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Genotypic Pattern of Sickle Cell Anaemia among Tribal Community of Dahod: A Tertiary Hospital Based Study

Dhiraj J Trivedi

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Abstract

Sickle cell anaemia (SCA) is a prevalent haemoglobinopathy worldwide, with Gujarat exhibiting a notably high prevalence. This study focuses on the frequency and genotype distribution of SCA within the tribal community of Dahod, aiming to enhance public health outcomes and contribute to a sickle cell-free India.

Study conducted at a tertiary care hospital, it analyses data from 12,259 individuals during June 2024 to August 2024.

The findings reveal a 17.15% prevalence of SCA among the tribal community. Among HbS positive individuals, SCT-AS genotype was observed in 14.55% of cases, while the SCD HbSβ0thal genotype was noted in 2.61% of cases. The frequency of SCT Hb-AS is approximately 5.6 times higher than that of SCD HbSβ0thal among the HbS-positive tribal population. Additionally, a gender disparity was observed, with a higher frequency in females.

Keywords: Hemoglobinopathies; Hemoglobin; Sickle Cell Anaemia; Sickle Cell Trait; Sickle Cell Disease; Hemoglobin S; Thalassemia; Tribal.

Author Affiliation: Professor, Department of Biochemistry, Director Central Clinical Laboratories, Zydus Medical College and Hospital, Dahod, Gujarat 389151, India.

Corresponding Author: Dhiraj J Trivedi, Professor, Department of Biochemistry, Director Central Clinical Laboratories, Zydus Medical College and Hospital, Dahod, Gujarat 389151, India.

E-mail: dhiraj99trivedi@gmail.com

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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 β-globin gene.[1] 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,[2] 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%.[3] In Gujarat, the prevalence of HbS among tribal populations ranges from 13% to 31%.[4] Specifically, surveys conducted in 22 districts of Gujarat reported a sickle cell carrier prevalence rate of 11.37%.[5] Additionally, some tribal groups in South Gujarat show a high prevalence of HbS (6.3% to 22.7%) and the β-thalassemia trait (6.3% to 13.6%).[6] 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-monthsranging from June 2024 to August 2024.

Participant registration

Our cohort studyuses 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 Dithionate qualitative solubility test (NESTROFT method).[7,8] 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.[9] 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

The present study, conducted at the tertiary care hospital of Dahod, aims to estimate the prevalence of sickle cell anaemia (SCA) 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.

Table 1: Preliminary over view of sample results

Category	Male	Female	Total
Population age range	01 year to 70 years	01 year to 72 years	-
Total number of cases	-	-	12,259 (100%)
Invalid Samples	-	-	475 (3.88%)
Valid Samples	-	-	11,783 (96.12%)
Gender Distribution	4,450 (37.77%)	7,333 (62.23%)	-
NESTROFT Test Result	Positive: 689 (34.09%)	Positive: 1,332 (65.91%)	Positive: 2,021 (17.15%)
	Negative: 3,761 (38.53%)	Negative: 6,001 (61.47%)	Negative: 9,762 (82.85%)

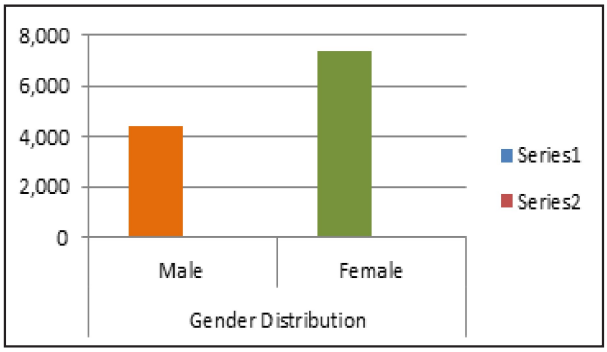


Chart 1: Gender Distrubution

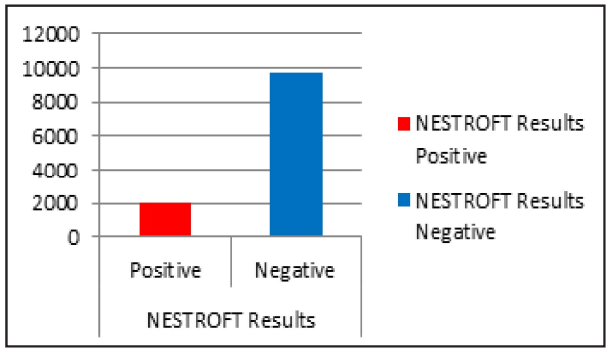


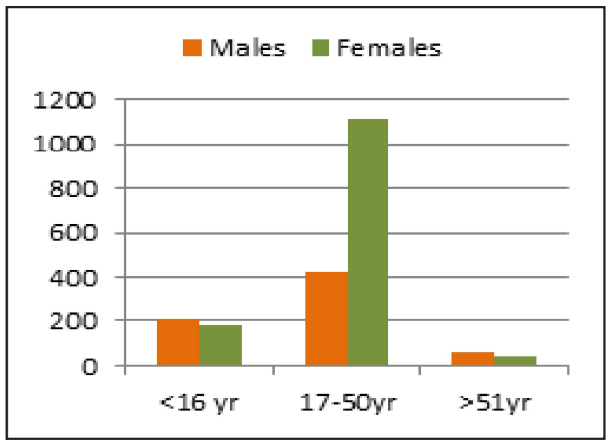
Chart 2: NESTROFT Result

As shown in **Table 1**, Preliminary results indicate 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 indicate 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. On comparing gender distribution, a higher percentage of positive results among females (65.91%) over males (34.09%) suggests a greater prevalence of the sickle haemoglobin among females. Present findings highlight the importance of gender differences while considering the prevalence and presence of the SCA condition. Although the positive prevalence 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.

Table 2: 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%)



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: ≤16 years, 17-50 years, and ≥51 years, with the number and percentage of samples for males and females within each group.

Inthe ≤16 years 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, indicatesgreater participation and a higher prevalence of sickle cell anaemia among females in this age range. The old age group ≥51 years has the small number of samples. The number of males is higher in this age group compared to females. The prevalence of the sickle haemoglobin among both genders of the old age group is almost same.

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

Table 3: Cellulose acetate hb electrophoresis results 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^{o}thal)	75 (36.59%)	50 (27.93%)
17-50 years	SCT (HbAS)	346 (81.22%)	1020 (91.48%)
	SCD (HbSβ^{o}thal)	80 (18.78%)	95 (8.52%)
≥51 years	SCT (HbAS)	53 (91.38%)	36 (94.73%)
	SCD (HbSβ^{o}thal)	5 (8.62%)	2 (5.27%)
01 to 75 years Both gender Total	SCT (HbAS)	1714/2021(84.81%)	
	SCD (HbSβ^{o}thal)	307/2021(15.19%)	

Notes: * SCT (HbAS): Sickle Cell Trait * SCD (HbSβothal): Sickle Cell Disease

As shown in **Table 3** the study found that the frequency of SCT (HbAS) was 84.81% and SCD (HbSβothal) was 15.19% among the total positive cases, but when total sample population are considered the frequency of SCT-AS emerged as 14.55% and SCD, HbSβothal as 2.61%.

When the distribution of sickle cell traits (SCT, HbAS) and sickle cell disease (SCD, HbSβothal) across different age groups and genderswas analysed, the results indicated a higher prevalence of SCT (HbAS) compared to SCD (HbSβothal) across all age groups and genders. The proportion of individuals with SCT increases with age, while the proportion with SCD decreases with advancing age. Females show a higher percentage of SCT compared to males in all age groups. Conversely, males have a slightly higher percentage of SCD in the younger age groups, but this trend reverses in the oldest age group. This distribution highlights the varying impact of sickle cell traits and disease across different demographics.

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

DISCUSSION

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 ahigh incidence of haemoglobin variants among some communities, causing a significant public health issue in India. (10,11) 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, which is in agreement with reports from Gujarat.[4-6] Based on the total number of samples analysed, the SCT-AS pattern was observed in 14.55% of cases, while the SCD HbSβ0thal pattern was noted in 2.61% of cases. The frequency of SCT Hb-AS is approximately 5.6 times higher than that of SCD HbSβthal 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.

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 various 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 variants among the tribal community of Dahod 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 HbSβthal 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.

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