ICMR - Centre for Research, Management and Control of Haemoglobinopathies, Chandrapur

# Newborn Screening Approaches for Sickle Cell Disease : Evaluation and Outcome

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## Introduction

- There is no national neonatal screening programme for Sickle Cell Disease (SCD)in India.
- Since many children get identified only when they become ill, hospitalized and symptomatic.
- There is high risk of morbidity and mortality during the first three years of SCD newborn's life.
- Early identification and intervention may reduce the morbidity and mortality rate and improves their quality of life.

## Aim and Objectives

- To **undertake a newborn screening program** for SCD in the tribal population in Central India for early detection to understand the **magnitude of the problem.**
- To measure the **benefit of early comprehensive care** of the affected babies
- To observe the role of **genetic modifiers with** disease severity.

## Method

- Prospective, Interventional, Follow up study from August 2019 to October 2022 in the Chandrapur and Gadchiroli, Maharashtra,Central India.

Identified SCD babies by cord samples and tested by HPLC and Molecular diagnosis

Family screening at home of SCD babies

Enrolment of SCD newborn for early intervention

Vaccination, Antibiotic Prophylaxis, Folic acid, HU, if required.

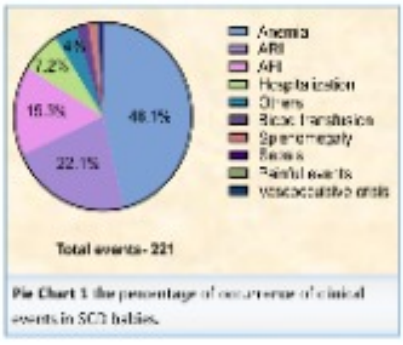
Complete Blood count, HPLC, LFT, RFT




## Results

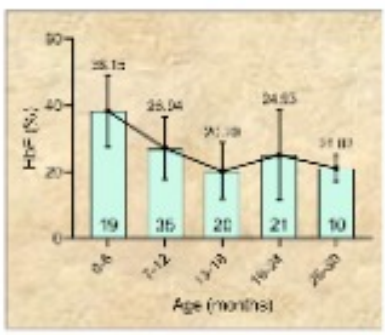
### Clinical Analysis

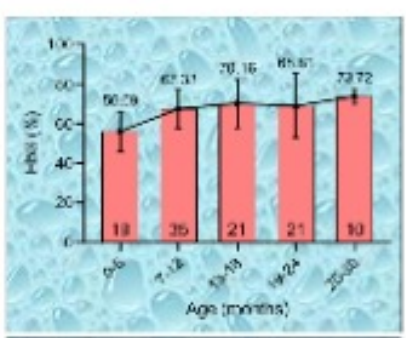
| | |
|---|---|
| Total screened0 | 14503 |
| AA | 13183 (91%) |
| AS | 1253 (8.6%) |
| Total SCD | 67 (0.4%) |
| SS | 58 (86.56%) |
| S β-THAL | 9 (13.43%) |
| Tribal | 37 (55%) |
| Non Tribal | 30 (45%) |
| M:F | 32:35 |

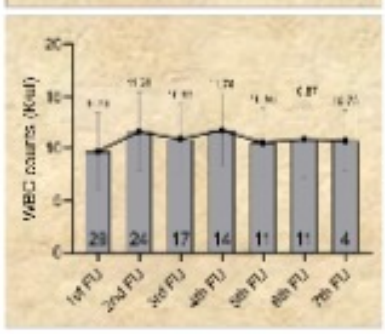


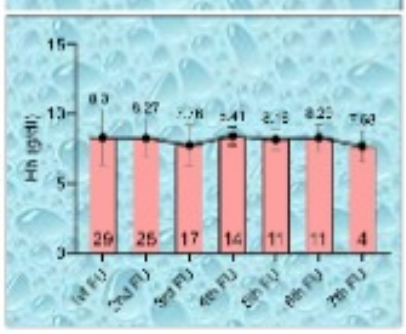

Pie Chart 1 the percentage of occurrence of clinical events in SCD babies.

### Laboratory Analysis

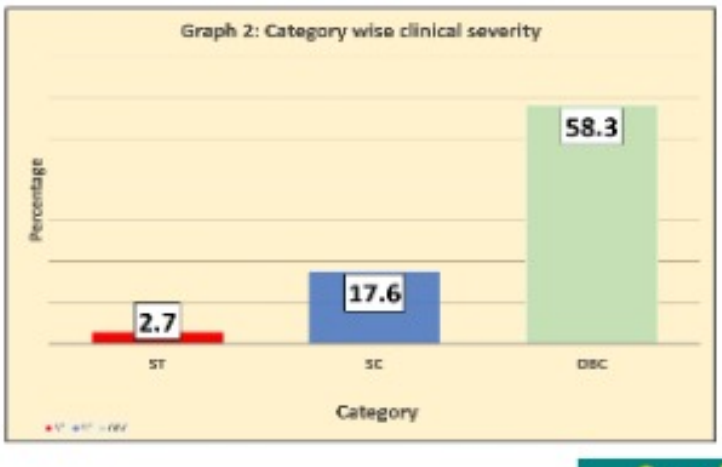


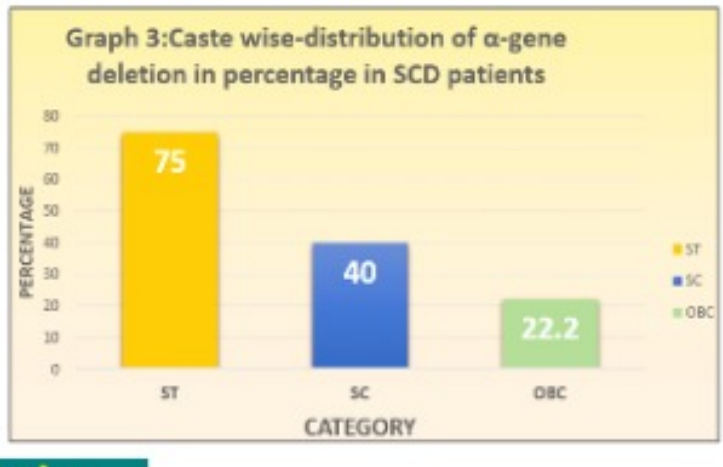




## Genetic Analysis


## Conclusion

- Total 67 (n=67) SCD newborn were identified out of 14503 newborn screening. HbSS-β-thalassemia - 14%
- The associated α-gene deletion was reported 58.5% of the cases. The associated α-gene deletion lead to amelioration of disease severity.
- As per the three years of observation of the study, there was no death reported. indicates the benefit of early intervention and babies.