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Recurrent Familial Normophosphatemic Tumoral Calcinosis: A Case Report

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Learning Point of the Article:

Normophosphatemic tumoral calcinosis with ophthalmic involvement.

Abstract

Introduction: Tumoral calcinosis (TC) is a rare disorder characterized by periarticular soft-tissue deposition of calcium around large joints.

Case Report: We report a recurrent familial case of a 63-year-old Indian female with progressive painless discharging mass over the left proximal thigh associated with extensive ophthalmological involvement in the form of perilimbal calcific deposits, angioid streak, pigment epithelial defects and choroidal neovascular membrane in both eyes. Hematological parameters revealed normophosphatemic. Radiological investigation suggested periarticular calcific deposits with sedimentation sign and the diagnosis was later confirmed on histopathological examination.

Conclusion: We report the case because of its rarity and suggest thorough ophthalmological and genetic evaluation in each and every patient of TC.

Keywords: Recurrent familial, normophosphatemic, tumoral calcinosis, ophthalmological manifestation.

Introduction

Tumoral calcinosis (TC) is a rare, benign condition characterized by deposits of calcific material in periarticular soft tissues [1]. It most commonly involves the larger joints such as the hip and shoulder, but also has been reported in joints such as the elbow, wrist, knee, scalp, larynx, spine, and sacrum [2]. This condition mostly occurs in adolescents and young adults, but familial forms affecting the infants are also described [3]. These lesions are slowly growing and progress over the years and may be associated with ulcerations of the overlying skin [4]. In 1899, Duret reported two such cases in two siblings and named it as calcified endothelium [5]. In 1935, Teutschlander labeled it as lipocalcino granulomatosis or Teutschlander disease. In 1943, the name was revised to TC by Inclan et al. [6].

The etiology of TC is inconclusive despite the proposal of many theories [4]. On the basis of underlying etiology, the disease can be primary or secondary to other conditions. Depending upon the phosphate level, primary TC can be normophosphatemic or hyperphosphatemic. The secondary TC is usually secondary to conditions such as chronic kidney diseases, hyperparathyroidism, and hypervitaminosis D [7]. In this article, we report a primary recurrent normophosphatemic familial variety of TC in a female patient.

Case Report

A 63-year-old female patient with no comorbidities presented to

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Author's Photo Gallery


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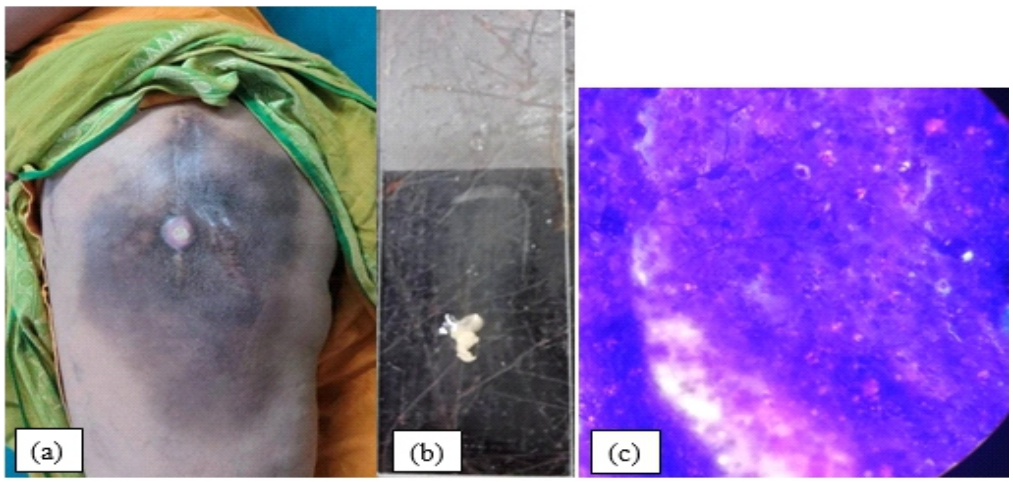

Figure 1: [a]: Clinical picture of the mass with ulceration and hyperpigmentation, [b]: Chalky whitish fluidic discharge from the ulceration. [c] Histological examination of discharge showing a fibrous connective tissue with amorphous calcified material boarded by a florid proliferation of macrophages and multinucleated osteoclast-like giant cells.

our department with a history of progressive swelling over the left upper thigh with discharge of whitish chalky material from the ulceration over it (Fig. 1a). She was aware of the mass for the last 6 months, associated with minimal dull aching pain following strenuous exertion. She had similar episodes twice in the past, for which she was operated on the first time when she was 16 years old and for the second time around 7 years ago. Among her family members, only one of her younger sisters has a similar mass over the left gluteus region. On examination, an irregular, hard, mildly tender, mobile mass of size 10 × 5 cm was present over the left lateral aspect of the proximal thigh associated with hyperpigmentation of the overlying skin with two linear scar marks. A single ulcer was present at the center of the mass with chalky white fluid discharge (Fig. 1b). The range of motions of the ipsilateral hip joint was normal, and the local draining lymph nodes were not enlarged.

Radiography revealed an irregular, calcified mass of size 10 × 5 cm over the left upper thigh without any involvement of the hip joint and proximal femur (Fig. 2a). Contrast-enhanced computed tomography (CT) of the pelvis revealed an ill-defined, multilobulated, amorphous and heterogenous calcified lesion in the subcutaneous plane in the left gluteal region into the left gluteus maximus with minimal involvement of the gluteus medius measuring 7.1 × 7.5 × 8.9 cm (Fig. 2b). The coronal CT scan (bone window and soft tissue window) shows a left periarticular multilocular calcified mass with a few fluid calcium levels suggestive of sedimentation sign (Figs. 2c and d). On post-contrast images, mild enhancement was seen in the non-calcified soft tissue, and there was no involvement of the bony structure. Magnetic resonance imaging (MRI) revealed a subcutaneous lobulated mass over the left greater trochanteric region of the femur, posterolaterally with fixity to the underlying deep fascia and gluteus maximus. Overlying skin showed focal thickening and scar formation (Fig. 3a, b, c).

Laboratory investigations showed normal phosphate value, without any hematologic value alterations of 1,25-dihydroxyvitamin D or parathyroid hormone. Renal function was normal. Histological examination of the discharge taken from the sinus showed a fibrous connective tissue with amorphous calcified material bordered by a florid proliferation of macrophages and multinucleated osteoclast-like giant cells (Fig. 1c).

Dental examination revealed no abnormality, whereas ophthalmological examination revealed best corrected visual acuity of 6/18 in both eyes and intraocular pressure of 17 mmHg in both eyes. Slit lamp examinations of the anterior segment detected perilimbal calcific deposits (Fig. 4). Ocular fundus examination of the left eye shows an angioid streak between the disc and macula, multiple pigment epithelial defects, and a choroidal neovascular membrane (CNVM) in the macular area with multiple calcific deposits in the periphery (Fig. 5b). The right eye showed pigment epithelial detachments with multiple

calcific deposits in the periphery (Fig. 5a). Optical coherence tomography showed multiple pigmented epithelial defects and CNVMs in both eyes (Fig. 5c).

Factoring in the patient history, clinical assessment, and diagnostic evaluation, the final diagnosis of recurrent familial normophosphatemic TC of the greater-trochanteric region was made and planned for wide excision.

The patient underwent a wide excision, keeping a 1 cm healthy margin. The resected mass was measured to be 15 cm in the largest diameter, whitish in color, and milky fluid came out during sectioning (Fig. 6a). Pathologic evaluation revealed deeply basophilic amorphous granular material consistent with calcium deposits surrounded by dense fibrous tissue, confirming the diagnosis of TC (Fig. 6b). No cellular atypia and other signs of malignancy were found. The patient was evaluated after 2 years post-discharge. The scar was healthy, and there was no sign of recurrence.

Discussion

Based on the underlying etiology, Smack et al. [7] divided TC

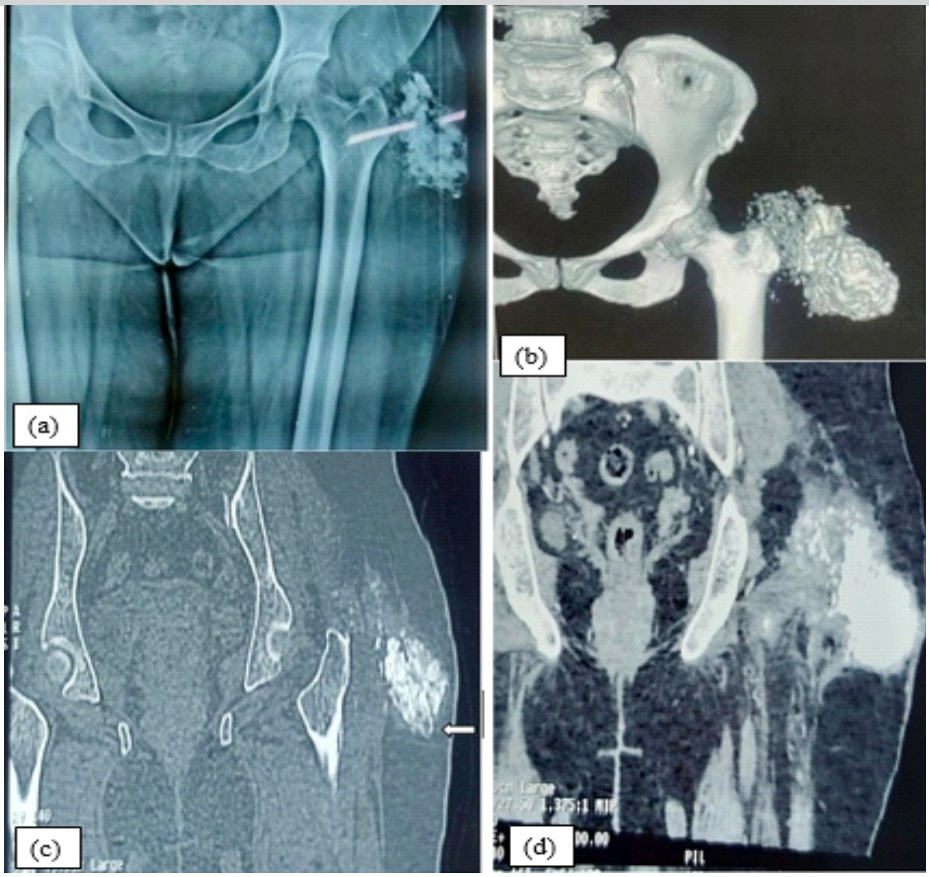

Figure 2: Fig 2[a]: X-ray picture showing a multiobulated periarticular calcified mass around the left hip joint. [b]: CT scan depicting the calcified mass around left hip joint. [c & d]: The coronal CT scan (Bone window & soft tissue window) shows left periarticular multilocular calcified mass with few fluid calcium level (suggestive of sedimentation sign) involving left gluteus maximus muscles with soft tissue extensions around the hips & the upper femur.

into primary or secondary to other conditions. In primary normophosphatemic types, both serum calcium and phosphorus levels are the same without any other hematological abnormalities. It usually presents before the 2nd decade of life, and this variant is familial, involving the gene encoding the SAMD9 protein [7]. In our reported case, repeated hematological investigation showed normophosphatemia, and the patient presented her first episode in the 2nd decade, for which she had undergone surgery somewhere else. The presence of a similar presentation in one of her sisters confirms the familial form of the condition. The second group is the primary hyperphosphatemic type, usually presenting during the first and second decades of life. This group of patients has a genetic predisposition with reduced urinary phosphate excretion caused by autosomal recessive mutations in GALNT3 and KLOTHO that cause inactivation of FGF23, a phosphaturic hormone. The third group encompasses secondary TC connected with chronic renal failure, primary hyperparathyroidism, hypervitaminosis D, sarcoidosis, etc. [7].

Despite its different etiopathogenesis, the clinical and histopathological features are almost similar to those of our reported case. Joints most commonly affected

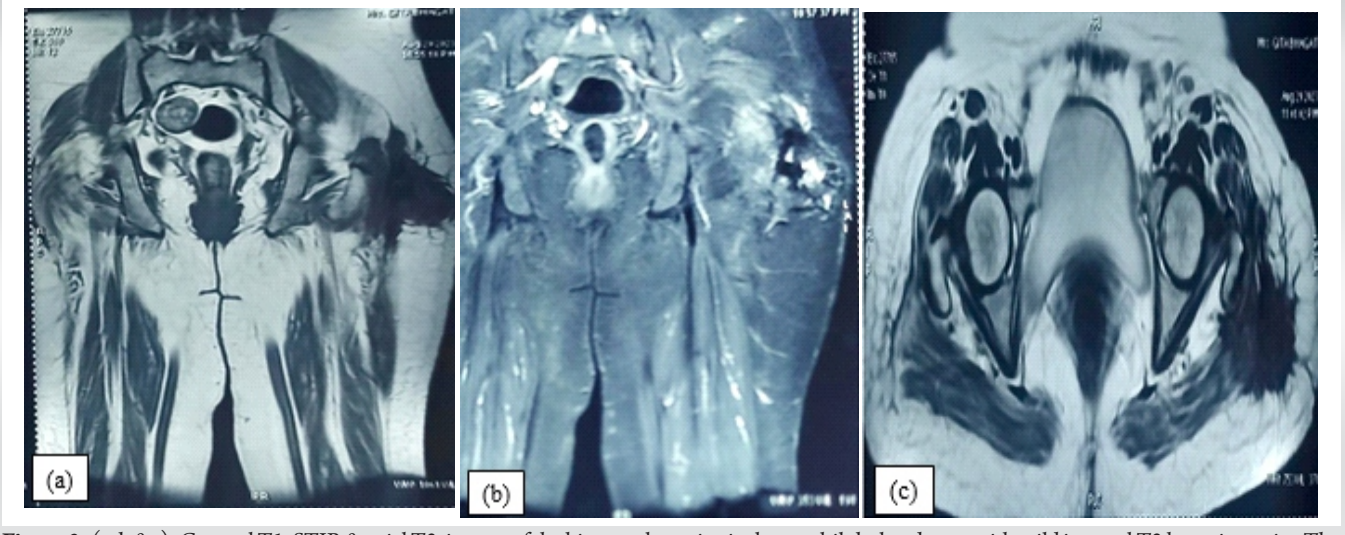

Figure 3: (a, b &c): Coronal T1, STIR & axial T2, images of the hip reveal a periarticular , multilobulated mass with mild internal T2 hyperintensity .The mass is well demarcated, lobulated and predominantly low signal on T2 weighted sequences suggestive of calcification noted in the left side.

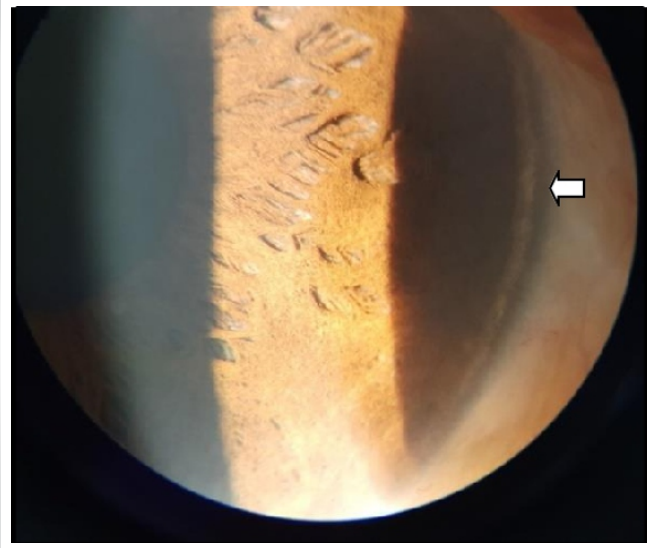

Figure 4: Arrow head showing perilimbal calcific deposit.

include the hip, elbow, shoulder, foot, and wrist [4, 8]. Apart from this, small joints of the hand, such as proximal and distal interphalangeal joints, are also involved [9]. These lesions are slow-growing and progress over the years. Lesions may be associated with ulceration of the overlying skin with characteristic chalky white liquid discharge material [10]. In children, bone marrow sclerosis and periosteal reactions have also been reported, suggesting its association with chronic recurrent multifocal osteomyelitis [3]. Ophthalmological findings include scleral calcification, limbal calcific deposits, angioid streaks, and subretinal neovascular membrane [11, 12]. Angioid streaks in the retina may be related to GALNT3 or FGF23 gene mutations [13]. However, we also noted a similar finding, although the above case represents familial normophosphatemic TC. In some cases, calcific deposits in the eyelid and conjunctiva are also seen. There are also possibilities of dental involvement in the form of pulp cavity calcification [14].

Preoperatively, TC is diagnosed by various radiological techniques. Findings of amorphous, multiloculated, and cystic calcifications in a periarticular location on plain radiographs are suggestive of TC [8]. This well-circumscribed, multilobular radiological appearance is described as a cobblestone or chicken wire appearance. CT serves as a guide for surgical excision. It detects the periarticular calcified mass with sedimentation sign (cystic loculi with fluid levels caused by calcium layering) [15]. In MRI, the lesion usually has non-homogeneous diffuse low signal intensity in T1-weighted sequences and alternating signal patterns on T2-weighted images with low signal intensity and cystic components with fluid levels (MRI sedimentation sign) [15].

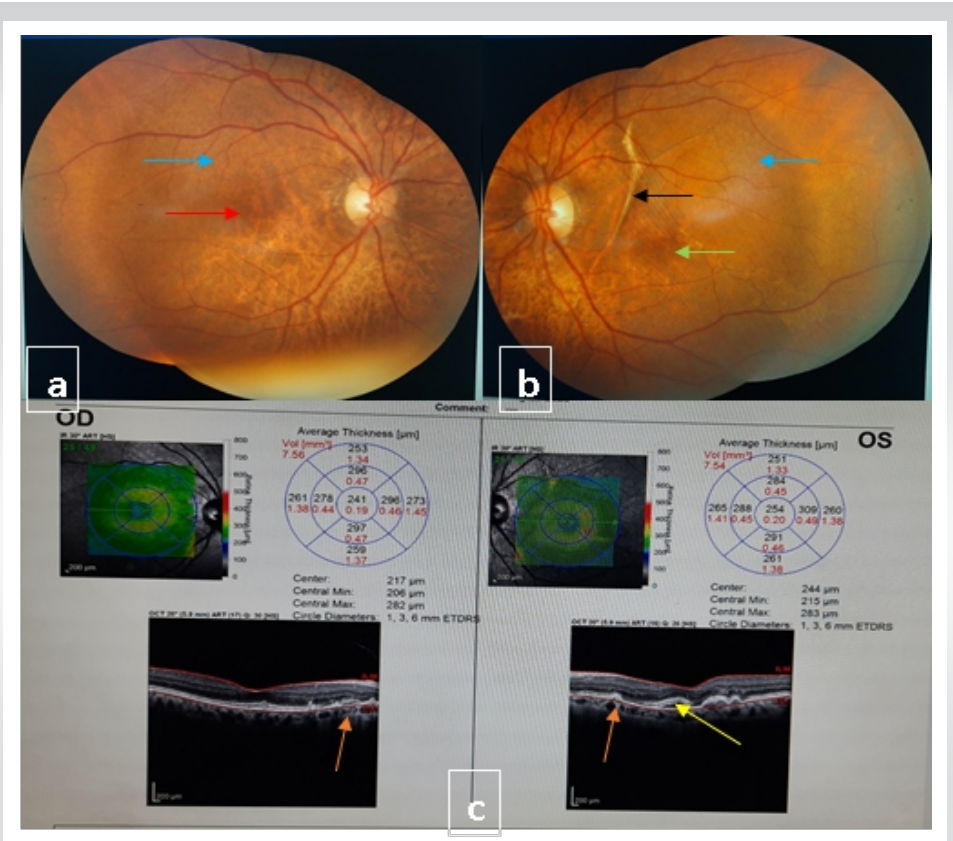

Figure 5: Fundus and Optical Coherence Tomography (OCT) picture of right and left eye (a) Red arrow showing pigment epithelial defects and blue arrow showing calcific deposits in right eye fundus (b) Black arrow shows angioid streak, green arrow representing pigment epithelial defects and blue arrow pointing dotted spot of calcifying deposits in left eye fundus. (c) OCT picture both eye showing multiple pigmented epithelial defects (yellow arrow) and choroidal neovascular membranes (Orange arrow).

Complete surgical excision has been classically described for the primary type, although it is associated with high recurrence, failure of complete excision, persistence of etiology, high complication rate, and secondary infection [16]. Even wider surgical excision, keeping an adequate healthy margin, has also not been proven to prevent recurrence [17]. Recurrence is managed by repeated excision. In hyperphosphatemic variants, in order to prevent recurrence, medical managements such as dietary phosphate restriction, use of phosphate binders, acetazolamide etc., are added postoperatively [18].

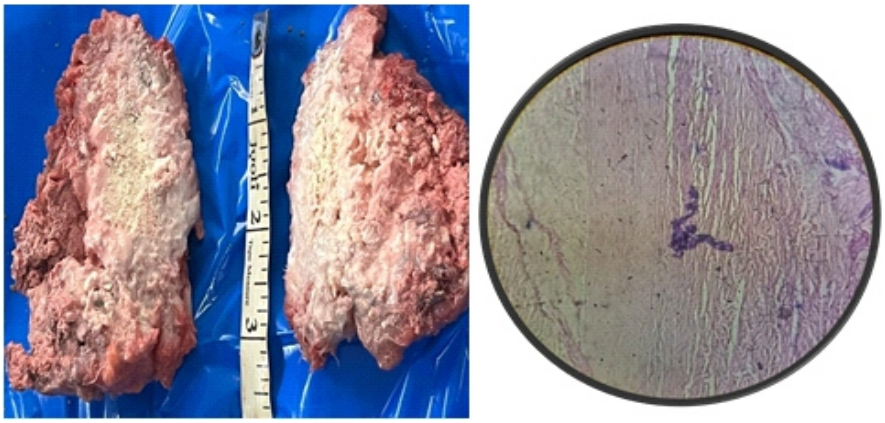

Figure 6: [a]: Gross appearance of the en bloc specimen, [b]: Microscopic section revealing calcification and fibrosis.

diagnosis, and ophthalmological findings, the present case has been reported. Clinical features, careful evaluation of imaging findings, and biochemical profile help in reaching the diagnosis. Biopsy is rarely indicated, only in suspicion of malignancy. Wide surgical excision with or without medical treatment can prevent recurrence.

Conclusion

TC is a rare diagnosis with poorly defined etiologies. We report an unusual recurrence with a metabolically active lesion in an adult female of familial variety following a long period of quiescence. Considering the rarity of the case, difficulty in

Clinical Message

Although TC is a rare entity, a high index of suspicion is needed to rule it out in patients presenting with periarticular calcific deposits. Once a diagnosis is made, all patients should be screened for possible ophthalmological involvement.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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