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Ocular manifestations of sickle cell anemia in central India's tribal population

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Abstract

Context: Sickle cell disease (SCD), the most prevalent hemoglobinopathy leads to many vision threatening ophthalmic complications which are frequently overlooked as compared to systemic issues despite their potential impact on quality of life.
Aims: To determine the prevalence of the ophthalmological manifestations in patients with sickle cell anemia at a tertiary healthcare centre located in a tribal area.
Materials and methods: A cross- sectional observational study conducted at tertiary care centre in tribal area. This study involved 182 patients, both genders, aged above five years. Detailed ophthalmic examinations included visual acuity, pupillary response, intraocular pressure (IOP), anterior and posterior segment examination.
Statistical analysis used: Chi square test
Results: The study revealed a significant prevalence of ocular manifestations in sickle cell anemia (SCA) patients. Ocular manifestations were identified in a total of 106 (58.24%) patients out of 182. Ocular manifestations are more prevalent in HbSS compared to HbAS (P=<0.0001). The highest proportion of patients with ocular manifestations was observed in the 21-30year age group (80%), followed by the 11-20 year age group (59%) with no gender predilection. Conjunctival sign (48.90%) was most common manifestations overall and venous tortuosity (32.96%) was most common fundus finding.
Conclusions: This study emphasizes the significant burden of ocular manifestations in SCA patients in tribal areas, stressing the importance of routine eye examination. Early detection of retinal changes allows for timely interventions, preventing vision loss and enhancing the quality of life for these patients.

Key-words: Sickle cell disease, visual acuity, Intraocular pressure, proliferative and non-proliferative retinopathy.

INTRODUCTION

Sickle cell disease (SCD) is one of the most common inherited hemoglobinopathies worldwide, characterized by significant morbidity and mortality.[1] In India, SCD is particularly prevalent among tribal populations residing in remote and hilly regions.[2] The disease results from a point mutation in the β-globin gene, leading to a single nucleotide substitution (GAG → GTG). This mutation replaces glutamic acid with valine at the sixth position of the β-globin chain, producing sickle hemoglobin (HbS).[3] Under hypoxic conditions, HbS undergoes polymerization, causing red blood cells to deform into a rigid, sickled shape. These deformed cells exhibit increased blood viscosity, reduced deformability, and a propensity for vascular occlusion, which contribute to the systemic and localized complications of the disease.[4]

Central India, often referred to as the "sickle cell belt," bears a disproportionately high burden of SCD, with patients in this region experiencing more severe clinical manifestations compared to individuals in other parts of India and developed countries.Vaso-occlusive crises and ischemic strokes are among the most prevalent and debilitating complications. Advances in medical care have led to increased life expectancy for individuals with SCD. However, this improved survival has been accompanied by the emergence of additional complications, including ocular manifestations, which significantly impact the quality of life in affected individuals.

Ophthalmic complications in SCD are primarily driven by vaso-occlusive crises caused by sickled red blood cells and their increased adhesion to vascular endothelium. Both the anterior and posterior segments of the eye can be affected by the pathological processes of the disease. Anterior segment manifestations include conjunctival sickling, iris atrophy, and neovascularization, which may lead to secondary glaucoma and hyphema. However, posterior segment involvement is more critical due to its higher prevalence and potential for severe visual impairment. Posterior segment findings include optic neuropathy, sickle cell retinopathy (both non-proliferative and proliferative), maculopathy, retinal hemorrhages, choroidopathy, and vascular abnormalities. These vascular changes are characterized by vessel tortuosity,

"silver-wire" arterioles, angioid streaks, and arterial or venous occlusions. Among these, proliferative sickle cell retinopathy (PSCR) is of particular concern due to its potential to cause irreversible vision loss, making it a critical focus of ophthalmologic care in SCD patients.[5]

This study aims to find the burden of ophthalmic findings in patients with SCD its corelation with genotypes, age and gender. By addressing ocular morbidity, it seeks to improve visual outcomes and quality of life in this underserved.

MATERIALS AND METHODS

This study included a total of 182 patients diagnosed with sickle cell anaemia. The sample size is calculated by using sample size calculation formula for a cross sectional study.

$n=z^2P(1-P)/d^2$

Z=Z statistic for a level of confidence,
n =Sample size,
P = Expected prevalence or proportion
and d= Precision (If the precision is 5%, then d=0.05).

Diagnosed cases of sickle cell anaemia and trait above five years who gave informed consent was included. Patient with other hemoglobinopathies and systemic diseases like hypertension, diabetes were excluded. Informed consent was obtained from all participants. Demographic and haematological data were collected, along with information on the reason for hospital visits and any previous hospitalizations. Each patient underwent a comprehensive systemic examination, including the recording of vital signs such as pulse rate, blood pressure, and respiratory rate. A full ophthalmological examination was performed. Visual acuity was measured using the Snellen chart, or the E chart for illiterate patients. Best corrected visual acuity for patients with poor vision. Anterior segment examination was done using a torch light and confirmed via APPASAMY'S slit-lamp bio microscopy. Pupillary reaction was assessed under controlled lighting. Both pupils were dilated with 0.5% tropicamide eye drops, and fundus examination was performed using HEINE'S direct and APPASAMY'S indirect ophthalmoscopes. Detailed evaluation was carried out using slit-lamp bio microscopy with 90-diopter VOLK'S lenses. Intraocular pressure (IOP) was measured using Goldmann applanation tonometry, with 0.5% proparacaine eye drops as topical anaesthesia. Fundus photograph was taken for selected patient. Diagnosed cases of sickle cell anaemia and trait above five years who gave informed consent was included. Patient with other hemoglobinopathies and systemic diseases like hypertension, diabetes were excluded.

RESULT

This observational study included 182 patients diagnosed with sickle cell disease (SCD), comprising 100 individuals with sickle cell anemia (HbSS) and 82 with sickle cell trait (HbAS). The mean age of the cohort was 25 years, with an age range spanning from 6 to 55 years. The prevalence of ocular manifestations attributable to sickling pathology was highest in the 21–30year age group (80%), followed by the 11–20year age group (59%). These findings suggests that young adults demonstrate the greatest burden of disease. Table no 1 shows age wise
distribution of ocular manifestations in sickle cell patients .

TABLE NO 1: AGE WISE DISTRIBUTIONS OF OCULAR MANIFESTATIONS

AGE	OCULAR MANIFESTATION		TOTAL
	PRESENT	ABSENT	
5 -10	4	13	18
11-20	29	20	49
21-30	52	13	65
31-40	12	18	30
41-50	5	7	12
51-60	4	4	8

Ocular manifestations were detected in 106 patients (58.24%)out of 182 patients. Among these, 69 patients (69%) had homozygous sickle cell anemia (HbSS), whereas 37 patients (45.12%) had heterozygous sickle cell trait (HbAS). A statistically significant association was observed between genotype and the prevalence of ocular manifestations ($P < 0.001$, Chi-square test), indicating a higher predisposition to ocular complications in HbSS compared to HbAS. These findings highlight the influence of genotype on the severity of ocular involvement in SCD.

There was no significant association between gender and ocular manifestations of sickle cell anaemia ($P = 0.5$, Chi-square test).

Most ocular manifestations observed in sickle cell patients did not result in significant visual impairment. The most common findings were conjunctival vessel tortuosity and the comma sign, occurring in 63% of sickle cell disease (SCD) patients and 31% of carrier (HbAS) patients. Yellowish scleral discoloration (jaundice) was present in 16% of SCD patients and 6% of carrier patients. Three SCD patients and one carrier patient presented with hyphema following trauma. Cataracts were observed in three carrier patients. Iris atrophy was not observed in any patient.

Posterior segment involvement, which may lead to visual impairment, was observed in one patient with HbSS who presented with vitreous haemorrhage and retinal detachment. Neovascularization was noted in three SCD patients. The salmon patch was found in 8% of SCD and 3.65% of HbAS patients. Venous tortuosity was the most common retinal finding in SCD, occurring in 45% of SCD and 18.26% of HbAS patients. Peripheral arteriolar occlusion was observed in 8% of SCD and 3.56% of HbAS patients. Mild disc pallor and disc signs were present in only four SCD patients. Maculopathy was not detected in any of the patients in this study. Table no 2 shows all ocular manifestations in sickle cell patients.

TABLE NO 2: OCULAR MANIFESTATIONS IN SICKLE CELL PATIENTS.

SIGN	SCA(n=100)	SCT(n=82)	TOTAL (n =182)
CONJUNCTIVAL SIGN	63	26	89
YELLOWISH DISCOLOURATION OF SCLERA	16	5	21
HYPHEMA	3	1	4
CATARACT	0	3	3
VENOUS TORTUOSITY	45	15	60
ARTERIOLAR OCCLUSION	8	3	11
SALMON PATCH	8	3	11
NEOVASCULARISATION	3	0	3
RD	1	0	1
VH	1	0	1
Disc changes	4	0	4

DISCUSSION

Sickle hemoglobinopathies result from a mutation in the β-globin gene, producing abnormal hemoglobin S (HbS), which causes chronic hemolytic anemia and recurrent vaso-occlusive crises. These crises lead to severe pain and extensive organ damage.[6] Reduced oxygen levels induce a conformational change in red blood cells containing HbS, transforming them from biconcave discs to elongated, crescent-shaped sickle cells.[6] The rigidity of these sickled cells obstructs capillary flow, exacerbating tissue hypoxia and further promoting sickling. This microvascular pathology can affect virtually any organ.[6]

The ocular structures are similarly affected by this microvascular pathology, resulting in a range of ocular manifestations in sickle cell anemia (SCA). While many of these manifestations do not lead to significant vision loss, some can cause severe visual impairment, highlighting the importance of early detection and management of ocular complications in SCA patients.

A 20-year prospective longitudinal study by Downes SM et al in Jamaica reported an increased incidence of ocular complications with age in both genotypes, with a higher prevalence observed in sickle cell disease (HbSS) patients by ages 24 to 26 years.[7] In our study also the prevalence increases with age and highest number of patients with ocular manifestations found in the 21-30 year age group (80%), followed by the 11-20 year age group (59%).

Our study identified conjunctival vessel tortuosity and the comma sign as the most common ocular manifestations, occurring in 63% of SCD and 31% of carrier patients. An observational study by AlRyalat SA et al involving 1904 patients with a mean age of 27.67 (±11.72) years found that the most common ocular finding on slit-lamp examination was vascular loops and segments, indicative of a positive conjunctival sign (54.1%). [8]

Mukaram Khan et al was conducted a prospective study in a tertiary care centre in North Maharashtra; found that ocular involvement was more common in patient with HBSS than HBAS. Similarly our study also shows similar finding. Venous dilatation and tortuosity to be the most common peripheral retinal change, present in 55.23% of SCD and 29.5% of HbAS patients.[9] Our study similarly found venous tortuosity to be the most common fundus finding in SCD, present in 45% of SCD and 18.26% of HbAS patients.

Another similar study from Odisha reported that 10% of patients had proliferative retinopathy, 30% had non-proliferative retinopathy, 26% had optic disc changes, 14% had retinal macular changes, and 4% had retinal detachment.[10] In contrast, our study found peripheral arteriolar occlusion in 8% of SCD and 3.56% of HbAS patients. Mild disc pallor and disc signs were seen in only four SCD patients, and no cases of maculopathy were observed. One patient was presented with vitreous haemorrhage and retinal detachment.

CONCLUSION

In conclusion, our study shows higher prevalence of sickle cell manifestations was found in HBSS than HBAS patients which increases with age and no gender correlation. It highlights the importance of routine ophthalmic screening in SCA patients to detect and manage ocular complications early, thereby improving visual outcomes and quality of life in this underserved population.

Limitations

This study has some limitations. It does not include other sickle cell genotypes, such as HbSC and sickle-thalassemia. Additionally, it does not compare the prevalence of ocular manifestations with the hematological status of sickle cell patients, limiting the ability to assess potential associations. Furthermore, the study does not analyse the correlation between ocular manifestations and systemic severity of disease.

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Conflicts of interest

There are no conflicts of interest

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