ICMR-Hemoglobinopathies Satelite Centre, Chandrapur

# Assessment of Neonatal Screening Approaches for Sickle Cell Disease and the Effect of Early Intervention in Management of the Disease in Tribal Population

## INTRODUCTION

- There was an immense need of the national neonatal screening programme for Sickle Cell Disease (SCD) as many children get identified only when they become symptomatic.
- There is high risk of morbidity and mortality during the first 3 years of SCD newborns life.
- Early identification and intervention may improve their quality of life.

## AIM AND OBJECTIVES

**Primary**

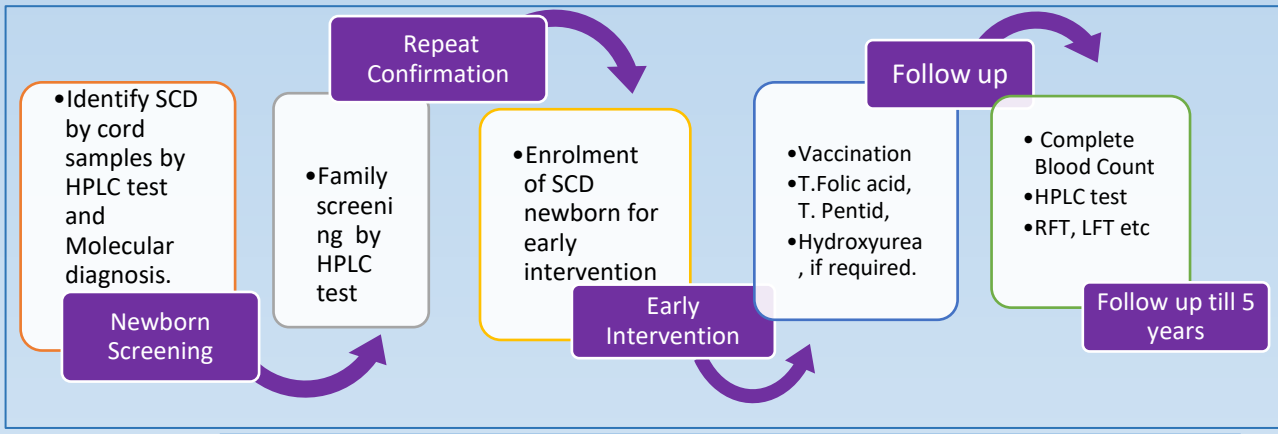
- To undertake a newborn screening program for SCD in tribal populations of different states for early detection, the magnitude of the problem and the barriers for undertaking such programme.
- To measure the benefit of early comprehensive care of affected babies.

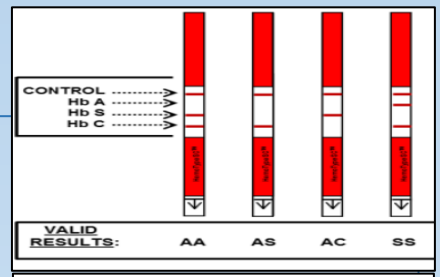
**Secondary**

- To evaluate the genotypic and phenotypic correlation to understand role of genetic modifiers for disease severity

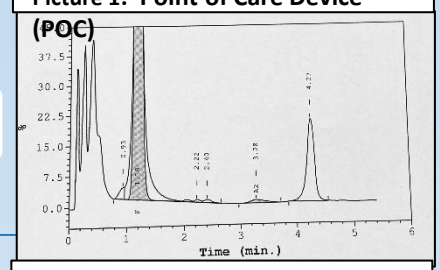
## MATERIAL AND METHODS

Multicentric, Prospective, Interventional, Follow up study from August 2019 to November 2021




Picture 1: Point of Care Device (POC)




Picture 2: HPLC Chromatogram

## RESULTS

- Newborn cord blood screening is done in two district hospitals **Chandrapur and Gadchiroli**

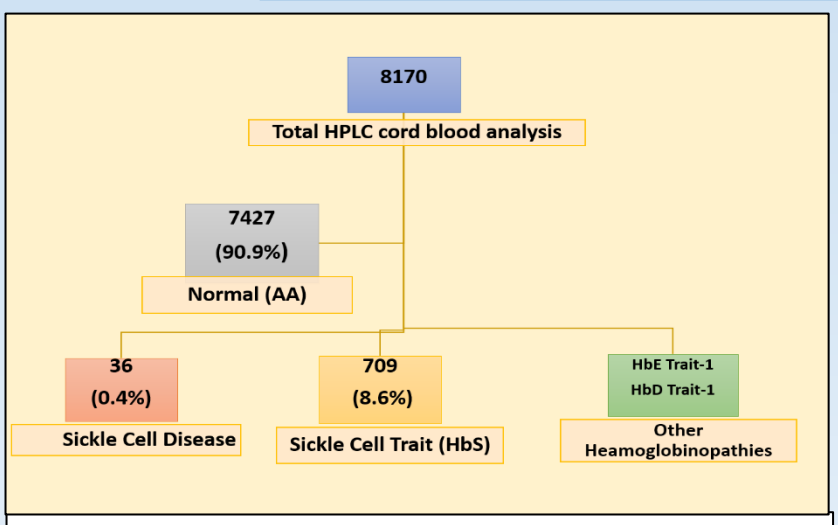
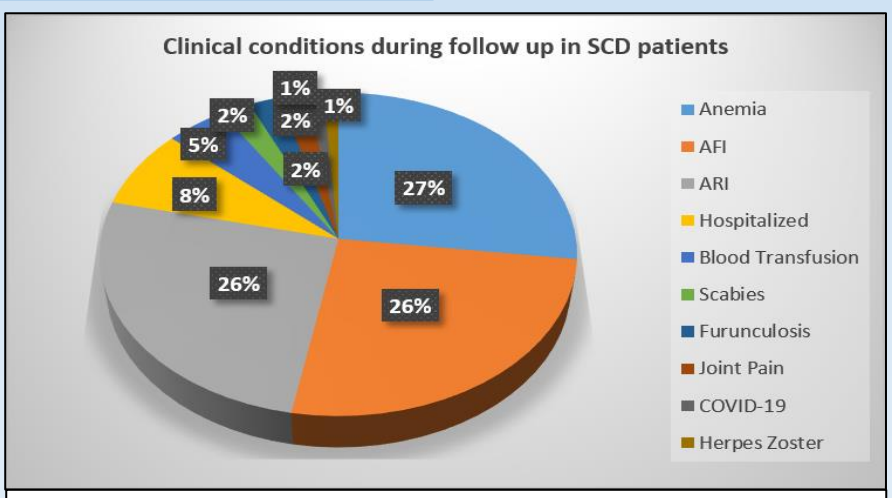



Figure 1: Detailing screening of the newborn cord blood HPLC analysis




Acute Respiratory Infection (ARI) and Acute Febril illness (AFI), HPLC- High Performance Liquid Chromatography, KFT- Kidney Function Test, LFT- Liver Function Test

Figure 2:Clinical conditions during follow up in SCD patients

## CONCLUSION

- 3/36 (8.3%) babies were compound heterozygous (sickle –β thalassemia), remaining 33/36 (91.7%) were homozygous.
- SCD newborns were found in both tribal and non- tribal population.
- The hospitalization rate was 8%, Blood Transfusion rate was 5%.
- Acute Respiratory Infection (ARI) and Acute Febril illness (AFI)-26% were more commonly seen.
- 80% of the babies are vaccinated as per their age. Prophylactic ,Tab. Folic acid, Tab. Pentid V 400 mg has shown lesser events of severe infections.
- Among the genetic modifiers associated α thalassemia was reported to be 47.2%. Xmn1(-158 C/T) polymorphism was reported to be +/+ was found to be prevalent (84.6%), followed by +/-(11.5%) and -/- (3.84%) in our cohort.