Rare disease

Recurrent hyperphosphatemic tumoural calcinosis

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Summary

Tumoural calcinosis (TC) is a benign gradually developing disorder that can occur in a variety of clinical settings, characterised by subcutaneous deposition of calcium phosphate with or without giant cell reaction. We describe a case of 11-year-old girl presenting with recurrent hard swellings in the vicinity of shoulder and hip joints associated with elevated serum phosphate and normal serum calcium levels. TC has been mainly reported from Africa, with very few cases reported from India. After the diagnosis of hyperphosphatemic TC was established, the patient was treated with oral sevelamer and is under constant follow-up to detect recurrence, if any. The present case highlights the fact that although an uncommon lesion, TC must be considered in the differential diagnosis of subcutaneous hard lump in the vicinity of a joint.

BACKGROUND

Tumoural calcinosis (TC) is a special form of idiopathic calcinosis of obscure aetiology that affects adolescents and young adults and is characterised by massive subcutaneous deposits of calcium phosphate near the joints such as hips, shoulders, elbows, wrists, feet and hands.^{1–3} The deposits consist of pleomorphic calcium phosphate (hydroxyapatite) crystals.³ Also known as Teutschlander disease, TC is a morphological diagnosis that can occur in a variety of clinical settings.⁴

CASE PRESENTATION

An 11-year-old girl presented with a bony hard swelling in the right gluteal region for 1½ year duration. The swelling first appeared as a small nodule and gradually grew to attain the present size. There was history of excision of similar swelling around the shoulder girdle, 3 years back. Examination revealed 12×10 cm subcutaneous mass located in the right gluteal region which was non-tender and firm to bony hard on palpation. The swelling was fixed to the surrounding structures, partially restricting the movements around the hip joint. The general and systemic examination was within normal limits except for the operative scar over the shoulder girdle. There were no symptoms suggestive of endocrinal abnormalities or connective tissue disorder and no history of similar swelling in other family members.

Roentgenogram of the pelvis disclosed a large, lobulated, radio-opaque soft tissue calcareous mass in the right gluteal region with septae extending into the juxta-articular region (figure 1). The contiguous bones and joint were unaffected. Haemogram, liver and renal function test were within normal limits. Biochemical examination revealed raised serum phosphate (7.2 mg%)

Figure 1 Roentgenographic appearance of right gluteal region (A) and right shoulder girdle (B) showing subcutaneous radio-opaque, lobulated, calcific masses without any abnormality of contiguous bones and joint.

Figure 2 Gross appearance of the excised gluteal specimen consisting of irregular cystic cavities filled with calcareous material.

with normal serum calcium, uric acid and alkaline phosphate levels. Hormone analysis for parathormone and calcitonin were within the normal range. Antinuclear antibody and rheumatoid factor were negative.

The excised mass was well encapsulated measuring 11×8×5 cm. The cut-surface was grey-white consisting of multiple cystic spaces containing granular grey white material. Microscopic examination showed irregular cystic spaces layered with granular, basophilic calcific material surrounded by histiocytes, fibroblasts, lymphocytes, plasma cells and numerous foreign body giant cells. Von-Kossa stain for calcium was positive.

OUTCOME AND FOLLOW-UP

The postoperative period was uneventful. The patient was treated with oral sevelamer to normalise the serum

phosphate level and is under follow-up to detect recurrences, if any.

DISCUSSION

TC is a heterogeneous disorder of obscure aetiology characterised by extensive non-osseous calcification especially in peri-articular soft tissue region of major joints¹ and is regarded as a special form of idiopathic calcinosis cutis.² Common sites are hip, shoulder, elbow and ankle, with the hip joint being the most commonly affected.⁵ TC most commonly affects adolescents, although occurrences in all age groups ranging from infancy to old age have been reported in the literature.⁵

TC is a gradually developing benign disorder⁴ with a characteristic juxta-articular calcification, progressive enlargement over time and a tendency to recur after surgical removal.⁵ The differential diagnosis includes other conditions with soft tissue calcification, that is, myositis ossificans, calcified fracture haematoma and systemic lupus erythematosus with secondary calcinosis.⁵ Morphologically, as in the present case, the lesions are cystic, multilocular and filled with calcareous material or chalky, milky fluid (figure 2). Polarising microscopic studies have shown that deposits in TC consist of calcium hydroxyapatite. The non-birefringence of these calcific deposits helps in differentiating it from birefringent calcium pyrophosphate dihydrate crystals of pseudogout (figure 3).⁵ Histopathology reveals calcium deposits in the dermis with or without giant cell reaction.¹

The aetiology remains obscure although the presence of lesions around pressure points may justify aberrant tissue reaction to local trauma as a possible aetiological mechanism.^{4 6} The disease may be sporadic, with patients having normal mineral levels and absence of underlying disease or; familial, an autosomal recessive hyperphosphatemic disorder with mutations in any of the fibroblast growth

Figure 3 (A) Microscopic appearance showing lesion comprising irregular cystic spaces heavily layered with basophilic calcium (H&E×100). (B) Higher magnification shows cystic space lined by fibroblasts, dense inflammatory infiltrate, foreign body giant cells and granular calcific deposits (H&E ×400).

factor gene, KL or GALNT3 gene.⁴ Defective function of these proteins results in defect in the proximal renal tubular cell, with elevated phosphate reabsorption and increased production of 1,25 dihydroxyvitamin D.^{3 4} Familial affection and association with underlying disorders of phosphate and calcium homeostasis-like chronic renal failure, primary hyperparathyroidism, hypervitaminosis D, milk-alkali syndrome and massive osteolysis are reported.² However, many cases like the present one are not associated with any biochemical derangement. Thus, TC is a morphological diagnosis characterised by tumour-like subcutaneous calcium salt deposition.²

Pathogenetically, TC has been divided into three distinct subtypes: (1) primary normophosphatemic TC; (2) primary hyperphosphatemic TC (PHTC); and (3) secondary TC wherein, patients have a concurrent disease capable of causing soft tissue calcification.⁷ The present case with normal calcium and elevated phosphate levels and no secondary disorder falls in the category of PHTC. Similar cases of recurrent PHTC have also been reported by Rangaswamy *et al*¹ and Jindal *et al*.³

Various treatment modalities like aluminium hydroxide or acetazolamide to increase urinary phosphate excretion have been tried, but none has been found to be effective.¹ Complete surgical excision has been recommended but recurrences are common.³ The present case highlights the fact that TC, although an uncommon lesion, should be considered in the differential diagnosis of a subcutaneous hard lump in the vicinity of a joint.

Learning points

- ▶ Tumoural calcinosis (TC) is an idiopathic disorder characterised by subcutaneous calcific deposits near the vicinity of a joint.
- ▶ The disease may be sporadic or familial; and may or may not be associated with derangement of biochemical parameter or other metabolic disorders.
- ▶ Though an uncommon lesion, TC should be considered in differential diagnosis of soft tissue calcific swellings.
- ▶ Lesions may recur and therefore the patient should be kept under constant follow-up.

Competing interests None.

Patient consent Obtained.

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