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Rickets secondary to lamellar ichthyosis in two Indian male siblings in a family

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Abstract: We report two cases of rickets in male siblings of a family secondary to a disorder of skin keratinisation, lamellar ichthyosis. Nine years old male child presented with knock knee and generalised scaling of skin all over body. His 6 year old younger sibling also had scaling all over body with genu valgus deformity. Clinical, laboratory, radiological and histopathological investigations confirmed diagnosis of rickets with lamellar ichthyosis. Both children were treated for rickets with parental vitamin D3 followed by daily calcium and vitamin D supplementation.
Keywords: Lamellar Ichthyosis, Rickets, Genu Valgus

INTRODUCTION

The word "rickets" is derived from "wrickken" which means to twist [1]. Rickets, a disease of growing bone, occurs due to defective mineralisation of growth plate before epiphyseal closure. Rickets is predominantly due to deficiency of vitamin D (most commonly nutritional disease). Rickets may rarely occur due to disorders of skin keratinisation [2, 3].

Lamellar ichthyosis is a disorder of skin keratinisation usually of autosomal recessive inheritance. Affected child can present at birth as collodion baby. After shedding of membrane child develops large scales all over body including flexural surfaces [2, 3]. These children are prone to develop rickets which can lead to severe bone deformities with disability, and simple parental education with calcium and vitamin D supplementation can prevent these complications. To the best of our knowledge no cases have been reported in 2 siblings in same family.

CASE REPORT

9 year old male child born of 2nd degree consanguineous marriage presented to pediatric out patient department with c/o deformity of bilateral lower limbs since 2 years and scaling of skin. Child was born of a full term vaginal delivery and admitted in NICU for collodion baby for 1 month. Membrane was shed after 2 months and mother noticed skin becoming dry and scaling all over the body. Developmental history was normal. Child had difficulty in walking due to knock knee since 2 years. Mother also gives history of minimal exposure to skin and covering whole body with clothes while going outside. There was no history s/o malabsorption or renal disorder.

On examination child has short stature. Dermatological examination revealed generalised scaling all over body predominantly involving flexural surfaces. Palms and soles were hyperkeratotic. There was minimal erythema of skin. Teeth, hair, nails and mucous membranes were normal. On skeletal system examination there was wrist widening, knock knee and cubitus valgus deformity.

Mother also gives history of similar complaints in younger male sibling of 5 years old. This child was absolutely normal at birth and he developed hyperkeratosis and scaling of skin since the age of 6 months. On examination child had wrist widening, cubitus valgus and genu valgus deformity.

Investigations of older child showed low serum calcium (6.8 mg/dl), low 25-OH vitamin D3 (10.2 ng/dl) and high alkaline phosphatise (845 IU/L). Younger child also had low serum calcium (7.4 mg/dl), low 25-OH vitamin D3 (11.8 ng/dl) and mildly elevated alkaline phosphatise (623 IU/L). Both children X ray of limbs showed cupping and fraying of metaphysis with osteopenia s/o rickets. Complete hemogram, urine examination, stool examination, renal function test and ultrasound abdomen were normal. Skin biopsy of both children confirmed diagnosis of lamellar ichthyosis.

Both children were given parental vitamin D3 600000 units followed by oral calcium (100 mg/kg/day) and vitamin D (400 IU/day) along topical emollients and keratolytics.

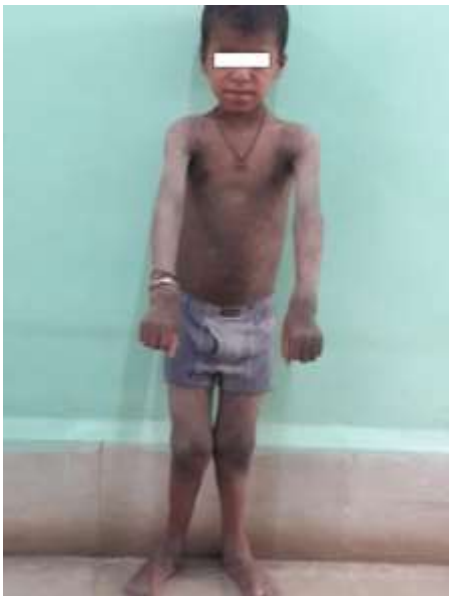

Fig-1: Child with lamellar icthyosis with genu valgus deformity

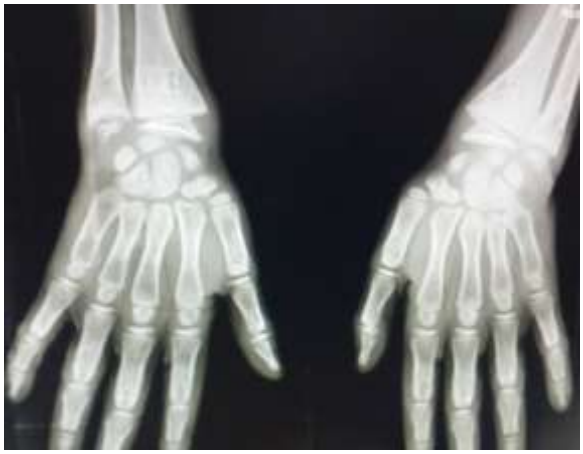

Fig-2: Wrist X ray suggestive of rickets

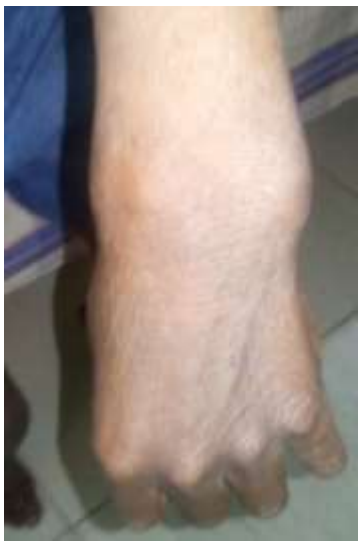

Fig-3: Wrist widening

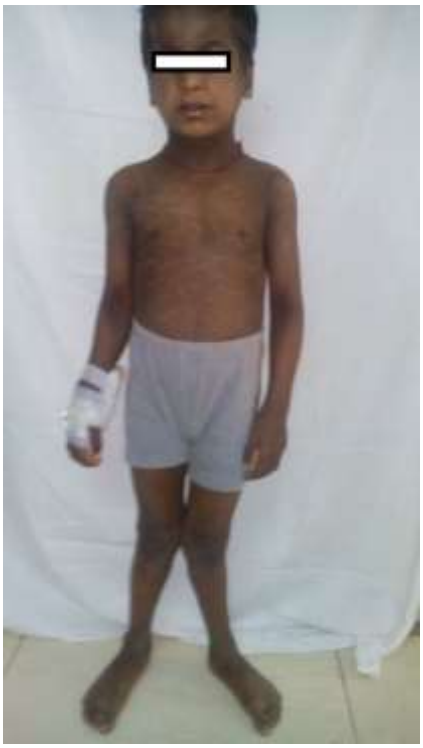

Fig-4: Younger sibling (Lamellar icthyosis with genu valgus deformity)

DISCUSSION

Rickets is a disorder of bone mineralisation commonly seen in young children. There is generalised softening of bones in this disorder. As the child starts walking it leads to a variety of bone deformities like bow legs, knock knees. It is most commonly due to nutritional deficiency of vitamin D or calcium [4]. Rickets secondary to ichthyosis is very rare [5].

Lamellar ichthyosis is an autosomal recessive disorder most commonly due to mutation of transglutaminase I. Rickets has been previously reported to occur in association with lamellar ichthyosis[6,7], epidermolytic hyperkeratosis[6,8,9], X-linked ichthyosis[10], Chouhan et al [11] conducted study of 45 Children and adolescents with ichthyosiform erythroderma, and reported these children especially with pigmented skin (types IV-VI), are at increased risk of developing vitamin D deficiency and clinical rickets. In a series of 41 Sudanese children with nutritional rickets, three were found to have ichthyosis[12]. Sethuram et al reported that severe skeletal involvement due to rickets in association with ichthyosis is exceptionally rare[5]. In our report both children had severe skeletal deformities.

Maintenance of healthy skeleton requires vitamin D, the main source of which is its biosynthesis in the skin. On sun exposure, 7-dehydrocholesterol present in keratinocytes is converted to cholecalciferol (vitamin D3) which undergoes hydroxylation to form

active vitamin D3. The keratinocytes are the only cells in the body having the entire pathway of vitamin D synthesis. Due to defective synthesis of vitamin D in the diseased epidermis and excessive loss of calcium through skin, stimulation of PTH secretion occurs, which in turn puts the children with ichthyosis at increased risk of rickets[13].

Contributing factors for development of rickets in patients with icthyotic skin disorder are

1. Poor or no penetration of skin by sunlight secondary to hyperkeratosis[14].
2. Lack of sun exposure to prevent heat intolerance[14].
3. Defective synthesis of vitamin D in diseased epidermis leading to vitamin D dependent rickets [11].
4. Calcium loss through skin shedding.
5. Alterations in epidermal cholesterol metabolism possibly involving vitamin D receptors.

In our case report two siblings are affected, so parents need genetic counselling to prevent recurrence of the disease. All these patients with icthyotic skin disorders needs to be started with vitamin D and calcium supplementation in infancy and should be continued lifelong.

CONCLUSION

In India because of social stigma and due to heat intolerance, child with ichthyosis will not be exposed to sunlight. Nutritional deficiency and malnutrition is rampant in India. Above two reasons predisposes Indian child with ichthyosis to rickets. Hence we conclude being a rare but crippling skeletal deformities of rickets can be prevented by health education to parents of icthyotic skin disorder.

Consent has been taken from the parents for publication.

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REFERENCES

1. Tiosano D, Hochberg Z; Hypophosphatemia: the common denominator of all rickets. J Bone Miner Metab, 2009;27:392-401.
2. Linda S, Nield., Kamath D; Fever. In: Nelson Textbook of pediatrics.(Kliegman RM, Behrman RE, Jenson HB, Stanton BF, eds), 19th ed.. Philadelphia: Saunders; 2011:200-8,2267-68
3. Fleckman P, DiGiovanna JJ; The Ichthyoses. In: Fitzpatrick's Dermatology in General Medicine (Wolff K, Goldsmith LA, Katz SI, Gilchrest BA, Paller AS, Leffell DJ, eds), 7th edn. USA: McGraw-Hill Inc; 2008:401-423.
4. Balasubramanian K, Rajeswari J, Gulab, Govil Y C, Agarwal AK, Kumar A, et al.; Varying role of vitamin D deficiency in the etiology of rickets in young children vs. adolescents in northern India. J Trop Pediatr., 2003;49: 201–206.
5. Sethuraman G, Khaitan B, Dash S, Chandramohan K, Sharma V, Kabra M, et al.; Ichthyosiform erythroderma with rickets: Report of five cases. Br J Dermatol., 2008;158:603-606.
6. Kothari D, Doshi B, Garg G, Khopkar US; Ichthyosis Associated with Rickets in Two Indian Children. Indian J Dermatol, 2013; 58(3): 244.
7. Ali R, Aman S, Nadeem M; Lamellar Ichthyosis with Rickets. Pak J Med Sci , 2013; 29(2):660-662.
8. Nayak S, Behera SK, Acharjya B, Sahu A, Mishra D; Epidermolytic hyperkeratosis with rickets. Indian J Dermatol Venereol Leprol, 2006;72:139–142.
9. Perosa A, Sardella L, Acquafredda A, Mazzotta F, Bonifazi E: Epidermolytic ichthyosis and rickets. Eur. J. Pediat. Dermatol. 2004; 14: 13-16
10. Milstone LM, Bale SJ, Insogna KL; Secondary hyperparathyroidism in patients with ichthyosis is not caused by vitamin D deficiency or ingestion of retinoids. Arch Dermatol. 1993;129:648.
11. Chouhan K, Sethuraman G, Gupta N, Sharma VK, Kabra M, Khaitan BK et al.; Vitamin D deficiency and rickets in children and adolescents with ichthyosiform erythroderma in type IV and V skin. Br J Dermatol., 2012 ; 166(3):608-615.
12. El Hag Al, Karrar ZA; Nutritional vitamin D deficiency rickets in Sudanese children. Ann Trop Pediatr,1995;15: 69-76.
13. Milestone LM, Ellison AF, Insogna KL; Serum parathyroid hormone level is elevated in some patients with disorder of keratinization. Arch Dermatol, 1992;128:926–30.
14. Griffiths WAD, Judge MR, Liegh IM; Disorders of keratinization. In: Champion RH, Burton JL, Burns DA, Breathnach SM, editors. Rook's Textbook of Dermatology. 6th ed. Oxford: Blackwell Science; 1998;1483–588.