Ocular involvement in tumoral calcinosis

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We report a 32-year-old male who presented with blurring of vision in the right eye since 1.5 years. He had history of swelling over the extensor surfaces of large joints which were migratory in nature. Few of them spontaneously subsided following suppuration of chalky white discharges except over the gluteal region. Ophthalmological examination revealed visual acuity of counting fingers (CF) at 1 m in the right eye and perilimbal conjunctival calcific deposits and retinal angiod streaks in both eyes. There was choroidal neovascular membrane with subretinal hemorrhage in right eye, confirmed by fundus fluorescein angiography (FFA) and optical coherence tomography (OCT). B scan ultrasonography and simultaneous vector A scan detected calcification of the subretinal neovascular membrane and the adjoining sclera.

Key words: Angiod streaks, perilimbal calcification, scleral calcification, subretinal neovascular membrane, tumoral calcinosis

Introduction

Tumoral calcinosis (TC) is a rare familial disorder of phosphate metabolism. It is characterized by single or multiple painless, lobular, well-demarcated periarticular soft tissue swelling/s. These are mostly distributed along the extensor surfaces of large joints due to ectopic deposition of calcium.[1] In TC serum phosphate level is elevated with or without increase in level of serum 1,25-dihydroxy vitamin D and usually with normal serum calcium and parathyroid hormone levels.[2] Genetically it is a posttranslational defect where autosomal recessive mutation had been reported to genes GALNT3[3] or fibroblast growth factor 23 (FGF 23).[4] Ocular involvement in TC has been reported, but in Indian literature such report was not found (PubMed search - ocular tumoral calcinosis). We report a rare case of ocular involvement in TC where scleral calcification was its unique feature in addition to other findings.

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Case Report

A 32-year-old male presented with a history of acute and painless blurring of vision in right eye since 1.5 years. He had been suffering from multiple, migratory large joint swellings. All the swellings spontaneously subsided following suppuration of chalky white material and scar formation except over the gluteal region.

Ophthalmological examination revealed best corrected visual acuity (BCVA) of counting fingers (CF) at 30 m in the right eye and 20/20 vision in the left eye. Slit lamp examination of the anterior segment detected perilimbal calcific deposits [Fig 1]. Ocular fundus examination revealed angiod streaks in both eyes and choroidal neovascular membrane with subretinal hemorrhage in the right eye [Fig 2]. Fundus fluorescein angiography (FFA) confirmed the above findings [Fig 3]. Spectral domain optical coherence tomography (OCT) also showed subretinal membrane and fluid in the macula in the right eye [Fig 4]. B scan ultrasonography with simultaneous vector A scan showed elevated macular area with high reflective echo, one each over the subretinal membrane and the sclera. These findings were suggestive of focal calcification of both the structures in the right eye [Fig 5].

Systemic examination revealed scoliosis, shortening, and deformity of the right upper limb and left lower limb. There were scars and multiple discharging cutaneous sinuses over both the gluteal folds [Fig 6]. A multilobulated swelling with shiny overlying skin was present over the right shoulder [Fig 7]. On physical examination, no findings suggestive of pseudoxanthoma elasticum, Ehlers-Danlos Syndrome, scleroderma, or dermatomyositis were found. X-ray pelvis and computed tomography (CT) scan of chest and pelvis

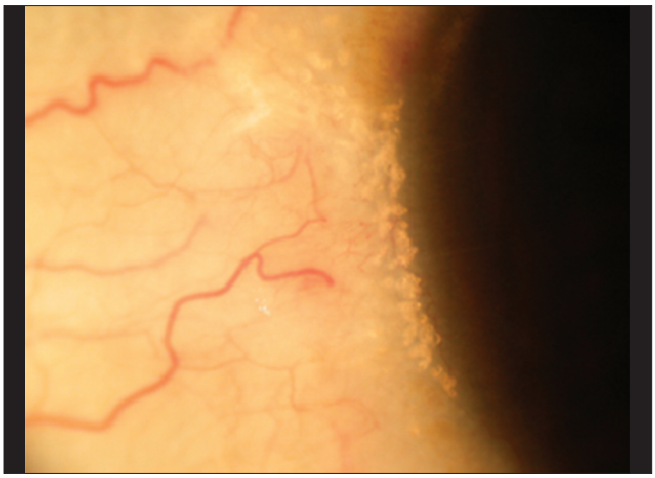

Figure 1: Perilimbal calcifications

Figure 2: Color montage photographs of the ocular fundi. (a) Right fundus showing angiod streaks with subretinal hemorrhage and neovascular membrane. (b) Left fundus showing the angiod streaks

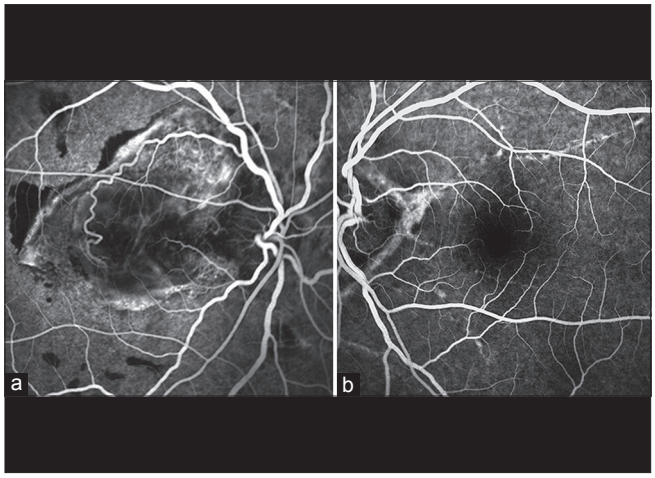

Figure 3: (a and b) Fundus fluorescein angiogram showing angiod streaks in both eyes. Retinal hemorrhage is the cause of blocked fluorescence and hypofluorescence is due to subretinal neovascular membrane in the right eye

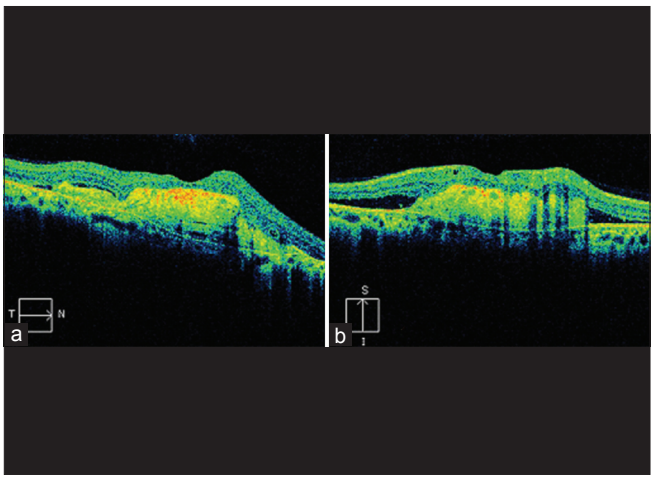

Figure 4: (a and b) Spectral domain optical coherence tomography (OCT) showing a big subretinal membrane and fluid in the macular region

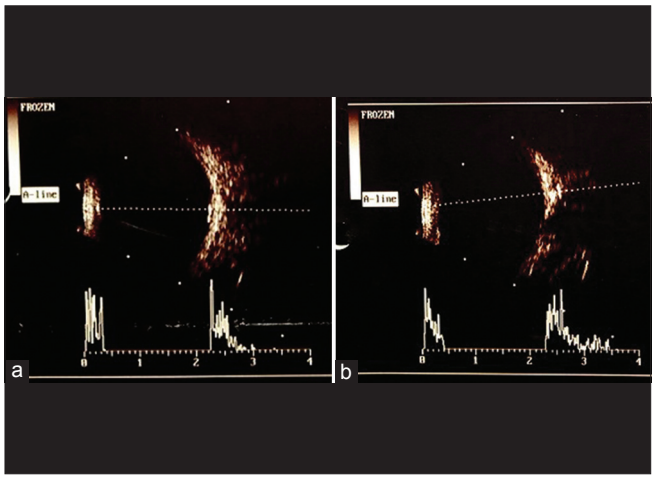

Figure 5: B scan ultrasonography with vector A scan showing elevated macular area with (a) a high reflective echo over the surface and (b) another high reflective echo on the outer surface of the sclera with shadowing affect, suggesting focal calcification in the membrane and the sclera, respectively

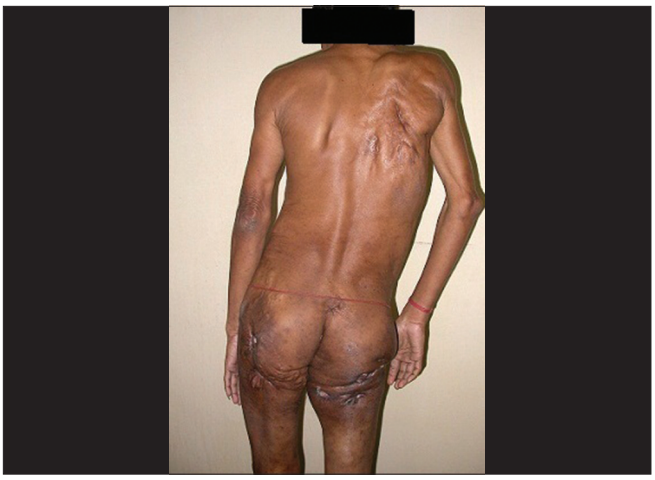

Figure 6: Photograph of the dorsal aspect of the patient showing scars over both the elbow and the hip joints and the right scapula and multiple cutaneous discharging sinuses over the gluteal folds

showed nodular calcifications in periarticular soft tissues, bilateral adrenal glands, and spleen [Figs 8-10].

On laboratory investigations, serum phosphate and serum calcium levels were 4.9 and 7.9 mg/dl, respectively. Renal function was normal.

Based on the clinical features and radiological and laboratory findings, the condition was diagnosed as TC with ocular involvement.

Discussion

TC is a rare systemic disorder. It is also known as Teutschlaender disease.[5] Inclan *et al*., differentiated the condition from dystrophic and metabolic calcifications and coined the term TC.[6] In the present case, the radiological findings were typical of TC. Subject had high serum phosphate levels with normal renal function. However, serum calcium level was low and cystic spaces showing fluid levels, usually seen in TC, were not found in our present case. This was probably because of the healing stage of the disease. The lesions were homogenous without any adjoining osseous destruction suggesting a reduced metabolic activity, thereby excluding neoplasm.

To our best knowledge (PubMed search), this is the first report of TC presenting with ocular manifestations in Indian literature. The case remained undiagnosed till the ophthalmic manifestations occurred. The systemic involvement started at 8 years of age and visual symptoms at 20.5 years of age, which is the usual age of ocular involvement.[7] Except for scleral calcification, all other findings found in the present case like limbal calcific deposits,[7] angiod streaks,[7,8] and subretinal neovascular membrane,[7] have been reported in literature. Angiod streak in the retina may be related to GALNT3 or FGF23 gene mutation. Since sclera is a connective tissue, similar mechanism may also be responsible for calcification in the periarticular as well as scleral connective tissue. The other known causes of sclerochoroidal calcification

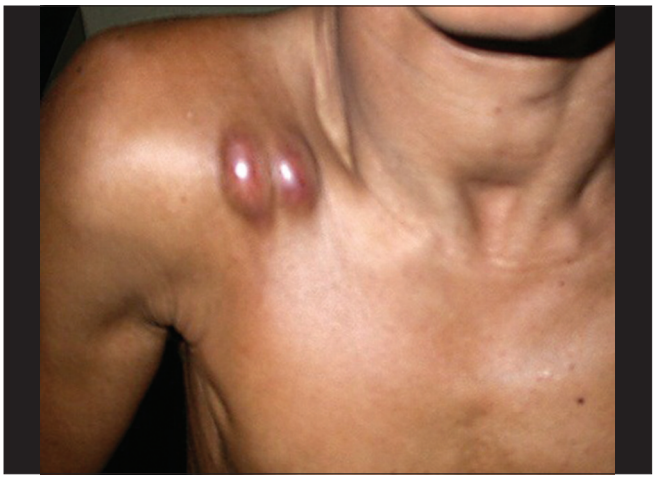

Figure 7: Photograph of the ventral aspect of the right shoulder joint showing a lobulated fresh lesion with shiny skin

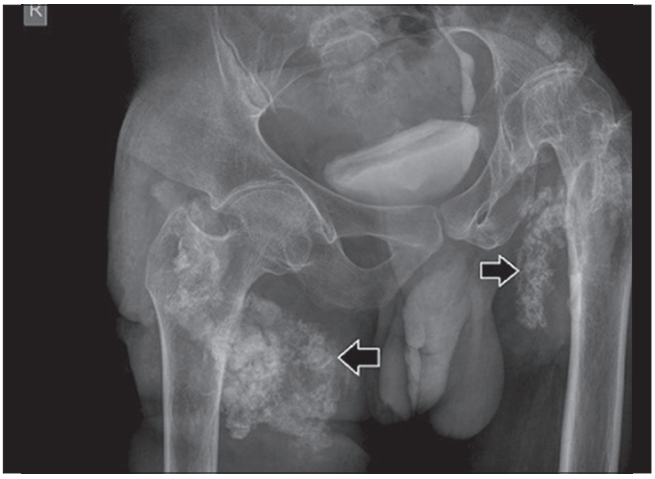

Figure 8: Plain X-ray pelvis showing multilobulated, well-demarcated, densely calcified masses (as indicated by arrows) confined to the soft tissue on the extensor surface of the hip joint and adjoining areas (both gluteal regions)

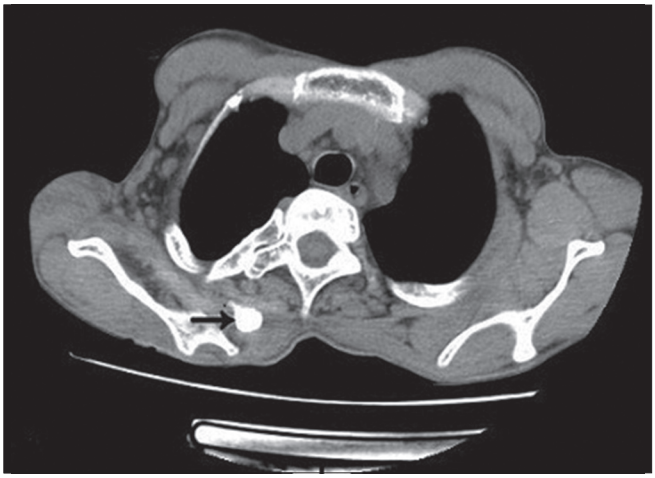

Figure 9: Axial computed tomography (CT) chest showing nodular calcification between chest wall and the scapula (as indicated by the arrow). The lesion is predominantly homogenous suggesting lower metabolic activity

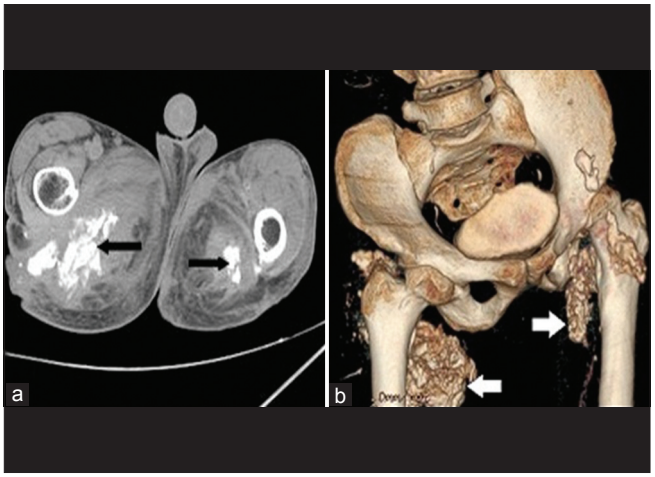

Figure 10: (a) Axial CT pelvis and (b) volume-rendering technique (VRT) image showing extensive nodular calcifications (as indicated by arrows) surrounding the hip joints

like hyperparathyroidism, pseudohypoparathyroidism, and renal tubular acidosis were carefully excluded.

Considering the rarity of the case, difficulty in diagnosis and scleral calcification, which was a new finding, the present case has been reported.

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