

Osteitis Fibrosa Cystica versus Malignancy & The role of Vitamin D in the diagnosis

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Introduction

- Osteitis Fibrosa Cystica is a skeletal disorder and is defined as as the classic skeletal manifestation of advanced hyperparathyroidism., it manifests clinically by bone pain, tenderness or fractures and radiologically as ground glass or moth eaten appearance.
- Osteitis Fibrosa Cystica may be confused with Malignancy and may present in a very similar way either clinically or after the conventional imaging modalities and thorough investigations are needed for its diagnosis
- This is a very interesting case of a middle aged female presenting with only left shoukler pain and all the imaging modalities were suggestive of malignancy but Vitamin D level, Parathormone hormone level were crucial in suspecting Ostetis Fibrosa cystica and the trephine bone biopsy confirmed the diagnosis.

Case Report

- Female patient named M.F., aged 37 years was complaining of Dull aching pain in her left shoulder, difficulty on lifting the left arm into right angle
- The patient had past history of generalised body aches
- The patient sought orthopaedic consultation because the pain in her left shoulder became severe
- X Ray on her left shoulder** revealed an intamedullary infiltrative lesion in the humerus of ground glass (Mouth eaten) appearance
- MRI of the left shoulder** which revealed abnormal infiltrative bone marrow signals
- CT chest and abdomen** revealed Bony skeleton marrow infiltration, Left lower lung nodule, Multiple hepatic focal lesions, Splenomegaly (Query Leukemia or Deposits) and Bone marrow biopsy was advised.
- Laboratory investigations** showed normocytic normochromic anaemia Serum Calcium: 7.7 mg/dl and Ionised Calcium 1 mmol/L
- Bone marrow aspiration biopsy** which revealed Hyper-cellular bone marrow with erythroid hyperplasia
- PET scan** revealed Multiple medullary based osteo-sclerotic lesions scattered all over the skeleton
- Other laboratory investigations** Vitamin D level: 7.46 ng/ml, Parathormone hormone: 198.9 pg/ml (normal range is 12-72)., Alkaline phosphatase: 315 U/L
- Patient underwent **Bone marrow trephine biopsy from right superior posterior iliac spine** which revealed Fibrovascular Bone marrow, consistent with Osteitis Fibrosa Cystica related to parathyroid hyperfunction and that was the final diagnosis
- The patient received treatment in the form of :Vitamin D supplements, Calcium supplements, Calcitonin inhalation, Alpha calcidol. The patient received this treatment for 4 months, the pain in left shoulder has improved and the body aches also improved .
- February 2015; Total calcium: 9.5 mg/dl. Actual calcium: 1.30 mmol/L, Vitamin D: 26 ng/ml. Parathormone hormone: 71 pg/ml & The most recent X-ray showed improvement and follow up investigations in January 2016 were all normal.

Discussion

- Vitamin D/ Calcium/ Parathormone hormone: **What is the starting point ??**
- If the low vitamin D is the starting point, thus cause the parathyroid to increase parathormone hormone production.
 - If the Vitamin D is low & the Calcium is high, this means 1ry Hyperparathyroidism.
 - If the Calcium is high and Vitamin D is normal this means 1ry Hyperparathyroidism.
 - Remember: 2ry Hyperparathyroidism due to low Vitamin D is never associated with high calcium level.
 - Low Vitamin D causes hypocalcaemia and can't be ever the cause of hypercalcemia.
 - Almost all patients with primary hyperparathyroid have low Vitamin D

Conclusions & Novel insights into our practice

Vitamin D, Parathormone level and alkaline phosphatase can be the Only key for diagnosis such a condition and must raise the suspicion for Osteitis Fibrosa Cystica and direct us toward confirming the diagnosis by invasive biopsy.

Treatment of Osteitis Fibrosa Cystica is directed towards achieving normal levels of Vitamin D and serum calcium and also trialling to decrease osteoclastic activity.

The improvement of the clinical condition may takes a long time especially with the widespread bone affection which precludes any rush for surgical intervention.

The generalised bone affection of the Osteitis Fibrosa Cystica on the different imaging modalities may mimic the picture of a primary tumour within the bone marrow or metastastizing bone secondaries and invasive maneuvers are required to differentiate between them

References:

- Gal-Moscovici A, Popovtzer MM. New worldwide trends in presentation of renal osteodystrophy and its relationship to parathyroid hormone levels. *Cin Nephrol* 2005; 63:284.
- D'Amour P. Acute and chronic regulation of circulating PTH: significance in health and disease. *Clin Biochem* 2012; 45:964.
- Brown EM, Pollak M, Seidman CE, et al. Calcium-ion-sensing cell-surface receptors. *N Engl J med* 1995; 333:234.