Case Report

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Vitamin D Resistant Rickets in a 4-Year-Old Male Child

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Abstract

Vitamin D resistant rickets is a rare disorder characterized by impaired bone mineralization despite adequate vitamin D levels. This case report describes a 4-year-old male with growth retardation and leg bowing. Clinical examination and laboratory findings suggested rickets, with normal vitamin D levels but hypophosphatemia and elevated 1,25-dihydroxyvitamin D. Radiographs confirmed rickets. Since the patient was non-affording so PHEX gene testing was not done however patient responded well to oral phosphate supplementation. Treatment with phosphate supplements and calcitriol improved the patient's condition. This case highlights the importance of recognizing genetic rickets forms and the role of targeted therapy in management.

Keywords:- X-linked hypophosphatemia, Vitamin D resistant rickets, PHEX gene, Hypophosphatemia

INTRODUCTION

Rickets is a condition characterized by impaired mineralization of the growth plate cartilage in children, leading to bone deformities and growth disturbances. Vitamin D plays a crucial role in calcium and phosphate homeostasis, which is essential for healthy bone development. While nutritional rickets due to vitamin D deficiency is common, vitamin D resistant rickets is a rare condition that persists despite adequate vitamin D levels. This condition is primarily due to genetic defects that affect the metabolism or action of vitamin D.[1]

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The pathophysiology of vitamin D resistant rickets involves mutations in genes responsible for the metabolism of vitamin D or the responsiveness of the vitamin D receptor. One of the most common forms is X-linked hypophosphatemic rickets (XLH), caused by mutations in the PHEX gene. These mutations lead to increased fibroblast growth factor 23 (FGF23) activity, which in turn decreases renal phosphate reabsorption and suppresses 1,25-dihydroxyvitamin D synthesis. This results in hypophosphatemia and inadequate bone mineralization. [2]

Clinically, vitamin D resistant rickets presents with similar symptoms to nutritional rickets, including growth retardation, bone pain, and skeletal deformities such as bowing of the legs and rachitic rosary. Diagnosis is based on clinical presentation, biochemical findings of hypophosphatemia with normal or elevated levels of 1,25-dihydroxyvitamin D, and genetic testing to identify specific mutations. Radiographic findings typically show widening, fraying, and cupping of the metaphyses of long bones. [3]

An important diagnostic finding in vitamin D resistant rickets is the presence of normal to elevated serum levels of 1,25-dihydroxyvitamin D despite clinical features of rickets. This differentiates it from nutritional rickets, where 1,25-dihydroxyvitamin D levels are usually low due to vitamin D deficiency. Additionally, hypophosphatemia and increased renal phosphate wasting are key biochemical features. [4]

CASE REPORT

A 4-year-old male presented to the pediatric clinic with a history of growth retardation and progressive bowing of the legs over the past year. His medical history was unremarkable, and there was no family history of metabolic bone disease. On physical examination, the child had frontal bossing, rachitic rosary, and significant bowing of the legs. His height and weight were below the 5th percentile for his age.

Initial laboratory investigations revealed serum calcium of 9.2 mg/dL (normal: 8.5-10.5 mg/dL), phosphate of 2.5 mg/dL (normal: 3.5-5.0 mg/dL), alkaline phosphatase of 450 U/L (normal: 100-320 U/L), and 25-hydroxyvitamin D of 30 ng/mL (sufficient: >20 ng/mL). Serum 1,25-dihydroxyvitamin D was elevated at 80 pg/mL (normal: 25-65 pg/mL). Urinary phosphate excretion was increased, indicating renal phosphate wasting.

Radiographs of the lower limbs showed widened, frayed, and cupped metaphyses of the distal femur and proximal tibia, consistent with rickets. Given the clinical and biochemical findings, a diagnosis of vitamin D resistant rickets was suspected. Since the patient was non-affording so PHEX gene testing was not done however patient responded well to oral phosphate supplementation.

Management included oral phosphate supplements and active vitamin D analogs (calcitriol). The patient was monitored regularly for growth parameters, biochemical markers, and skeletal deformities. After six months of treatment, there was noticeable improvement in the child's growth velocity and reduction in the severity of leg bowing.

Lab Investigation	Result	Reference Range
Serum calcium	9.2 mg/dL	8.5-10.5 mg/dL
Serum phosphate	2.5 mg/dL	3.5-5.0 mg/dL
Alkaline phosphatase	450 U/L	100-320 U/L
25-hydroxyvitamin D	30 ng/mL	>20 ng/mL (sufficient)
1,25-dihydroxyvitamin D	80 pg/mL	25-65 pg/mL

Table 1 : Lab investigation in the studied case.

DISCUSSION

Vitamin D resistant rickets, particularly XLH, is a genetic disorder with significant implications for bone health in children.[5] This case highlights the importance of considering genetic forms of rickets in patients who do not respond to conventional vitamin D supplementation. The identification of elevated 1,25-dihydroxyvitamin D levels in the context of hypophosphatemia and clinical rickets is a crucial diagnostic clue. [6]

Similar cases have been reported in the literature. Studies have described a cohort of children with XLH who presented with similar clinical and biochemical features and responded well to treatment with phosphate supplements and calcitriol . Some case reports have highlighted the genetic basis of the disease and the role of PHEX

mutations in the pathogenesis of XLH . A recent review discussed the long-term management and outcomes of patients with XLH, emphasizing the need for early diagnosis and treatment to prevent growth retardation and skeletal deformities . [7]

In this case, the combination of clinical presentation, biochemical findings, and genetic testing was essential for accurate diagnosis and management. The improvement in the patient's condition following appropriate treatment underscores the effectiveness of targeted therapy in vitamin D resistant rickets. [8]

.CONCLUSION

Vitamin D resistant rickets, especially XLH, requires a high index of suspicion for diagnosis. The combination of hypophosphatemia, elevated 1,25-dihydroxyvitamin D levels, and specific genetic mutations guides the diagnosis and management. Early and appropriate treatment is crucial to improve clinical outcomes and prevent long-term complications.

Conflict of interest

None

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