Sickle Cell Disease and Newborn Screening Information Resource

Sickle cell disease (SCD) is a genetic condition which causes abnormal haemoglobin (HbS) and results in complications such as vaso-occlusion, infection, and organ damage.[1] [2] Sub-Saharan Africa (SSA) carries the greatest burden of SCD globally, with recent estimates suggesting that approximately 2.75 million (1.5%) children under 5 years were living with SCD in the region in 2023.[3] It is a significant contributor to mortality rates in children under 5 years of age in Africa, accounting for about 7.3 deaths per 100 child-years.[4] Despite this burden, newborn screening (NBS) and access to comprehensive SCD care remain limited across much of the region.[3] [4]

The World Health Organisation recognises SCD as a major public health priority. Through its 2006 resolution, countries were urged to implement NBS programmes and integrate SCD care into maternal and child health services.[5] Countries such as the United States of America and the United Kingdom have universal NBS programmes for SCD, leading them to achieve survival rates into adulthood of over 90%.[6]

The Consortium on Newborn Screening in Africa is a multi-country initiative led by the American Society of Hematology to expand and strengthen NBS for SCD across SSA, such as Ghana, Kenya, Nigeria, and Tanzania.[7]

Purpose of NBS

Why screen newborns?

- Early screening of newborns (ideally within 24-72 hours of birth or at the earliest opportunity) helps to detect SCD before symptoms develop, as most affected infants appear healthy at birth. This will, in turn, enable early, life-saving interventions that significantly reduce morbidity and mortality.

Key benefits of NBS:

- Early initiation of preventive treatment:
    - Penicillin prophylaxis: Infants with SCD are at high risk of bacterial infections. Penicillin prophylaxis initiated in early childhood reduces the risk of infections and significantly lowers mortality.[8]
    - Vaccination: Children with SCD require both routine childhood immunisations and additional targeted vaccines due to increased infection risk. Vaccination schedules include pneumococcal, *Haemophilus influenzae* type b (Hib), and meningococcal vaccines.
- Improved treatment adherence and clinical outcomes:
    - Early enrollment into comprehensive care programmes. This may include regular clinic follow-ups, vaccination programmes, infection prophylaxis (e.g., with penicillin), hydroxyurea therapy, pain management, laboratory monitoring, patient and/or caregiver education, and multidisciplinary care involving physicians, nurses, and pharmacists, delivered through specialist clinics. This leads to a reduction in severe infections, hospitalisations and further complications.[4]
    - Caregivers are taught to recognise early warning signs (e.g., fever, lethargy, pallor, difficulty breathing, and left-sided abdominal swelling or pain suggestive of splenic enlargement) and seek prompt medical care. Caregiver education increases understanding of the disease, the importance of the medications, and vaccinations, which also improves treatment adherence and long-term outcomes.

Impact:

- In high-income countries, survival into adulthood in patients with SCD now exceeds 90% as a result of universal NBS.[6] In low- and middle-income countries (LMICs), however, the lack of screening means children are often diagnosed late or not at all. Implementation of NBS programmes can dramatically reduce early deaths by enabling early detection and linkage to care.

NBS Process

Figure 1: **Heel prick blood sample collection**[9]

A small blood sample is collected using a heel prick (or a fingerstick in some settings) (Fig. 1). Blood is either:

- Placed on a filter paper for laboratory testing, or
- Used directly for point-of-care testing (POCT)

Timing:

- This is ideally performed 24-72 hours after birth.
- It can also be done before discharge from the hospital or during early postnatal visits (if birth occurs at home).
- In practice, timing may vary depending on local healthcare systems, birth setting, and access to postnatal services.
- In LMIC settings, particularly where home births or delayed postnatal attendance occur, screening may be performed at the earliest available healthcare contact (e.g., during early postnatal or immunisation visits).

Testing methods:

Purpose of test	Screening tests	Confirmatory reference tests (used to confirm SCD diagnosis)		
Feature	**POCT (rapid tests)** e.g., HemoTypeSC™, Sickle SCAN®, Gazelle™ Hb Variant Test[10 11]	**Hb electrophoresis**	**High-performance liquid chromatography (HPLC)**	**Isoelectric focusing (IEF)**
Principle	Immunoassay detecting Hb variants	Separation by electrical charge	Separation by retention time (chromatography)	Separation by isoelectric point (pH)
Accuracy	Requires confirmatory testing	High	Very high	Very high
Key advantage	Fast, portable	Widely available, reliable	Quantitative and highly sensitive	Excellent resolution, ideal for newborns
Limitation	Not definitive, needs a confirmatory test	May have lower resolution than IEF	Expensive equipment	Requires specialised equipment

NB: Sickle cell solubility (sickling) test is NOT suitable for NBS because high levels of foetal Hb interfere with the detection of HbS.

POCT provides rapid initial screening in primary care settings, while confirmatory reference tests are laboratory-based and provide a definitive diagnosis.

Figure 2: **POCTs for rapid sickle cell screening**

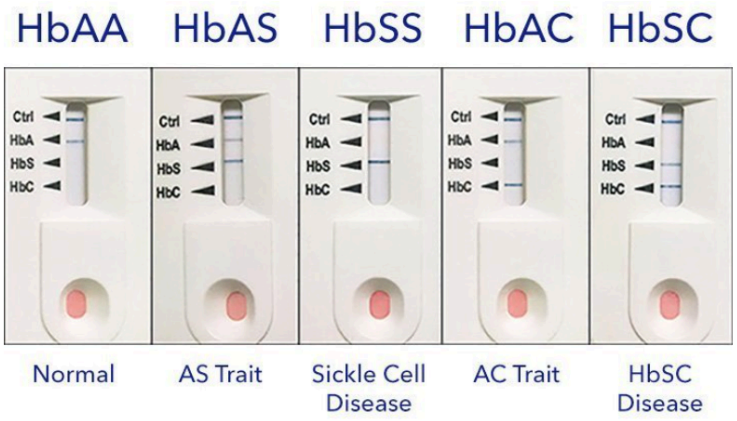

Figure 2a: **Sickle SCAN®**[12]

Figure 2b: **Gazelle™ Hb Variant Test**[13]

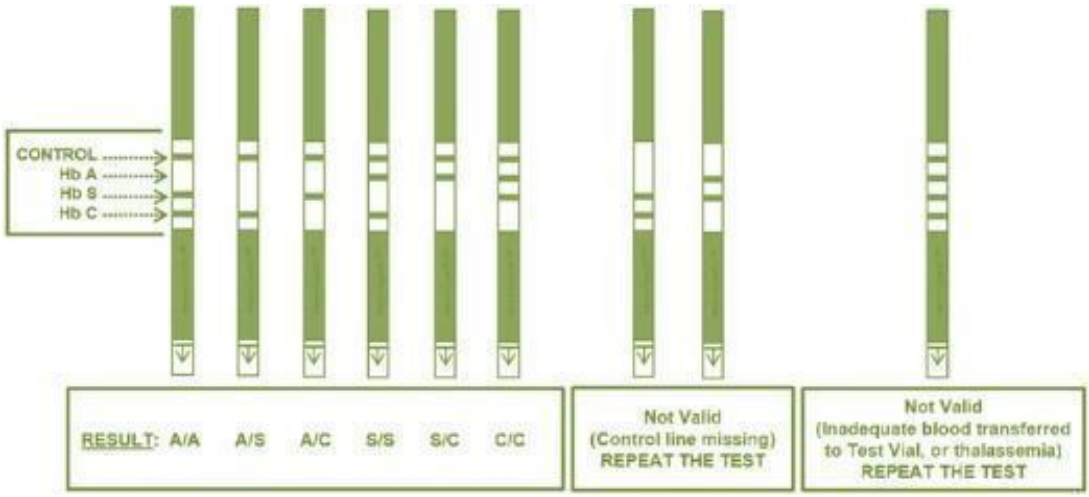

Figure 2c: **HemoTypeSC™**[14]

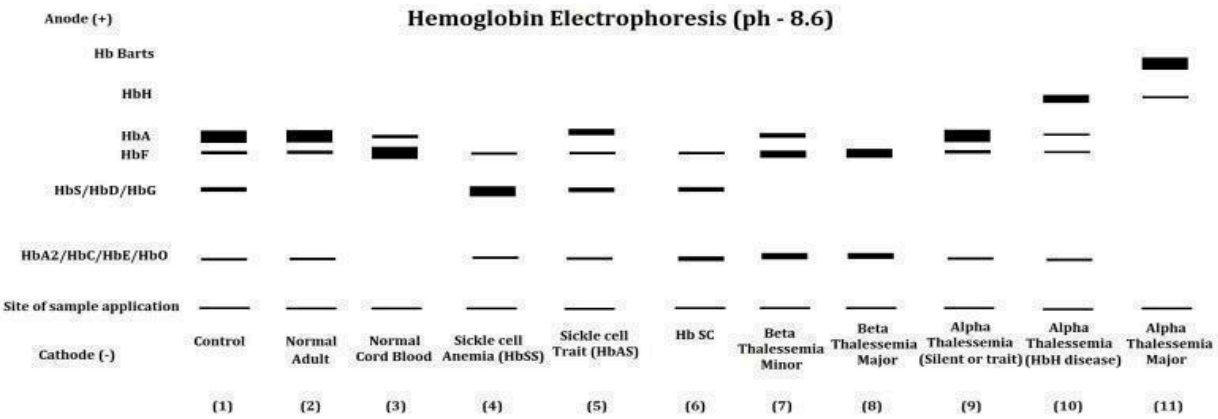

Figure 3: **Hb electrophoresis**[15]

Interpretation of the results:

Normal (HbAA)	**No disease**
Sickle cell trait (HbAS)	**Carrier (no disease, but genetic counselling needed)**
SCD (HbSS or others)	**Possible SCD, confirmatory testing and referral for comprehensive care required**

Follow-up care:

- Following a positive POCT result (e.g., HbSS, HbSC, HbSβ-thalassaemia), patients should undergo confirmatory testing (eg, HPLC or Hb Electrophoresis) and be referred to an SCD care team, which may include a paediatrician, haematologist, primary care clinician/family doctor, specialist SCD clinic, nurses, pharmacists, and laboratory services.
- Following confirmation of diagnosis, early interventions such as penicillin prophylaxis, vaccination, malaria prophylaxis (where applicable), and folic acid supplementation are required. Hydroxyurea therapy may form part of longer-term comprehensive SCD management where clinically appropriate and in accordance with national guidelines, local protocols, and monitoring capacity.
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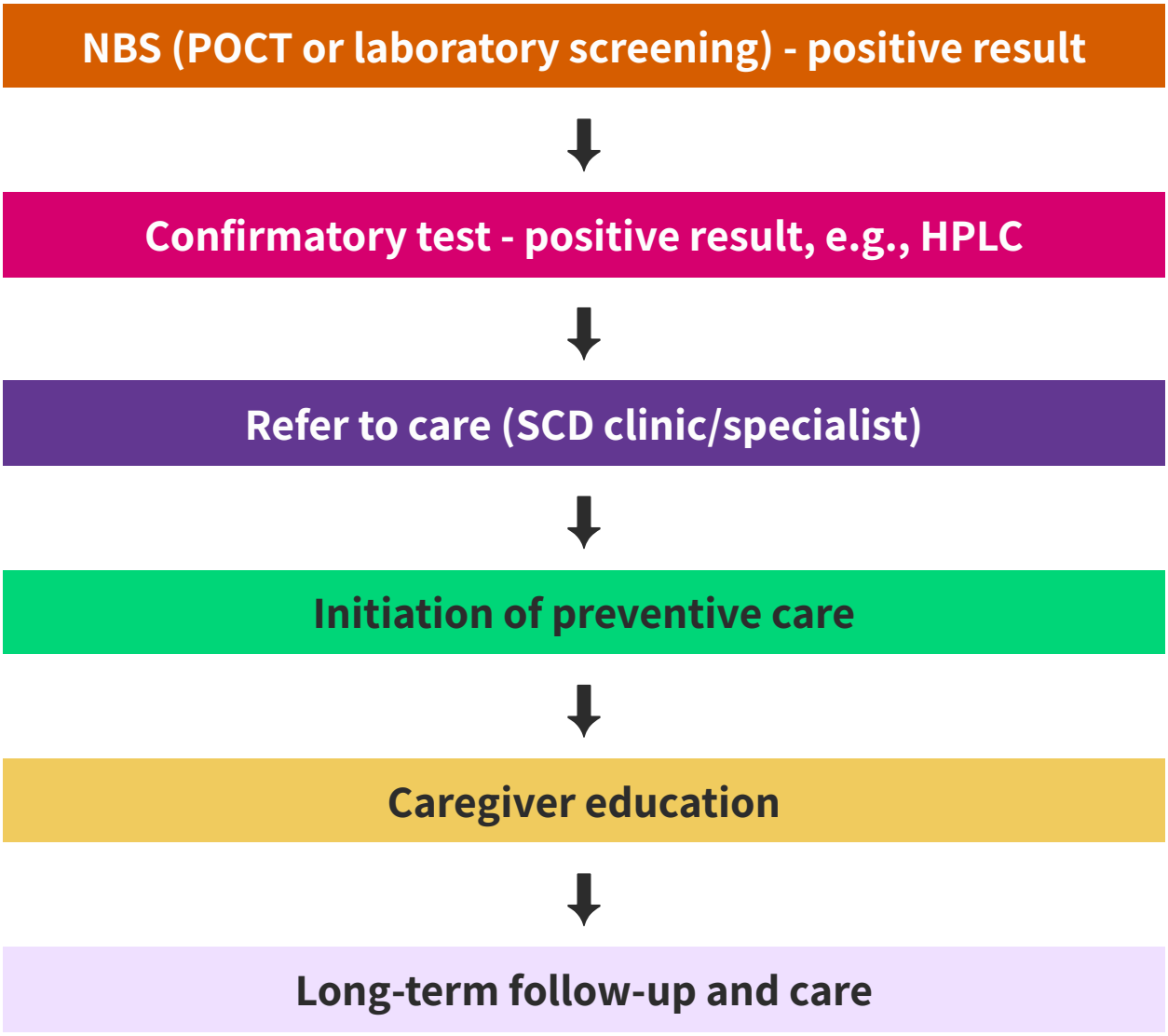
NBS (POCT or laboratory screening) - positive result
Confirmatory test - positive result, e.g., HPLC
Refer to care (SCD clinic/specialist)
Initiation of preventive care
Caregiver education
Long-term follow-up and care

Role of the Pharmacist

- Promote awareness of NBS within the communities.
    - Early identification and referral: Recognise infants, children or families who may benefit from sickle cell screening and refer them for further assessment.
- Medicine optimisation and adherence support:
    - Support safe and effective use of medicines such as penicillin prophylaxis, folic acid supplementation, malaria prophylaxis (where applicable), and hydroxyurea treatment.
    - Monitor for potential adverse effects, drug interactions, and adherence challenges. Work collaboratively with other healthcare professionals to optimise therapy and address medicine-related problems.
- Medication counselling and the importance of adherence:
    - Educate caregivers on correct dosing, administration, adverse effects, and storage of medication.
    - Reinforce the importance of adherence to prophylactic therapies, clinic appointments, and vaccination schedules, and address barriers such as missed doses or misconceptions.
- Caregiver education and counselling:
    - Educate caregivers about SCD: how it is inherited, the importance of screening, early diagnosis, and follow-up.
    - Teach patients and caregivers to recognise early warning signs or red flags such as fever, lethargy, breathing difficulty, severe pain, pallor, or splenic enlargement.
- Improve access to essential medicines and vaccines:
    - Help ensure availability and appropriate storage of essential medicines and vaccines through effective inventory management, forecasting, and supply chain monitoring.
    - Participate in innovative medicine access programmes such as revolving fund pharmacies, which help maintain a sustainable supply of affordable medicines in resource-limited settings.

NBS is a key strategy for the early identification of SCD, allowing affected infants to be diagnosed before symptoms develop. Positive screening results should be followed by confirmatory testing and timely referral to comprehensive SCD care services.

Early preventive interventions, including penicillin prophylaxis, vaccination and caregiver education, can significantly reduce morbidity and mortality. Long-term management requires coordinated multidisciplinary care, regular monitoring, and ongoing support for patients and families. Pharmacists contribute to this process through patient or caregiver counselling, medicines optimisation, adherence support, referral, and by helping to ensure ongoing access to essential medicines and vaccines.

Awareness:

- **World Sickle Cell Day** (June 19)
  2026 theme: **"Closing the Survival Gap: Equity in Sickle Cell Care"**

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Document Revision and Approval

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