Delay in the Diagnosis of Osteomalacic Myopathy Secondary to Vitamin D Deficiency Due To Coexistent Thoracic Intradural Arachnoid Cyst: A Case Report

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Abstract: The patient with vitamin D deficiency may manifest with osteomalacic myopathy which has been reported in females with poor nutrition and inadequate sun exposure, especially in community with Purdah practice in India. Clinical features of osteomalacic myopathy are painful symmetric proximal muscle weakness, waddling gait and brisk tendon jerks. We report a case of osteomalacic myopathy in a 25 year-old female which was initially missed due to presence of thoracic intradural arachnoid cyst. Patient showed rapid improvement with Calcium and vitamin D supplementation.

Keywords: Osteomalacic, myopathy, arachnoid cyst, brisk jerks, vitamin D

1. Introduction

Osteomalacic myopathy is a disabling painful myopathy. It occurs due to vitamin D deficiency. Major source of the Vitamin D is the exposure of sun over the skin. In spite of adequate sun exposure in India throughout the year, social practice of Purdah system especially in females results in insufficient sun exposure. With poor nutrition, pregnancy or continued lactation they are at risk of developing osteomalacic myopathy. Clinical features of osteomalacic myopathy are bone tenderness, painful proximal weakness, waddling gait and brisk tendon jerks. The Diagnosis is based on the radiological (pseudofractures in adults) and biochemical findings (elevated serum alkaline phosphatase, low serum calcium and low phosphorus). There are very few case reports of the osteomalacic myopathy in the literature.

2. Case Report

A 25 year-old female came to our clinic with the history of painful muscle weakness of all four limbs for last 2 years. She had difficulty in carrying out her daily activities of life like climbing stairs and rising from sitting position without support. On clinical examination we found bone tenderness, in addition to the muscle weakness, muscle wasting and brisk tendon jerks. The power was grade 3 in proximal muscles and grade 4 in distal muscles in all the four limbs. Deep tendon jerks were brisk in all four limbs. There was no sensory involvement. She had normal bowel and bladder habits. Her routine blood investigation showed low haemoglobin (8.7gm%), low serum calcium(7mg%), low serum phosphorus(2.1mg%) and elevated serum alkaline phosphatase (291mg%). This along with pseudo-fractures of both femoral neck led to the diagnosis of osteomalacia. Her 25-hydroxy vitamin D was deficient (12.28 ng/ml). Other investigation like blood sugar, total creatinine kinase (CPK), serum electrolytes, thyroid, kidney and liver function tests were normal. Based on the clinical and laboratory tests, diagnosis of osteomalacic myopathy secondary to vitamin D deficiency was made. She was given 60,000 IU cholecalciferol weekly with 1500 mg elemental calcium daily for 12 weeks followed by maintenance therapy of 2000 IU daily. In 12 weeks her pain and weakness improved rapidly and she was able to do her daily activities of life without any support.

Patient was initially investigated with MRI spine in other hospital in view of brisk tendon jerk. Based on the clinical evaluation and MRI of the spine which showed thoracic intradural arachnoid cyst extending from D11 to L1 level, she was advised surgery in the previous hospital. MRI of cervical spine was normal. Because of inability to explain brisk tendon jerks in upper limbs with generalized bone pain and painful proximal muscle weakness, we investigated her further and found above findings, that were consistent with osteomalacic myopathy. Figure 1a & 1b Saggital & coronal section of MRI showed arachnoid cyst from D11 to L1 with no significant mass effect over spinal cord.

Figure 2: Plain radiograph of pelvis with both hip showed bilateral pseudofractures of both femoral neck.
3. Discussion

Arachnoid cysts are most commonly located in the thoracic spine and may present with spinal cord compression with upper motor neuron signs like paraparesis, neuropathic pain, gait ataxia, or incontinence.8,9

In our case, bone pain with more proximal muscle weakness, preservation of sensation and exaggerated tendon jerks in upper limb led us to consider pathology other than the thoracic intradural arachnoid cyst. MRI of the spine showed no significant compression due to cyst. So it was considered asymptomatic.

In the literature osteomalacic myopathy and thyrotoxic myopathy are reported as the causes of brisk tendon jerks with proximal muscle weakness. In our case pseudofractures in the femoral neck was the specific radiological feature of the osteomalacia and she was euthyroid.10, 11, 12

In adults, severe vitamin D deficiency can result osteomalacia, which is associated with diffuse bone pain, muscle weakness, and difficulty in walking.13

Vitamin D deficiency is measured by serum 25-hydroxyvitamin D level in patients at risk. Concentration of at least 30 ng/mL (75 nmol/L) is needed to prevent impaired calcium absorption. Adult with vitamin D deficiency should be treated with cholecalciferol 50,000 IU once weekly for 8 weeks or 6,000 IU daily for 8 weeks with maintenance therapy 1500-2000 IU daily.14

4. Conclusion

In patients with musculoskeletal symptoms such as bone pain, muscle pain, and weakness all over body, a routine test for Vitamin D deficiency should be considered. Osteomalacic myopathy due to Vitamin D deficiency has very good prognosis and on repletion of vitamin D stores rapidly resolves bone pain, muscle pain and weakness.

References