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Osteomalacic Myopathy Masquerading Bilateral Femoral Neck Fracture: A Rare Case

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Abstract

Vitamin D deficiency is prevalent worldwide and has many consequences ranging from generalized aches, seizures, irritability, tetany, pathological fractures and osteomalacic myopathy. We discuss a rare case of an adolescent girl who presented with bilateral lower limb weakness and initially misdiagnosed as neuromuscular disorder but eventually found to have severe vitamin D deficiency leading to osteomalacic myopathy which masqueraded the features of bilateral femoral neck fractures. Severe osteomalacia as a cause of myopathy is underdiagnosed and may mimic a primary neuromuscular disorder. Any patient with generalized muscle pains should be first screened for common aetiologies such as VDD before proceeding to extensive investigations.

Keywords: ***Vitamin D deficiency; Bilateral neck of femur fracture; Osteomalacic myopathy***

1. Introduction

The prevalence of Vitamin D deficiency (VDD) in India ranges from 37% to 99% [1] and is about 15.7% worldwide [2]. Vitamin D deficiency accounts for the most common nutritional deficiency among children and adults and is the most common

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cause of osteomalacia. The clinical symptomatology of VDD can demonstrate a wide spectrum ranging from generalized body ache, seizures, irritability, tetany, and pathological fractures. Osteomalacic myopathy is an uncommon manifestation of VDD and is usually seen in adolescents [3,4]. The simultaneous bilateral neck of femur fracture due to severe vitamin D deficiency is a known entity but rarely encountered in clinical practice. We describe a rare case of an adolescent girl who presented with bilateral lower limb weakness and initially misdiagnosed as neuromuscular disorder but eventually found to have severe vitamin D deficiency leading to osteomalacic myopathy which masqueraded the features of bilateral femoral neck fractures.

2. Case Report

A 16-year-old previously healthy girl presented with acute onset bilateral lower limb weakness for 5 days gradually progressing to cause inability to walk or stand independently associated with difficulty in rising from supine or sitting position and restriction of neck movements. There was associated pain in the lower limbs and generalized myalgia. This was preceded by fever 10 days ago with no other constitutional symptoms.

She was fourth born to third degree consanguineous parents at term gestation via normal vaginal delivery with no neonatal issues. She goes to high school with reasonable scholastic performance. She was partially immunized up to 5 years of age according to Indian National Immunization Schedule. She attained menarche at the age of 13 years. Her Tanner SMR breast stage and pubic hair stage were both 5. She weighed 35 kg (<3rd percentile) and stood 158 cm tall (<15th percentile) indicative of severe malnourishment with history of maximum indoor dwelling and inadequate exposure to sunlight. On examination, there was marked proximal weakness of both upper and lower limbs with power 2/5 at hip and shoulder joints and 3/5 at all other joints along with generalized muscle tenderness. Muscle tone and sensation were normal with no signs of bowel and bladder involvement. Chvostek's sign and trousseau sign were positive. Her vital signs were normal.

Baseline investigations showed hemoglobin 12.8 g/dL, with a normal white cell count and C-reactive protein (<6 mg/L). The Renal function tests, sodium, potassium, chloride and bicarbonate levels were normal. Liver function tests were normal except for elevated alkaline phosphatase levels of 483 U/L.

On the basis of history and clinical examination, differential diagnosis included:

1. Acute inflammatory demyelinating polyneuropathy
2. Infectious Myositis
3. Autoimmune polymyositis

Further work-up to look for the above differentials was performed. The Erythrocyte sedimentation rate was raised at 75mm (1st hour) and 100 mm (2nd hour). CPK NAC was 1007 IU/L international units. Nerve conduction studies were normal. Her ANA profile and myositis profile were negative with no trace of myoglobin in urine but raising titers of CPK NAC (1347 on day 3 of admission). MRI thigh STIR images and MRI screening spine were normal, however, incomplete bilateral neck of femur fracture with marrow edema and bilateral pubic rami fracture with and severe osteopenia with loosers zone (FIG. 1) were also noted.

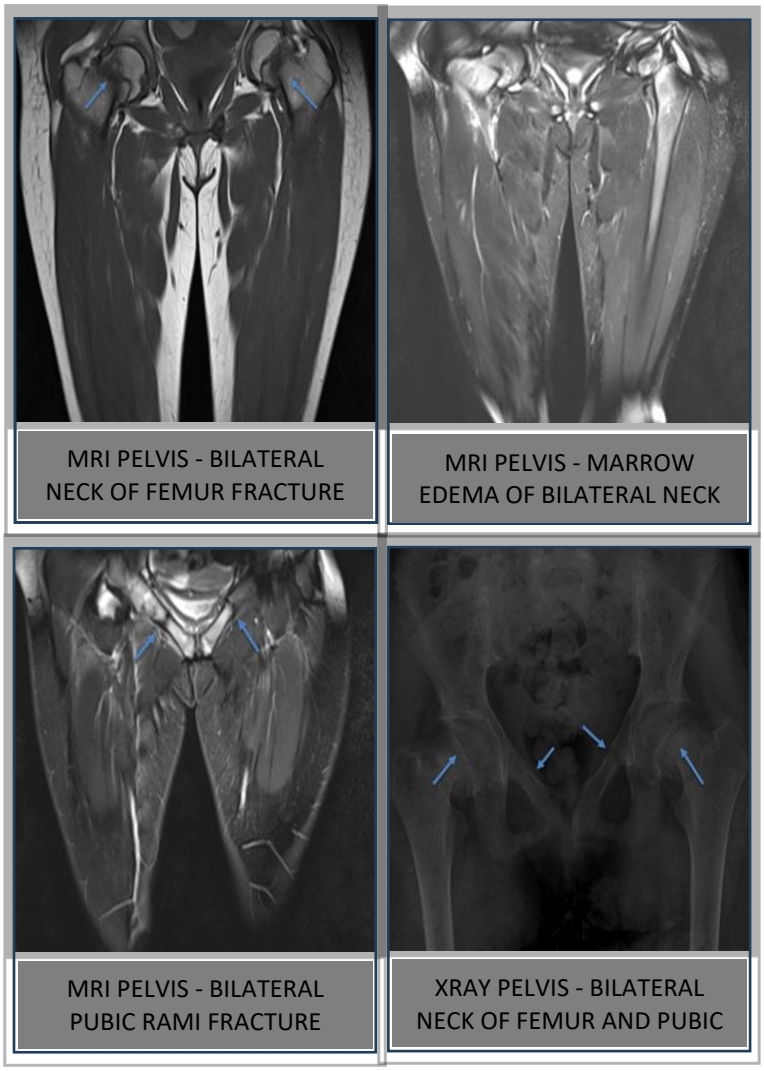

FIG. 1.

On probing into detailed history, mother revealed history of trivial fall 15 days back following which child complained of some pain in bilateral hip region which responded to analgesics. Her serum calcium and vitamin D levels were extremely low (4.2 mg/dl and 8.1 ng/dl respectively). Corrected calcium was low (4.8). Serum phosphorus was 4.62 mg/dl and PTH was 306.6 pg/ml with normal thyroid function.

Based on these findings the patient was diagnosed with pathological fracture of bilateral neck of femur secondary to osteomalacia and grade IV osteopenia due to severe VDD. The etiology of proximal muscle weakness was attributed to osteomalacic myopathy. She was started on symptomatic IV calcium supplementation at admission, and this was changed to enteral calcium once the chvostek's sign and trousseau sign were negative. She was also started on vitamin D therapy, and she had internal fixation of the fractures after the metabolic optimization was achieved. Child is on regular follow up and is able to walk on her own and perform daily activities.

3. Discussion

Simultaneous bilateral fracture neck of femur is a rare condition which is usually associated with high energy trauma, intense seizure activity, high voltage electric injury, collagen disorders, cerebral palsy, disorders of bone metabolism such as vitamin D deficiency, renal osteodystrophy, primary hyperparathyroidism and osteomalacia. Severe VDD can cause significant osteopenia resulting in low-impact fracture which is defined as a fracture occurring from a fall no greater than standing height or occurring spontaneously [5]. Adolescent boys are found to have a higher rate of bone fracture as compared to girls, with a peak incidence between the ages of 13 and 14 years [6]. Fracture risk is also related to rapid pubertal growth - a period of increased cortical weakness [7].

VDD associated myopathy, called osteomalacic (or rachitic) myopathy, is a well-known but often underdiagnosed entity. The clinical features are similar to other primary muscle diseases and affect predominantly proximal muscles and lower limbs. Muscle weakness may be severe enough to render a patient wheelchair-bound [8]. Body pains secondary to osteomalacia usually co-exist. The mechanism includes VDD induced secondary hyperparathyroidism which induces muscle catabolism and leads to the development of myopathy [9]. Osteomalacic myopathy is relatively more common in adolescents which can be attributable to their increased vitamin D requirement related to growth spurt and hormonal modulation during puberty [10]. Owing to its overlapping features and absence of signs of rickets in adolescent age group, osteomalacic myopathy continues to be misdiagnosed as rheumatological disorder or neuromuscular disorder [3]. Therefore, the diagnosis of osteomalacic myopathy requires a very high index of clinical suspicion. Once suspected, the diagnosis can easily be established by appropriate laboratory and radiographic investigations. However, the final confirmation is made after myopathic symptoms have resolved following treatment with vitamin D [11].

Our patient presented with pseudo-paralysis of both lower limbs which prompted us to first investigate for a systemic cause such as neuromuscular disorders including acute inflammatory demyelinating polyneuropathy, post infectious myositis and autoimmune polymyositis, rather than suspecting local cause such as bilateral fractures. MRI spine, which was done to look for the aetiology of paraparesis paved way for the diagnosis of severe VDD by demonstrating severe osteomalacia and bilateral femoral neck fractures. In this case, the local symptoms of bilateral neck of femur fracture were masked by predominant myopathy features misleading the diagnosis towards neuromuscular and rheumatological disorder. Therefore, any patient with generalized muscle pains should be first screened for common aetiologies such as VDD before proceeding to extensive investigations. Presence of the constellation of biochemical alterations should limit invasive and costly neuromuscular workup.

4. Conclusion

Severe osteomalacia as a cause of myopathy is underdiagnosed. High index of suspicion is mandatory to clench the diagnosis owing to its overlapping features with myositis and neuromuscular disorders. Any patient presenting with muscle weakness should first be evaluated for VDD especially in tropical countries like India. Regular testing and vitamin D supplementation is advisable in areas endemic for VDD.

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