

## SICKLE CELL ANAEMIA AMONG DAHOD'S TRIBES: A GENETIC PERSPECTIVE

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### ABSTRACT

Sickle cell anaemia (SCA) is a widespread haemoglobinopathy, particularly high prevalence is observed in Gujarat. This study investigates the genotype distribution of SCA within the tribal community of Dahod District. The aim is to map genetic pattern among the tribal community so as to improve public health outcome and contribute towards a sickle cell-free India. Study was conducted at a tertiary care hospital which analysed 12,259 individual's data during June 2024 and August 2024 from ongoing study. The result indicates a 17.15% prevalence of SCA among the tribal community of Dahod District. A 14.55% Sickle cell trait (SCT-AS) genotype and 2.61% Sickle cell Disease (SCD-HbS- $\beta$ 0thalassemia genotype was noted in the study population. The frequency of SCT Hb-AS is approximately 5.5 times higher than that of SCD HbS- $\beta$ 0thalassemia among tribal population. Additionally, a gender disparity was observed, with a higher prevalence in females.

**KEYWORDS:** Hemoglobinopathies, Hemoglobin, Sickle Cell Anaemia, Sickle Cell Trait, Sickle Cell Disease, Hemoglobin S, Thalassemia, Tribal.

### INTRODUCTION

Sickle cell anaemia (SCA) is one of the most prevalent hemoglobinopathies globally, characterised by the presence of abnormal hemoglobin S (HbS) due to a genetic mutation in the  $\beta$ -globin gene.<sup>[1]</sup> This mutation leads to the production of sickle-shaped red blood cells, which can cause severe health complications. In India, the prevalence of SCA is notably high among tribal populations<sup>[2]</sup>, making it a significant public health concern.

The tribal communities in Dahod district of Gujarat, exhibit a high prevalence of sickle cell anaemia. Previous studies have documented varying prevalence rates of sickle cell carriers among different tribal groups, ranging from 1% to 40%.<sup>[3]</sup> In Gujarat, the prevalence of HbS among tribal populations ranges from 13% to 31%.<sup>[4]</sup> Specifically, surveys

conducted in 22 districts of Gujarat reported a sickle cell carrier prevalence rate of 11.37%.<sup>[5]</sup> Additionally, some tribal groups in South Gujarat show a high prevalence of HbS (6.3% to 22.7%) and the  $\beta$ -thalassemia trait (6.3% to 13.6%).<sup>[6]</sup> Given these statistics, there is a lot of disparity and no such study was conducted on the tribal community of Dahod, hence, it is crucial to study the prevalence and genotype of sickle cell anaemia in the tribal community of Dahod district.

Studying the genotype of sickle cell anaemia in the tribal community of Dahod district is not only scientifically significant but also crucial for improving public health outcomes. By addressing the genetic and epidemiological aspects of SCA, this research can contribute to better planning of genetic counselling and healthcare strategies, ultimately improving the quality of life for affected individuals.

## **MATERIALS AND METHOD**

The ongoing analytical study aims to screen and genotypically analyse positive cases of hemoglobinopathies in the tribal community of Dahod district. The study is being conducted at the Central Clinical Biochemistry Lab of Zydus Medical College and Hospital, Dahod. This ongoing study collected data over a period of three-months ranging from June 2024 to August 2024.

### **Participant registration**

Our cohort study uses a random sampling method for enrolling participants who possess records of the tribal community and are visiting our tertiary care hospital for their routine check-up or any other illness. The study population comprised 12,259 participants from the tribal community of Dahod district.

### **Inclusion criterion**

Individuals from both genders, above 01 year of age, having documented evidence of tribal community of Dahod District, Gujarat, and volunteered to participate in the study.

### **Exclusion criterion**

The participants not belonging to the tribal community of Dahod district, Gujarat. Participants with a history of blood transfusion in the past three months were excluded.

### **Sample Collection**

After obtaining informed written consent from each participant, 1.8 ml of venous blood was collected in EDTA vacuette by taking proper antiseptic precaution. The blood vacuette was kept on the rotator for proper mixing with anticoagulant to avoid clotting.

Whole blood samples were first screened for sickling by the Dithionite qualitative solubility test [NESTROFT method].<sup>[7,8]</sup> The presence of any haemolysis was recorded visually.

This was followed by the assessment of HbA0, HbF, HbS and HbA2 levels of haemoglobin fractions by preparing haemolysate and subjecting it on cellulose acetate paper based microchip Gazelle hemoglobin variant electrophoresis machine at alkaline pH.<sup>[9]</sup> Bands obtained were quantified using the inbuilt software Gazelle Reader GZ-100 provided by the Company.

The results obtained were tabulated in MS Excel file. Qualitative data are presented as frequencies and percentages. All the data were analysed using Microsoft Excel 2013.

## RESULTS AND DISCUSSION

The present study, conducted at the tertiary care hospital of Dahod, aims to estimate the prevalence of sickle cell anaemia (SCA) and type of genetic pattern among the tribal population of Dahod district of Gujarat. This ongoing study collected data over a three-month period from June 2024 to August 2024.

As shown in Table 1 and Chart 1 & 2 Preliminary results indicated that out of 12,259 samples from the tribal community of Dahod, 11,783 (96.12%) samples were valid, comprising 4,450 (37.77%) males and 7,333 (62.23%) females. A total of 475 (3.88%) samples were excluded under various lab rejection criteria.

The study analysed a total of 11,783 valid samples, representing 96.12% of the total collected samples. The gender distribution results observed a higher proportion of females (62.23%) compared to males (37.77%) among the valid participants. The positive NESTROFT test results show prevalence of 17.15% sickle cell anaemia amongst the total samples tested from the tribal community of Dahod District. On comparing gender distribution, a higher percentage of positive results among females (65.91%) over males (34.09%) suggest females are having a greater prevalence of the sickle haemoglobin. Although the positive result among males is low, the higher positive test rate among females suggests that targeted screening and intervention programs can be more beneficial for the female population.

The Table: 2 provides a detailed breakdown of the total NESTROFT Positive samples (n=2021) across different age groups and genders. The data is categorised into three age groups:  $\leq 16$  years, 17-50 years, and  $\geq 51$  years, with the number and percentage of samples for males and females within each group.

In the  $\leq 16$  year age group, males constitute a higher percentage of the samples compared to females. This suggests a greater participation of younger males in the study. The age group 17-50 years has the highest number of samples. A higher number of females in this age group compared to males, indicates greater participation and a higher frequency of sickle cell anaemia among females in this age range. The old age group  $\geq 51$  year has the small number of samples. The number of males is higher in this age group compared to females. The percentage of positive results for the sickle haemoglobin among male gender is higher over females among the old age group.

Overall higher presentation of males in the younger and older age groups may indicate some different demographic health conditions affecting these populations. Increased occurrence of sickle cell anaemia among females in 17 to 50 year of reproductive age group is of concern for national goal of sickle cell free India.

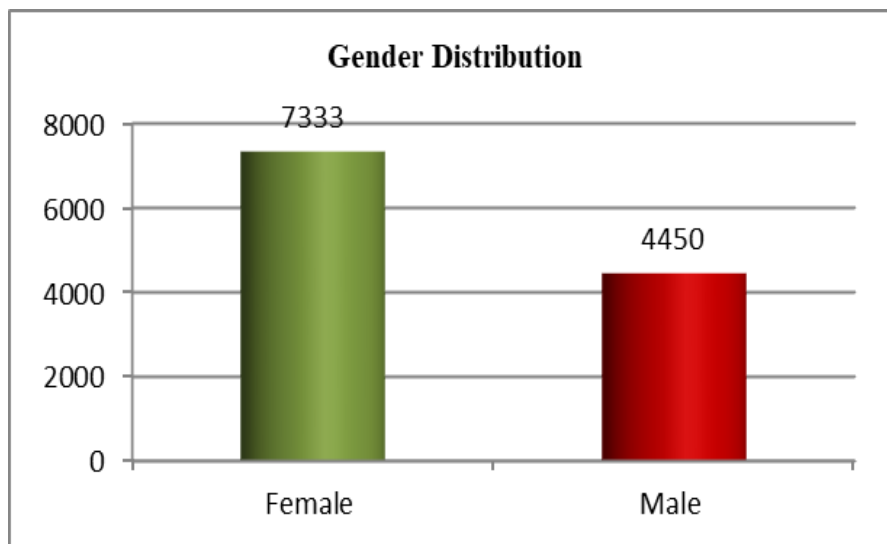
As shown in **Table 3** the study found that the frequency of SCT (HbAS) was 84.81% and SCD (HbS $\beta$ thal) was 15.19% among the total sickle haemoglobin positive cases, whereas among the total sample population (11,783) the frequency of SCT-AS emerged as 14.55% and SCD, HbS $\beta$ thal at 2.61%.

The distribution of sickle cell traits (SCT, HbAS) and sickle cell disease (SCD, HbS $\beta$ thal) across different age group and gender results indicated a higher prevalence of SCT (HbAS) compared to SCD (HbS $\beta$ thal) across all age groups and genders. The frequency of individuals showing SCT increases with age, while the frequency of individuals having SCD decreases with advancing age. Females show a higher percentage of SCT as compared to males in different age

groups. Conversely, a slightly higher percentage of SCD is observed in the younger age males group, when compared with females but, decreasing trend is noted with advancing age. This distribution highlights the varying impact of sickle cell traits and disease across different demographics.

Hemoglobinopathies are the most common recessive single-gene disorders worldwide, and their management poses a significant global health challenge. India's ethnic diversity is mirrored in the variety of haemoglobin variants found across different ethnic groups. Unrestricted global migration of people, identifying carriers and conducting prenatal diagnoses of hemoglobinopathies is essential, even in regions where these conditions are not endemic. Migration has led to the mixing of populations from various regions, while consanguinity, caste, and area endogamy have resulted in a high incidence of haemoglobin variants among some communities, causing a significant public health issue in India.<sup>[10,11]</sup> Although many of these abnormal variants are clinically insignificant in their heterozygous state, they can cause severe disease when combined with other variants. Hence, sickle cell syndrome (including HbS/HbS, HbS/HbC, and HbS/Beta thalassemia) which is one of the four hemoglobinopathies requires genetic counselling and prenatal diagnosis.

The present study found over all 17.15% of sickle cell anaemia prevalence among the tribal community of Dahod District, which is in agreement with reports from Gujarat.<sup>[4-6]</sup> Based on the total number of samples analysed, the SCT-AS pattern was observed in 14.55% of cases, while the SCD HbS $\beta$ 0thal pattern was noted in 2.61% of cases. The frequency of SCT Hb-AS is approximately 5.5 times higher than that of SCD HbS $\beta$ 0thal among the tribal population with HbS positivity. Though many reports state there is no gender disparity but, our study found female gender has a higher frequency; this may be due to the higher participation of females in the present study.



**Chart 1: Gender Distribution for sample population.**

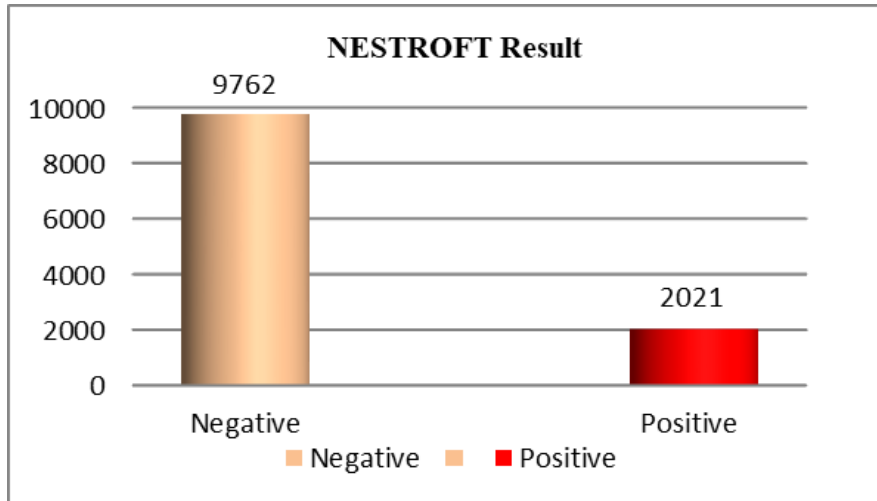


Chart 2: NESTROFT Result Among total samples (11,783).

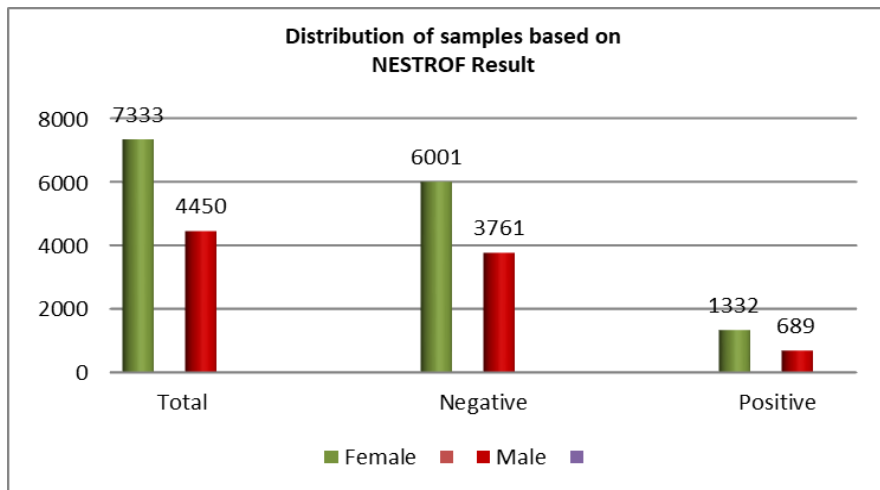


Chart 3: Distribution of samples based on NESTROFT result.

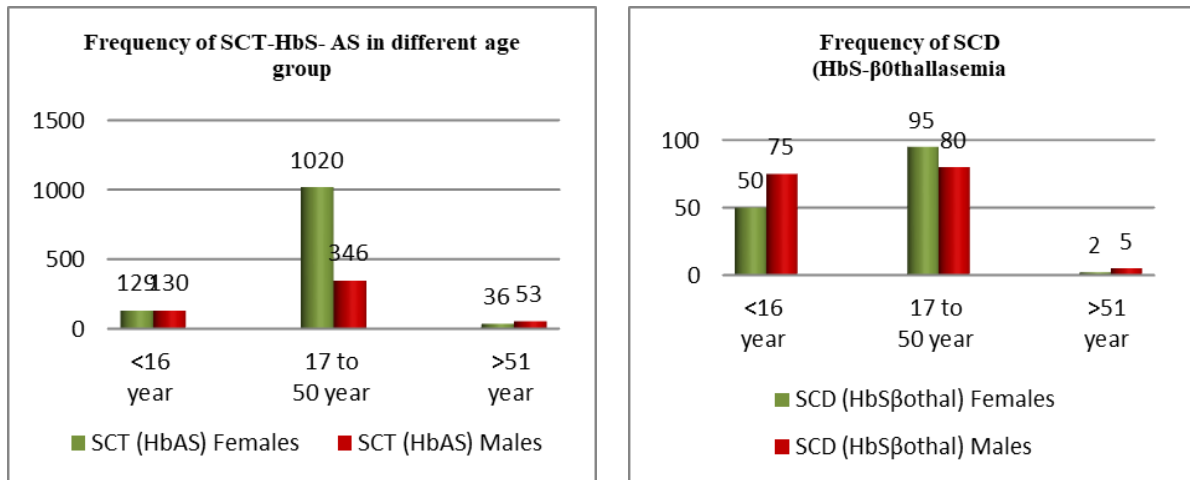
Table 1: Distributions of NESTROFT Positive Samples by Age Group and Gender.

Age Group	Total Samples (n=2021)	Male (n=689)	Female (n=1332)
	n (%)	n (%)	n (%)
≤16 yrs	384 (19.00%)	205 (29.75%)	179 (13.44%)
17-50 yrs	1541 (76.25%)	426 (61.83%)	1115 (83.71%)
≥51 yrs	96 (4.75%)	58 (8.42%)	38 (2.85%)

Table 2: Distribution Cellulose Acetate Hb Electrophoresis Results among positive cases by Age Group and Gender.

Age Group	Type of Sickle	Male (n=689)	Female (n=1332)
		n (%)	n (%)
≤16 years	SCT (HbAS)	130 (63.41%)	129 (72.07%)
	SCD (HbSβ <sup>o</sup> thal)	75 (36.59%)	50 (27.93%)
17-50 years	SCT (HbAS)	346 (81.22%)	1020 (91.48%)
	SCD (HbSβ <sup>o</sup> thal)	80 (18.78%)	95 (8.52%)
≥51 years	SCT (HbAS)	53 (91.38%)	36 (94.73%)
	SCD (HbSβ <sup>o</sup> thal)	5 (8.62%)	2 (5.27%)
01 to 75 years Both gender Total	SCT (HbAS)	1714/2021(84.81%)	
	SCD (HbSβ <sup>o</sup> thal)	307/2021(15.19%)	

Notes: \* SCT (HbAS): Sickle Cell Trait \* SCD (HbSβ<sup>o</sup>thal): Sickle Cell Disease



**Chart 4: Frequency of Sickle cell trait / Sickle cell disease in different age group.**

### Limitations of the Study

This study has several limitations that should be acknowledged. Firstly, the sample analysed consists solely of individuals who visited our hospital for their illnesses. This may introduce a selection bias, as the sample may not be representative of the general population. Secondly, the instrument used for haemoglobin electrophoresis has inherent limitations, as it is primarily designed for screening purposes.

### CONCLUSION

The present study, utilising Hemoglobin electrophoresis, highlights the significant prevalence of abnormal hemoglobin variant HbS among the tribal community of Dahod District who are visiting a tertiary care hospital. The findings suggest that the analysed population has a high frequency of SCT HbS-AS genotype along with minor SCD HbSβ0thal genotype. This may represent only a small fraction of the actual burden. Therefore, comprehensive mass screening covering every individual is essential to achieve the goal of a sickle cell anaemia-free India.

### Implication of study

The Overall findings emphasise the critical need for comprehensive screening programs on large population followed by genetic counselling and targeted healthcare interventions in the tribal population of Dahod to manage and lessen the impact of SCA specifically in future generations. Further research involving large sample size is justified to explore the underlying factors contributing to the observed gender disparities and to develop effective prevention and treatment strategies.

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**REFERENCES**

1. WHO Report, Community control of hereditary anaemias. Memorandum from a WHO meeting. Bull World Health Organ, 1983; 61: 63-80.
2. Kanjaksha Ghosh, Roshan B. Colah, and Malay B. Mukherjee. Haemoglobinopathies in tribal populations of India: Indian J Med Res, 2015; 141(5): 505-508.
3. BhatiaHM, RaoVR. Genetic atlas of Indian Tribes, Bombay: Institute of Immunohematology (ICMR), 1987.
4. RaoVR. Genetics and epidemiology of sickle cell anemia in India. Indian J Med Sci, 1988; 42: 218-22.
5. Patel AP, Naik MR, Shah NM, Sharma N, Parmar P. Prevalence of common hemoglobinopathies in Gujarat: An analysis of a large population screening programme. Natl. J Community Med, 2012; 3: 112-6.
6. Patel AG, Shah AP, Sorathiya SM, Gupte SC. Hemoglobinopathies in South Gujarat population and incidence of anaemia in them. Indian J Hum Genet, 2012; 18: 294-8.
7. Diggs, L. W, Walker, R: A Solubility Test for Sickle Cell Hemoglobin I. Aggregation and Separation of Soluble and Insoluble Components With-out Centrifugation. Lab. Med, 1973; 4: 27-31.
8. Huntsman RG, Barclay GP, Canning DM, Yawson GI. A rapid whole blood solubility test to differentiate the sickle-cell trait from sickle-cell anaemia. Journal of Clinical Pathology, 1970; 23(9): 781-3.
9. Muhammad Noman Hasan et al. Paper-based microchip electrophoresis for point-of-care hemoglobin testing. Analyst, 2020; 145(7): 2525–2542.
10. Balgir RS. The burden of hemoglobinopathies in India and the challenges ahead. Current Sci. Association, 2000; 79(11): 1536-47.
11. Balgir RS. The genetic burden of haemoglobinopathies with special references to community health in India and the challenges ahead. Indian J Hemat. Blood Transfuse, 2002; 20: 2-7.