Original Article Indian J Pediatr 1997; 64 : 73-77

Clinical Profile of Sickle Cell Disease in Orissa

Bimal Chandra Kar and Satyabhama Devi

Department of Medical Aid and Research Center, Burla, Orissa

Abstract. Children comprised 52% of patients with Sickle Cell Disease (SCD). Types of Sickle Cell Disease encountered were SS (92.7%). SB thalassaemia (6.7%) and SD disease (0.7%). The disease was widespread in almost all castes and communities in the society; largest number of patients (20%) belonging to scheduled castes and only 1.4% were from scheduled tribes. Maximum number of cases were in the age group 2-4 and 4-6 years, many of whom died around this age. Besides attacks of pain, jaundice and anemia, frequent attacks of fever with anemia or only anemia in childhood were a predominant presenting feature. Splenic sequestration was frequent (10.1%). The patients usually had a steady state hemoglobin level of 6-10 g/dl, with which they thrived well. Fetal hemoglobin was 5-30%. Blood transfusion was not a frequent requirement, but prophylactic long acting penicillin was helpful in preventing frequency of crisis. **(Indian J Pediatr 1997; 64 : 73-77)**

Key words. *Sickle cell disease; Children; Orissa.*

Sickle Cell Disease (SCD) is not rare in India and has been found in most communities in certain parts of the country. Detailed epidemiological, hematological, clinical and genetic profile of SCD patients, both children and adults, as observed at Burla has been published earlier[1-5]. The problems in the pediatric age group are vastly different from those in adults and thus deserve special attention.

Material and Methods

Of 800 consecutive cases of SCD seen at the sickle cell clinic of MARC, pediatric patients upto 14 years of age were 417 (52%). The diagnosis was established by sickling test, hemoglobin electrophoresis in both alkaline (tris EDTA borate buffer on CAM or strach-agarose) and acid (citrate buffer on agar gel) media. Besides a detailed history of specific symptoms from birth, a thorough clinical examination was made and routine hematological investigations were done by standard methods. In most cases genotype was determined by examination of both parents and in some by DNA analysis[4]. Efforts were made for follow-up at 3 months intervals during which clinical examination and hematological investigations were repeated and the events during the interval documented in records which were being updated from 1986 onwards.

Reprint requests : Dr. B.C. Kar, Retired Professor of Medicine, Director, Medical Aid and Research Centre, Burla-768 017, Orissa.

Results

Amongst 417 SCD children there were 376 (92.6%) SS (homozygous SCD), 28 (6.7%) SB thalassaemia (double heterozygous state for sickle gene and B thalassaemia gene) and three (0.7%) SD disease (a double heterozygous state for sickle and D

Punjab gene). Clinically and hematologically the three types of disease were indistinguishable. Multiple cases were seen in many families (Table 1).

The patients were mostly Hindus (79.1%) belonging to almost all subcastes in the society, while 19.6% were scheduled castes and 1.4% were scheduled tribes (Table 2).

TABLE 1. Family Distribution of SCD Children

No. of affected person	No. of families	%
2 Sibs	42	10.7
3 Sibs	6	1.4
4 Sibs	1	0.2
Child & parent	6	1.4

TABLE 2. Caste Distribution of SCD Children

Caste	No. of patients	%
Kulita (farmer)	96	23.0
Scheduled caste	82	19.6
Chasa (farmer)	54	12.9
Agharia	48	11.5
Milkman/Dumal	45	10.7
Oilman	33	7.9
Fisherman, fluorists, Kumuti, oriha, ahir	15	3.6
Kshatriya/Rajput	13	3.1
Scheduled tribes	6	1.4
Muslim	4	0.9
Brahmin	3	0.7
Khandayat	3	0.7
Weavers	1	0.2
Total	417	100

Age distribution of SCD children is shown in Fig. 1. Maximum number of patients were in the age group 2-4 and 4-6 years; the proportion declined thereafter suggesting thereby that with growing age many children probably die. Thus, Sickle Cell Disease is one of the causes of death

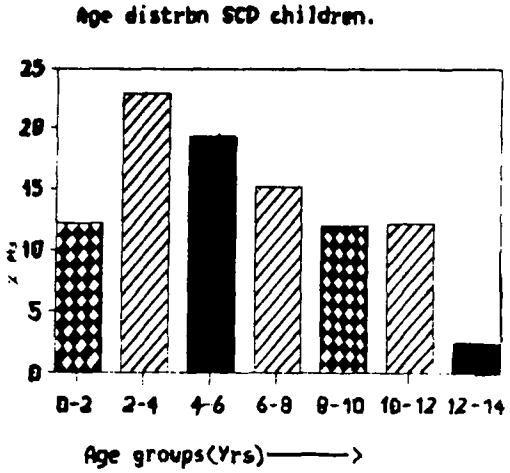

Fig. 1. Age distribution of SCD children (n = 417).

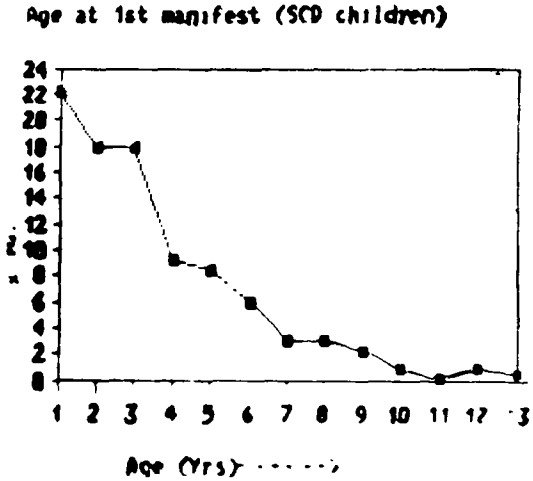

Fig. 2. Age at first manifestation in SCD children (n =390).

in early childhood.

This is further corroborated by considering the age at first manifestation (Fig. 2) More than half (58.3%) of the children manifested specific features before the age of 3 years, though there were about 6.5% SCD children who presented only at 13 years or more. Clinically the dominant features fell into six groups (Table 3) which occurred in recurrent episodes. Attacks of pain were characterised by hand-foot syndrome, generalised musculoskeletal pain or abdominal pain. Acute chest syndrome with respiratory distress or cerebrovascular episodes observed frequently in other countries were relatively rare. Respiratory involvement was mainly seen in the form of acute respiratory tract infection with fever and responded well to appropriate antibiotics such as amoxycyllin, trimethoprim sulphonamides etc. Such episodes were frequently associated with anemia. But many cases presented with only short fever (without any specific detectable cause) with severe anemia. Such episodes in a sickle cell area arouse suspicion of SCD. The cause of aggravation of anemia in these situations is either hyperhemolysis or transient bonemarrow hypoplasia (aplastic crisis). Recurrent severe anemia without evidence of preceeding hyperhemolysis was also seen in many children, the cause of which is not known. But acute severe anemia with rapidly enlarging spleen and pain over the spleen was a frequent phenomenon in children and was due to sequestration. Though, this is a transient event, it may endanger the life of the child unless blood transfusion is given in time. Such recurrent episodes are an indication for splenectomy.

Other clinical features commonly observed in children patients of SCD are listed in Table 4. Epistaxis usually occurred in summer and was sometimes very severe. Cardiomegaly and jaundice due to other causes *e.g.* rheumatic heart disease and viral hepatitis can also occur in SCD patients and need not always be

TABLE 3. Dominant Clinical Features in Recurring Episodes in SCD Children (n = 417)

	No.	%
Attacks of pain	277	66.4
Jaundice, anemia	42	10.1
Splenomegaly, fever with anemia	59	14.0
Splenic sequestration	42	10.1
Only jaundice	6	1.5
Only anemia	5	1.4

TABLE 4. Common Clinical Features in SCD Children (n = 417)

Features	No	%
Epistaxis	65	15.6
Bone necrosis	3	
Osteomyelitis	2	
Jaundice		
Persistent	121	26.8
During attacks of pain	88	21.1
Viral hepatitis	4	
Hepatic crisis	2	
Biliary stones sludge	2	
Cardiomegaly	52	14.9
Rheumatic heart disease	1	
Congestive heart failare	1	
Functional murmur	152	36.7

Nephrotic syndrome, Stroke, Fainting attacks (Recurrent), Fever with allergic rashes (recurrent) Leg ulcer

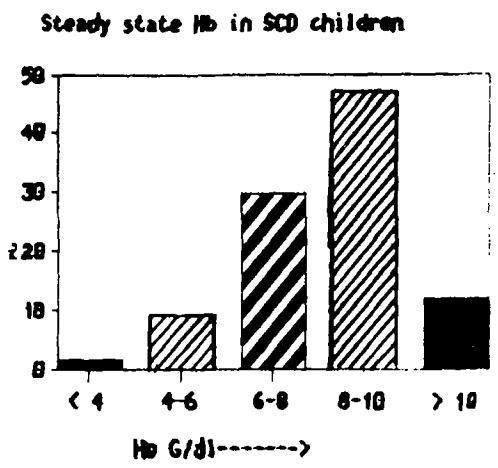

Fig. 3. Steady state hemoglobin level in SCD children (n = 389).

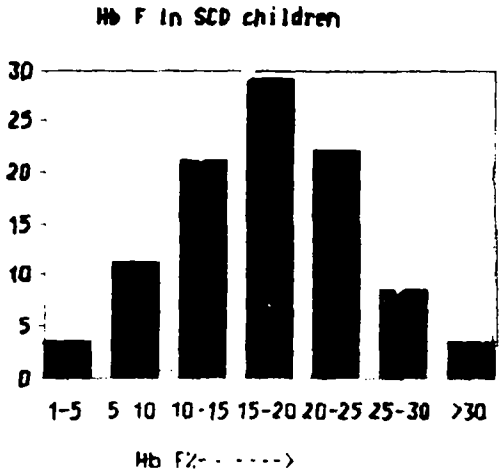

Fig. 4. Fetal hemoglobin in SCD children (n = 301)

due to SCD. Recurrent fever with allergic rashes and recurrent fainting attacks in one case each were unusual features, cause for which was not apparent. Palpable spleen was found in 25% of children in 0-1 year age group. The proportion of cases with palpable spleen, sometimes 6-7 cm below the costal margin gradually increased with age and was present in 97% of children by 12 years of age.

Total hemoglobin was between 6-10 g/dl in most of the children (77%). It was more than 10 g/dl in 12% of children (Fig. 3). With such hemoglobin level they thrived and grew well. No attempt was made to raise their hemoglobin with blood transfusion or iron therapy, though they needed extra folate supply regularly.

In most children fetal hemoglobin (HbF) was 5-30% (Fig. 4). It was less than 5% in less than 5% children. Significance of high HbF on clinical and hematological features of SCD is not clear[6].

About 65% of SCD children required hospitalisation some time or the other, while 15% needed it twice or more. This was required for severe attacks of pain, respiratory infection or severe anemia requiring blood transfusion. Unlike thalassaemia, transfusion was required only once in 26.6%, twice in 5.5% and more than twice in 5% of children only when anemia was life threatening.

Discussion

Sickle Cell Disease is not a disease of only the scheduled castes and scheduled tribes in India and should be suspected in children belonging to any caste or religion especially in the central belt of India. Recurrent attacks of musculoskeletal pain, anemia, jaundice and splenomegaly in a child are typical features which should arouse suspicion of SCD and promptly sickling test and hemoglobin electrophoresis should be done. Frequent respiratory infections, fever and anemia with or without jaundice or splenomegaly in a young child are also strong case for inves-

tigation for SCD. Regular use of long acting penicillin upto an age of 6-8 years is helpful in preventing such attacks which can precipitate a sickle crisis. Similarly, prompt treatment of any infection with adequate hydration and antibiotic is an essential part of management to prevent a crisis. Unlike thalassaemia blood transfusion is not a frequent requirement and should be used only as a life saving measure whenever hemoglobin level falls below 5 g/dl. Simple analgesics *e.g.* paracetamol alone or in combination with ibuprofane are adequate to give relief from musculoskeletal pains, which are rather very resistant to therapy in older children and adolescents. Regular folic acid supplement and maintaining good nutrition help in ensuring normal physical and mental growth and normal activity of the child inspite of a persistent hemoglobin level of 7-9 g/dl.

In fact attempts should not be made to raise the hemoglobin to near normal levels by repeated blood transfusion or iron therapy which may prove detrimental. In view of the large number of cases in the society and multiple cases in families there is a strong case for appropriate genetic counselling to the parents by pediatricians.

References

1. Kar BC, Devi S, Dash KC *et al.* The sickle gene is widespread in India. *Trans Royal Soc Trop Med Hyg* 1987; 81 : 275-278.
2. Kar BC, Satpathy RK, Kulozik AE *et al.* Sickle cell disease in Orissa state, India. *Lancet* 1986; 11 : 1198-1201.
3. Kar BC. Sickel cell disease in India. *J, Ass Phys India* 1991; 39 : 954-960.
4. Kulozik AE, Wainscot JS, Serjeant GR *et al.* Geographical survey of B-globin gene haplotypes, evidence for an independent Asian origin of the sickle cell mutation. *Am J Hum Genet* 1986; 39 : 239-244.
5. Kulozik AE, Bail S, Kar BC *et al.* Sickle cell B^+ thalassaemia in Orissa state, India. *Brit J Hematol* 1991; 77 : 215-220.
6. Kar BC, Devi S, Pattnaik G *et al.* HbF levels do not determine the clinical course of 'SS' disease. *Indian J Hematol Blood Transf* 1994; 12 : 225-229.