Brief Reports

Double Heterozygosity for Hemoglobin S and E

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Sickle cell disease (SCD) is widely prevalent in different parts of the country(1). The applies especially to Madhya Pradesh, Orissa, Andhra Pradesh, Gujarat and Vidharba district of Maharashtra. Scattered cases are seen and reported from each part of the country(2). Hemoglobin-E(Hb-E) is also commonly described from India especially from Bengal and Assam(3). Once again, occasional cases are reported from each part of the country(4). However, combination of Hb-S and Hb-E has never been reported from India. In fact, we could trace only 4 families for such a combination even in the English literature(5-8). We encountered a case with this combination and, due to its rarity, the same is being reported here.

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Case Report

An 18-year-old male was seen for vague abdominal discomfort, pain in lower extremities and loss of weight for 4 months. He gave no history of jaundice or blood transfusions. There was nothing to suggest recurrent infection, or painful episodes in the past. There was no history of consanguinity. Father expired during patient's childhood and mother was asymptomatic. He had one brother aged 24 years who was healthy.

Physical examination showed mild pallor, no icterus and just palpable spleen. His built and nutrition were average. Investigations revealed Hb 9 g/dl, PCV 28.3%, RBC 3.7 $\times$ 10^{12}/L, MCV 76 fl, MCH 24.3 pg, MCHC 31.8%, Total WBC count 9 $\times$ 10^9/L, P-63%, L-29%, E-5%, M-2%, B-1%, platelet count 262 $\times$ 10^9/L, reticulocyte count 3.3%, RBC morphology hypochromia +, microcytoses +, target cells +, poikilocytosis +, S. bilirubin (T) 1.3 mg/dl, (D) 0.4 mg/dl, SGOT 24 IU/L, SGPT 12 IU/L, S. creatinine 0.8 mg/L, S. protein (T) 7.2 g/L, S. albumin 4.4 g/dl, S. globulin 2.8 g/L, S. iron 72 μg/dl, total iron binding capacity 277 μg/dl, transferrin saturation 26%. Hb electrophoresis at alkaline pH (8.6) using Beckman paragon Hb gel system showed HbS + E patterns (*Fig.*) with HbS of 68% and HbE of 29.9%. HbF (Betke's technique) was 2.1%. Sickling test (sodium metabisulphite technique) was positive. Presence of HbE was confirmed by Agaros Electrophoresis at acidic pH, where Hb-C and Hb-E separate out. Other investigations showed: RBC G-6-PD screen normal, Coomb's test (D)

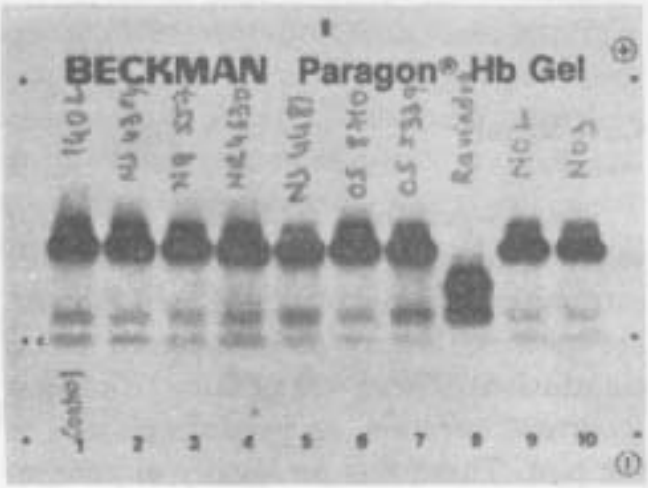

Fig. Hemoglobin electrophoresis at pH 8.6 using Beckman Paragon Hb gel system showing hemoglobin E and hemoglobin S bands (No. 8) in a case of double heterozygosity for hemoglobin S & E.

negative, *X*-ray chest normal, and ECG normal. Other family members were not available for work up.

Discussion

Various types of sickle cell disease (SCD) have been described. Of these, the commonest are homozygous sickle cell disease (Hb-SS), Sickle/B-Thalassemia and HbSC disease. Rarely one comes across cases of double heterozygosity for HbS with HbD, HbO Arab, HbE and hereditary persistence of Hb-F. As stated earlier, both Hb-S and Hb-E have been well described from India(1-4). However, no case of hemoglobin-S/E has been published so far. In fact, we could trace only 5 cases from 4 families of SE disease being reported in the English literature(5-8).

HbE and HbS are prevalent in different regions. HbS is far more common in tribal population while HbE is essentially restricted to North East part of the country. Inspite of the jet age, it is uncommon for the tribal population to travel to other parts of the country or mix with other communities. This probably, is the reason for rarity of Hb-SE disease in India. Similarly, in the world as a whole HbE is common in the far East where Hb-S is relatively uncommon. Also, even if such combination occurs, the resultant disease is mild. This is obvious from the details of 5 cases of SE disease published so far (*Table I*). Most of the patients maintain reasonably good hemoglobin, normal or slightly low RBC indices. Occasionally, subjects have mild

TABLE I–*Hematological Features in Cases of HbS-E Diseases*

Case No.	Age/Sex	Hb (g/dl)	PCV %	MCHC %	RBC $\times 10^{12}$/l	MCV (fl)	Retic %	S. bilirubin (mg/dl)	Ref No.
1.	70/F+	11.4	45	25	4.75	95	4	1.2	5
2.	30/M+	13.2	52	25	5.60	93	4	1.6	5
3.	63/F	84.0	31	27	3.20	97	4	1.3	6
4.	20/F	12.7	34	37	4.66	71	–	–	7
5.	26/F	12.7	38	34	5.29	71	2	–	8
6.	18/M	9.0	28.3	31.8	3.70	76	3.3	1.3	Present report

+ Mother and son

hyperbilirubinemia and reticulocytosis. The peripheral blood film is usually unremarkable. It is also well known that higher HbF protects against sicklings and that could be an additional factor producing mild clinical picture.

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Hemophagocytic Syndrome

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Virus-associated hemophagocytic syndrome (VAHS) is characterized by benign hyperplasia of histiocytes showing hemophagocytosis in the reticulo-endothelial system. The patients usually present with high grade fever, failure to thrive, massive hepatosplenomegaly, lymphadenopathy and bleeding tendencies. Since its first report in 1979 by Risdall *et al.*(1), the understanding of this syndrome has improved considerably. We report a similar case, admitted to the pediatric unit of the hospital.

Case Report

A 7-year-old boy presented with fever of 7 days duration, headache and blood stained vomiting. He was treated with parenteral chloramphenicol, two days prior to admission, for suspected enteric perforation. On admission the child was toxic but responding to commands and had signs of meningeal irritation. He had scattered ecchymotic spots, bleeding from intravenous sites, subconjunctival and retinal hemorrhages, abdominal distension and bleeding

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