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RESEARCH ARTICLE

CASE OF REFRACTORY RICKETS -VITAMIN D DEPENDANT RICKET TYPE I.

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ABSTRACT

There are two types of vitamin D dependent rickets (VDDR) that cause rickets in children. Both occurs due to metabolic defects in vitamin D metabolism,Vitamin D dependent rickets type 1 (VDDR-I) is caused by defect in renal conversion of calcidiol (25OHD) to calcitriol (1,25(OH)2D) by the enzyme 1alpha-hydroxylase. Vitamin D dependent rickets type 2 (VDDR-II) is caused by a defect in the vitamin D receptor (VDR). We report a case affected by VDDR-I, in developing country like INDIA where vitamin D deficiency rickets is still prevalent. Although it is difficult to differentiate between vitamin D dependant rickets & Vitamin D deficiency rickets, its important As VDDR causes more morbidity and mortality.

INTRODUCTION

Vitamin D3 (cholecalciferol), synthesized in the epidermis in response to UV radiation, and dietary vitamin D2 (ergocalciferol, synthesized in plants) are devoid of any biologic activity. Vitamin D hormonal activity is due primarily to the hydroxylated metabolite of vitamin D3, 1-alpha, 25-dihydroxyvitamin D3 (calcitriol), the actions of which are mediated by the vitamin D receptor (1; 2). In the liver, vitamin D 25-hydroxylase catalyzes the initial hydroxylation of vitamin D at carbon 25; in the kidney, 1-alpha-hydroxylase catalyzes the hydroxylation and metabolic activation of 25-hydroxyvitamin D3 into 1,25-dihydroxyvitamin D3. The active metabolite 1,25(OH)2D3 binds and activates the nuclear vitamin D receptor, with subsequent regulation of physiologic events such as calcium homeostasis and cellular differentiation and proliferation (3). Disorders of vitamin D metabolism or action lead to defective bone mineralization and clinical features including intestinal malabsorption of calcium, hypocalcemia, secondary hyperparathyroidism, increased renal clearance of phosphorus, and hypophosphatemia. The combination of hypocalcemia and hypophosphatemia causes impaired mineralization of bone that results in rickets and osteomalacia (2).

Case

18 month old male came with c/o repeated LRTI, not gaining weight; developmental delay. On examination vitals were stable. Anterior fontanel was wide open, bossing of forehead, ricketic rosary; generalised hypotonia, pot belly, doubling of malleoli and widening of wrist were present. Patient was mildly pallor with no icterus & no sign of other vitamin deficiency, no signs of hepatic cell failure. Mother's examination was done for bowing of legs and any other sign of rickets but it was perfectly normal. Child had received vitamin D & calcium three times in dose of 6 lakh unit but there was no improvement clinically or radiologically. Provisional diagnosis was made of refractory rickets. On investigations we found that X ray suggestive of rickets, Alkaline phosphatase was raised, serum calcium was slightly on lower side, phosphorus was normal.ABG was normal. LFT and KFT were normal, no history suggestive of malabsorption.PTH levels were raised showing that it was Hypocalcemic rickets and we found that 25OHD level was normal but 1,25(OH)2D was reduced. Diagnosis was made of vitamin D dependant type 1 rickets and treated with 1,25 Vit D (Calitriol) 0.25-2 µg/day& Oral Calcium.
On follow up X ray showed ZPC after 4 weeks.

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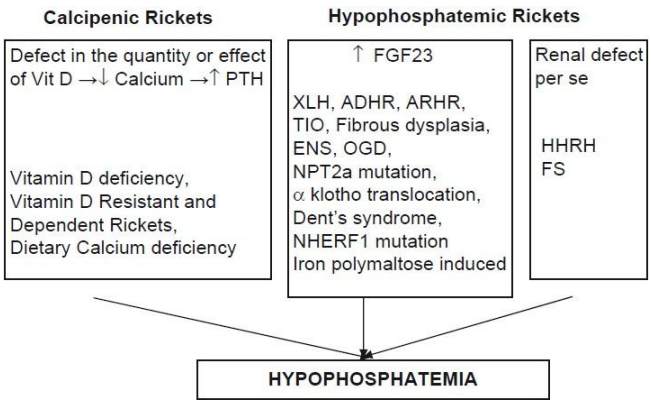
DISCUSSION

Hypophosphatemic rickets, vitamin D dependent rickets (VDDR) and renal tubular acidosis (RTA) are important causes of non-azotemic refractory rickets. VDDR characterized by impaired synthesis (type I) or resistance to the action of 1, 25 dihydroxyvitamin D3 (type II) (4). In our patients, the presence of hypocalcemia and elevated blood levels of PTH were sufficient to differentiate VDDR from other causes, including hypophosphatemic rickets.

Approach to refractory rickets has been described in following table



Response to treatment with potent vitamin D analogs (alpha-hydroxyvitamin D or 1, 25dihydroxyvitamin D) is useful for differentiating between VDDR types I and II.(5) Still we have performed 25OHD level which turned normal, 1,25(OH)2D was low, so it was confirmed that it was type I VDDR. VDDR also known as pseudovitamin d-deficiency rickets. It is caused by mutation in the gene encoding 25-hydroxyvitamin D3-1-alpha-hydroxylase (CYP27B1; 609506) on chromosome 12q13.

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