

Case Report

Hyperphosphatemic tumoral calcinosis: a rare differential of periarticular swelling

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ABSTRACT

Tumour calcinosis is a rare clinical and histopathological syndrome characterised by deposition of calcium deposits in different periarticular soft tissue regions of the body. It mainly manifest in childhood/ adolescence as a painless, firm to hard tumour like mass around the joints. Most common regions involved: Shoulder, elbow and hip. An 18 year old male patient presented to the opd with a history of pain and swelling over his left hip since 2 months. On examination, there was a diffuse tender swelling over the left greater trochanter, skin over the swelling was normal with no discharge, no dilated/ engorged veins. Range of motion of left hip was normal, no limb length discrepancies. X-ray: Showed a well define calcified mass over the greater trochanter with no osseous involvement. MRI revealed an encapsulated hypointense mass present posterior to the greater trochanter, mostly in the muscular plane. Lab findings revealed mild hyperphosphatemia. An aspirate from the swelling showed casseousmaterial. En mass removal was done and sent for biopsy. Biopsy showed features suggestive of tumoral calcinosis. Tumoral calcinosis is a distinct clinico-radiopathological entity characterised by soft tissue periarticular calcinosis which mimics a true neoplasm, associated with elevated levels of serum phosphate. It is an extremely rare condition which is seen in the adolescence and requires more studies regarding the surgical and medical management of the same.

Keywords: Hyperphosphatemia, Myositis ossificans, Perarticular calcinosis, Tumour calcinosis

INTRODUCTION

Every once in a while authors come across diseases with very obvious clinical manifestations that could be recognised easily and yet very often misdiagnosed. Tumoral calcinosis is one such condition which is characterised by massive periarticular calcium deposits in soft tissues.¹ It mainly manifests in childhood or adolescence as a painless, firm, tumour like mass around the joints that might lead to restriction of movements if it is very large in size. Most common regions involved include: shoulder, elbow and hip. Spine, TMJ, metacarpals/metatarsals and popliteal space involvement are not uncommon. Tumoral Calcinosis was first stated by Inclan et al, as early as 1943 for a disease which was

characterised by periarticular calcified mass without involvement of skin or viscera². The characteristic features of these lesions were, the presence of multiple cysts filled with calcified deposits lined by histiocytes, gaint cells and xanthomatous histiocytes. It was also studied by Teutschlaender between 1930 and 1950 and was called the Teutschlaenders disease.³ In 1960s it was established that tumoral calcinosis had a familial tendency without any sex predominance and that it was significantly higher in among the African population.²

The lesions primarily show proliferation during the first 2 decades of life, although most of patients have normal serum calcium levels, a small group of them have found to have mild hyperphosphatemia.³ The lesions are

characterised by lobular, densely calcified mass confined to the soft tissue especially found over the extensor of the joints where there is anatomical distribution of bursa.

The clinical diagnosis of the disease has always been dependent upon the presence of periarticular calcified mass. Laboratory findings include elevated serum phosphate levels and an increased serum 1,25 dihydroxy vitamin D concentration. The radiological findings include typical calcified masses with fluid levels.

However the current diagnosis is based mostly on light microscopic findings of the excised mass which showed cysts containing deposits of calcium lined by granulation tissue or dense fibrous tissue. These Histopathology findings have helped in classification of the disease into active and inactive stages. Salvin et al, conducted a study wherein he divided the disease into 3 stages, stage 1 included cellular lesions without calcification, stage 2 included cellular cystic with calcification and the third stage was inactive calcifying mass.⁴ In this case report, authors describe the clinical and pathological findings and their diagnostic importance.

CASE REPORT

A nineteen year old boy presented to orthopaedic OPD with swelling over his left hip since 2 months. The patient gave a history of trivial trauma at the gym following which he noticed a swelling in proximal aspect thigh initially was associated with pain for a period of 2 days then the pain subsided, but the size of swelling gradually increased. His medical history was insignificant, he had no fever, no history of haematuria, abdominal pain or renal insufficiency or significant family history. Physical examination showed a tender mass over posterolateral aspect of greater trochanter of size 7x5cm, which was firm in consistency. The mass was sessile, was not attached to the under lying bone. The texture and temperature of the overlying skin was normal. There was no restriction of hip movement. On investigation, the serum calcium, serum albumin, vitamin D and renal function tests were normal while the phosphate level was high (5mg/dl). His haemogram was normal. Plain radiograph showed large lobulated calcified mass over the lateral aspect of greater trochanter (Figure 1).

MRI showed heterogeneously enhancing lesion in the intramuscular plane involving the gluteus medius and minimus muscle in the left thigh: likely myositis ossificans (Figure 2). Provisional diagnosis of myositis ossificans was made and decision was taken to excise the mass in view of pain and increasing size of mass.

Surgical Procedure

A linear incision of 10cm was made over the greater trochanter. Skin, fascia and gluteus muscle were incised,

the swelling was a well -defined encapsulated mass below gluteus maximus. Mass was removed en-mass.

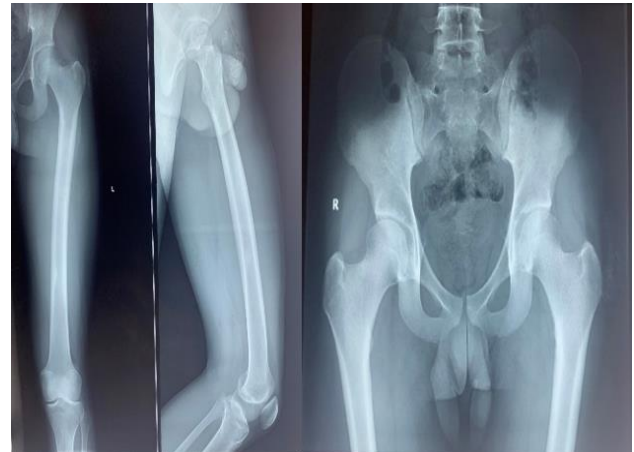


Figure 1: Amorphous cystic, lobular well demarcated calcifications in the periarticular regions without involvement of bone or joint.

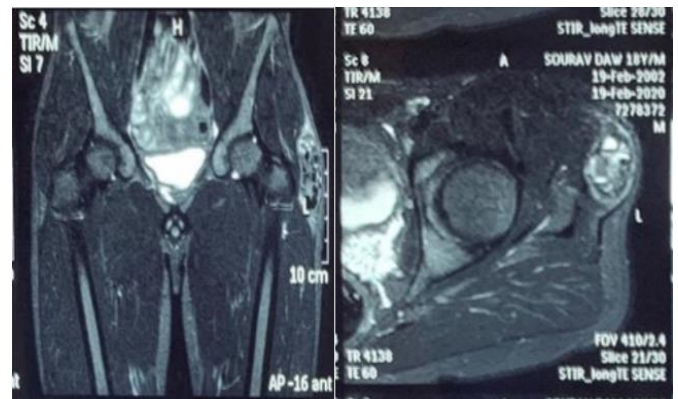


Figure 2: Heterogeneously enhancing lesion in the intramuscular plane involving the gluteus medius and minimus muscle in the left thigh: reported as likely myositis ossificans in the sagittal and axial sections.



Figure 3: Excision of mass.

Pathological findings

Gross

The tumor comprises of a well-defined encapsulated globular soft tissue mass. The 70x50x30 mm in dimension. The cut section of the tumor was grey- white and gelatinous with areas of calcification. Mass was also filled with white caseous material (Figure 3,4).

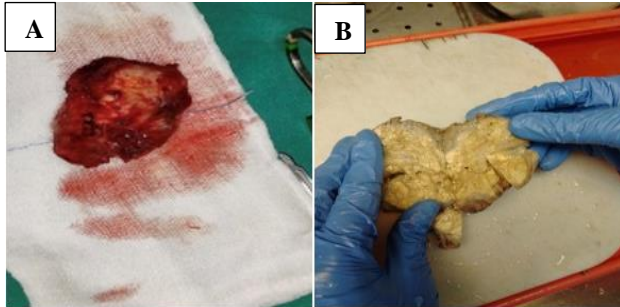


Figure 4: a) Excised specimen showing well-defined encapsulated globular soft tissue mass of size 70x50x30 mm in dimension. b) The cut section of the tumor was grey- white and gelatinous with areas of calcification.

Microscopy

The H and E stained tissue showed multiple cystic spaces with large geographic areas of calcification surrounded by palisaded histiocytic and numerous foreign body type of giant cells. The intervening fibrocartilaginous stroma was infiltrated by lymphocytes (Figure 5).

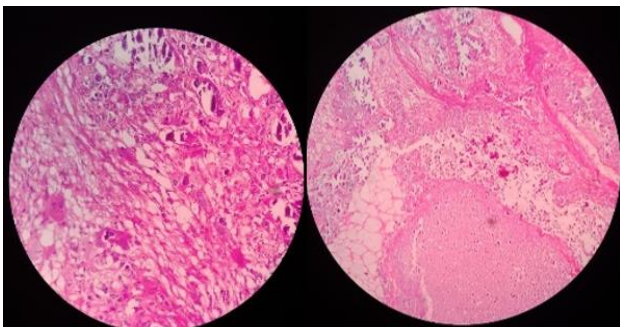


Figure 5: Multiple cystic spaces with large geographic areas of calcification surrounded by palisaded histiocytic and numerous foreign body type of giant cells. The intervening fibro cartilaginous stoma was infiltrated by lymphocytes.

Follow up

Patient was serially followed at 6 weeks, 3 months and 6months, 1 year for signs of recurrence and other complications. At one year followup, patient did not have any signs of recurrence. Hip movements were normal without any abductor insufficiency.

DISCUSSION

Tumoral calcinosis is a rare entity characterized by metabolic dysfunction of an unknown etiology. Manifests with large calcium deposits near large joints with a slow progressive growth. Tumoral calcinosis differs from other disorders such as hypervitaminosis D, Milk-alkali syndrome, dystrophic calcification, myositis ossificans as it is typically characterised by amorphous, cystic and multilobulated calcification in the periarticular region.² Dystrophic calcifications more superficial, smaller and do not progress in size unlike tumoral calcinosis About 1/3rd of the cases of tumoral calcinosis are familial, and show about autosomal dominant and recessive inheritance patterns.⁵⁻⁷ Lyles K. W et al, conducted a study on genetic transmission of tumoral calcinosis in a single generation of kinders and concluded that tumoral calcinosis is transmitted as an autosomal dominant condition with variable clinical expressions.⁷ In this case there no significant family history. The primary defect responsible for tumoral calcinosis appears to be hyperphosphatemia with and increased capacity of renal tubules to reabsorb filtered phosphate.^{8,9} In another study conducted by Lyles KW et al, on correlation of serum concentrations of 1,25 dihydroxy vitamin D, phosphorus and parathyroid hormone in tumoral calcinosis in 9 affected patients, it was found that both serum phosphorus and renal phosphate reabsorption threshold (TmP/GFR) were positively correlated with serum 1,25-(OH)2D levels.⁸ As tumoral calcinosis is a disorder with abnormal renal phosphate transport, a comparison was done between the TmP/GFR and serum 1,25-(OH)2D levels and a significant correlation between TmP/GFR and 1,25-(OH)2D levels was found suggesting that in these diseases 1,25-(OH)2D production is regulated in some manner by phosphate transport.⁹ Serum calcium, parathormone, ALP levels and renal function are generally normal. In this case, all blood parameters were normal except for raised phosphate levels. The complications include compression of neural structures and ulceration of the overlying skin with drainage of chalky white material with a risk of secondary infection.¹⁰ Tissue trauma associated with joint movement may be associated with periarticular calcifications. In this case, patient did not have any compressive symptoms or any skin changes, there was history of a trivial trauma was probably made him notice the swelling. But the swelling started to increase in size following trauma. This made us think in terms of myositis ossificans but classic gross and histopathological findings gave us the diagnosis of tumour calcinosis. The treatment of choice of this condition is complete excision. Excision of large masses could be associated with recurrence, secondary infection and abscess formation. In one of the studies conducted by Verma S bhansali it was concluded that chronic restriction of phosphorus was beneficial.¹⁰ Lufkin EG et al, conducted a study on phosphorus excretion in two related subjects and two controls, and it was found that the TC patients had normal sensitivity to PTH and normal response to acetazolamide with normal vitamin D levels.

Hence concluded TC was not due to abnormal hormonal response or due to disturbance in vitamin D distribution. Since acetazolamide aids in excretion of phosphorus it could also be used as a treatment modality in Tumoral calcinosis.¹¹⁻¹³

In this case authors did not use any phosphate lowering drugs as the levels were only marginally high. In serial followup of upto 10 months there was no signs of any recurrence of tumour and phosphate levels had returned to normal.

CONCLUSION

Tumoral calcinosis was described by Inclan as a disease of its own², with more than 800 cases of described in the literature under different names. Patients with this condition present at an early age with complaints of swelling with or without restriction of movements, with elevated phosphorus levels. Tumoral calcinosis also has very distinct radiological and pathological features. Hence this diagnosis should be kept in mind while treating patients with periarticular calcifications. Excision of mass is a reliable treatment modality with low chances of recurrence.

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