Vitamin D deficiency rickets with Lamellar ichthyosis

Sir,

A five-year-old boy presented with history of multiple fractures involving both upper and lower limbs and progressive bony deformities leading to inability to walk over the past two years.

He was the first child of non-consanguinous parents and was born normally. He was noticed to have thick skin right from birth. He was immunized appropriately and his dietary intake (including that of vitamin D) was adequate. There was no history suggestive of malabsorption, renal disorder or decreased exposure to sunlight.

His weight was 13 kg (< 50th centile), height 87 cm (< 3rd centile) and head circumference 50 cm. He had signs of rickets in the form of frontal bossing of the skull, widening of the wrists, rachitic rosary and protuberant abdomen without organomegaly. His upper arm and lower limb bones showed marked lateral bowing. Both fontanelles were closed. He had generalized thickening and hyperpigmentation of the skin with scaling.

As shown in Table 1, investigations confirmed the presence of rickets. The serum level of 25 OH Vitamin D_3 was less than 5 ng/ml (Normal: 9-37.6 ng/ml). Serum creatinine levels and arterial blood gas analysis were within normal limits. Radiographs of the wrists also corroborated the diagnosis of rickets as they showed cupping and fraying of the distal ends of the radius and ulna with growth plate widening [Figure 1A]. Radiograph of upper arms and lower limbs showed bowing and shortening with multiple fractures. The skin biopsy showed a markedly thickened stratum corneum and epidermal thickening consistent with lamellar ichthyosis.

It was postulated that the patient's vitamin D deficiency was secondary to lamellar ichthyosis as a result of poor penetration of skin by ultraviolet rays. The child showed clinical, radiological and biochemical response [Table 1, Figure 1B] to oral vitamin D_3 granules 30,000 units daily. He was advised regular application of liquid paraffin over the body. He underwent bilateral femoral closed wedge osteotomy and bilateral tibial osteotomy with rush pin nailing of both tibia for correction of deformities. With the aid of calipers, he was able to walk [Figure 2]. His height velocity was 6cm/year over the next two years of follow-up. He was advised to continue vitamin D_3 supplementation lifelong to prevent recurrence of the deficiency and development of further complications.

Cutaneous hyperproliferative states like ichthyosiform dermatoses are uncommon causes of rickets in children.[1] Lamellar ichthyosis is an autosomal recessive disorder that is apparent at birth and is present throughout life. The following factors are proposed for development of rickets in skin disorders, (i) alterations in epidermal cholesterol metabolism possibly involving vitamin D receptors, (ii) increased keratinocyte proliferation resulting in poor or no penetration of skin by sunlight, (iii) associated vitamin D dependent rickets and (iv) limited sun exposure to prevent sunburn and sunstroke.[2] Milstone *et al* reported elevated parathyroid hormone and low-to-normal 25-hydroxyvitamin D values in patients with various disorders of keratinization, including three adult patients with lamellar ichthyosis.[3]

In our case vitamin D deficient rickets is most likely to be due to poor penetration of skin by sunlight resulting from increased keratinocyte proliferation. A low serum 25-hydroxyvitamin D_3 level in the absence of other causes of vitamin D deficiency supported our diagnosis. This boy showed marked improvement with vitamin D supplements, clinically and biochemically.

Although the ichthyosis did not improve with resolution of

Table 1: Summary of the biochemical parameters before and after replacement therapy in a child with lamellar ichthyosis and vitamin-D-deficient rickets

Parameter	Reference value	Pre-treatment	Post- treatment		
			4 weeks	8 weeks	12 weeks
S. Calcium (mg/dL)	8.8-10.8	7.7	8.7	9.0	9.7
S. Phosphorus (mg/dL)	3.7-5.6	2.2	6.2	5.2	4.3
S. Alkaline phosphatase (U/L)	145-420	2310	1082	400	240

Reference value: 16th edition Nelson textbook of Pediatrics

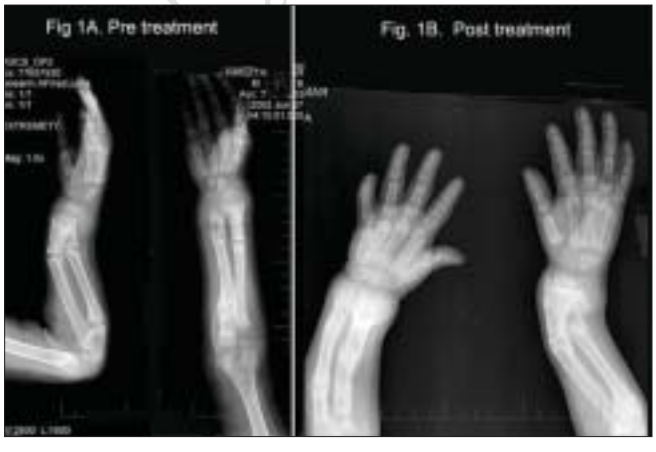

Figure 1: (A) Radiograph of upper arm showing bowing and shortening with multiple fractures and cupping and fraying of distal ends of radius and ulna with growth plate widening. (B) Radiograph of wrists showing good healing of rickets after vitamin D supplementation

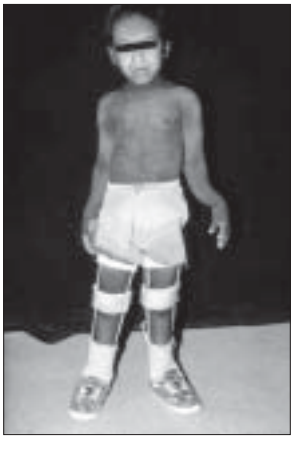

Figure 2: Clinical photograph of a 5 year old boy with lamellar ichthyosis and vitamin D deficiency rickets after vitamin D supplementation and surgery. He is able to walk with special shoes.

vitamin D deficiency and rickets, one of two children treated with topical calcipotriene showed improvement in the treated areas of skin. Calcipotriene does not seem to be effective in reversing systemic vitamin D deficiency but can be effective in improving the severity of skin disease in children with ichthyosis.[4] Children with vitamin D deficiency secondary to skin disorders need lifelong supplementation with vitamin D to prevent its deficiency and consequences.[5]

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References

1. Dayal D, Kumar L, Singh M. Non-bullous ichthyosiform erythroderma with rickets. Indian Pediatr 2002;39:207-8.
2. Griffiths WA, Judge MR, Leigh IM. Disorders of keratinization. *In*: Rook, Wilkinson, editors. Ebling Textbook of Dermatology, 6th ed. Blackwell Science: Oxford; 1998. p. 1483-8.
3. Milstone LM, Ellison AF, Insogna KL. Serum parathyroid hormone level is elevated in some patients with disorders of keratinization. Arch Dermatol 1992;128:926-30.
4. Thacher TD, Fischer PR, Pettifor JM, Darmstadt GL. Nutritional rickets in ichthyosis and response to calcipotriene. Pediatrics 2004;114:e119-23.
5. Holm AL, Goldsmith LA. Impetigo herpetiformis associated with hypocalcemia of congenital rickets. Arch Dermatol 1991;127:91-5