

The man who can't run: a case of tumoral calcinosis

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ABSTRACT A 26-year-old white man presented with a several-year history of inability to run, a slowly enlarging lump on the left olecranon, and increasing stiffness at the back of both ankles. Physical examination showed a firm, non-tender, freely mobile mass on the left olecranon. Each Achilles tendon was surrounded by a firm oblong subcutaneous mass extending to the mid calf. X-rays of the left elbow showed a lobulated calcified mass over the olecranon; each Achilles tendon was surrounded by a large, lobulated calcified mass overlying and encasing the tendon. The calcified deposits were surgically removed from the elbow and each Achilles tendon was surgically debulked. The histology was typical of tumoral calcinosis. Primary tumoral calcinosis is an unusual benign condition characterised by the presence of slow-growing calcified periarticular soft tissue masses composed of calcium salts and usually located around the large joints. The exact cause is unknown.

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KEYWORDS Tumoral calcinosis

LIST OF ABBREVIATIONS Anti-nuclear antibody(ANA), double-stranded deoxyribonucleic (dsDNA), erythrocyte sedimentation rate (ESR), infrared (IR), tumoral calcinosis (TC)

DECLARATION OF INTERESTS No conflict of interests declared.

CASE REPORT

A 26-year-old white man presented with a several-year history inability to run, a slowly enlarging lump on the left olecranon and increasing stiffness at the back of both ankles. Aged 10 years, he had presented with stiffness of the fingers and right ankle. There were no abnormal physical signs on examination. He was followed up for six years and synovitis or limitation of range of movements were never observed. During this period, his ESR varied from 4 to 10 mm/hr, rheumatoid factor, ANA and dsDNA antibodies were repeatedly negative. X-rays of the hands, feet and the right ankle were also normal.

He first noticed a lump on the left elbow when he was aged 18. Soon after, he became aware of the increasing stiffness at the back of both heels.

Physical examination shows a firm, non-tender, freely mobile mass on the left olecranon (see Figure 1A). Each Achilles tendon is surrounded by a firm oblong subcutaneous mass extending to the mid-calf (see Figure 1B). X-rays of the left elbow show a lobulated calcified mass over the olecranon (see Figure 2A); each Achilles tendon is surrounded by a large, lobulated calcified mass overlying and encasing the tendon (see Figure 2B). A small lump of calcification is also seen in the right heel.

The calcification deposits were surgically removed from the elbow and each Achilles tendon was surgically debulked. His mobility improved significantly.

Both surgical specimens showed similar histological features. Within the connective and fibrofatty tissues, there were foci of amorphous and/or granular calcified material surrounded by proliferating mono- or multinucleated macrophages with chronic inflammatory cells and admixed with fibroblasts, and osteoclast like giant cells (see Figure 3). In other areas, the calcified material is surrounded by dense fibrous material extending into adjacent tissues.

Analysis of the calcified material in the tissues removed from our patient using IR spectroscopy shows the material to be 99% calcium phosphate as carbonate apatite $[\text{Ca}_{10}(\text{PO}_4\text{CO}_3\text{OH})_6(\text{OH})_2]$. Our patient has no family history of any calcinosis.

DISCUSSION

Tumoral calcinosis is an unusual benign condition characterised by the presence of slow-growing calcified periarticular soft tissue masses composed of calcium salts and usually located around the large joints. It was first defined in 1943 as a specific disease with concomitant elevated serum phosphate but normal



FIGURE 1A shows a firm, non-tender, freely mobile mass on the left olecranon. **FIGURE 1B** shows the right Achilles tendon surrounded by a firm oblong subcutaneous mass extending to the mid calf.

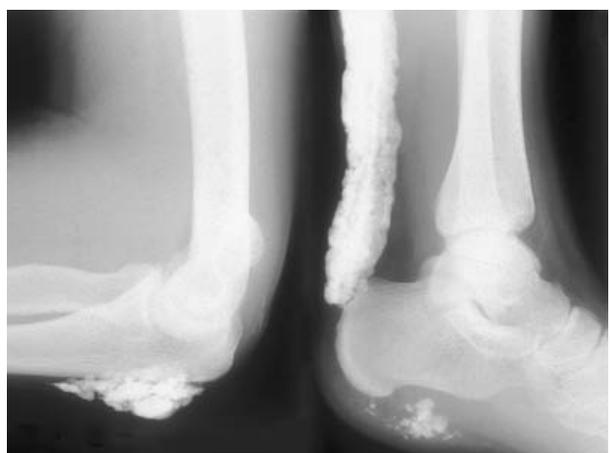


FIGURE 2A is an X-ray of the left elbow showing a lobulated calcified mass over the olecranon. **FIGURE 2B** is an X-ray showing the right Achilles tendon surrounded by a large, lobulated calcified mass overlying and encasing the tendon and a small lump of calcification in the right heel.

serum calcium levels, in the absence of renal, metabolic, or collagen vascular disease.¹ However, the term TC has gradually been used to describe all tumor-like multilobulated periarticular calcific deposits. As a result, there is a more recent pathologically based classification: primary normophosphataemic TC, primary hyperphosphataemic TC and secondary TC.² Primary normophosphataemic TC occurs predominantly in otherwise healthy children and young adults. The lesions are often multiple. There are generally no demonstrable abnormalities in calcium metabolism. Our patient was a white Englishman with no family history and had normal serum calcium, phosphate and calcitriol. He had primary normophosphataemic TC.

Primary normophosphataemic TC is more common in black Africans.³ Less common is primary

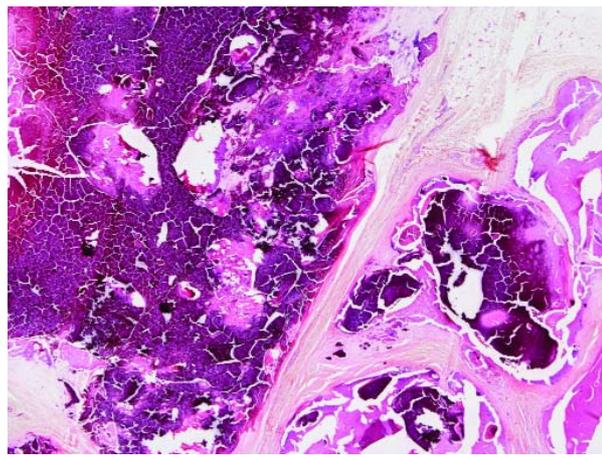


FIGURE 3 Photomicrograph at low power shows amorphous calcified material adjacent to areas of dense fibrosis and chronic inflammatory reaction.

hyperphosphataemic TC which is usually familial (hence the term 'familial TC'). Familial TC is a rare autosomal recessive disorder characterised by hyperphosphataemia due to an increase in proximal tubular phosphate transport, often in association with increased serum calcitriol concentrations.⁴ The underlying defect is a mutation in the *GALNT3* gene, which encodes a glycosyltransferase.⁵ Mutations in fibroblast growth factor *FGF23* gene have also been implicated.^{6,7} It is not clear how these mutations cause hyperphosphataemia. It is postulated that mutations in either the *GALNT3* gene or in *FGF23* each could lead to deficiency of circulating *FGF23*, a promoter of renal phosphate excretion.⁵ Familial TC related to the *GALNT3* gene mutation appears to be the mirror image of X-linked and autosomal dominant hypophosphataemic rickets in which increased activity of phosphatonin appears to cause a primary increase in renal phosphate excretion. Lowering the serum phosphate concentration by dietary restriction and antacids, which impair phosphate absorption, often results in resolution of these deposits.^{4,8} If this is ineffective, increasing urinary phosphate excretion by the chronic administration of acetazolamide may be beneficial.⁹

Calcinosis circumscripta is a similar condition which can also occur in a primary form or be associated with vitamin D intoxication, scleroderma and chronic renal failure but it is usually restricted in distribution to the articular region of the phalanges and the extensor aspects of the knees and the elbows.¹⁰ The calcinosis in the circumscripta variety can achieve tumoral size.¹¹ Calcinosis universalis is the term used to describe extensive calcification of the soft tissues of the thoracic and abdominal walls and usually occurs in patients with connective tissue diseases especially juvenile dermatomyositis.¹²

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A ROLE MODEL OF THE REAL KIND

I was saddened to read in the December issue of the *Journal* of the demise of Professor Gemmel Morgan. My association with him dates back to 1963–64 when I was a house officer in the dermatology department at Dundee Royal Infirmary under the late Dr W Frain-Bell.

Since I was preparing for the MRCP examination, I thoroughly worked-up every inpatient, even though they were admitted only for skin problems. A 70-year-old woman was admitted for eczema of both legs. History revealed that she felt weak, had lost weight and was constipated. I ordered a chest X-ray, which showed a recent rib fracture, but there was no history of trauma or severe cough. Having just read about parathyroid disorders in Charles Dent's *Metabolic Diseases*, I

obtained a urine sample and performed the Sulkowitch test. To my surprise it showed a thick curdy precipitate. I consequently sent her blood sample for calcium estimation with a provisional diagnosis of parathyroid adenoma on the requisition slip.

The next day, as I was having a quick lunch in the hospital canteen, a slightly built man came charging in waving a paper and wanting to know who Dr Yesudian was. Someone pointed me out and he came to my table and said 'Dr Yesudian, you must add a feather to your cap.' When I gave him a puzzled look he showed me the requisition slip I had sent the previous day and my patient's serum calcium was a whopping 19 mg! He then met my chief and had the patient transferred to the surgical ward under Mr Sturrock. Unfortunately, on the day of her operation, I was admitted to the

infectious diseases hospital with chicken-pox. When I returned, I was told that the patient indeed had a large parathyroid adenoma.

I am writing this anecdote to bring out the fine qualities of the man. Firstly, his humility – as the head of the department he could have easily summoned me, a mere house officer, to his department. Secondly, his bubbling enthusiasm shown by his rushing down to the canteen as soon as he saw the result. Thirdly, though he was in a non-clinical department, his care and concern for the patient, in immediately starting the ball rolling for her surgery. It is for these excellent qualities that I remember Professor Gemmel Morgan. Even though four decades have elapsed, I will continue to remember him for the rest of my life.

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