

Hyperphosphaturic Mesenchymal Tumor induced Osteomalacia– A Case Report

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ABSTRACT

Hyperphosphaturic Mesenchymal Tumor (HMT) is a very rare benign tumor of the soft tissue or bone which produces tumor induced osteomalacia, also called as oncogenic osteomalacia. This activity can only be stopped by the surgical removal of the tumor. We present a 23 years old man who presented with long standing bony pains without any relief by a variety of medications. The clue to the diagnosis was taken from pelvis skiagram, Magnetic Resonance Imaging (MRI) of the body, PET scan and the blood chemistry.

Keywords: *Hyperphosphaturic Mesenchymal Tumor (HMT), Hypophostemia, Oncogenic osteomalacia*

Hyperphosphaturic Mesenchymal Tumor (HMT) was first described in 1947 by Mc Cance and Prader et al in 1959 further discovered its association with metabolic disorder [1]. HMT is a rare benign tumor of soft tissue or bone which produces tumor induced osteomalacia, or oncogenic osteomalacia. This paraneoplastic syndrome acts by secreting fibroblast growth factor-23 (FGF-23). These act by inhibiting renal tubular reabsorption of phosphate along with conversion of 25 hydroxyvitamin D₃ to dihydroxy vitamin D₃, resulting in phosphaturia and hypophostemia. These tumors are often of the mesenchymal in origin.

There is paucity of literature about the detailed pathophysiology of these tumors because of their rarity [2]. Children are less affected by tumor induced osteomalacia as compared to the elderly age group. The pediatric patients are usually affected by genetically linked syndromes and diseases.

CASE REPORT

23 years old male presented with generalized bony pains of five year duration unrelieved with medication. He had progressive difficulty in walking and limping. He was treated by multiple physicians without much improvement; therefore, he was referred to us for further management. There was no history of trauma, fever or surgery. On examination, he was afebrile, conscious with stable vital signs. Tenderness was present over left hip joint. His systemic examination showed no abnormality.

Plain X-ray pelvis AP view had shown mildly decreased bone density with loser's zone. The pelvis has shown triradiate configuration with transcervical fracture of left femoral neck (Figure 1). Blood chemistry has shown normal calcium levels with decreased serum phosphate (2.9 mg/dL), increased serum alkaline phosphatase (1200 IU/L) and normal parathyroid levels.

Renal and liver function tests were normal. MRI shows well defined soft tissue lesion adjacent to the left sacroiliac joint measuring 17x11x14 mm. The lesion was hypointense on T1W (Figure 2a) and shows intense homogenous post contrast enhancement (Figure 2b). Lesion showed hyperintensity on T2W (Figure 2c) and STIR images (Figure 3).



Figure 1 - X ray Pelvis AP view shows osteomalacia related changes along with looser's zone and trans-cervical fracture of the neck of the left femur (white arrow).The pelvis is of triradiate configuration (white cross).

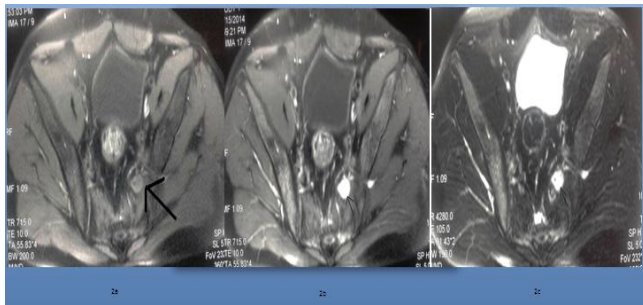


Figure 2 (a) - Non contrast T1W fat sat image shows a mixed intensity well defined lesion present on the medial aspect of the left sacroiliac joint (black arrow). (b) - Contrast T1W image shows the intense enhancement (thin black arrow) of the lesion. (c) - T2W fat sat axial section shows hyperintense (black arrow) lesion at the same location.

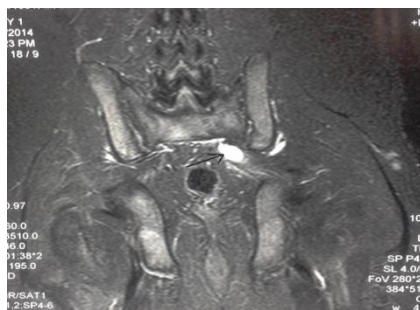


Figure 3 - STIR Coronal image shows the hyperintense well defined lesion (black arrow).

Diagnosis of tumor induced osteomalacia was made and Positron Emission Tomography (PET) (DOTA-NOC or 1,4,7,10-tetraazacyclododecane-1,1,7,10-tetraacetic acid)-1NaI(3)-octreotide scan was performed which showed increased uptake adjacent to the left sacroiliac joint. Plasma fibroblast growth factor-23 level was increased to 282.9 RU/mL (normal - 180 RU/mL). The complete tumor was surgically resected from the pelvic region. The tumor was totally separated out from the adjoining areas. The post operative period was uneventful.

Histopathologic examination (HPE) of the specimen confirmed the diagnosis which showed benign mesenchymal tumor rich in blood vessels. The tumor is arranged predominantly around the thin walled small vascular channels and interspersed thick walled medium sized vessels with medial hypertrophy. The tumor cells are bland appearing and show oval to plump nucleus with mild degree of pleomorphism, fine chromatin and moderate amount of cytoplasm. At places there is new bone. The blood parameters returned to normal in six months after the resection and there was no further deterioration of the symptoms.

DISCUSSION

Hyperphosphaturic Mesenchymal Tumors cause debilitating osteomalacia with resultant long standing bone pains and pathological fractures. The diagnosis can be confirmed first by blood chemistry and then by radionuclide studies or imaging modalities. Blood tests often reveal low phosphate levels, elevated alkaline phosphatase and low 1,25 dihydroxy vitamin D₃ levels. In our case, the long standing bony pains was the main feature and also resulted in transcervical fracture of the left femoral neck because of underlying osteomalacia. Clue can be obtained from increased urine phosphate excretion.

Fibroblast Growth Factor (FGF) 23, secreted by mesenchymal tumors, is the factor responsible for all the physiological changes seen in HMT [3]. It restricts the reabsorption of phosphate from the proximal renal tubules resulting in high urinary phosphate levels [4]. Plasma FGF 23 was markedly raised in our case. The diagnosis can be missed if the increase in urine phosphate got unnoticed as sometimes this may be the only clue to suspect the tumor [5]. Other similar condition like familial hypophosphatemic rickets and Fanconi syndrome can be ruled out by the negative family history.

Radiological modalities like plain radiography, MRI and PET scan help in diagnosing these types of rare tumors. The localization of small tumors is easy by multiplanar and multisectional imaging by MRI as this has high specificity of tissue characterization. Management can be done by surgical resection of the underlying tumour, which usually leads to immediate cure [7]. The patients are treated with 1-3 microgram /day Calcitriol and 1-4 g/day of Phosphorus in divided doses after the surgery till symptoms subside. The correct and timely diagnosis of the pathology can prevent many complications of the disease.

CONCLUSION

The cause of suspicious osteomalacia must be investigated by biochemical, radionuclide and imaging studies. Radiological Imaging modalities like X-ray, PET and MRI are of great importance to locate the tumors in the cases of tumor induced osteomalacia.

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