

# Prevalence, clinical, and hematological profile of sickle cell disease in South Rajasthan

Sanjay Mandot, Gaurav Ameta

From Department of Pediatrics, Geetanjali Medical College, Udaipur, Rajasthan, India

**Correspondence to:** Dr. Sanjay Mandot, Department of Pediatrics, Geetanjali Medical College, Udaipur - 313 002, Rajasthan, India.

Phone: +91-9462022900. E-mail: drmandot@gmail.com

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## ABSTRACT

**Objectives:** To know the prevalence of sickle cell disease in 0-15 years old children of tribal area and to study its clinical and hematological profile. **Materials and Methods:** This cross sectional study was conducted between May 2014 and December 2015 in children below 15 years of age from tribal area of Sirohi and Udaipur district in South Rajasthan. **Results:** A total of 1090 children in tribal area were screened for sickle cell disease. Prevalence of sickle cell disease was found to be 8.53% of which 0.77% children were suffering from sickle cell anemia (HbSS), whereas 7.7% were having sickle cell traits (HbAS). Common morbidities were anemia, pain, infection, and splenomegaly. **Conclusion:** Sickle cell disease is prevalent in this area. Screening for sickling in this area is suggested for early diagnosis and to promote preventive measures so as to decrease morbidity and mortality.

**Key words:** Age, Hemoglobin, Morbidities, Sex, Sickling test, Splenomegaly

Sickle cell disease is an autosomal recessive hemoglobinopathy and is due to the replacement of valine for glutamic acid in position 6 of the beta globin chain of hemoglobin (Hb). This alteration leads to an unstable sickle-shaped red blood cell under hypoxic condition with a shortened life span of 10-20 days instead of the normal 120 days. Sickle cell disease refers not only to patients with sickle cell anemia (SCA) (HbSS, sickle cell Hb [HbS] >90%), but also to compound heterozygotes (HbS >50%) with a gene mutation other than the sickle cell mutation such as HbC,  $\beta$  thalassemia, HbD and HbO. In individual with sickle cell trait (SCT), the HbS level is <50%, thus having mild or no clinical manifestations [1].

The sickle cell gene in India was first described among tribal groups in South India but is now recognized to be widespread and prevalent in many parts of India, especially in central India, where the prevalence in different communities has ranged from 9.4% to 22% [2-4]. Clinically, SCA in India is moderate to severe with high HbF level. It is milder than SCA in Africa and similar to in central Asia [5].

Since little recent published data are available on this disease from Southern Rajasthan, we undertook the present study. Sickle cell disease in this tribal area poses difficulty in diagnosis and management as the sign and symptoms overlap with other prevalent common diseases. Our emphasis was to sensitize health care providers about the same.

## MATERIALS AND METHODS

During May 2014-December 2015, health checkup camps for tribal children were organized by tertiary care hospital in Sirohi

and Udaipur district of Rajasthan. Only tribal (Garasia) children under the age of 15 years were included in the study, who attended the health camps on their own. Children with known case of other hemolytic diseases such as thalassemia, G6PD, etc. were excluded from the study. Sickle cell disease is endemic in tribal parts of India, assuming its prevalence 12%, minimal sample size for a random sample at 95% confidence level with absolute precision of 2%, was computed as 1014 (according to Daniel 1999 formula). Multistage sampling was used to cover all geographical areas of both districts.

Children were screened by sickling test (with freshly prepared sodium metabisulfite), and Hb estimation (by Sahli's acid hematin method) was done on blood sample collected by finger prick. Sickling positive children were further evaluated by Hb electrophoresis (on cellulose acetate membrane) to confirm the diagnosis and to classify them as SCA (Hb SS) or SCT (Hb AS) [6]. Participants' details were recorded including age, sex, relevant history, clinical examination findings, and laboratory parameters. Statistical analysis was done by simple proportions and Chi-square test. The study was carried out after getting clearance from Institute Ethics Committee.

## RESULTS

A total of 1090 children were screened. The prevalence of sickle cell disease among tribal (Garasia) children of Sirohi and Udaipur district was found to be 8.53% (93/1090) of which 0.77% (9/1090) were homozygous (SCA, Hb SS), whereas 7.7% (84/1090) were heterozygous (SCT, HbAS). Out of 1090

children, 687 (63.02%) were males, and 403 (36.97%) were females. 57 (8.29%) male children and 36(8.9%) female children were having sickle cell disease. The difference in sex wise prevalence was not found statistically significant ( $\chi^2=0.1317$ ;  $df=1$ ;  $p=0.7167$ ).

A total of 29 (6.56%) children in the age group of 0-4 years, 56 (10.03%) in the age group of 5-9 years, and 8 (8.88%) in the age group of 10-15 years were suffering from sickle cell disease. The difference in age wise prevalence of sickle cell disease was found to be not significant ( $\chi^2=3.8318$ ;  $df=2$ ,  $p=0.1472$ ). Among SCA (Hb SS) cases, 4 were among 0-4 years age group, 3 from 5 to 9 years age group, and 2 were from 10 to 15 years age group. Among SCT (Hb AS) cases, 25 were from 0 to 4 years age group, 54 from 5 to 9 years age group, and 6 were from 10 to 15 years age group (Table 1).

Among sickle cell anemia (Hb SS) cases, main complaints were pain in 9 (100%), weakness/fatigability in 7 (77%), and fever in 6 (66.6%) patients. Pallor was the most common clinical sign observed in all 9 (100%) patients followed by splenomegaly in 7 (77%) patients. Hepatomegaly was observed in 5 (55.5%) patients. Jaundice was observed in 3 (33%) patients and gall stones in 1 patient. Two patients presented with pneumonia. Severe anemia (Hb <7 g%) was observed in 5 (55%) patients. Vasoocclusive crisis was observed in 4 patients (44.4%) (Table 2).

Among children with SCT main symptoms were weakness/fatigability 63 (75%), fever 34 (40.47%), and abdominal pain 28 (33.3%). Pallor was the most common sign observed in 74 (88%) patients followed by splenomegaly in 17 (20.2%) patients. Severe anemia (Hb <7 g%) was observed in 6 (7.14%)

patients. In addition to anemia and splenomegaly, ARI (20.8%), severe acute malnutrition (16.7%) and vitamin deficiencies (6%) were encountered. Malaria parasite was not detected in any of them (Table 2).

## DISCUSSION

The childhood diseases in the tribal area pose difficulty for diagnosis and management as the sign and symptoms of sickle cell disease overlap with other common prevalent diseases. Only high suspicion and screening can guide the health-care providers for better management of cases and prevent morbidities and mortalities. This study highlights the epidemiological and clinical picture of Sickle cell disorder in the tribal children.

The prevalence of Sickle cell disease in this study (8.53%) is similar to that reported from Orissa, Madhya Pradesh, Chhattisgarh, and Maharashtra, i.e., between 3.2% and 22.5% [2,3,5,7-9]. There was no significant difference in sex wise prevalence of sickle cell disease, and it is observed by other studies also [7-9]. The ratio of homozygous and heterozygous disease is very less as compared to observed by Kamble and Chatruvedi, who showed SCA in 61.6% whereas, SCT in 38.4% [10]. A similar finding was also observed in a study done by Patra et al., i.e. SCA in 59.1% and SCT in 40.9% [11].

In this study, among SCA (HbSS), 4 were diagnosed before 3 years of age, and total 7 (77.7%) were diagnosed before 6 years of age. About 66.6% cases with SCT presented before the age of 5 years. An earlier study from Orissa found 50% patients to be symptomatic before 5 years of age, whereas in a study from Jamaica, 90% were symptomatic by the age of 5 years [8,12]. According to Kar, 75% of the children had their, first manifestation within 10 years of age [8].

Pain, fatigability, and fever were the most common symptoms. Anemia was the most common association. The majority of the case had mild to moderate anemia. Besides sickle cell disease nutritional deficiency and infections may also contribute to anemia. Mean Hb value was found to be 7.8 g% which is similar to other studies [5,13]. Splenomegaly was found in 25.8% of children and was more in SCA children (77%). Splenomegaly was reported by many workers (24-70%) [7-9,11].

Neonatal screening for sickle cell disease in this high-risk population (tribal scheduled caste) might be very helpful. Once the cases are identified, counselors should explain to the parents about general supportive and preventive measures such as maintaining hydration, avoiding stress and adverse climatic conditions, early treatment of infections, and immunization.

We acknowledge that a community-based study is ideal to know the true prevalence of the disease, but because of the ethical/social issues involved, it is difficult to conduct a community-based study involving invasive procedure (blood collection), especially in pediatric age group. However, this hospital-based study is important for sensitization and can serve as a baseline for generating more data.

**Table 1: Age and sex wise distribution of sickle cell disease**

Age group (years)	SS type (n=9)			AS type (n=84)			Total
	Male	Female	Total	Male	Female	Total	
0-4	3	1	4	16	9	25	29/442
5-9	1	2	3	31	22	53	56/558
10-15	1	1	2	4	2	6	08/90
Total	5	4	9	52	32	84	93/1090

Age wise prevalence ( $\chi^2=3.8318$ ;  $df=2$ ,  $p<0.05$ ), Sex wise prevalence ( $\chi^2=0.1317$ ;  $df=1$ ;  $p<0.05$ ). SS: Homozygous Sickle cell disease, AS: Heterozygous Sickle cell disease

**Table 2: Clinical manifestations of sickle cell disease**

Parameters	Symptom/signs	SCA (%)	SCT (%)
Presenting symptoms	Bone pain	9 (100)	5 (5.9)
	Abdominal pain	3 (33.3)	28 (33.3)
	Generalized weakness	7 (77.7)	63 (75)
	Fever	6 (66.6)	34 (40.47)
Presenting signs	Pallor	9 (100)	74 (88)
	Splenomegaly	7 (77.7)	17 (20.8)
	Hepatomegaly	5 (55.5)	6 (7.1)
Other manifestations	Jaundice	3 (33.3)	0
	Gall stones	2 (22.2)	0
	Pneumonia	2 (22)	3 (3.57)
	Vasoocclusive crisis	4 (44.4)	0

SCT: Sickle cell trait, SCA: Sickle cell anemia

## CONCLUSION

Sickle cell disease is prevalent in this area. Recurrent attacks of musculoskeletal pain, anemia, frequent respiratory infections, jaundice, and splenomegaly, are the features which should arouse the suspicion of sickle cell disease. Early detection of sickle cell disease is useful for improving the quality of life in these tribal children.

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