Letters to the Editor

Hereditary Spherocytosis in a Family from Tamil Nadu

Hemolytic anemias are generally more prevalent in north India than south, and among the tribal population throughout the country. Of the hemolytic anemias, thalassemia, sickle cell anemia and G6PD deficiency are more prevalent than hereditary spherocytosis (HS)(1-3). Literature search did not reveal any report on this disorder from Tamil Nadu. We are describing a family with this disorder from Virudhachalam in Tamil Nadu.

Three siblings, two males aged twelve and nine years, and one female aged eleven years who were brought to our hospital for the complaints of breathlessness on exertion, puffiness of face and abdominal pain were observed to have mild jaundice, anemia and splenomegaly. The past history was unremarkable except for recurrent episodes of jaundice in all of them. Since the jaundice showed fluctuation in severity and several members of their family also had jaundice and were doing well they did not show further interest in the symptom. The family tree is shown in the *Fig. I*

From the history and clinical findings a diagnosis of familial hemolytic anemia was made. Investigations were suggestive of hereditary spherocytosis. Ultrasound examination of the abdomen did not show any evidence of cholelithiasis. Screening of other family members could not be done because of practical difficulties. Based on the laboratory findings in these children and from the history, it was concluded that other family members with jaundice also probably had HS.

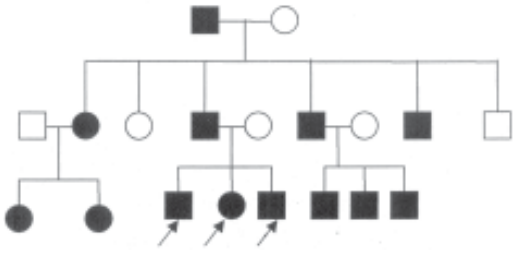

Fig. 1. Pedigree chart.

About 80% of patients with HS show an autosomal dominant pattern of inheritance(4) as seen in the present family.

In the dominantly inherited form, the deficiency is mild and hence the anemia is mild, while in the non dominant forms the deficiency is greater and the anemia severe. The mild nature of the disorder in our children could be because of the dominant pattern of inheritance in them.

Splenectomy results in significant improvement in the clinical condition and prevents complications such as cholelithiasis. However, the advantages of splenectomy have to be weighed against the complications of surgery. In mild cases splenectomy may not be indicated. Since our children had only a mild disorder, splenectomy was not advised. They were put on folic acid suppplementation and are doing well on follow up.

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Hypophosphatemic Rickets with Epidermal Nevus Syndrome

Epidermal nevus syndrome (ENS) is defined as combination of a sebaceous nevus with any single significant skeletal, neurologic or ocular abnormality(1). There are 11 cases of ENS in association with hypophosphatemic rickets reported in English literature (2).

A 7½-year-old boy presented with difficulty in walking due to progressive deformity of the lower limbs since 2 years of age, poor height gain and weakness with wasting of the left upper and lower limb. The patient was born at term of non-consanginous marriage. Motor and mental milestones were normal. The family history was non-contributory. He had been treated with high dose parenteral vitamin D with no significant clinical benefit. On examination he was normotensive, with widened bilateral wrists, right knock knee and mild scoliosis. He was prepubertal in his development. There was left spastic hemiparesis involving both the left upper and lower limbs. He had hyperkeratotic, hyperpigmented nevus on the scrotal skin and right big toe (*Fig. 1*).

Investigations revealed serum calcium: 9.6 mg/dL, serum phosphate: 1.8mg/dL; alkaline phosphatase: 982 U/L. Acid load test was negative. TMP/GFR: 0.5, Magnetic resonance imaging of the brain showed interdigitation of gyri in the right frontal lobe. *X*-ray showed changes of rickets. Biopsy of the skin lesion was suggestive of epidermal nevus. He was started on phosphate mixture

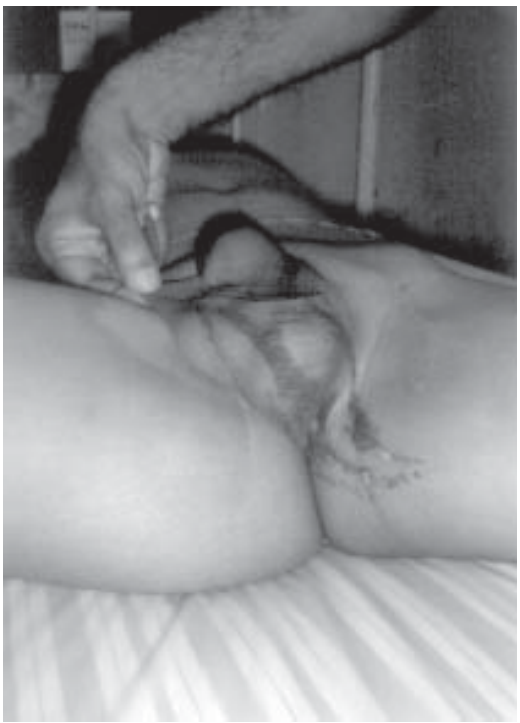

Fig. 1. Epidermal nevus on scrotal skin