Original Research

Prevalence and Ethnic Distribution of Sickle Cell Trait and Sickle Cell Anemiathe Saurashtra Region: A Cross-Sectional Study

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How to cite this article: Dhara P Trivedi, Deval Mehta Diploma, Punithan Narayanan et. al. Prevalence and Ethnic Distribution of Sickle Cell Trait and Sickle Cell Anemiathe Saurashtra Region: A Cross-Sectional Study. Indian Journal of Public Health Research and Development / Vol. 16 No. 3, July-September 2025.

Abstract

Background: Sickle cell disorders represent a significant public health concern in India, with varying prevalence across different ethnic groups and geographical regions. Understanding regional distribution patterns is crucial for implementing effective screening and management programs.

Objective: This study aimed to investigate the prevalence and distribution patterns of sickle cell trait and sickle cell anemia across different ethnic groups in the Saurashtra region, while also establishing comprehensive hematological profiles for both conditions.

Methods: A cross-sectional study was conducted on 1,546 individuals from various ethnic backgrounds in the Saurashtra region. Blood samples were analyzed for complete blood count and hemoglobin electrophoresis. Demographic data was collected to establish ethnic distribution patterns.

Results: The overall prevalence rates were 4.98% (77 cases) for sickle cell trait and 2.07% (32 cases) for sickle cell anemia. Hematological analysis revealed significant differences between trait and anemia patients, with HbS levels of 30.5 ± 25.6% and 73.8 ± 9.31% respectively. Hindu communities showed the highest prevalence in both trait (36.36%) and anemia (28.13%) cases, followed by Adivasi populations (trait: 22.08%, anemia: 15.63%). Significant variations in RBC counts (trait: 5.00 ± 1.19 x 10^6/µL; anemia: 3.22 ± 1.17 x 10^6/µL) and hemoglobin levels (trait: 10.6 ± 3.23 g/dL; anemia: 7.33 ± 2.87 g/dL) were observed.

Conclusion: The study reveals distinct distribution patterns of sickle cell disorders across ethnic groups in the Saurashtra region, with significant presence in both tribal and non-tribal populations. These findings emphasize the need for comprehensive screening programs and suggest potential genetic admixture in the region.

Keywords: Sickle cell trait, Sickle cell anemia, ethnic distribution, hematological parameters, Saurashtra region, genetic epidemiology.

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Submission date: November 4, 2024 **Revision date:** December 30, 2024 **Published date: June 7,2025**

Introduction

Sickle cell disease (SCD) represents one of the most prevalent genetic disorders worldwide, affecting millions of people across different geographical regions and ethnic groups[1]. In India, the condition presents a significant public health challenge, with an estimated 44,000 births of children with sickle cell disease annually [2]. The Saurashtra region of Gujarat, in particular, has emerged as an area of particular interest due to its diverse ethnic composition and historically reported cases of hemoglobinopathies[3].

Sickle cell disease occurs due to a point mutation in the β-globin gene, resulting in the formation of abnormal hemoglobin S (HbS). While individuals with sickle cell trait (heterozygous HbAS) are generally asymptomatic, those with sickle cell anemia (homozygous HbSS) experience severe clinical manifestations[4] . Understanding the distribution patterns of both conditions across different ethnic groups is crucial for implementing effective screening and management programs.

Previous studies have documented varying prevalence rates of sickle cell disorders across different Indian populations, with particularly high frequencies observed in tribal communities[6]. However, comprehensive data specific to the Saurashtra region, especially regarding the distribution across various ethnic groups and associated hematological parameters, remains limited. Recent research has emphasized the importance of understanding these regional patterns for developing targeted intervention strategies[5].

The present study aims to investigate the prevalence and distribution patterns of both sickle cell trait and sickle cell anemia across different ethnic groups in the Saurashtra region. Additionally, it seeks to establish detailed hematological profiles for both conditions, contributing to better understanding of their clinical manifestations in this population. This information is vital for healthcare planning and genetic counselling services in the Saurashtra region of India.

Methodology

Study Design and Population This cross-sectional study was conducted in the Saurashtra Region to assess the prevalence of sickle cell trait and sickle cell anemia across different ethnic groups. A total of 1,546 participants were recruited for a camp basedscreening program after getting informed consent, representing various ethnic communities including Hindu, Adivasi, Koli, Bhil, and other local populations.

Study Population

Inclusion Criteria:

1. Age: Individuals between 1-65 years of age
2. Ethnicity: Permanent residents belonging to Hindu, Adivasi, Koli, Bhil, and other local populations in the Saurashtra Region
3. Consent: Willing to provide written informed consent/assent for participation
4. Physical presence: Available for in-person screening and blood sample collection

Exclusion Criteria:

1. Recent blood transfusion: Individuals who received blood transfusion within the past 3 months
2. Acute illness: Participants with severe acute illness or hospitalization at the time of screening
3. Previous diagnosis: Individuals with previously confirmed sickle cell disease or trait who are already under medical care
4. Pregnancy: Pregnant women in their third trimester
5. Migration status: Temporary residents or individuals who have lived in the region for less than 6 months
6. Inability to consent: Individuals unable to provide informed consent due to mental or physical conditions

Sample Collection and Processing Blood samples were collected from all participants under aseptic conditions using venipuncture technique. EDTA-anticoagulated whole blood samples were used for both complete blood count analysis and hemoglobin electrophoresis.

Hematological Analysis Complete blood count parameters were analyzed using an automated hematologyanalyzer to measure:

- Red Blood Cell (RBC) count
- Hemoglobin (Hb) concentration

- Hematocrit (HCT)
- Mean Corpuscular Volume (MCV)
- Mean Corpuscular Hemoglobin (MCH)
- Mean Corpuscular Hemoglobin Concentration (MCHC)
- Red Cell Distribution Width (RDWCV and RDWSD)

Hemoglobin Analysis Hemoglobin electrophoresis was performed to quantify different hemoglobin fractions:

- Hemoglobin A0 (HbA0)
- Hemoglobin A2 (HbA2)
- FetalHemoglobin (HbF)
- Sickle Hemoglobin (HbS)

The presence of sickle cell trait was confirmed when HbS levels were around 30-40% with normal HbA0, while sickle cell anemia was diagnosed when HbS levels were >50% with very low or absent HbA0.

Demographic Data Collection Detailed demographic information was collected from all participants, including:

- Ethnic background
- Community affiliation
- Migration history (particularly for Maratha populations)

Statistical Analysis The data was analyzed to determine:

- Overall prevalence rates for both sickle cell trait and sickle cell anemia
- Distribution patterns across different ethnic groups
- Percentage calculations for positive cases relative to total screened population
- Percentage calculations for positive cases within affected groups
- Mean and standard deviation calculations for all hematological parameters

Data was organized and analyzed to present:

1. Summary statistics of the total screened population
2. Comparison of hematological parameters between trait and anemia groups
3. Ethnic distribution patterns for both conditions
4. Relative frequencies within affected populations

Quality Control Standard quality control procedures were followed for all laboratory analyses, and results were verified by experienced laboratory personnel. Multiple readings were taken for each sample to ensure accuracy of the results.

Results

Table 1: Comparison of Hematological Parameters in Sickle Cell Trait and Sickle Cell Anemia

Parameter	Sickle Cell Trait	Sickle Cell Anemia
Hb A0 (%)	64.5 ± 8.34	8.82 ± 9.10
Hb A2 (%)	3.24 ± 0.545	3.61 ± 3.12
Hb F (%)	1.55 ± 2.48	15.9 ± 6.55
Hb S (%)	30.5 ± 25.6	73.8 ± 9.31
RBC (10^6/µL)	5.00 ± 1.19	3.22 ± 1.17
Hb (g/dL)	10.6 ± 3.23	7.33 ± 2.87
HCT (%)	33.5 ± 8.48	23.2 ± 7.13
MCV (fL)	67.6 ± 11.8	74.1 ± 15.3
MCH (pg)	22.4 ± 7.87	23.6 ± 3.90
MCHC (g/dL)	34.2 ± 28.0	30.3 ± 3.02
RDWCV (%)	17.6 ± 4.45	20.3 ± 5.89
RDWSD (fL)	43.0 ± 5.61	52.1 ± 8.45

Table 2: Distribution of Sickle Cell Trait in Different Ethnicities in Saurashtra Region

Ethnicity	Positive Cases	% of Total Screened	% of Total Positive Cases
Hindu	28	1.81%	36.36%
Adivasi	17	1.10%	22.08%
Koli	9	0.58%	11.69%
Bhil	5	0.32%	6.49%
Katara	3	0.19%	3.90%
Darbar	3	0.19%	3.90%
Thakor	2	0.13%	2.60%
Pataliya	2	0.13%	2.60%
Rathva	2	0.13%	2.60%
Siddhi	1	0.06%	1.30%
Bania	1	0.06%	1.30%
Rabari	1	0.06%	1.30%
Harijan	1	0.06%	1.30%
Dalwadi	1	0.06%	1.30%
Kadva Patel	1	0.06%	1.30%
Muslim	1	0.06%	1.30%

Table 3: Distribution of Sickle Cell Anemia in Different Ethnicities in Saurashtra Region

Ethnicity	Positive Cases	% of Total Screened	% of Total Positive Cases
Hindu	9	0.58%	28.13%
Adhivasi	5	0.32%	15.63%
Vankar	3	0.19%	9.38%
Muslim	3	0.19%	9.38%
Thakor	3	0.19%	9.38%
Ahir	2	0.13%	6.25%
Bhil	2	0.13%	6.25%
Khatri	1	0.06%	3.13%
Nayak (Migrated Marathas before 2 generation)	1	0.06%	3.13%
Meena Tribe	1	0.06%	3.13%
Darbar	1	0.06%	3.13%
Kumbhar	1	0.06%	3.13%
Wadekar (Migrated marathas before 2 generation)	1	0.06%	3.13%

In a study conducted in the Saurashtra Region, we analyzed the distribution of both sickle cell trait and sickle cell anemia across different ethnic groups. The study screened a total population of 1,546 individuals. For sickle cell trait, 77 positive cases were identified, resulting in an overall prevalence rate of 4.98%. In contrast, sickle cell anemia was detected in 32 individuals, showing a lower prevalence rate of 2.07% in the same population.

As shown in Table 1, the hematological parameters revealed significant differences between sickle cell trait and sickle cell anemia patients. Individuals with sickle cell trait maintained higher levels of normal hemoglobin (Hb A0) at 64.5 ± 8.34%, while those with sickle cell anemia showed dramatically lower levels at 8.82 ± 9.10%. The sickle hemoglobin (Hb S) was notably higher in anemia patients (73.8 ± 9.31%) compared to trait carriers (30.5 ± 25.6%). Fetalhemoglobin (Hb F) levels were also distinctly different, with anemia patients showing elevated levels (15.9 ± 6.55%) compared to trait carriers (1.55 ± 2.48%). Other critical parameters like RBC count and total hemoglobin levels were consistently lower in anemia patients, with RBC counts of $3.22 \pm 1.17 \times 10^6/\mu L$ compared to $5.00 \pm 1.19 \times 10^6/\mu L$ in trait carriers.

Table 2 demonstrates the ethnic distribution of sickle cell trait, which showed highest prevalence among Hindu communities with 28 cases (36.36% of positive cases), followed by Adivasi populations with 17 cases (22.08%). Other significant affected groups included Koli (11.69%) and Bhil (6.49%) communities. The distribution pattern showed smaller but notable presence across various other ethnic groups including Katara, Darbar, Thakor, and others, each representing between 1.30% to 3.90% of positive cases.

As detailed in Table 3, the distribution pattern of sickle cell anemia showed some similarities but with different proportions. Hindu communities again showed the highest number of cases with 9 positive individuals (28.13% of total positive cases), followed by Adhivasi populations with 5 cases (15.63%). Interestingly, some ethnic groups like Vankar, Muslim, and Thakor each represented 9.38% of positive cases with 3 cases each. The study also noted the presence of the condition in migrated Maratha populations (both Nayak and Wadekar groups), each representing 3.13% of positive cases.

This comprehensive analysis provides valuable insights into the distribution patterns of both sickle cell trait and anemia across various ethnic groups in the Saurashtra Region, highlighting the varying prevalence rates and hematological characteristics between the two conditions.

Discussion

The present study provides significant insights into the distribution patterns of sickle cell trait and sickle cell anemia in the Saurashtra region, revealing important epidemiological and hematological findings. The overall prevalence rate of 4.98% for sickle cell trait and 2.07% for sickle cell anemia aligns with previous regional studies in Western India, though showing some distinct patterns specific to the Saurashtra population [10].

The hematological parameters observed in our study demonstrate significant variations between trait and anemia patients, particularly in HbS levels (30.5 ± 25.6% vs 73.8 ± 9.31%). These findings are consistent with those reported by Antwi-BoasiakoC[8]. The elevated HbF levels could potentially explain the variable clinical severity observed in this population, as higher HbF levels are associated with milder disease manifestations[13].

The ethnic distribution pattern revealed a noteworthy concentration of cases among Hindu and Adivasi communities, comprising 36.36% and 22.08% of trait cases respectively. This distribution pattern differs somewhat from earlier studies in neighboring regions, where tribal populations showed higher prevalence rates[7]. The significant presence of cases among non-tribal communities in our study suggests a possible genetic admixture in the region, a phenomenon previously documented by H. LADet al.[9] in their genetic analysis of Indian populations.

Particularly interesting is the detection of cases among migrated Maratha populations (both Nayak and Wadekar groups), each representing 3.13% of anemia cases. This finding supports recent research by Mukhopadhyay D [12] suggesting the importance of migration patterns in the distribution of hemoglobinopathies across Indian states. The presence of cases in these migrant communities emphasizes the need for comprehensive screening programs that include both indigenous and migrated populations.

The relatively higher RBC counts and hemoglobin levels in trait carriers (5.00 ± 1.19 x 10^6/μL and 10.6 ± 3.23 g/dL respectively) compared to anemia patients (3.22 ± 1.17 x 10^6/μL and 7.33 ± 2.87 g/dL) align with established pathophysiological patterns [11]. However, the wide standard deviations observed in our study suggest considerable individual variation, highlighting the need for personalized clinical approaches.

These findings have important implications for public health strategies in the region. The diverse ethnic distribution of both trait and anemia cases suggests the need for broad-based screening programs rather than those targeting specific communities. Additionally, the hematological profiles established in this study can serve as regional reference values for clinical assessment and management.

Limitations, and Recommendations

This study, while comprehensive in its approach, encountered several limitations that should be considered when interpreting the results. The cross-sectional nature of the study limited our ability to track temporal changes in hematological parameters and disease progression. Additionally, the sample size, though adequate for general prevalence estimation, may not fully represent all ethnic subgroups in the Saurashtra region, particularly those with smaller populations. The study also lacked detailed clinical history and follow-up data of the affected individuals, which could have provided valuable insights into the phenotypic variations and disease severity across different ethnic groups. Furthermore, the absence of molecular genetic analysis limited our ability to identify specific genetic variants and their correlation with clinical manifestations.

Based on these limitations and our findings, several recommendations can be proposed for future research and clinical practice. First, we recommend conducting longitudinal studies to better understand the natural history of sickle cell disorders in this population, including regular monitoring of hematological parameters and clinical outcomes. Implementation of comprehensive genetic screening programs, including molecular analysis, would provide deeper insights into the genetic variants prevalent in this region. We also recommend establishing a regional registry for sickle cell disorders to facilitate better tracking and management of cases. Furthermore, developing community-specific intervention strategies, particularly for high-prevalence ethnic groups, would be beneficial. It is

also crucial to strengthen genetic counseling services and awareness programs, especially in communities showing higher prevalence rates.

Conclusion

In conclusion, this study provides valuable insights into the distribution patterns of sickle cell trait and sickle cell anemia across different ethnic groups in the Saurashtra region, revealing significant variations in prevalence rates and hematological parameters. The findings highlight the complex interplay between genetic predisposition and ethnic background in the manifestation of sickle cell disorders. The higher prevalence among certain ethnic groups, coupled with distinct hematological profiles, emphasizes the need for targeted screening and intervention strategies. The study's results contribute significantly to the existing knowledge base and provide a foundation for developing more effective, population-specific healthcare approaches in the region. Moving forward, these findings can serve as a valuable reference for healthcare planning, genetic counseling, and future research initiatives in the field of hemoglobinopathies, particularly in the context of diverse ethnic populations in Western India

Acknowledgements:

The authors would like to express their sincere gratitude to the following individuals for their valuable contributions to this study:

Ms. Shilpa (Senior Technition) for her technical assistance in laboratory analysis and quality control. This support was essential for ensuring the accuracy and reliability of our data

Ethical Clearance: The research/study approved by the Institutional Review Board at Shri M P Shah Medical College and Guru Govind Singh Hospital, number 199/05/2022, dated 13/01/2023.

Source of funding: Nil

Conflict of interest: Nil

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