

METABOLIC BONE DISEASE: UNCOMMON CAUSE OF QUADRIPARESIS WITH REMARKABLE RECOVERY

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Submission date: 06-Aug-2025 12:10PM (UTC+0700)

Submission ID: 2690327030

File name: IJAR-53128.docx (5.32M)

Word count: 1769

Character count: 9754

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ABSTRACT

¹⁷ Vitamin D is essential for the skeletal health along with its role in autoimmune disorders and cancers. Its deficiency classically presents as proximal weakness, muscle aches and waddling gait. Truncal involvement is rare. We present a case with gradually progressive quadri-paresis with truncal involvement in a young female due to social and cultural norms of covering the whole body leading to limited sunlight exposure. This highlights the need for considering a differential ¹⁶ of vitamin d deficiency in young females presenting with myopathy with gait abnormality.

KEY WORDS: VITAMIN D, PROXIMAL MYOPATHY, GAIT ABNORMALITY.

INTRODUCTION: Vitamin D deficiency despite its high prevalence is still undiagnosed and misdiagnosed for a prolonged duration before its actual diagnosis. It is usually misdiagnosed as a neuromuscular disorder rather than metabolic bone disease. According to the estimates ² Vitamin D deficiency or insufficiency affects around 1 billion population worldwide(1). In Indian population prevalence of insufficient dietary intake leading to vitamin D deficiency is approximately 50 to 90 %(2). At the same time 60 to 75% of patients are reported to have proximal myopathy secondary to hypovitaminosis (1). Due to such a high prevalence this i.e vitamin D deficiency should be kept as a plausible diagnosis before proceeding to higher costly and invasive procedure. Sunlight exposure for adequate duration help in synthesis of vitamin D from cholesterol using ultraviolet spectrum. It is more common in elderly bedbound population, as this group has limited sunlight exposure; and they usually presents with muscle aches and proximal symmetrical weakness (3). Other risk factors along with dietary insufficiency and poor sunlight exposure includes malnutrition, malabsorption, severe obesity, sedentary indoor working lifestyles, use of cytochrome P450 inducing enzymes, chronic kidney disease. We report a young female with gradually progressive quadri-paresis due to hypovitaminosis D probably due to religious norms of covering the whole-body area.

CASE PRESENTATION:

17 years old young female school student, born out of non-consanguineous marriage presented with chief complaints of gradually worsening walking ability with abnormal gait from last 2 years. Complaints started with gradually progressive weakness in the form of lethargy and multiple joints pain predominantly involving lower back and ankle. There were multiple tender points over the whole body which could have been easily misdiagnosed with fibromyalgia and patient was initially treated with multiple over the counter medications. Over the next one year she had complaints of difficulty in getting up from squatting position, low seated chairs, and using stairs. Due to worsening symptoms, she started using support of the wall or accompanying person for walking. At the same time, she developed waddling gait. Similar complaints she developed in upper limbs after 1 year of progression in the form of difficulty lifting heavy loads over the shoulder. Also, she had complaints of difficulty getting out of the bed and used to take support of bed railings for getting out as well as turning in the bed. This affected her routine work to such a extent that she was accompanied by father to school due to disabling gait and also she started taking half the number of books as against her usual routine because she could not lift heavy back bag.

There were no complaints of distal weakness in upper as well as lower limbs as history of difficulty in writing, wearing footwear, tremors or abnormal movements in the limb was absent. Sensory and bowel and bladder involvement was also absent. There was no significant family history or past history in the form of malabsorption, trauma, steroids use or any other drug use. Diet was lacto-nonvegetarian. She used to cover her whole body while going out since childhood as per her religion norms. Dietary habits were nonvegetarian with inclusion of milk and milk products.

On examination she was having normal vitals with no neurocutaneous markers. Cranial nerve examination was normal. Power in the proximal muscles of both upper and lower limbs was grade 3/5 and in distal muscles it was 5/5. Gowers's sign was positive. No fasciculations or atrophy was present. There was hyperreflexia in upper as well as lower limbs. There was tenderness over the lower back and hip region. Sensory examination was normal. There was waddling gait as well. There was no involvement of other systems.

Lab investigations revealed hemoglobin of 11.6 g/dL, total leukocyte count TLC- 11800, differential leukocyte count DLC- P-68, L28, M-02, E-02. Kidney and Liver function tests were normal. She was found to have low normal serum calcium levels 8.4 mg/dl. Serum phosphorus levels were also low 1.6 mg/dl with raised serum alkaline phosphatase levels-

1282.95 IU/dl. Vitamin D was very low less than 3 ng/dl (reference range 75-250ng/ml). serum intact PTH was elevated up to 96 pg/ml. (10-65 pg/dl). Urine was negative for protein and glucose. Antinuclear antibodies, thyroid function test and creatine phosphokinase levels were within normal limits. Imaging of MRI spine showed incomplete displaced fracture lines at lateral and medial aspect of right femoral neck. Fracture line also seen in bilateral sacrum at the junction of superior 2/3rd and inferior 1/3rd. Also, minimal anterior wedge collapse with end plate irregularity and subarticular fatty infiltration in lower dorsal and lumbosacral vertebrae from D11 to S3 level. These changes were suggestive of multiple end plate microfractures. These radiographic findings were giving clue towards osteomalacia with insufficiency fractures.

On the basis of history, examination and investigations diagnosis of osteomalacia with hypovitaminosis D with secondary hyperparathyroidism was made. Patient was treated with weekly dose of cholecalciferol 60000k IU once a week along with calcium carbonate 500 mg twice daily.

Follow up after 1 month revealed a gradual improvement with complete regain of motor power. She could do her activities of daily living independently. Patient was not willing for repeat biochemical parameters.

DISCUSSION:

Prevalence of vitamin d deficiency is very high in India reaching up to 90%. Lack of sunlight exposure is considered as one of the risk factors for vitamin d deficiency. However, some of the individuals despite having adequate exposure have low levels of vitamin d probably because of skin pigmentation with higher melanin content which is more common in Asians(4). Our patient had insufficient sun exposure due to their norms of covering their whole body while going out since childhood. Similar results were shown by a study done by Al-Said et al in 47 female patients from Saudi Arabia and concluded that traditional clothing prevented adequate exposure to sunlight and caused moderate to severe osteomalacia myopathy(5).

There are various other factors contributing to vitamin d deficiency like insufficient intake, malabsorption, certain drugs and chronic systemic conditions like CKD.

Clinical symptomatology of vitamin d deficiency is variable. Most patients present with proximal weakness. Osteomalacia leading to myopathy is common and presents as initial symptom in up to 30% patients (6). In a study done by Kate A Ward et al on 99 post menarchal 12 to 14 years old females concluded that vitamin D levels were significantly associated with muscle power and force(7).

‘How vitamin d deficiency leads to myopathy?’ is explained by the presence of vitamin D receptors on the muscle that modulate various transcription factors in the muscle cells which ultimately leads to differentiation into mature type 2 muscle cells(8). Not only in muscle maintenance but also in its function vitamin D plays an important role as it helps in muscle contraction with transport of calcium inside sarcoplasmic reticulum(9).

A serum level below 50 nmol/l is associated with increased body sway and below 30 nmol/l leads to decreased muscle strength(9). So, chronic vitamin d deficiency influences skeletal integrity by affecting the harmony between the muscle and skeleton.

Our patient had very low level of less than 3 nmol/l and patient had presented with quadriparesis as well as multiple micro fractures in spine and sacrum. Patient had complete resolution of symptoms after treatment. So, it is imperative to consider this as a differential even in young children presenting with muscle aches and proximal weakness along with gait disturbance. This would prevent expenditure in higher expensive and invasive investigations. Although the classical cardinal features of vitamin d deficiency are proximal weakness and waddling gait with pain and discomfort but it can vary from generalized weakness and fatigue to making patient bedbound, but because of its complete reversibility it should not be missed and should be screened in all patients. Replacement with ergocalciferol and calcium resulted in immediate complete recovery in our patient.

CONCLUSION:

- ❖ Vitamin d deficiency is the common cause of osteomalacia.
- ❖ Limited skin exposure to sunlight is one of the risk factors for vitamin d deficiency.
- ❖ Not only in elderly population, but also in young patients presenting with proximal muscle weakness with waddling gait, vitamin d deficiency should be suspected and managed with replacement as early as possible.
- ❖ It can be misdiagnosed as a neuromuscular disorder, fibromyalgia; so, awareness among clinicians about vitamin d deficiency clinical presentation is important.

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FIGURE 1: MRI SPINE: T1 SAGGITAL SPINE MRI SHOWING MINIMAL ANTERIOR WEDGE COLLAPSE WITH END PLATE IRREGULARITY AND SUBCORTICAL FATTY INFILTRATION IN LOWER DORSAL AND LUMBO-SACRAL VERTEBRAE FROM D11-S3 LEVEL SUGGESTIVE OF END PLATE MICRO FRACTURES.

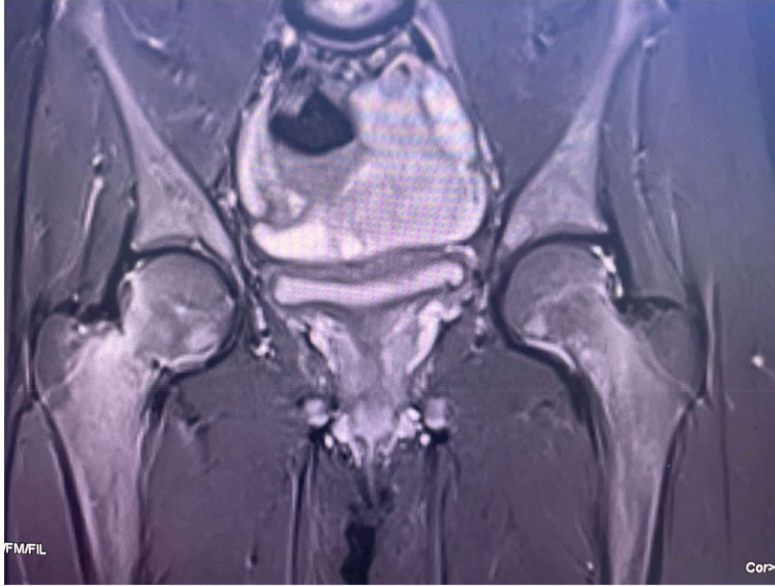


FIGURE 2: MRI PELVIS SHOWING INCOMPLETE UNDISPLACED FRACTURE LINES AT LATERAL AND MEDIAL ASPECT OF RIGHT FEMORAL NECK AND MEDIAL ASPECT OF LEFT FEMORAL NECK.

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