CASE REPORT

Tumoural Calcinosis

D Clarke, S Franklin, S Mullings, G Jones

ABSTRACT

Para-articular calcified masses remain a relatively common occurrence where it is most often due to chronic renal failure. The underlying aetiology is usually due to a disorder of calcium metabolism, chronic inflammation, or malignancy. In a small subset, it is due to hyperphosphatemia from an underlying disorder of phosphate metabolism, tumoural calcinosis. Identifying this subset of patients is paramount for effective management as medical management of the associated hyperphosphataemia is critical in lowering the incidence of recurrence.

Keywords: Para-articular calcified masses, tumoural calcinosis.

INTRODUCTION

Tumoural calcinosis is a rare benign idiopathic condition characterized by the presence of tumour like para-articular soft tissue masses (1, 2). These masses may be progressive and affect joint motion. Complete excision of the mass with medical management of phosphate metabolism is usually indicated (3).

A case of tumoural calcinosis in a 14-year-old male is presented here highlighting its progressive nature and management.

CASE REPORT

A 14-year-old boy presented to our outpatient department with a 6 month history of masses to the posterior aspect of his right knee and left elbow. There was no history of trauma, and no family history of similar occurrences. The masses gradually increased in size with tenting of skin. There were no neurological symptoms.

Examination revealed a firm mass in the posterior lateral aspect of popliteal fossa that was not attached to overlying skin. No neurovascular deficits were appreciated. Examination of the left arm revealed a mass to the posterior lateral aspect of the distal arm that was mobile and not attached to overlying skin. Biochemical markers were normal.

Sequential radiographs revealed a progressive increase in size (Figs. 1–4). A decision for surgical excision was made. At surgery, a lobulated mass measuring 15 × 7 × 5 cm was excised from the right knee, there was no attachment to surrounding soft tissue and the inner cavity consisted of a white amorphous substance. A mass similar in description was excised from the left elbow. His postoperative period was uneventful. Six months post-op, there were no signs of recurrence.

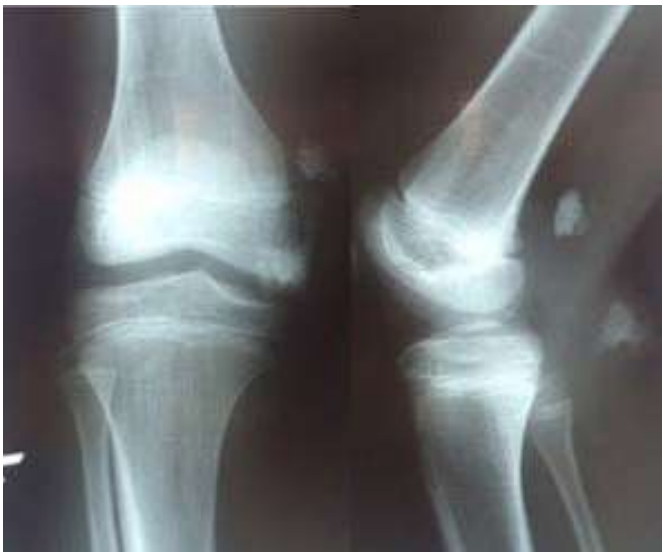

Fig. 1: Anterior posterior and lateral radiographs of knee on presentation.

From: Department of Orthopaedic Surgery, Mandeville Regional Hospital, Manchester, Jamaica.

Correspondence: Dr D Clarke, Department of Orthopaedic Surgery, Mandeville Regional Hospital, Manchester, Jamaica.
Email: daineoclarke@gmail.com

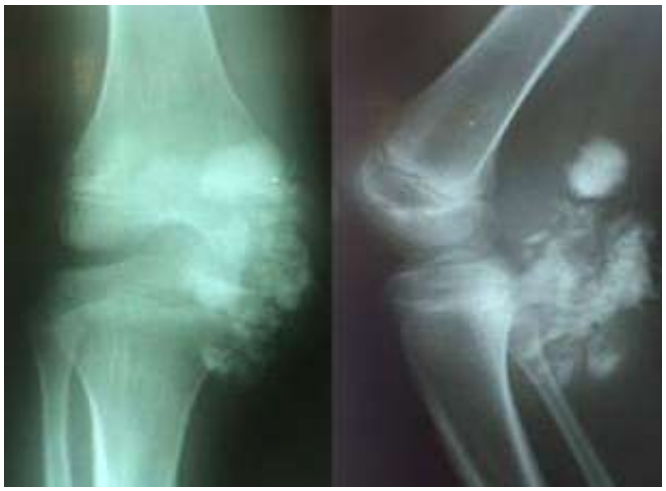

Fig. 2: Anterior posterior and lateral radiographs of the right knee 6 months after presentation.

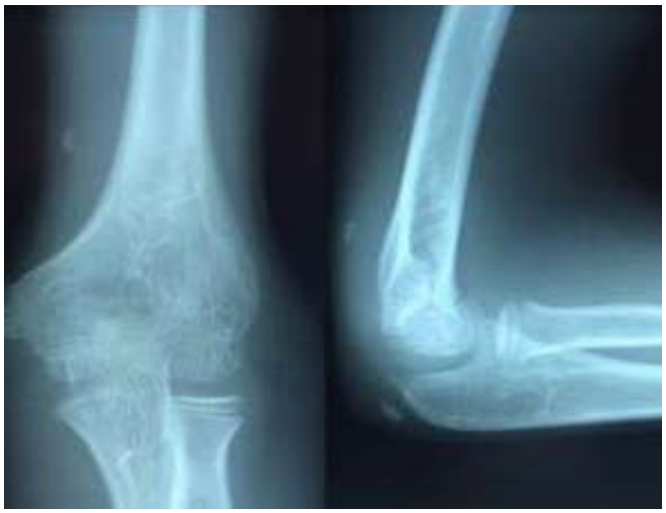

Fig. 3: Anterior posterior and lateral radiographs of left elbow on presentation.

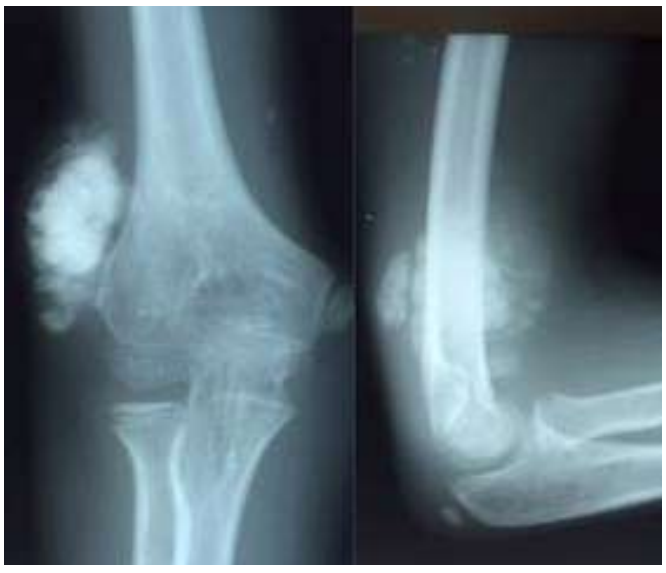

Fig. 4: Anterior posterior and lateral radiographs of left elbow 6 months after presentation.

DISCUSSION

Tumoural calcinosis is a rare condition characterized by the presence of painless firm circumscribed para-articular calcified masses. It was first described by Giard in 1898 and later by Duret in 1899 (4, 5). The term was coined by Inclan *et al* in 1943 (2). In their pivotal article, they defined metabolic criteria and differentiated tumoural calcinosis from other forms of soft tissue calcification. Inclan noted a normal calcium and phosphate level in all three of his reported cases, absence of collagen vascular disorders, and infections inclusive of tuberculosis. The authors made note of the progressive nature of the condition tendency to spread beyond bursal and muscular structures as well as its anatomic distribution in relation to gliding surfaces (2).

The condition has no sexual predilection and commonly occurs in the first or second decade of life as in the index case (6). There is an increased incidence in patients of African descent in keeping with our case (7–9). The condition may be sporadic or familial. Despite early reports of an autosomal dominant inheritance in the familial form by Lyles *et al*; more recent identification of the underlying genetic defect points to an autosomal recessive pattern of inheritance (10–13). Tumoural calcinosis has been linked to loss of function mutations in GALNT3, FGF23, and KLTHO gene that result in the inactivation of FGF-23 (11, 12). In effect, it may be seen as the clinically converse of hypophosphatemic rickets, which is due to a gain of function mutation in FGF23. Biochemically, this mutation is manifested by hyperphosphatemia due to increased renal reabsorption of phosphorus (14, 15). Other biochemical features of tumoural calcinosis include an elevated vitamin D level with normal calcium, parathyroid hormone, and renal function test (14–16). Thus serum calcium, serum phosphorous, urinary calcium (24 hours), serum parathyroid hormone, and serum vitamin D levels are indicated in the evaluation of these patients.

The clinical presentation is usually characterized by the presence of painless para-articular masses. The hip is the most commonly afflicted region followed by the elbow, shoulder, foot, and wrist (17). The lesions tend to progressively increase in size and may ulcerate and discharge a white calcific material (1). As the masses enlarge, they may also cause compressive symptoms and may affect joint motion, thus necessitating surgical excision (18). The characteristic radiographic appearance of tumoural calcinosis is that of multi-lobulated calcified densities separated by radiolucent bands with the absence of osseous destruction as seen in the radiographs of the

presented case (1). Plain radiographs may also demonstrate the characteristic dental lesions, root enlargement, and pulp stones (19). These represent calcific deposits that occupy and obliterate the pulp space. Martinez *et al* also demonstrated the radiographic evidence of calcific myelitis and periosteal reaction in three of their five patients (1). In their review of radiographic imaging in tumoural calcinosis in their patient set, bone scan offered the greatest sensitivity among all imaging modalities (1). Features on magnetic resonance imaging are rather unique with increased signal intensity on T2 weighted films (1). This seems rather paradoxical in lieu of the abundant calcific component. Computed tomography may show the 'sedimentation sign', representing layering of the calcification (20). Plain radiographs in conjunction with the history and biochemical parameters are, however, often sufficient to make the diagnosis.

The presence of para-articular calcified masses on plain radiographs is, however, not an infrequent occurrence. When present, the most common cause of this para-articular soft tissue calcification is chronic renal failure. Other differentials include chronic tophaceous gout, osteoma cutis, calcific myonecrosis, myositis ossificans, calcific tendonitis, synovial chondromatosis, and sarcomas: osteosarcoma and synovial sarcoma.

The underlying aetiology can be classified or stratified according to history and serum chemistry profile into metabolic, dystrophic, and idiopathic forms. Iatrogenic metabolic forms are due to elevated serum calcium levels with or without an associated elevated phosphate level. Examples are calcinosis of chronic renal failure and hyperparathyroidism. Dystrophic causes result from calcification in the presence of a normal calcium and phosphate level and are usually due to an underlying inflammatory disorder. Idiopathic calcification is characterized by normal calcium with elevated or normal phosphate levels. The latter is the group to which tumoural calcinosis exists.

The mainstay of management of tumoural calcinosis is complete excision of soft tissue masses where it may combined with medical management of phosphate dysregulation, *ie* phosphate bending antacid (*eg* aluminium hydroxide) in combination with acetazolamide (21, 22). Complete excision may prove challenging at times due to finger-like projections into surrounding soft tissue increasing the risk of recurrence (3). In cases of recurrence, the lesion tends to be more aggressive (3).

CONCLUSION

Tumoural calcinosis represents a rare form of idiopathic periarticular calcification due to a loss of function mutation resulting in an inactivation of FGF-23. Differentiating tumoural calcinosis from other conditions that may cause soft tissue calcification is critical to the management to prevent the added morbidity of over treatment.

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