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PREVALENCE OF SICKLE CELL DISORDER IN CENTRAL INDIA

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ABSTRACT

Background: Sickle cell disease (SCD) is the most prevalent monogenic inherited blood disorder worldwide, and is increasingly recognized in developing countries like India. SCD mostly occurs in rural tribal regions and low socio-economic group population

Aim: this study evaluate the prevalence of sickle cell disorder in central Indian population

Material & Methods: A cross-sectional study was carried out among 1 to 55 years age group population in a tertiary care center, Madhya Pradesh, India Detailed information regarding the family history, socio-demographic profile was inquired. Thorough general and systemic examination was done. Haemoglobin estimation, sickling test and haemoglobin electrophoresis was performed

Results: Out of total 286 suspected patients, 277 (96.8 %) were diagnosed sickle cell disease. majority of the patients (52%) was 16-25 years age group. No significant difference between the sickling test and HPLC electrophoresis. Heterozygous state sickle cell trait (HbAS) was 206 (72%), whereas homozygous state sickle cell disease (HbSS) was 80 (28%) on Electrophoretic pattern.

Conclusion: The overall higher prevalence of sickle cell trait was found to be 96.8%. Mainly in 16-25 years age group population.

Key words: Sickle cell disease, HPLC electrophoresis, HbSS, HbAS

INTRODUCTION

Sickle cell disease (SCD) is the most prevalent inherited blood disorder worldwide, characterized by sickling of red blood cells due to structural disorder of haemoglobin leading to complications due to vaso-occlusion and anemia among affected individuals [1-2].

Sickle cell disease represents a major public health problem in India, mainly among tribal populations of central and Southern parts of India varying from 1-40%. [3-4]

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Prevalence of SCD has been well documented across Gujarat, Maharashtra, Madhya Pradesh, Andhra Pradesh, Chhattisgarh, and western Odisha with a smaller focus in the southern states of Tamil Nadu and Kerala [5]

SCD is a monogenic disorder caused by a point mutation in the beta globin gene on chromosome 11 and is responsible for the production of sickle haemoglobin (HbS). However, the clinical phenotype among SCD patients varies from one individual to another due to factors like β-globin gene haplotype, alpha-thalassaemia mutation(s), foetal haemoglobin etc [6-7].

Patients with SCD often present with acute complications (eg, bone pain crisis, acute abdominal pain, acute chest syndrome, visceral sequestration crisis, aplastic crisis, acute anemia, cerebro-cardiovascular complications, and priapism). Chronic morbidities in SCD (eg, chronic pain syndromes, immunological and infectious complications, chronic lung disease, hepatobiliary complications, renal complications, leg ulcer, musculoskeletal complications, and psychosocial or psychiatric issues) are often encountered [8-9].

Out of the many methods available for hemoglobin analysis, Electrophoretic techniques are used for routine clinical purposes. Diagnosis of SCD is confirmed by hemoglobin electrophoresis and the sickling test [10]

Several simple, cost-effective interventions have dramatically reduced morbidity and mortality from sickle cell disease in developed countries, including: early diagnosis by neonatal screening, preventive care with prophylactic penicillin and pneumococcal vaccination, and regular follow-up and treatment with transfusion and hydroxyurea for severe cases [11].

Access to care for sickle cell disease is limited in India, especially among tribal communities that bear the highest burden of disease [12].

Aim of the study to determine the prevalence of sickle cell disease in central Indian population.

MATERIALS AND METHODS:

This prospective study was carried out in the department of pathology, tertiary care hospital, Madhya Pradesh, central India. Clinically suspected Patients of sickle cell disorder were referred to the pathology laboratory. The clinical history, family history and hematological investigations were recorded in a structured Performa after obtaining informed written consent. Complete blood count and red cell indices were measured by automated analyzer. The presence of sickle haemoglobin was identified by sickling test with 2% sodium metabisulphite and confirmed by membrane electrophoresis. Haemoglobin electrophoresis was also done on both parents of registered patients to ascertain their genetic zygosity. They were categorized as SCA, if both parents were heterozygous for sickle haemoglobin.

High-Performance Liquid Chromatography (HPLC) is the test of choice today to screen all hemoglobinopathies.

All identified patients are also entered into the SCD registry, which includes information on demographics, disease history, laboratory results, clinic visits, treatment, complications, transfusions, hospital admissions, and mortality.

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RESULTS:

A Total of 286 clinically suspected patients of sickle cell disease were screened for SCD**.** Our study enrolled 0-55 year age group patients. Majority of the patients (52%) were 16-25 year age group followed by (24%) were age group of 26-35 year (Table: 1.).

Sickling test used as a screening and HPLC as a confirmatory test for the diagnosis of SCD.

Out of 286 patients screen for SCD, 277 (96.8%) were true positive for sickle cell disease by HPLC method, whereas 276 (96.5%) positive by sickling test. there was no significant difference between the HPLC and sickling test for diagnosing SCD. The prevalence of SCD in our study was 96.8%, which was very high. Heterozygous state sickle cell trait (HbAS) was 206 (72%), whereas homozygous state sickle cell disease (HbSS) was 80 (28%) on Electrophoretic pattern.

Table 1: Age wise distribution

S.NO.	AGE (IN YEAR)	TOTAL	Percentage (%)
1	0-5	11	3.8
2	6-15	35	12.2
3	16-25	149	52.0
4	26-35	71	24.8
5	36-45	15	5.2
6	46-55	5	1.7
TOTAL		286	100%

Table.2 Sickling Test as a screening method

SN	Variable	Sickling test	HPLC (Gold Standard)
1	True negative for sickle cell	8	9
2	False negative for sickle cell	1	0
3	False-positive for sickle cells	1	0
4	True positive for sickle cells	276	277
	Total	286	286

Table 3: Hemoglobin Electrophoresis results

S.NO.	Electrophoretic pattern	MALE	FEMALE	TOTAL
1	HbSS	46	34	80
2	HbAS	36	170	206
TOTAL		82	204	286

DISCUSSION:

Early diagnosis of SCD can save many lives; proper preventive measures taken early can minimize disease morbidity to a significant extent. To this extent pre-marital diagnosis, pre-natal diagnosis and pre-implantation genetic diagnosis can contribute in minimizing the burden of SCD [13].

In our study Hb solubility (sickling) test used for screening of sickle cell anaemia, observed 96.5% true positive for sickle cells, comparable with the other study [14], but this test does not distinguish sickle cell trait (HbAS) from SCD and therefore requires further testing.
In the present study, was have found sickle cell trait (HbAS) was very common (72%) on Electrophoretic pattern, similar pattern observed by Kamble M. et al [15] and Charuhas V et al [16], reported sickle cell train was the very common cause of anaemia.
In the current study, most of the patients (52%) were 16-25 year age group presented with sickle cell disease, concordance with the study conducted by Ahmad, et al [17]. Incidence of the sickle cells carriers was more in this age group.
On Hb electrophoresis pattern our study observed sickle cell trait (HbAS) were common (82.6%) in female as compared to male, accordance to the Desai G et al [18].
Prevalence of sickle cell disease or trait was very common among tribal population, rural areas and low socio-economic status population Roshan B et al [19] and Serjeant et al [20].

RECOMMENDATIONS:

- Pre-marital counseling, pre-pregnancy counseling, and proper ante-natal check-ups should be established.
- Establish Screening set-up at the community level in places like colleges before admission, offices before placement and screening in schools on a rotational basis.
- Increase awareness among the mass and to give proper health facilities to those already suffering from the disease
- Workers trained for counseling can be provided in each state as per the requirement

CONCLUSION:

In this prospective study, it was found that overall prevalence of sickle cell disease was very high (96.8%). SCD most commonly occurs in 16-25 years age group population. Heterozygous state of sickle cell trait was higher in female whereas homozygous state of sickle cell anaemia was higher in male.

Conflict of interest: none

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