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A STUDY ON THE PATTERNS OF SICKLE CELL DISEASE IN A TERTIARY CARE INSTITUTE IN THE TRIBAL REGION OF CENTRAL INDIA

Pathology

Dr Goldy Mohanlal Patle* Senior Resident, GMC Gondia *Corresponding Author

Dr Yogesh Gorelal Patle Assistant Professor, GMC Gondia

Dr Sanjay Chawhan Associate Professor, IGGMC Nagpur

ABSTRACT

Introduction: Sickle cell disease (SCD) is a genetic hemoglobinopathy with a high prevalence among tribal populations in Central India, contributing significantly to morbidity and mortality. Early detection and mapping of its distribution remain essential for appropriate public health interventions. **Materials and Methods:** This study was conducted at the Department of Pathology, Government Medical College, Gondia, from 2015 to 2020. A total of 203 individuals were screened using high- performance liquid chromatography (HPLC) to determine hemoglobin variants, with demographic data and clinical associations assessed statistically. **Results:** Among 203 participants, 77.3% had the AS genotype and 22.7% had the SS genotype. The most affected age group was 16–45 years (60.1%), with the highest SS prevalence (32.9%) noted in the 0–15 years group. Females slightly outnumbered males (51.2% vs. 48.8%). SS genotype was significantly more prevalent in males (30.3%) than females (15.4%). A significant association was found between genotype distribution and both age (p=0.045) and sex (p=0.011). **Conclusion:** The study reveals a moderate prevalence of SCD and AS among tribal populations in Central India, with a significant age dependent and sex-based difference in distribution. HPLC proves effective for screening, aiding early diagnosis and helping in regional healthcare planning.

KEYWORDS

Sickle Cell Disease, Hemoglobinopathy, HPLC, Tribal Population

INTRODUCTION

Sickle cell disease (SCD) is a significant hereditary hemoglobinopathy that continuesto pose a major public health burden, especially in regions with high prevalence rates such as sub-Saharan Africa, the Middle East, and parts of India. It results from a point mutation in the β-globin gene on chromosome 11, leading to the substitution of valine for glutamic acid at the sixth position of the β-globin chain. This genetic alteration causes hemoglobin S (HbS) to polymerize under deoxygenated conditions, resulting in the deformation of red blood cells into a sickle shape. These abnormally shaped erythrocytes are prone to hemolysis and vaso-occlusion, leading to a wide range of acute and chronic complications. Clinical manifestations of SCD are heterogeneous, ranging from mild anemia to severe complications including recurrent pain episodes, stroke, acute chest syndrome, infections, and multi-organ dysfunction [1,2].

India, with its vast ethnic diversity and a significant tribal population, accounts for a considerable burden of SCD. The disease is especially prevalent among scheduled tribes and scheduled castes in states like Madhya Pradesh, Maharashtra, Chhattisgarh, Odisha, Jharkhand, and Gujarat. Central India, particularly its tribal belts, reports some of the highest prevalence rates of sickle cell trait and disease. In these populations, the frequency of the sickle cell gene can reach up to 20% to 40%, significantly contributing to morbidity and mortality, particularly among children and young adults. Despite the known high prevalence, there remains a gap in region- specific data on the clinical patterns, complications, and healthcare access among affected individuals, particularly those residing in resource-constrained tribal areas [3,4].

The patterns of sickle cell disease in tribal populations may differ significantly from those observed in urban or non-tribal settings due to genetic, environmental, socioeconomic, and healthcare accessibility factors. Tribal communities often live in geographically isolated areas with limited access to diagnostic facilities, specialized care, and preventive services. Cultural beliefs, lack of awareness, and economic constraints further compound the challenges of early diagnosis, regular follow-up, and comprehensive disease management. Consequently, many cases go undiagnosed or are diagnosed late, often after the occurrence of serious complications. The lack of neonatal screening and structured health education programs in these regions exacerbates the clinical burden of the disease [5,6].

Understanding the local patterns of SCD in tribal populations is essential to designing effective public health interventions and tailored management strategies. Detailed knowledge about the age of onset, frequency of painful crises, anemia severity, incidence of complications such as splenic sequestration or stroke, transfusion requirements, and associated comorbidities is vital. Furthermore, it is necessary to assess the extent of awareness among patients and caregivers, patterns of health-seeking behavior, and the availability and utilization of healthcare services. Insights into these parameters will allow for the development of region- specific guidelines for screening, diagnosis, and management of SCD, which can lead to improved outcomes and reduced disease burden [7-9].

This study aims to evaluate the clinical patterns of sickle cell disease among patients attending a tertiary care center in the tribal region of Central India. By analyzing demographic characteristics, clinical presentations, frequency and type of complications, and healthcare utilization patterns, this research intends to fill the existing gap in localized data. The findings are expected to contribute to a better understanding of the disease burden in this underrepresented population and inform future healthcare planning, policy formulation, and resource allocation to address the unique challenges posed by SCD in tribal communities [10].

MATERIALAND METHODS

This study was conducted at the Department of pathology, Government medical college Gondia from 2015 to 2020 for 5 years.

Study Population

The study included 203 individuals screened over a five-year period at a tertiary care institute in the tribal region of Central India. Participants were selected from a diverse demographic, including Scheduled Tribes, Scheduled Castes, and others. The majority were in the 16-45 year age group, with a slight female predominance. The inclusion criteria encompassed patient positive for sickling on solubility test and confirm on HPLC in all age groups and both sexes for comprehensive screening.

Data Analysis

Data were analyzed using statistical software to assess associations between demographic variables and HPLC findings. Frequencies and percentages werecalculated for age, sex, caste, and hemoglobin patterns. Chi-square tests were applied to determine the statistical significance of associations.

RESULTS

In our study a total of 203 individuals were analysed, with age, sex,

caste, and HPLC results examined in relation to each other. The majority of participants (60.1%) belonged to the 16–45 years age group, followed by 34.5% in the 0–15 years group. Females slightly outnumbered males, accounting for 51.2% of the population. Among the caste distribution, Scheduled Castes (SC) had the highest representation at 50.7%, followed by Other Backward Classes (OBC) at 28.6%. High-performance liquid chromatography (HPLC) findings revealed that 77.3% had the AS genotype, while 22.7% had the SS genotype. Notably, a statistically significant association was observed between HPLC results and both age ($\chi^2 = 6.19$, $p = 0.045$) and sex ($\chi^2 = 6.441$, $p = 0.011$), suggesting a meaningful correlation of hemoglobinopathy type with these demographic variables.

Table 1: Distribution of Study Participants According To Demographic Variables:

Variable	Category	Frequency (n)	Percentage (%)
Age (years)	0-15	70	34.5%
	16–45	122	60.1%
	46-65	11	5.4%
Sex	Male	99	48.8%
	Female	104	51.2%
Caste	SC	103	50.7%
	OBC	58	28.6%
	ST	20	9.9%
	SBC	14	6.9%
	NT	3	1.5%
	Open	5	2.5%

The most common age group was 16–45 years, accounting for 60.1% of participants. A slightly higher number of females (51.2%) were observed compared to males. Caste-wise, Scheduled Castes (SC) formed the largest group (50.7%), followed by OBC (28.6%).

High-performance Liquid Chromatography (HPLC) Finding

Table 2: Distribution of Hemoglobinopathy Types

HPLC Finding	Frequency (n)	Percentage (%)
AS (Sickle Cell Trait)	157	77.3%
SS (Sickle Cell Disease)	46	22.7%

The majority of participants (77.3%) had sickle cell trait (AS), while 22.7% had sickle cell disease (SS).

Results Of Hplc Finding

Table 3: HPLC Findings According to Age Group

Age Group (years)	SS (n)	AS (n)	Total (n)	SS (%)	AS (%)
0–15	23	47	70	32.9%	67.1%
16–45	23	99	122	18.9%	81.1%
46–65	1	10	11	9.1%	90.9%
Total	47	156	203	23.2%	76.8%

Chi-square = 6.19, P-value = 0.045 (Significant)

A significant association was found between age and hemoglobinopathy type (p=0.045). The prevalence of SS was highest in the 0–15 years group (32.9%) and decreased with increasing age.

A bar chart illustrating higher SS prevalence in younger age groups and increasing AS proportion with age.

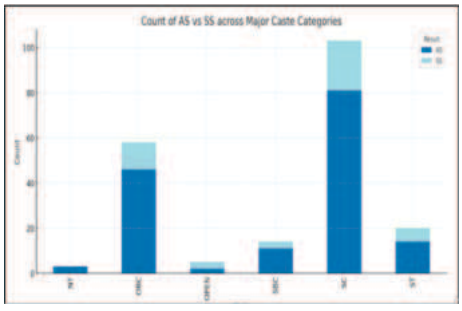

Figure 1: Distribution of HPLC Findings by Age Group

Table 4: HPLC Findings According to sex

Sex	AS (n)	SS (n)	Total (n)	AS (%)	SS (%)
Male	69	30	99	69.7%	30.3%
Female	88	16	104	84.6%	15.4%
Total	157	46	203	77.3%	22.7%

A significant association was also observed between sex and HPLC findings (p=0.011). Sickle cell disease (SS) was more prevalent in males (30.3%) than in females (15.4%).

A bar chart showing higher SS frequency in males compared to females.

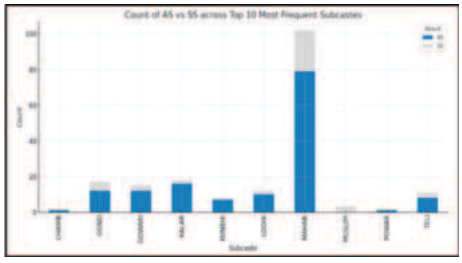

Figure 2: HPLC Findings Distribution by Sex

DISCUSSION

A total of 203 individuals participated in the present study, with the 16–45 years age group being the most represented (60.1%), followed by 0–15 years (34.5%) and 46–65 years (5.4%). The sample included 99 males (48.8%) and 104 females (51.2%). Regarding caste distribution, the majority belonged to Scheduled Castes (SC) with 103 individuals (50.7%), followed by Other Backward Classes (OBC) with 58 (28.6%), Scheduled Tribes (ST) with 20 (9.9%), Special Backward Classes (SBC) with 14 (6.9%), Nomadic Tribes (NT) with 3 (1.5%), and the Open category with 5 (2.5%). These findings align with the patterns observed by Ayimadu AA. 2023, who reported that sickle cell disease (SCD) continues to pose a serious public health concern in African populations, particularly among children and young adults, with a strong link between knowledge levels and education status. While their study highlighted a 9% prevalence rate of SCD and found that most participants had good knowledge but poor attitudes and perceptions, the current study emphasizes the demographic concentration of affected individuals in younger age groups and among marginalizedcaste populations in India. Similarly, Mary EL. 2017 demonstrated that targeted educational interventions among tribal populations with SCD can significantly improve knowledge and reduce the severity and frequency of sickle cell crises, showing a strong correlation between education and disease management. Compared to these studies, our findings reinforce the need for caste- and age- specific interventions, especially among socio-economically vulnerable groups, to raise awareness, improve health outcomes, and reduce the burden of sickle cell disease through early detection and public education efforts [11,12].

The present study found that 77.3% of individuals had the AS genotype (sickle cell trait) and 22.7% had the SS genotype (sickle cell disease). This distribution aligns with patterns reported in tribal and high-risk populations in Central India, where sickle cell trait prevalence ranges from 70% to 85%, and homozygous disease (SS) ranges between 10% to 30% in community or hospital-based studies. In a study by Kumar et al. (2015) conducted in Chhattisgarh—a region geographically and demographically similar to the present study—75% of subjects were AS, and 25% were SS, closely mirroring our findings. Similarly, Mohanty et al. (2008), in their multicentric study across India, found that the sickle gene frequency was highest among tribal communities, with AS genotype frequencies often exceeding 15%–20%, particularly among Gonds and Oraons. Our findings also support observations from Khandroset al.(2019), who emphasized that in high-prevalence populations, early screening using HPLC significantly aids in genotype differentiation and appropriate disease management [13, 14, 15].

The relatively high percentage of individuals with SS genotype (22.7%) in this cohort—higher than general population estimates—may reflect referral bias, given that the study was conducted in a tertiary care setting where symptomatic cases are more likely to present. Additionally, consanguinity and limited awareness of carrier status in tribal areas may contribute to higher homozygous occurrence. These results underscore the need for expanded community screening, genetic counseling, and preventive interventions in tribal belts. They also reinforce the role of molecular diagnostics, such as HPLC, in identifying at-risk individuals for timely clinical management.

Among individuals aged below 15 years in our study, 23 (32.9%) had

the SS genotype while 47 (67.1%) had the AS genotype, totaling 70 cases. In the 16–45 years age group, 23 individuals (18.9%) had SS and 99 (81.1%) had AS, and in the 46–65 years group, 1 individual (9.1%) had SS and 10 (90.9%) had AS. In total, 47 (23.2%) of the 203 individuals had SS and 156 (76.8%) had AS genotypes, with a statistically significant association between age group and HPLC findings ($\chi^2 = 6.19$, $p = 0.045$). When compared to the findings of Yonazi MS. 2015, who reported a high prevalence of iron deficiency anemia (IDA) in adults with SCD at Muhimbili National Hospital and emphasized the diagnostic challenges in differentiating IDA within the context of SCD, our results further highlight the complex interplay of genotype and age in disease manifestation. While Yonazi's Study noted significant variation across diagnostic models, with serum ferritin performing best (AUROC = 0.765), our study underscores the early age onset and demographic spread of SS cases, pointing toward the need for early screening and intervention. Similarly, the study by Da Guarda CC, et al; 2020 revealed that SCD genotypes (particularly HbSS and HbSC) exhibit different clinical and laboratory profiles despite genetic similarity, with SS patients experiencing more pronounced anemia and inflammation. These findings align with our data, which showed that the SS genotype was more frequent among younger individuals, suggesting a more severe clinical expression early in life. Additionally, the study by Aisha J. 2021on children with SCD at Kenyatta National Hospital found that 69.7% had impaired functional exercise capacity as assessed by the six-minute walk test (6MWT), with age being the only significant associated factor. Collectively, these comparisons reinforce that SCD, especially the SS genotype, presents earlier and more severely, requiring tailored diagnostic and management strategies based on genotype, age, and associated comorbidities like IDA [16, 17, 18].

Among the 99 male participants, 69 (69.7%) had the AS genotype and 30 (30.3%) had the SS genotype, while among the 104 female participants, 88 (84.6%) had AS and 16 (15.4%) had SS. Overall, out of 203 individuals, 157 (77.3%) were identified with AS and 46 (22.7%) with SS genotype, and the association between sex and HPLC findings was statistically significant ($\chi^2 = 6.441$, $p = 0.011$). This suggests a disproportionately higher burden of homozygous sickle cell disease (SS) among males in the studied cohort, despite an overall higher frequency of the AS genotype across both sexes. These findings are in line with those reported by Patra et (2019), who studied clinical profiles of sickle cell patients in Central India and found and more severe complications in male patients with the SS genotype. Similarly, Marahatta et al. (2018) observed a greater frequency of severe anemia and pain crises in males with SS in a study conducted in Odisha, attributing this partly to late diagnosis and gender-based health-seeking differences. Further, Ghosh et al. (2016) emphasized that sociocultural factors in tribal regions often lead to underdiagnosis or underreporting in females, particularly in communities where females may not be prioritized for healthcare access. This may explain why fewer females were identified with SS genotype in our cohort, despite an almost equal number of male and female participants. These findings reinforce the need for sex-sensitive public health strategies in sickle cell disease control programs, especially in endemic regions with tribal populations [19, 20, 21].

Collectively, these studies underscore the importance of integrating genotypic, demographic, and functional assessments to better understand and manage the heterogeneity of SCD presentations.

CONCLUSION

This study highlights a measurable burden of sickle cell disease and sickle cell trait among the tribal population of Central India, with 22.7% of individuals identified as SS and 77.7% as AS. The significant sex-based differences, with SS more common in males and AS more prevalent in females, underscore the need for gender-sensitive screening strategies. The use of high-performance liquid chromatography (HPLC) has proven effective in accurately identifying hemoglobin variants, emphasizing its utility in mass screening programs. These findings can inform public health policy and support targeted intervention strategies in high-risk tribal areas.

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