Case Report

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A rare case of congenital lamellar icthyosis and rickets with pathological fractures

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Abstract:
Lamellar ichthyosis is an autosomal recessive disorder that is apparent at birth and is present throughout life. Lamellar ichthyosis is rare disease. Many factors are proposed for development of rickets in skin disorders, alterations in epidermal cholesterol metabolism possibly involving vitamin D receptors, increased keratinocyte proliferation and limited sun exposure to prevent sunburn and sunstroke. Here we report a case of five-year-old boy presented with history of multiple fractures involving both upper and right lower limb and ichthyosis.

Key words: Lamellar ichthyosis, vitamin D, ichthyosiform dermatoses

Introduction:
Cutaneous hyper proliferative 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. Incidence of lamellar ichthyosis is 1 in 3, 00,000 population. 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 [3,4]

Case Report
A five-year-old boy presented with history of multiple fractures involving both upper and right lower limb, leading to inability to stand without support over the past 1 month. He was the fourth child of consanguineous parents (second degree) and first male child and was born normally. He was noticed to have thick skin right from birth all over the body [Figure1], with delayed milestones. He was immunized appropriately and his dietary intake

history was adequate. There was no history suggestive of malabsorption, renal disorder or decreased exposure to sunlight.

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, scaling and, hyper pigmentation.

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 30-100 ng/ml). Serum urea, creatinine levels and arterial blood gas analysis were within normal limits. Radiographs of the wrists and knee also corroborated the diagnosis of rickets as they showed cupping and fraying of the distal ends of the radius, ulna, distal end femur and upper end tibia with growth plate widening and generalized osteopenia. Radiograph of forearms and lower limbs showed both ulna fractures with evidence of callus formation and fracture shaft of right femur [Figure 2]. He also had eye lesions in the form of bilateral cicatricial ectropion with Conjunctival keratinisation [Figure 3] and difficulty in opening the mouth, loss of eyebrows which are usually seen as associated features of lamellar icthyosis and the same was treated with Lacryl PF eye ointment tid (BE), Chlorochol eye ointment bd (BE) .The skin biopsy showed a markedly thickened stratum corneum and epidermal thickening consistent with lamellar ichthyosis.

The child showed clinical and radiological response [Table 1], [Figure 4] to 6,00,000 IU of vitamin D 3 IM single dose with calcium supplements daily for 4 weeks. He was advised regular application of liquid paraffin over the body. Fracture shaft of femur was immobilised in a plaster cast for 6 weeks. He was advised to continue oral vitamin D3 supplementation after 4 weeks for lifelong to prevent recurrence of the deficiency and development of further complications.

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.

However, ichthyosis did not improve with resolution of vitamin D deficiency and rickets. Children with vitamin D deficiency secondary to skin disorders need lifelong supplementation with vitamin D to prevent its deficiency and consequences.

Discussion

Cutaneous hyper proliferative 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. Incidence of lamellar ichthyosis is 1 in 3,00,000 population. 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]. Milestone et al reported elevated parathyroid hormone and low-to-normal 25-hydroxyvitamin D values in patients with various disorders of keratinisation, including three adult patients with lamellar ichthyosis [3].

Conclusion

This case report emphasizes on early recognition and treatment of lamellar ichthyosis to prevent pathological fracture and associated complication.

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Table 1: Routine Blood Investigations

	observed values	**Normal Values**
RBC	4.55 millions/cu mm	3.9-5.3 millions/cu mm
Hb	9.7 g/dl	10.5-14 g/dl
HCT	34.5%	33-42%
MCV	75.8 fl	70-74 fl
MCH	21.3 pg	24-30 pg
MCHC	28.1 g/dl	31-37 g/dl
WBC	17,880 cells/dl	5,500-15,500 cells/dl
NEUTROPHILS	40 %	54-62%
LYMPHOCYTES	**50.8 %**	25-33%
MONOCYTES	4.3 %	3-7%
BASOPHILS	0.5%	0-0.75%
EOSINOPHILS	4.4%	1-3%
ESR	20 at the end of 1 hour	0-13 at the end of 1 hr
RECTIC COUNT	1%	1-3 %
BLOOD GROUP	'O' positive	
PERIPHERAL SMEAR	Dimorphic anaemia with lymphocytosis	

Table 2:Biochemical Investigations

	observed values	**Normal Values**
SERUMCALCIUM	10.6 mg%	8.8-10.8 mg%
SERUM PHOSPHORUS	2.9 mg%	3.7-5.8 mg%
SERUM ALKALINE PHOSPHATASE	1403 IU	145-420 IU
SERUM INTACT PTH	70 pg/ml	15-65 pg/ml
SERUM 25-OH VITAMIN -D	4.8 ng/ml	30-100 ng/ml

Table 3: Renal Function Test

	observed values	**Normal Values**
BLOOD UREA	18 mg%	5-18 mg%
SERUM CREATININE	0.6 mg%	0.3-0.7 mg%

Figure 1: Pathological fracture

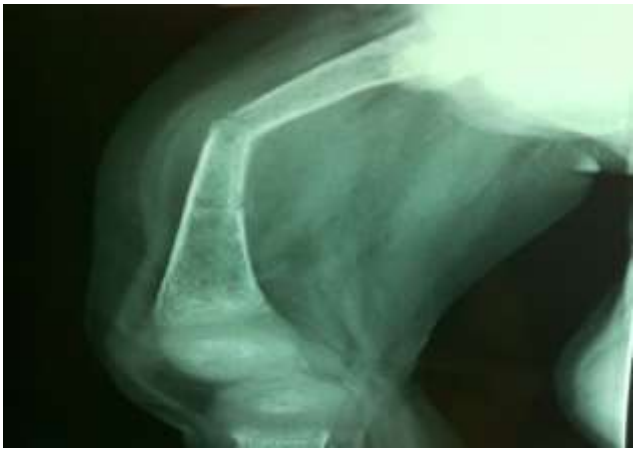

Figure 2: Generalised icthyosis of skin all over the body

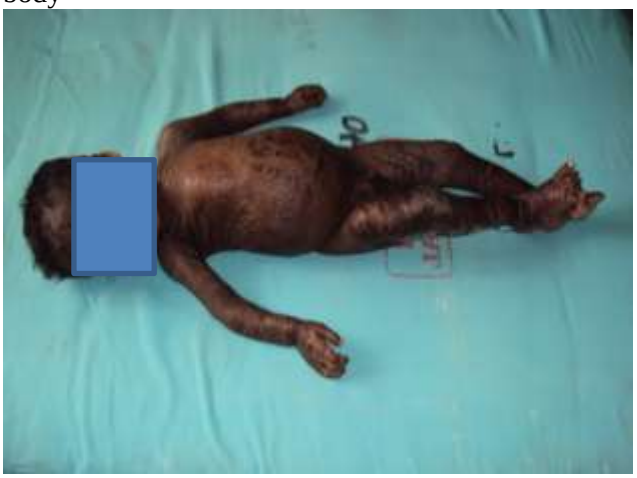

Figure 3

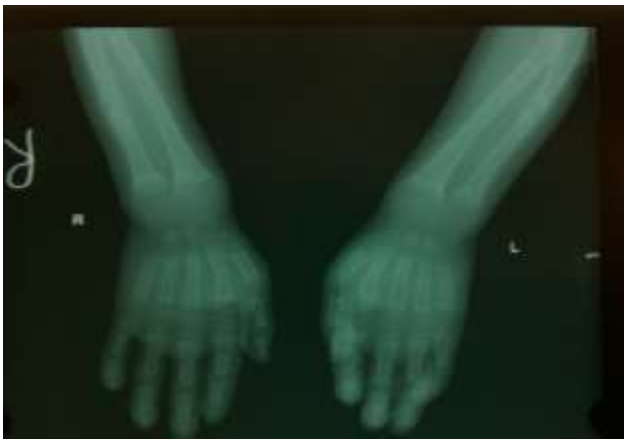

Figure 4:

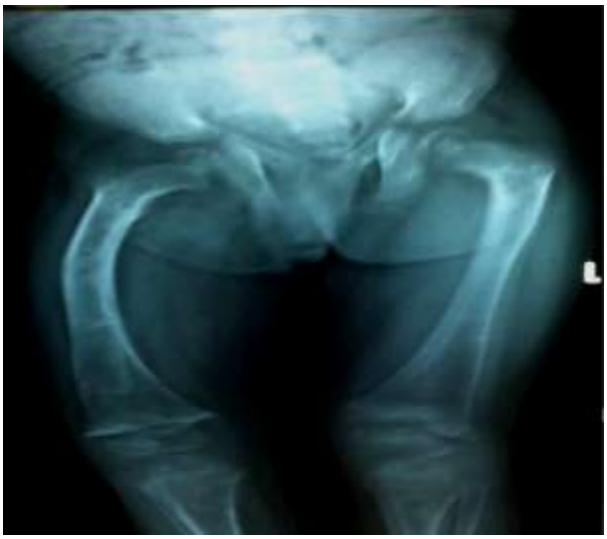

Figure 5: