Rickets in the Tropics: not always nutritional

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DESCRIPTION

Vitamin D-dependent rickets type I

A 6-year-old girl belonging to the lower socio-economic class of a Muslim community, product of consanguineous marriage presented in the paediatric outpatient department with a history of progressive bowing of limbs, recurrent chest infections requiring hospitalisations in the past and failure to thrive. Calorie and protein consumption was just one-third of the requirement. Family history of similar problems was present in the mother. The mother had frank rickets beginning at the age of 1 and progressively increasing with age. The mother was treated at a private clinic for her disease with 1,25-dihydroxycholecalciferol (Calcitriol). A clinical examination revealed florid manifestations of rickets (figure 1 A–D). Investigations showed near normal 25-hydroxy vitamin D and a low concentration of 1,25-dihydroxyvitamin D supporting the diagnosis of vitamin D-dependent rickets type-I

(VDDR-I; table 1 and figures 2 and 3). VDDR-I also called as pseudo-vitamin D deficiency rickets, is an autosomal recessive disorder occurring due to impaired activity of renal 25-hydroxyvitamin D 1 α -hydroxylase associated with raised 25-hydroxyvitamin D concentrations and markedly low to undetectable concentrations of 1,25-dihydroxyvitamin D.¹

Vitamin D-dependent rickets type-II in siblings

Two male siblings of age 9 and 2.5 years, respectively, belonging to a poor Hindu family presented with progressive deformity of the limbs, not gaining height, hairfall beyond the neonatal period preceded by folliculitis and premature falling of teeth. Younger sibling had painful lower limbs and did not start walking. On examination, all the growth parameters were below third centile. Both the children had classical manifestations of severe rickets along with peculiar features

Figure 1 (A) Features of florid rickets: marked bowing of forearms and legs. (B) Features of florid rickets: narrow bell-shaped pigeon chest with rachitic rosary and marked bowing of forearms. (C) Yellow enamel changes and (D) scoliosis.

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Table 1 Investigations of three cases of pathological rickets

Investigations	Normal	VDDR-I	VDDR-II (sibling1)	VDDR-II (sibling 2)
S.calcium (mmol/L)	2.25–2.75	1.52	2.07	2.25
S.phosphorus (mmol/L)	1.13–1.45	0.58	1.19	1.16
Alkaline phosphatase (U/L)	300	1001	1457	548
Parathormone (pg/mL)	11.1–79.5	98.8	–	–
25 (OH) vitamin D (μ g/L)	30	29.6	–	–
1,25 (OH) ₂ vitamin D (nmol/L)	47.04–130.32	21.6	–	–

Figure 2 (A) Vitamin D-dependent rickets type II (VDDR-II) sibling 1 showing genu valgum and alopecia totalis. (B) VDDR-II sibling 2 showing folliculitis (precedes hair loss) and alopecia. (C) VDDR-II sibling 1 showing premature loss of tooth.

of VDDR-II in the form of alopecia totalis, epidermal cyst and premature falling of teeth^{2 3} (figure 2A–C). Owing to the financial constraints of the patients' family, 1,25-dihydroxy-vitamin D and parathyroid hormone could not be performed but alopecia and premature falling of teeth were the strong pointers to this rare diagnosis.^{2 3} Investigations are

shown in table 1. VDDR-II also known as hereditary 1,25-dihydroxyvitamin D resistant rickets, is a rare autosomal recessive disease that arises as a result of mutations in the gene encoding the vitamin D receptor. It usually presents with early onset severe rickets and alopecia with elevated circulating levels of 1,25-dihydroxyvitamin D3.⁴

Figure 3 Radiological features of vitamin D-dependent rickets type I: bowing, cupping, fraying and widening of the metaphyses, severe osteopenia and pathological fractures.

Learning points

- ▶ All cases of rickets are not nutritional in origin even in tropical countries.
- ▶ Genetic causes should be strongly considered if patient presents beyond 2 years of age with severe rickets, pathological fractures, positive family history and failure to respond to vitamin D.
- ▶ Alopecia and premature falling of teeth are important clinical pointers to vitamin D-dependent rickets type II.

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and AP verified the diagnosis and other scientific facts. All the authors were responsible for clinical follow-up of the case. All authors read and approved the final version of the manuscript.

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