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Case Report

A case report on an unusual case of gait abnormality

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ABSTRACT

Vitamin D3 (cholecalciferol) is a fat-soluble vitamin that is essential for the skeletal health. Here, we discuss a case of a child from an upper middle-class family who presented with gradually progressive proximal myopathy and gait abnormality over 3 years. She was found to have severe vitamin D deficiency, resulting in proximal myopathy and pubic arch fractures with slipped capital femoral epiphysis, leading to the gait abnormality. In our country in spite of ample sunlight, vitamin D3 deficiency is rampant. The patient highlights the need of considering vitamin D deficiency as one of the differentials in myopathy and gait abnormalities in children.

Key words: *Cholecalciferol, Gait abnormality, Proximal myopathy*

Vitamin D deficiency and its ill effects have come into focus over the past several years. In spite of this, it is still underdiagnosed [1]. The role of vitamin D has been well established not only in relation to the skeletal system but also in relation to immune deficiency, cancers and autoimmune diseases. Cases of severe vitamin D deficiency leading to myopathy have been reported primarily in females from Arabic countries. Vitamin D deficiency causing significant myopathy in children has been rarely reported in our country. We present one such case of severe proximal myopathy which responded to replacement therapy.

CASE REPORT

A 14-year-old female child, the first product of a non-consanguineous marriage belonging to a Jain family, was brought with the complaints of gradually progressive generalized weakness. It started with the weakness and lethargy in the child, and soon, she found it difficult to rise from a chair and to climb stairs. Over the next few months, there was difficulty in combing hair and she also developed a waddling gait. However, there was no tremors, abnormal movements, difficulty in writing, or wearing footwear suggestive of no distal muscle involvement. Parents also reported that she was depressed and introverted due to her incapacities. The parents sought treatment from various clinicians including a naturopath; without relief. Detailed history revealed no significant past or family history. Diet was lactovegetarian with the consumption of around 200 ml of milk daily and other milk products on some occasions.

On examination, she was an obese child with body mass index of 30, vitals and blood pressures were normal. No neurocutaneous markers or bone pains were noted. Cranial nerve examination and facial muscles were normal. The power in the proximal muscles of upper and lower limbs was Grade 3, and that in the distal muscles was Grade 4+. Gower's sign was positive. No fasciculations or atrophy or hypertrophy of muscle groups was noted. She had a waddling gait. Sensory system and deep tendon jerks were normal. Other systems were normal.

With a provisional diagnosis of proximal myopathy and a differential of limb-girdle dystrophy, endocrine myopathy, late-onset Pompe's, and late-onset metabolic myopathies, she was investigated. A normal serum creatinine phosphokinase (CPK) ruled out primary muscle disease, which in muscular dystrophies, it would be remarkably increased. Nerve conduction velocity (NCV) and electromyogram (EMG) as well ruled out most of the neuromuscular diseases. A muscle biopsy was, hence, not attempted.

Other diseases which may present with late onset proximal muscle weakness are late-onset Pompe's disease and metabolic myopathies. A normal α-glucosidase and β-galactosidase and normal lactate levels ruled out the late onset Pompe's disease (which may have normal EMG, NCV and CPK levels) and mitochondrial myopathies (which can have normal CPK and EMG), respectively. She was also worked up for endocrine myopathies associated with hypothyroidism and hypercortisolism by checking her free thyroid profile and morning (8 am) cortisol levels, respectively. Her serum calcium and vitamin D3 levels were low as shown in Table 1. In view of significant hypovitaminosis D and hypophosphatemia, serum electrolytes, serum magnesium, fibroblast growth factor, 24 h urinary calcium, and tubular reabsorption of phosphorus were done to rule out familial hypophosphatemia and renal tubular defects; thus, excluding causes of resistant rickets (Table 1).

Table 1: Clinical and laboratory investigations

Parameters	Values (normal range)
CRP	<5 mg/L (<5)
Serum Na/K/Cl	138/4.5/101 mg % respectively
Serum magnesium	1.96 mg % (1.7-2.4)
CPK	36 IU/L (34-145)
SGOT	25 IU (14-36)
Serum calcium	7.8 mg % (8.6-10.2)
Serum phosphorus	2.87 mg % (2.5-4.5)
Serum alkaline phosphatase	857 IU (53-141)
Vitamin D3	6.7 IU (30-100 IU)
Serum cortisol	17.68 mcg/dl (3-21)
Free thyroid profile	Normal
FGF23	133.4 (N 0.00-150)
24 h urinary calcium excretion	4.5 mg in 24 h (5-40)
Tubular reabsorption of phosphorus	90%
EMG, NCV	Normal
Serum lactate	0.8 mmol/L (0.9-1.7)
α-glucosidase	Normal
β-galactosidase	Normal

CPK: Creatinine phosphokinase, EMG: Electromyogram, NCV: Nerve conduction velocity, CRP: C-reactive protein, FGF23: Fibroblast growth factor, SGOT: Serum glutamic oxaloacetic transaminase

X-ray of the pelvis with both hips showed fractures of both pubic arches and a slipped capital femoral epiphyses responsible for the waddling gait. These were attributed to the osteomalacia due to vitamin D deficiency. They needed no active intervention (Figs. 1 and 2). On the basis of these findings, she was diagnosed as "severe vitamin D deficiency with rickets/osteomalacia with proximal myopathy," caused by nutritional deficiency and aggravated by obesity. She was given 600,000 U of vitamin D intramuscularly as a single dose and treated with maintenance calcium and vitamin D supplements, equivalent to elemental calcium 1200 mg and cholecalciferol 2000 IU daily for 6 months during which she was on close follow up. There was a significant improvement in her biochemical parameters, after 3 months (vitamin D3 - 25.3 ng/ml, PO_4 - 3.78 mg/dl, Ca - 9.4 mg/dl, and alkaline phosphatase - 102 IU). Her power, gait, and psychological issues had improved on a 6 month follow-up and were completely normal at 1 year. She was advised to continue cholecalciferol 1000 IU daily as maintenance therapy.

DISCUSSION

Rickets and osteomalacia are an important problem even in tropical India, where paradoxically there is a high prevalence of vitamin D deficiency, with a study reporting 90% prevalence in adults and 84% prevalence in pregnant women [1]. Asians require approximately three-fold longer periods of sunlight exposure because of the protective pigmentation in their skins as compared to the Caucasian population [2]. Furthermore, adiposity, as existed in this child is seen to have a positive correlation with hypovitaminosis D [3,4].

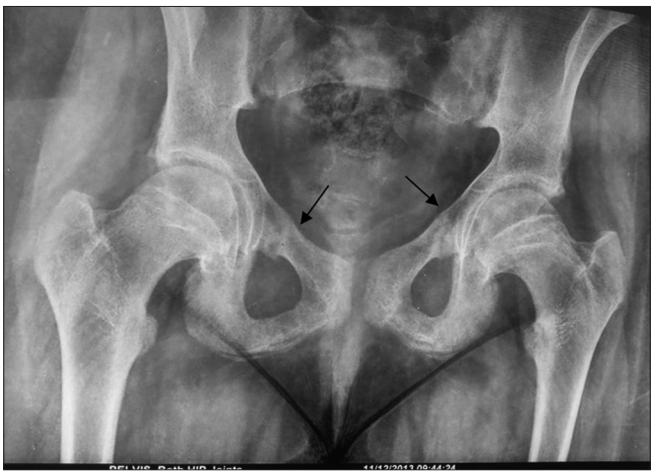

Figure 1: X-ray both hips with pelvis showing fractures of pubic arches and mild slipped capital femoral epiphysis

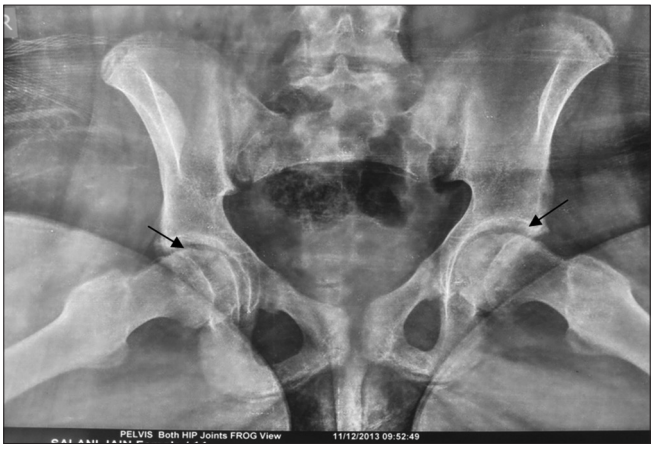

Figure 2: Frog view of pelvis with hips showing slipped capital femoral epiphysis

The incidence of proximal myopathy in patients of osteomalacia may be as high as 70% [5]. This case was reported to highlight the fact that obesity amplifies the problem of vitamin D deficiency. In young children, this proximal muscle weakness can lead to non-participation in physical activities and sports causing suboptimal performance at school leading to the lack of confidence and emotional issues as we noted in our case. Muscles play a crucial role in bone development and integrity [6]. Chronic vitamin D deficiency influences skeletal integrity through its role in calcium and phosphorus metabolism and also by affecting optimal muscle action on the skeleton.

Most of the cases of vitamin D deficiency with severe myopathy have been reported in females from the age group of 15 years onward from Arab countries [7]. Traditional clothing that prevents adequate exposure to sunlight, as reported by Al-Said et al. in 47 female patients aged 13-46 years, caused moderate to severe osteomalacia myopathy [7]. The case presented here had severe muscle weakness, in addition to the fractures of the pubic arches and slipped femoral epiphysis which contributed to the significant waddling gait. Her diet was lactovegetarian; also she was obese, and there was minimal exposure to sunlight due to a sedentary lifestyle. Obesity is known to affect vitamin D levels

due to higher rates of deposits in the fat reserves [8,9]. The doses recommended for treatment and maintenance is much higher than the standard recommended doses [10].

A study conducted in the UK by Ward et al. revealed that in a group of postmenarchal adolescents, serum 25 (OH) D was positively related to muscle power, force, velocity, and jump height [9]. There are certain hypotheses as to how vitamin D contributes to muscle weakness. The basis of this hypothesis is that skeletal muscle contains vitamin D receptors that modulate various transcription factors in muscle cells [11], mediating differentiation into mature Type 2 muscle fibers. Furthermore, vitamin D is responsible for the active transportation of calcium into the sarcoplasmic reticulum assisting muscle contraction essential for maintenance of postural equilibrium [12]. Furthermore, vitamin D itself in association with secondary hyperparathyroidism is the primary cause of muscle tissue abnormalities.

It is imperative that we consider this condition as one of the differentials of proximal myopathy and gait disturbance. A routine test for 25 (OH) D3 can pick up hypovitaminosis D and hence, prevent expensive and elaborate neurological investigations in children presenting with proximal muscle weakness. We would go a step further to suggest that children presenting with excessive fatigue and avoidance of physical activities, especially obese ones should be screened and then treated for vitamin D deficiency so as to optimize their performance in school and hence prevent psychological issues resulting from low self-esteem.

CONCLUSION

Vitamin D deficiency is highly prevalent in the Indian population particularly due to increased skin pigmentation, pollution, and poor dietary habits. Among the urban rich obesity is also a contributory factor to be kept in mind while treating so as to give adequate doses to obtain expected therapeutic results.

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