CASE REPORT

HYPERPHOSPHATAHEMIC TUMORAL CALCINOSIS SUCCESSFULLY TREATED WITH SURGICAL EXCISION AND ACETAZOLAMIDE

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ABSTRACT:
Tumoral Calcinosis (TC) is a rare disease of obscure aetiology. In its classic form, it is characterised by solitary or multiple large foci of mineralisation in the soft tissue adjacent to the bone around large joints in the absence of disorders of calcium metabolism and visceral calcification. We present a rare case of tumoral Calcinosis associated with hyperphosphataemia in a 27-year old Sudanese woman. Histological findings confirmed the diagnosis of tumoral calcinosis. Laboratory investigations showed hyperphosphataemia with normal levels of serum calcium and parathyroid hormone (PTH). The patient was treated successfully with surgical excision and acetazolamide.

Key words: Hyperphosphataemia, Tumoral Calcinosis, Acetazolamide
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INTRODUCTION:
Soft tissue calcifications are found in different diseases such as milk alkali syndrome, hypervitaminosis D, primary and secondary hyperparathyroidism and tumoral calcinosis (TC) [1]. Among these conditions, TC remains a poorly understood disease in which either solitary or multiple benign calcifications are usually found near large joints, without any involvement of the synovium itself or the adjacent bone in patients with normal serum calcium levels.

Although the aetiology of TC is unknown, some authors have associated this disease with metabolic disorders or with trauma [2]. TC
seems to be more common in the tropics [3]. A case of TC associated with hyperphosphataemia in a 27-year old Sudanese female who had been followed up for 8 months is presented.

**CASE REPORT:**
A healthy 27-year old Sudanese woman presented in October 2012 with a 4-month history of a painless swelling in the right upper lateral aspect of the hip region. It was insidious in onset, gradually increasing in size.

The patient had no fever and no pain, numbness, or weakness of the leg. There was no family history of any similar disease. Whereas it was not possible to recall any cause of the swelling (particularly trauma) the patient affirmed that she tended to lie on her right side while sleeping.

Physical examination revealed an oval, firm, subcutaneous tumour measuring about 5x6 cm. The skin over the mass exhibited an orange-peel appearance. Her right hip had a normal range of motion. The anteroposterior radiograph of the right hip revealed a lobulated, calcified soft tissue mass (Fig. 1). There was no fracture or periosteal change, and the soft-tissue thickness was normal. Fine needle aspiration cytology was done, but it showed no abnormality. The patient was planned for surgical excision of the mass. Wide local excision was done (Figure 2) with primary closure.

Histopathological examination revealed diffuse and extensive areas of spotty calcification in the subcutaneous tissue surrounded by fibro-fatty connective tissue and interspersed with macrophages and mixed inflammatory cell infiltrate (Figure 3). These findings confirmed the diagnosis of Tumoral Calcinosis. The margins were free and there was no evidence of malignancy.

Chemical analysis showed calcium phosphate crystals. Laboratory investigation of the blood revealed; serum calcium level of 2.43 (normal range: 2.20–2.70) mmol/l, serum phosphate level of 6.83 (normal range: 0.81–1.94) mmol/l, serum parathyroid hormone level of 2.8 (normal range: 1.6–6.9) μmol/l.

Hyperphosphataemia was treated with acetazolamide 500 mg/day [4]. Serum phosphorus of 1.6mmol/l was obtained within 4 weeks. During the follow-up period of 8 months there was no recurrence.
Figure 1: Anteroposterior radiograph of the right hip showing a lobulated calcific mass in the lateral region of the right hip. No joint or bone involvement is seen.

Figure 2: Macroscopic appearance of the completely excised mass: the lesion consists of an encapsulated, firm, lobulated soft tissue mass (5x6 cm) consisting of fibrous septa surrounding areas containing chalky material.

Figure 3: A photomicrograph of the biopsy-specimen showing areas of calcification surrounded by fibro-fatty connective tissue.
DISCUSSION:
The term tumoral Calcinosis (TC) define a condition in which either single or multiple tumour-like calcified masses are present without any associated calcium or phosphate metabolism disorder [5]. The calcifications normally adhere to the surrounding tissue and frequently appear in the vicinity of joints. Although hyperphosphaternia has been described in few patients, TC in the presence of a normal circulating phosphate concentration is the rule rather than the exception [6].

TC is a rare disease which has been recognised as a clinical entity since 1899 [7]. The pathogenesis of TC remains unclear and several theories have been proposed. Some authors consider it a hereditary metabolic dysfunction of phosphate regulation (with normal serum calcium levels) and distinguish it from calcification associated with renal osteodystrophy [8]. The presence of lesions around pressure points suggests the possibility of local trauma as a causative factor [9]. Cases of familial tumoral calcinosis generally are observed in younger patients, who tend to have multiple areas of calcification. A positive family history is encountered in 30% to 40% of cases [10]. The literature includes about 100 reports of familial tumoral calcinosis. In these reports, the disease results from disruptions in phosphate metabolism and is characterized by a high serum phosphate level. Recently, the genetic basis of familial tumoral calcinosis has been clarified [11]. The kidneys’ ability to excrete excess phosphorus from the body depends on the phosphaturic factor known as fibroblast growth factor 23 Nacetylgalactosaminyltransferase 3 (GalNAc-T3) isoform. A mis-sense mutation in the GalNAc-T3 gene is thought to constitute the genetic basis of this disorder [12].

The clinical presentation and radiographic features in our patient were quite typical, although the diagnosis could only be confirmed by a histological study. Even though some authors have shown that, in addition to radiography, other diagnostic modalities such as computed tomography and magnetic resonance imaging can also be helpful in making a correct preoperative diagnosis of TC [13], these examinations could not be performed in our case due to lack of sophisticated facilities at our small hospital.

A complete surgical resection of the calcium phosphate deposits thus remains the treatment of choice in patients where TC is not associated with any metabolic disorder, yet recurrence is frequent if removal is incomplete [14]. Our patient, who demonstrated hyperphosphstemia, was therefore treated with a wide local excision of the calcified mass in addition to acetazolamide and no recurrence was evident during 8 months of follow-up.
CONCLUSION:
The young age of our patient together with the presence of subcutaneous calcified mass, high serum phosphorus and histopathology allowed us to make the diagnosis of Tumoral Calcinosis and treat the patient successfully. Surgical excision should be complete in order to avoid local recurrence. This diagnosis should be kept in mind when dealing with subcutaneous calcified masses.

REFERENCES: