Hyperphosphatemic Multifocal Tumour Calcinosis in a 14 year old boy

Sandesh Madi[1], Monappa Naik[2], Sandeep Vijayan[3], Sharath Rao[4]

ABSTRACT

Background: Tumour Calcinosis is a condition wherein amorphous calcium phosphate crystals get deposited in the soft tissues around the joints. Hyperphosphatemic type of Tumour Calcinosis is predominantly seen in Afro-American / Afro-Caribbean descent. It creates a diagnostic dilemma because of its rare and sporadic incidences. **Case report:** We report the case of hyperphosphatemic multifocal Tumor Calcinosis in a teenage boy, which was entirely unilateral affection, who underwent several futile surgeries before the actual diagnosis was made. **Conclusion:** Tumour Calcinosis is a rare differential diagnosis for soft tissue calcification in the periarticular region. Though the Hyperphosphatemic type has a familial tendency, diagnosing the index case without positive family history can be challenging. This leads to delay in the diagnosis of the condition and mismanagement. We report a case of hyperphosphatemic multifocal Tumour Calcinosis in a fourteen year old boy and briefly discuss the epidemiology, etiology, clinical, and radiological features of this condition.

Keywords: drainage, excision, hyperphosphatemia, multifocal, Tumor Calcinosis

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Conflict of interest: None declared

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INTRODUCTION

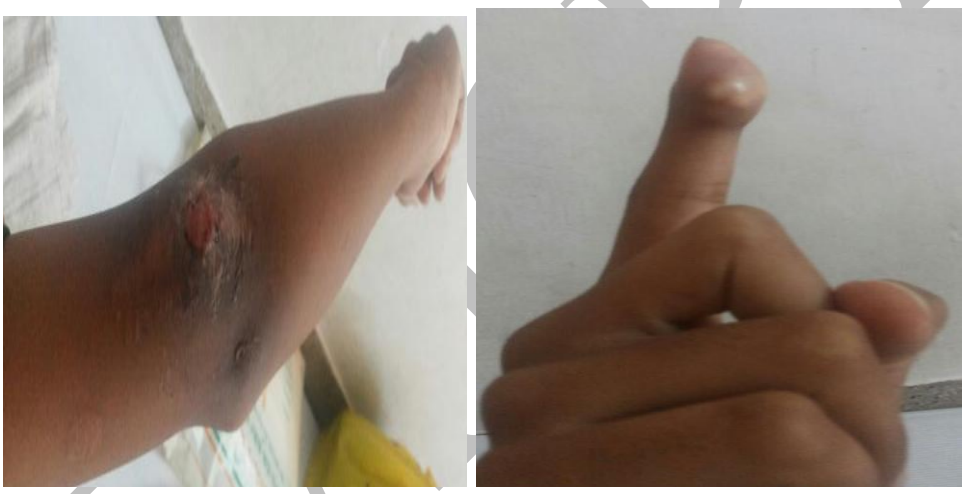
Tumour Calcinosis was first described by Giard in 1898; however the term was coined by Inclan in 1943 [1]. Clinical and radiological appearances mimic soft tissue malignancy and hence the name. Multifocal involvement is not uncommon, in which the large joints are mainly affected. There are three types of idiopathic Tumor Calcinosis: primary normophosphatemic, primary hyperphosphatemic, and the secondary. The hyperphosphatemic type is characterized by

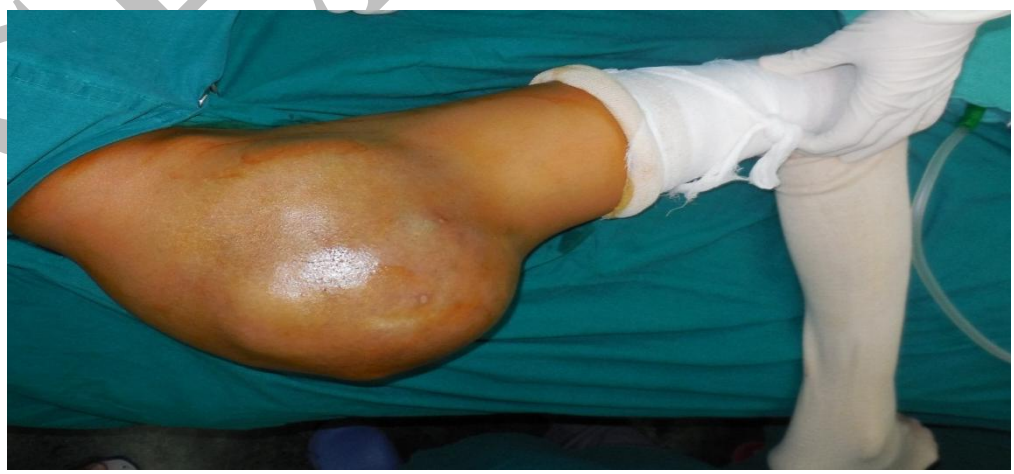
a comparatively high familial incidence, onset before 20, black race and multiple lesions [1]. Despite being a benign condition, it is however, notorious for recurrence (0-33%) [2].

CASE REPORT

A 14 year old boy presented to our Paediatric Orthopaedic clinic with a swelling over right elbow, right index finger and right hip region [Figure 1].

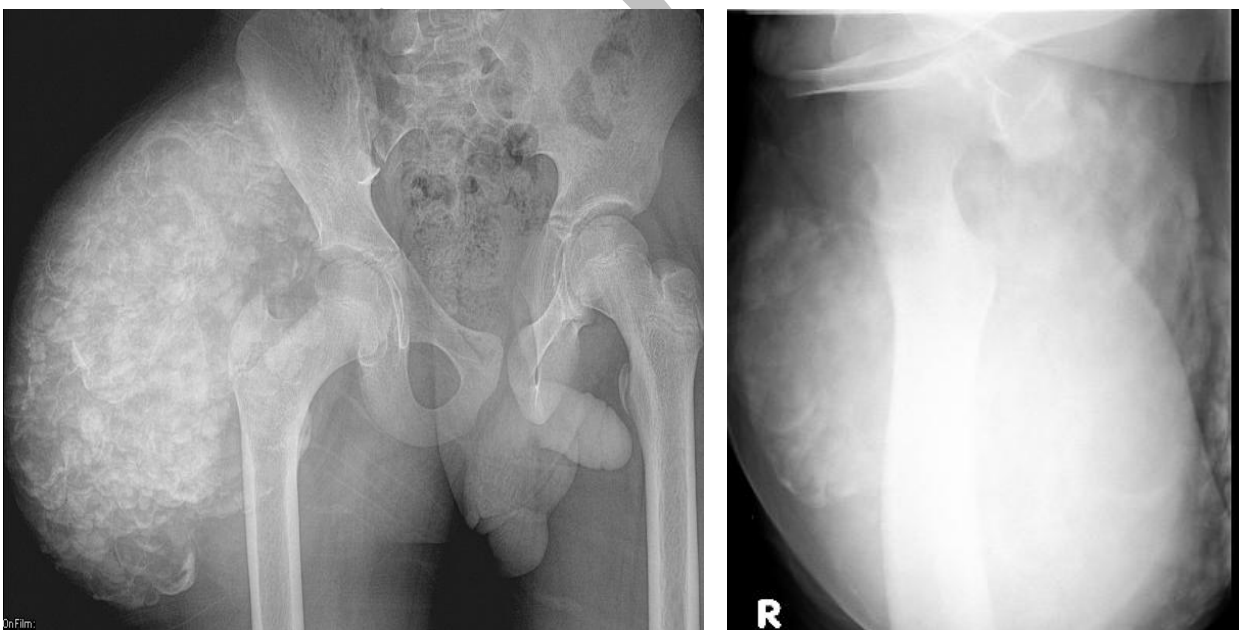
Figure 1: Clinical images of right elbow, right index finger and right hip

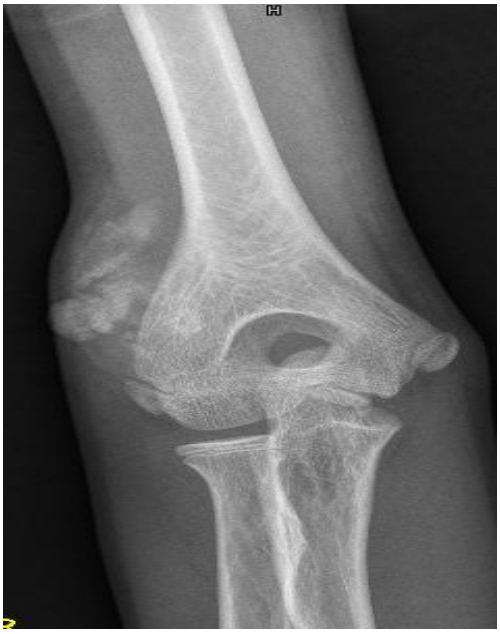


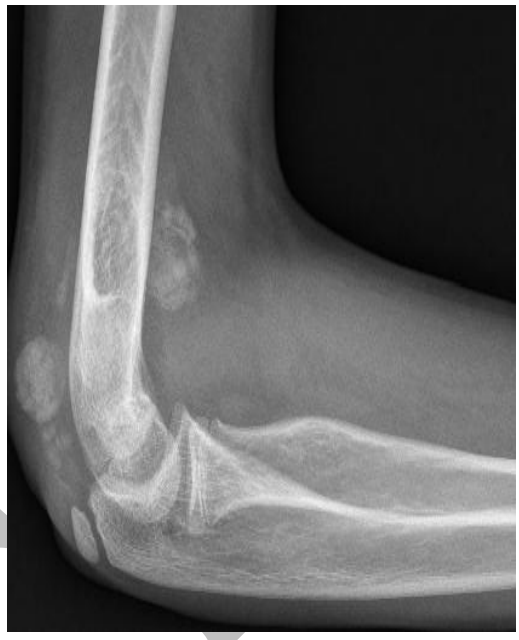


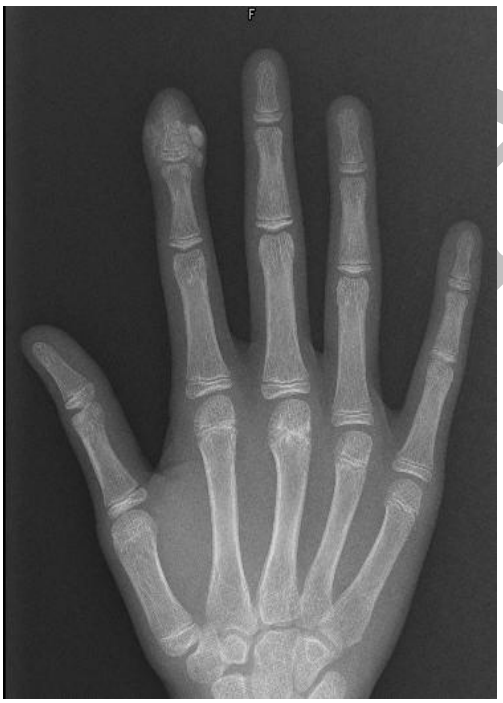
The swellings were non traumatic origin, painless, insidious onset and gradually progressive. There was no history of fever or weight loss. There was no similar condition in the family. The swelling first started in the elbow region 1 year back for which incision and drainage was done three times by a general practitioner assuming it to be an abscess. Next the pulp of right index finger got involved. Parents also noticed swelling around the right hip since 2 months which started to grow rapidly. All the lesions were firm in consistency and located in subcutaneous plane. No neuro-vascular deficits were seen. Range of motion of these joints were unaffected. Plain radiographs revealed cystic multi-lobulated calcifications around the hip, the elbow and distal phalanx index finger [Figure 2].

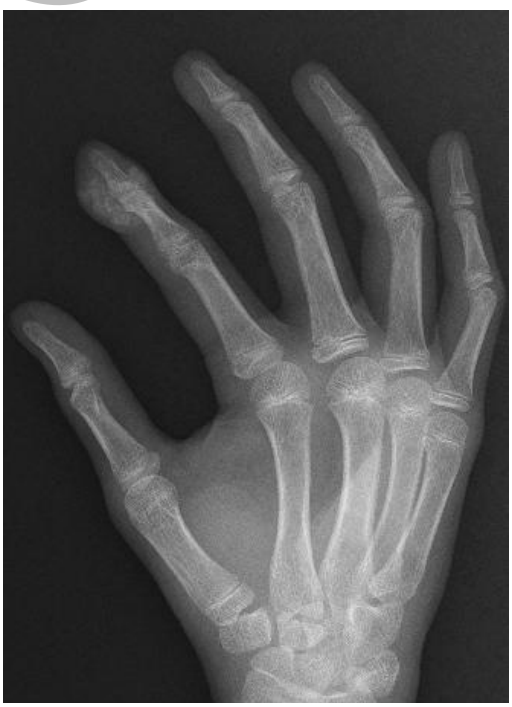
Figure 2: plain x-rays (antero-posterior and lateral views) of the right hip, elbow and index finger (oblique view).





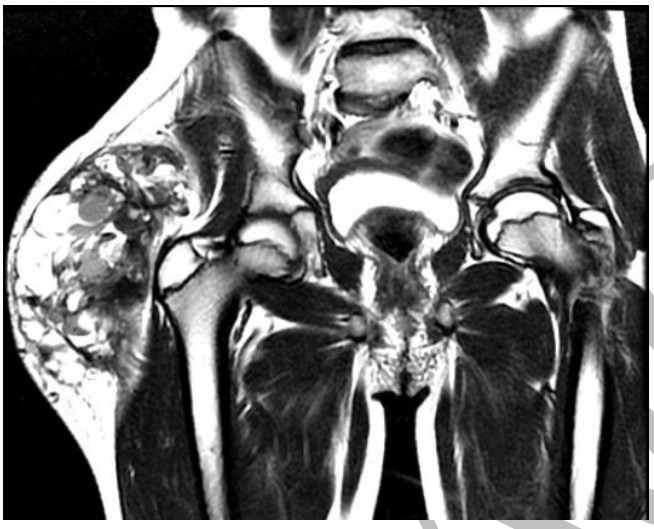


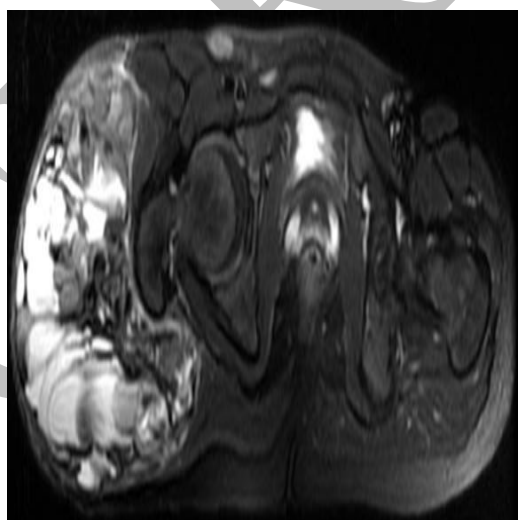




His serum Calcium profile revealed hyperphosphataemia with normal serum calcium, alkaline phosphate, vitamin D and parathyroid hormone levels. MRI hip reported a large multi-lobulated septated mass lesion measuring 12.3x12x15.5cms with multiple fluid levels seen infiltrating Gluteus Medius and Maximus [figure 3].

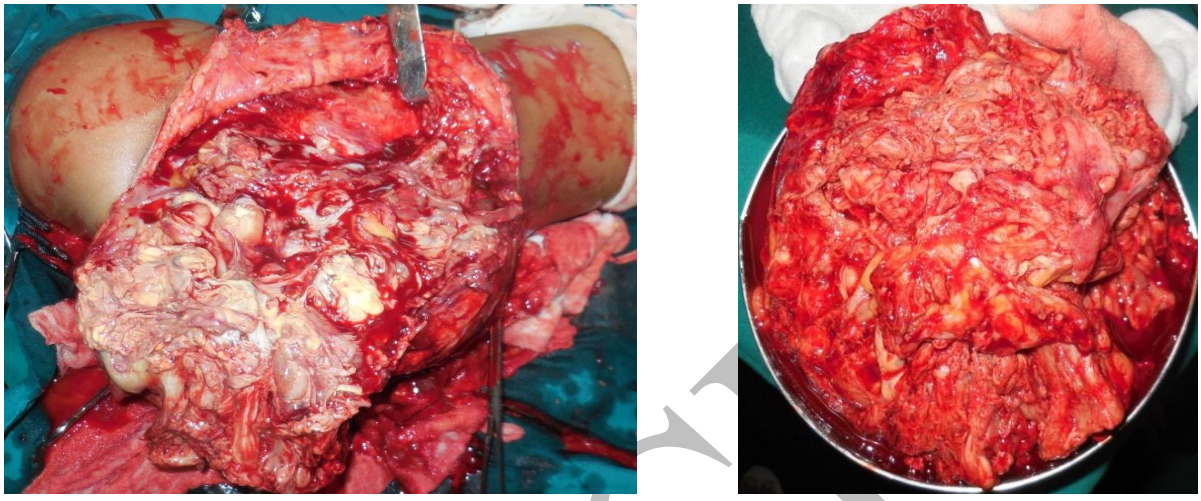
Figure 3: MRI images of the hip defining the level and extent of the lesion (coronal and axial views).





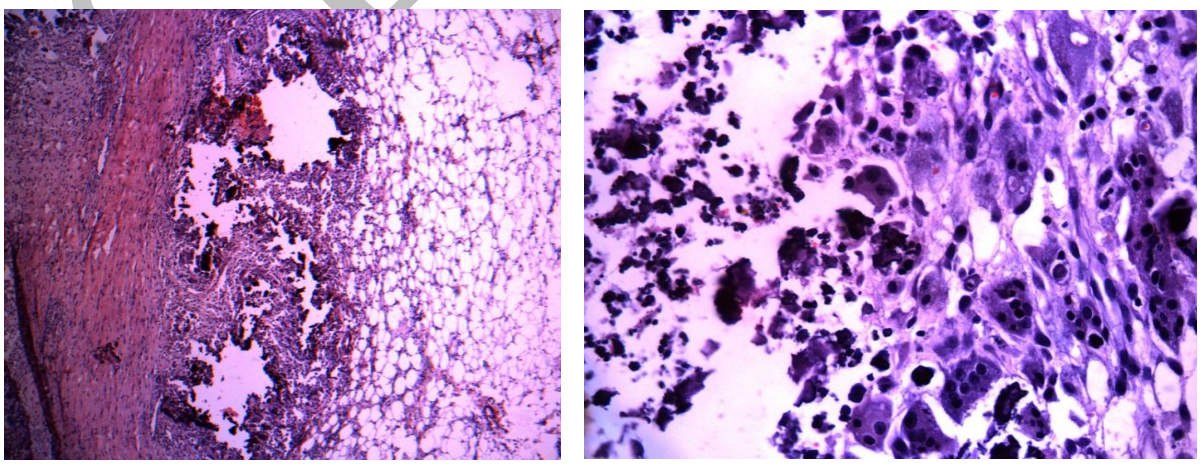
The elbow lesion was biopsied which showed focal aggregates of calcified areas surrounded by fibrous connective tissue with inflammatory changes suggestive of calcinosis cutis. In view of rapid growth and structures at risk of compression around the right hip, the lesion was excised. It was noted that the lesion was adherent to the gluteal maximus muscle [Figure 4].

Figure 4: Intra operative images of the hip lesion and gross appearance of the lesion.



Histopathology sections from the hip lesion showed fibrocollageneous stroma with multiple cystic spaces containing amorphous calcification [figure 5]. The post-operative days were uneventful. Patient was discharged with oral phosphate binding agents and a diet restricted in calcium and phosphate. There is no recurrence of the swelling in the hip or elbow for last 1 year; surprisingly, the finger lesion remains asymptomatic despite being his dominant hand.

Figure 5: Histopathlogy showing multiple cystic spaces filled with calcium salts and a fibro colageneous stroma [10X & 40X power].



DISCUSSION

Tumour Calcinosis usually presents in first two decades of life; males and females are apparently equally affected. The genetic transmission in the familial type is still unclear. Both autosomal dominant and autosomal recessive transmissions with variable expressivity are seen. Joints commonly involved are, in the decreasing order, hips, elbows, shoulders, and knees, mainly on the extensor aspect. Small joints of hands and feet are rarely involved [3].

Although the condition is classically described to be common in Afro-American/Afro-Caribbean population, more than 800 sporadic cases have been reported in the literature till date under different names [4] worldwide and yet the condition is frequently misdiagnosed. McClatchie et al in 1969 reported 26 cases of Tumor Calcinosis in the Kenya –Uganda region. Most of the cases were wrongly diagnosed as calcified Oncocercariasis which was endemic in the region sat that time. In the same paper they mention of 50 additional cases reported from Rhodesia and Nigeria [5]. After the publication of this paper, three more cases were reported from west of Ireland. The author refuted that the condition is under recognized and believed that it can affect the white race as well [6]. In the same year, R.A.Cooke claimed that the condition is also common in inhabitants from Papua-New Guinea and reported 16 cases [7]. Riemenschneider and Ecker, in 1952 from U.S.A described an unusual tumour calcinosis of spine presenting as sciatica [8]. Jin Soo Suh reported about three Korean women, where only the small joints of hands were involved [9]. Che Zhang et al in 2011 first reported the condition in two members of Han Chinese family [10]. More than 40 isolated and sporadic cases have been reported from the Indian subcontinent in the

last 35 yrs. Several isolated cases have also been reported from Middle East region.

Disease manifests with multiple non-synchronous painless growths in the periarticular region. The swellings are firm to hard, lobulated, and firmly attached to underlying fascia and muscles. The rate of progression is highly variable with some showing rapid growth and others being indolent for several years. There is no predilection to any particular sites for the rapid growth to occur. It is uncertain whether the growth of these lesions occur more rapidly when multifocal involvement is encountered. Solitary lesions too have proven the potential to grow rapidly and become symptomatic. Sometimes, due to massive growth, the overlying skin tends to break down and ulcerate which can get secondarily infected. Over a period of time, this can develop into a sinus from which a milky fluid containing calcium salts keeps discharging. Underlying neurovascular structures may also be at risk of compression.

The exact etio-pathogenesis is still unclear, however two theories have been speculated: (a) an anomalous tissue response to trauma leading to dystrophic calcification, especially around the pressure points. (b) An inborn error of phosphorus and vitamin D metabolism characterized by an increase in proximal tubular phosphate transport. This results in formation of the calcium phosphate complex which gets deposited in the skin and subcutaneous tissues. Soft tissue calcification also occurs frequently in patients with secondary hyperparathyroidism due to renal failure. The other conditions leading to ‘secondary’ Tumour Calcinosis are Hypervitaminosis D, Primary Hyperparathyroidism, Sarcoidosis, Milk-Alkali syndrome, Calcinosis Universalis, Collagen Vascular diseases,

Pseudogout and Paraneoplastic syndromes. In all the above mentioned conditions hypercalcemia is seen but primary Tumour Calcinosis is typically characterized by normocalcemia.

Clinically the disease is frequently mistaken for an abscess/parasitic infestations and futile attempts of drainage are done, especially by local practitioners in remote places who are unaware of the condition. When there is no response to multiple drainage attempts, patients have been inadvertently started on anti-Tuberculosis regime also, which is not uncommon in Tuberculosis endemic belts [3].

CONCLUSION

Tumour Calcinosis is rare differential diagnosis for soft tissue calcifications in periarticular region. Clinical and radiological picture are usually characteristic, however, in some cases it can masquerade as soft tissue malignancy or infections which leads to delay in diagnosis. More and more cases being reported from all over the globe highlights that the condition is not restricted to a particular ethnicity/region and some other underlying factors may also be involved for its etiopathogenesis. Complete surgical excision of the 'symptomatic' swelling is the treatment of choice; but, vigilance is must for recurrence. Surgical indications include pressure symptoms, restricted range of motion of the joints, significant deformity or rarely for cosmetic purposes. Medical line of treatment may be helpful in the form of phosphate binding antacids and phosphaturic agents, but only in the 'hyperphosphatemic' type.

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