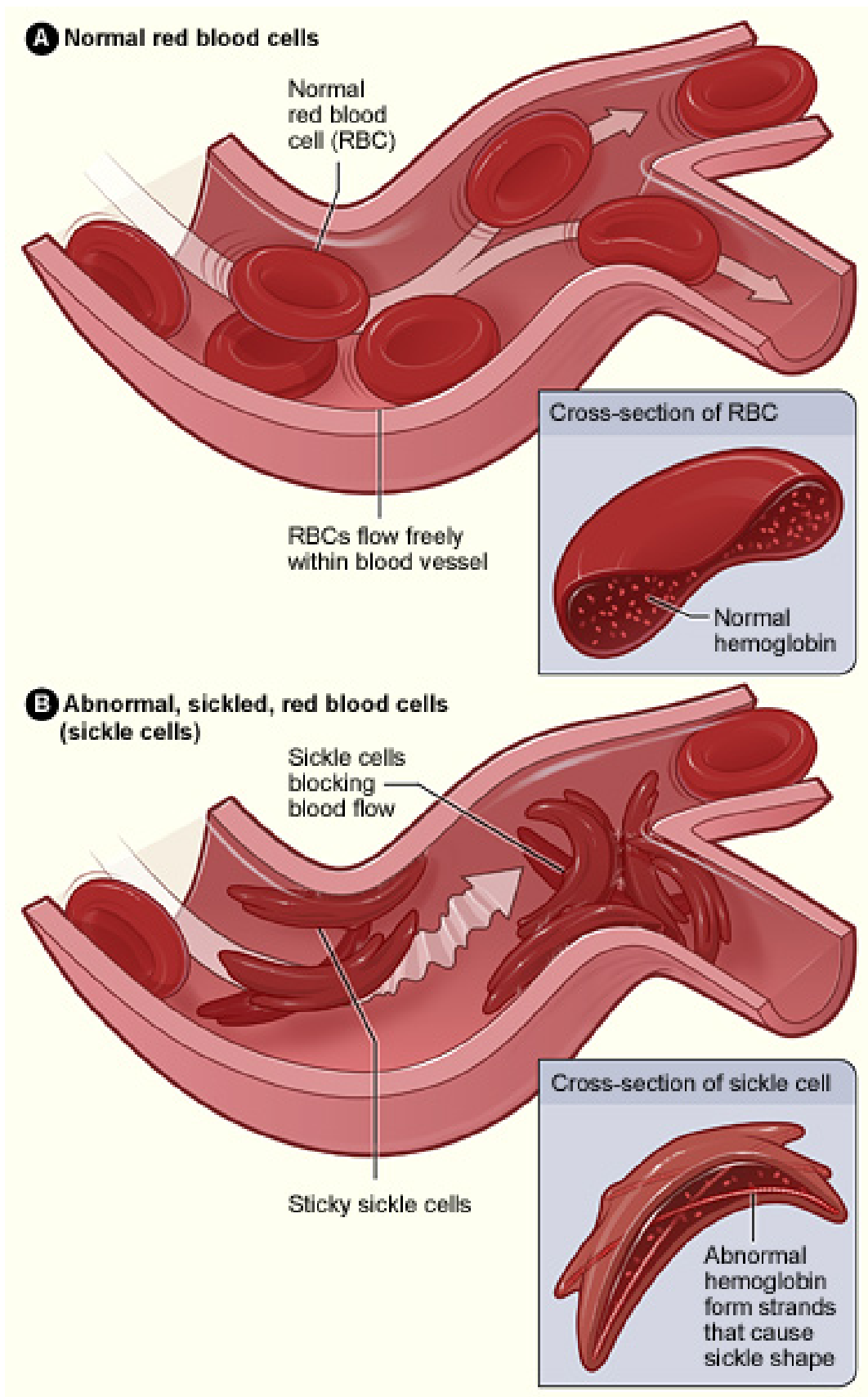
#### ▪ Symptoms: Symptoms of sickle cell disease can vary, but some **common symptoms** are-

- [Chronic anaemia](#) which leads to fatigue, weakness, and paleness.
- Painful episodes (also known as **sickle cell crisis**) cause sudden and intense pain in the bones, chest, back, arms, and legs.
- **Delayed growth and puberty.**

#### ▪ Treatment Processes:

- **Blood Transfusions:** These can help relieve anaemia and reduce the risk of pain crises.
- **Hydroxyurea:** This medication can help **reduce the frequency of painful episodes** and prevent some of the disease's long-term complications.
- **Gene Therapy:** It can also be treated by bone marrow or [stem cell](#) transplantation by methods like [Clustered regularly interspaced short palindromic repeats \(CRISPR\)](#).

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## What is India's Current Status of Sickle Cell Disease (SCD)?

- **India ranks third globally** in terms of the **number of SCD births**, following Nigeria and the [Democratic Republic of the Congo](#).
- Regional studies indicate that an **estimated 15,000 to 25,000 babies** with SCD are born in India annually.

- Most of **these births occur in tribal communities**, highlighting the geographical and socioeconomic disparities in healthcare access and awareness.

## What are the Challenges Related to the Treatment and Accessibility of SCD?

- **Limited Awareness:** There is a **lack of understanding about SCD** among the public and healthcare providers, leading to delayed diagnosis and inadequate treatment.
- **Inadequate Healthcare Infrastructure:** Many rural and tribal areas lack specialised healthcare facilities and trained medical personnel for managing SCD.
- **High Treatment Costs:** The long-term management of SCD can be financially burdensome for many families due to the cost of medications, regular check-ups, and potential hospitalizations.
  - For Example, **Treatments like CRISPR cost \$ 2-3 million**, and it's difficult to find bone marrow donors.
- **Limited Access to Medications:** Inconsistent availability of essential medications for SCD treatment, such as **hydroxyurea and pain relievers**, is a concern in certain regions.
- **Inadequate Screening Programs:** The **absence of systematic newborn screening** and early detection initiatives results in missed opportunities for early intervention and genetic counselling.
- **Geographical and Socioeconomic Barriers:** Rural, **remote, and tribal communities** face challenges in accessing quality healthcare due to geographical isolation, lack of transportation, and socioeconomic factors.
  - **Stigma and Discrimination** further hinder access to healthcare services.

## What are the Government Initiatives Regarding SCD?

- **National Sickle Cell Anaemia Elimination Mission:**
  - Aimed at enhancing the care for all **Sickle Cell Disease (SCD)** patients and reducing the disease's prevalence through an integrated approach **encompassing screening and awareness campaigns**.
  - Targeting **complete elimination of sickle cell disease as a public health concern by 2047**.
    - Under the Sickle Cell Anaemia Mission, **the Council of Scientific and Industrial Research (CSIR)** is developing gene-editing therapies for SCD.
- **National Health Mission (NHM) 2013:**
  - It is, a flagship programme of the Indian government, that encompasses provisions for disease prevention and management, with a **specific focus on hereditary anomalies such as sickle cell anaemia**.
  - Dedicated programs within NHM focus on **raising awareness, facilitating early detection, and ensuring timely treatment** of sickle cell anaemia.
  - NHM facilitates drugs like hydroxyurea to treat SCD in its **“essential medicines List”**.
- **The National Guidelines for Stem Cell Research 2017:**
  - It **restricts the commercialisation of stem cell therapies** to clinical trials, except for Bone marrow transplantation (BMT) for SCD.
  - Gene editing on stem cells is permitted only for in-vitro studies.
- **National Guidelines for Gene Therapy Product Development and Clinical Trials 2019:** It provides guidelines for the development and clinical trials of gene therapies for inherited genetic disorders.
  - India has also approved a **five-year project** to develop CRISPR techniques for sickle cell anaemia treatment.
- **State Haemoglobinopathy Mission of Madhya Pradesh:**
  - It aims to address the challenges in screening and management of the disease.
- **Rights of Persons with Disabilities (RPwDs) Act, 2016:**
  - **SCD is included in the 21 disabilities** that provide for benefits such as **reservation in higher education (minimum 5%), government jobs (minimum 4%), and allocation of land (minimum 5%)**, for persons with benchmark disabilities and those with high support needs.
  - **Free education** is guaranteed for every child with a benchmark disability between 6 and 18 years.

## Note

- Recently, the **US Food and Drug Administration (FDA)** approved **two gene therapies** designed for sickle cell disease.
- The approved therapies include **Lyfgenia** and **Casgevy**.
  - Both treatments received clearance for individuals aged 12 years and above.
  - **Casgevy also approved in the U.K.**, is the **first CRISPR-based therapy** to have received regulatory approval.
  - **Lyfgenia** doesn't use CRISPR **but depends on a viral vector to change blood stem cells**.
- **Both treatments** entail collecting a patient's blood stem cells, modifying them, and **administering high-dose chemotherapy to destroy the damaged cells** in the bone marrow.
- The modified cells are then infused into the patient through a hematopoietic stem cell transplant.

## Way Forward

- **Early Detection and Screening:**
  - Strengthen and expand **genetic counselling and testing programmes**.
  - **Prioritising basic treatments like hydroxyurea** is essential for urgent needs.
  - Identify carriers at an early stage to provide essential information to affected families.
  - **Ensuring equitable access** to clinical trials is important to address the issue from the grassroots level.
- **Public Education and Awareness:**
  - Implement sustained public awareness initiatives.
  - Educate communities about the disease's hereditary nature and the importance of genetic testing.
  - Public involvement in regulatory discussions is necessary.
- **Research and Development:**
  - Allocate resources for ongoing research.
  - Gain deeper insights into the genetic and molecular aspects of SCD to develop more effective treatment options and potential cures.
  - Comprehensive healthcare access is vital for better long-term health outcomes.

## UPSC Civil Services Examination Previous Year Question (PYQ)

### Prelims:

**Q. Consider the following statements in the context interventions being undertaken under Anemia Mukt Bharat Strategy : (2023)**

1. It provides prophylactic calcium supplementation for pre-school children, adolescents and pregnant women.
2. It runs a campaign for delayed cord clamping at the time of child-birth.
3. It provides for periodic deworming to children and adolescents.
4. It addresses non-nutritional causes of anaemia in endemic pockets with special focus on malaria, hemoglobinopathies and fluorosis.

**How many of the statements given above are correct?**

- (a) Only one
- (b) Only two
- (c) Only three
- (d) All four

**Ans: (c)**

**Mains:**

**Q.** What are the research and developmental achievements in applied biotechnology? How will these achievements help to uplift the poorer sections of society? **(2021)**

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