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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-consanguineous 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

and calcitriol, the doses of which were adjusted based on clinical response, parathyroid hormone levels and radiology. Phosphate was maintained between 1.7 and 3.2 mg/dL with this treatment. Follow up evaluations did not show hypercalciuria or nephrocalcinosis. Close clinical follow up was kept anticipating precocious puberty which our patient did not develop.

In the last 4 years, he has grown at the rate of 10.8 cm /year. There is significant radiological improvement in the lesions. He still has genu valgum for which surgical intervention is planned after his growth is complete.

This case illustrates the rare association of hypophosphatemic rickets with epidermal nevus and the associated skeletal and neurological abnormalities. In ENS, skeletal involvement has been reported to include bone cysts, kyphoscoliosis, joint deformities, cranial involvement and resistant rickets(3). In 2 patients, surgical excision of the tumor resulted in substantial remission of the biochemical and clinical parameters(4). The causal association between hypophosphatemic rickets and ENS has been suggested by the demonstration of a phosphaturic substance extracted from the skin lesions of a child with ENS and rickets(4).

Epidermal nevus is essential for the diagnosis of ENS. Other skin lesions that can occur in ENS are vascular nevi, hypopigmented macules, café au lait macules and multiple pigmented nevi(5). The nevi can be complicated by development of secondary malignancies and adnexal tumors(5). The

central nervous system abnormalities in patients with ENS include seizures, hemiparesis, developmental delay, mental retardation, abnormal cerebral gyration, underdeveloped temporal lobe and sensorineural deafness(1,5). Precocious puberty rarely observed in association with ENS(3). Identifying rickets at an early age in patients with ENS would lead to optimal treatment to realize their height potential.

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