A CASE REPORT: TUMORAL CALCINOSIS IN THE ELBOW REGION

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ABSTRACT

Tumoral calcinosis is a rare condition in clinical practice. We report a clinical case involving an unusually large mass in the elbow region, with postoperative histopathological findings confirming tumoral calcinosis. This case adds to the understanding of the pathogenesis, treatment options, and recurrence rates following surgical intervention.

Keywords: *tumoral calcinosis, soft tissue calcificatio, surgical excision*

I. INTRODUCTION

Author Alberto Inclan introduced the term "tumoral calcinosis" in his 1943 report on three black patients with abnormal calcified tumor growths, for which the cause could not be determined at the time[1]. Large soft tissue tumors present significant challenges in treatment, particularly in determining the nature of the tumor, deciding on the appropriate therapeutic approach, and predicting outcomes for clinicians, especially those new to the field of orthopedic trauma. In this article, we report a clinical case involving an unusually large mass in the elbow region. Postoperative histopathological findings confirmed the diagnosis of tumoral calcinosis. This case aims to contribute to the understanding of the pathogenesis, treatment options, and recurrence rates following surgical intervention.

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II. CLINICAL CASE SUMMARY

Patient N.T.N.D, female, 54 years old, with a history of chronic renal failure undergoing long-term hemodialysis, was admitted on October 7, 2024, due to an abnormal mass in the left arm-elbow region. According to the patient, the mass had been present for over 11 months, initially small but progressively enlarging. The patient sought admission because the mass was affecting her daily activities and aesthetics.

Clinical Examination Findings: A large mass was observed in the left arm-elbow region, measuring approximately 20 cm x 20 cm. The mass was soft, with a smooth surface, poorly mobile, non-tender, and with increased superficial vascularization. No warmth or redness was noted, and the margins were indistinct. Shoulder, wrist, and hand movements were preserved, but elbow movement was limited due to compression from the large mass. The radial pulse on the left side was palpable.

Blood Biochemistry Results: Creatinine: 589.2 µmol/L and eGFR: 6.9 ml/min

Ultrasound Findings: A large soft-tissue mass under the skin of the left arm with multiple internal septations and minimal vascularity. The mass was partially cystic with dense calcifications. The left upper extremity arteries showed scattered small atherosclerotic plaques without narrowing and normal flow patterns.

X-ray Findings: The mass showed a mixed-density lesion involving the soft tissues of the left elbow, with possible bone involvement (Figure 1). CT Scan Findings: the CT scan revealed a mass in the posteromedial region at the junction of the distal third of the arm and the elbow, evaluated using RadiAnt DICOM Viewer software (https://www.radiantviewer.com) (Figure 2). The mass was multiloculated with calcifications. Pathological Analysis: Fine-needle aspiration suggested a diagnosis of a brown tumor (Figure 3).

We explained to the patient that surgery would be performed to remove the tumor as completely as possible, and the obtained specimen would be sent for histopathological analysis. Postoperatively, the wound showed no signs of inflammation or fistula formation. The histopathological results concluded that the tumor was calcified (Figure 4).

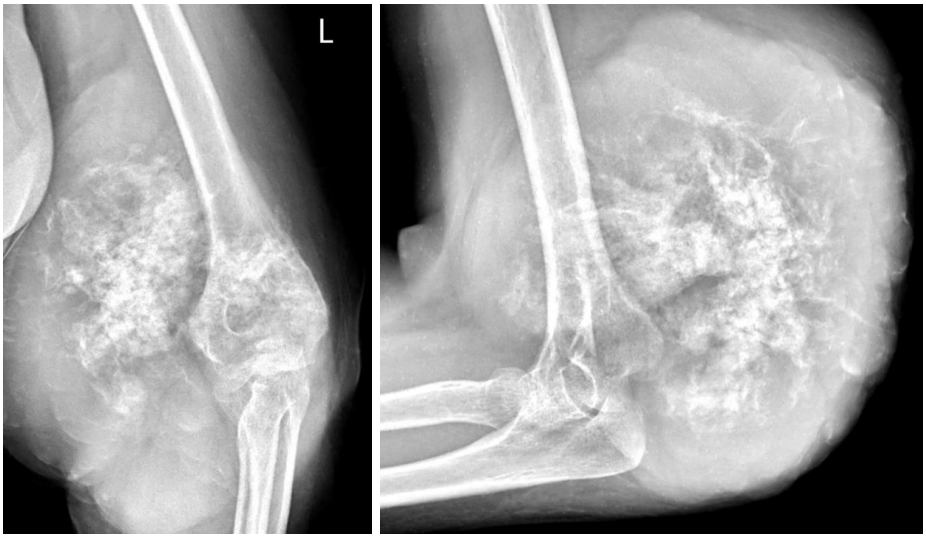

Figure 5: X-ray of the patient's left elbow joint

(Source: Author's research materials)

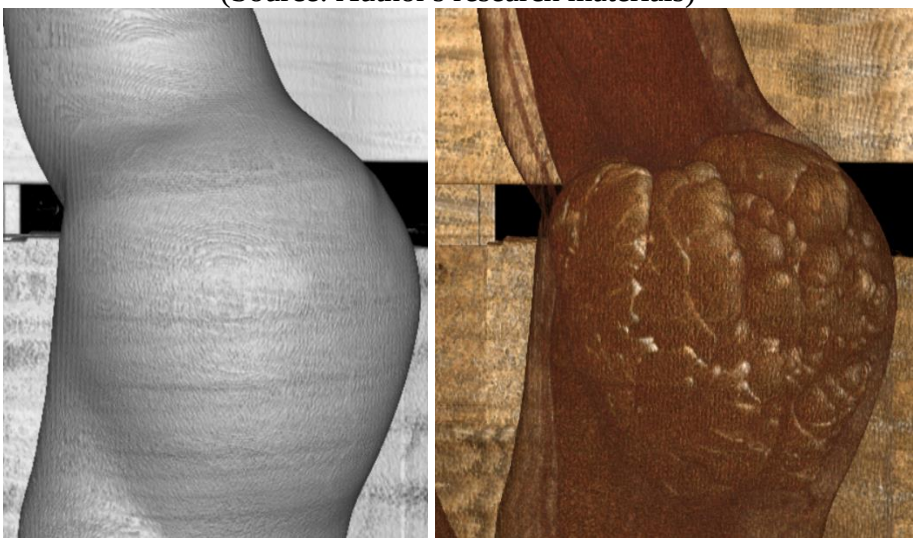

Figure 6: A computed tomography scan reconstructs the tumor in three-dimensional space

(Source: Author's research materials)

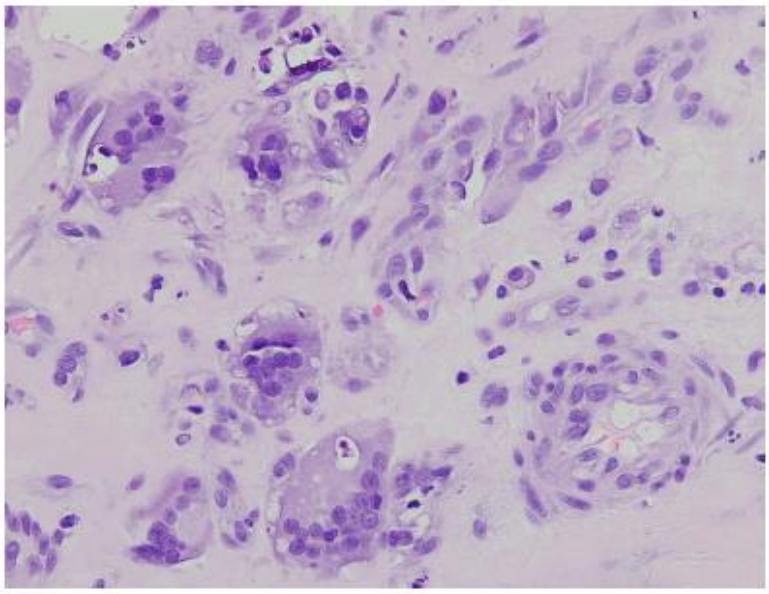

Figure 7: Histopathological results from pre-surgical fine-needle aspiration of the tumor
(Source: Author's research materials)

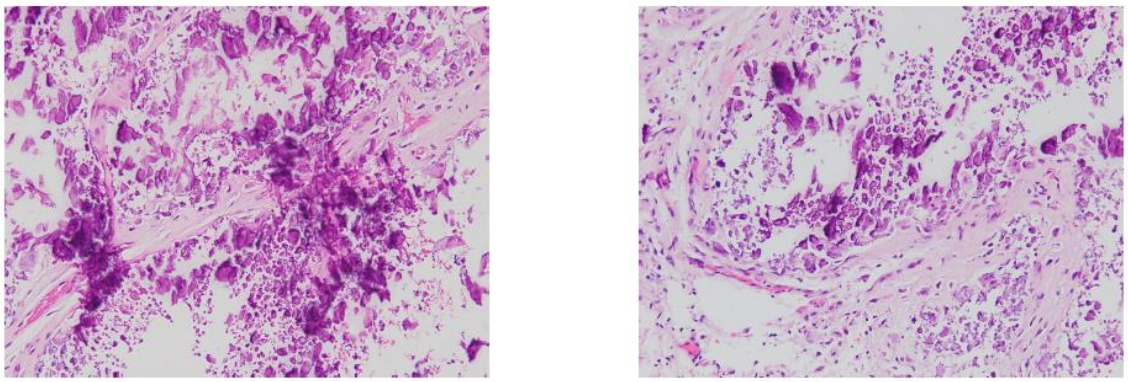

Figure 8: Histopathological results of the tumor following surgical excision
(Source: Author's research materials)

III. DISCUSSION

3.1. Causes of Pathogenesis and Common Locations of the Tumor

Tumoral calcinosis is an uncommon benign disorder marked by the deposition of calcium phosphate crystals within the soft tissues surrounding major joints.

While the precise mechanisms underlying its development are not fully understood, Smack et al. [2] introduced a classification system in 1996, following their analysis of 121 cases, to categorize the condition based on its pathogenesis:

(1) Primary tumoral calcinosis without hyperphosphatemia (normophosphatemic). This form of tumoral calcinosis is defined by normal serum calcium and phosphate levels, without any history of conditions linked to soft tissue calcification. It most commonly occurs in the hip (31%), elbow (24%), and knee (16%).

According to Smack, complete removal of calcified tumor tissue significantly reduced the likelihood of recurrence. When full excision was not possible, a combination of partial removal and dietary management—featuring oral aluminum hydroxide and a diet low in calcium and phosphate—proved to be a beneficial approach..

Recent studies have provided evidence that this condition is associated with mutations in the gene encoding the SAMD9 protein [3],[4], this protein is involved in controlling cell proliferation and functions as a tumor suppressor in specific types of cancer.

(2) Primary tumoral calcinosis with hyperphosphatemia (hyperphosphatemic). This form of tumoral calcinosis is distinguished by normal serum calcium levels alongside increased serum phosphate, calcitriol concentrations, and enhanced renal phosphate reabsorption. There is no history of conditions linked to soft tissue calcification. It commonly affects areas such as the hip (37%), elbow (26%), and shoulder (23%).

There are reports linking primary tumoral calcinosis with hyperphosphatemia to mutations in various genes: GALNT3, FGF23, and α-Klotho. The GALNT3 gene codes for the glycosyltransferase enzyme UDP-N-acetyl-α-D-galactosamine: polypeptide N-acetylgalactosaminyl transferase 3 (ppGalNacT3), which plays a crucial role in initiating the O-glycosylation of FGF23 [3], Mutations in GALNT3 disrupt this glycosylation process, making FGF23 more vulnerable to enzymatic cleavage [5] This increased susceptibility leads to its degradation, resulting in lower levels of active FGF23 in the bloodstream. Or, inactivating mutations in the KL gene, which encodes α-Klotho, can occur. α-Klotho is an essential co-receptor for the function of the FGF23 receptor in phosphate regulation. [5]. Mutations in the FGF23 gene itself can also occur. These mutations lead to the production of either inactive FGF23 or truncated, non-functional FGF23 proteins.

The FGF23 gene, located on chromosome 12p13.3, encodes a protein composed of 251 amino acids [5], This protein is crucial in regulating 1,25-dihydroxyvitamin D metabolism, promoting phosphate excretion by the kidneys, and lowering serum phosphate levels. When the secretion of intact FGF23 is disrupted, phosphate reabsorption in the kidneys continues unabated, and 1,25-dihydroxyvitamin D production is not inhibited, even in the presence of hyperphosphatemia. This imbalance can increase calcium and phosphate absorption in the intestines, fostering a biochemical environment that promotes the deposition of calcium-phosphate crystals in soft tissues and blood vessels.

In this classification, author Smack noted that both complete and partial surgical excision in cases where complete removal of calcifications is not possible are ineffective, with an 84% recurrence rate. Slower tumor growth following dietary changes and the use of aluminum hydroxide supplementation was also observed and documented by the author..

(3) Secondary tumoral calcinosis. This type of calcification is characterized by detectable abnormalities in laboratory tests, depending on the underlying condition, such as hypercalcemia. Conditions frequently associated with this disorder, listed in descending order of prevalence, include chronic kidney disease, pseudoxanthoma

elasticum, granulomatous conditions, primary hyperparathyroidism, scleroderma, vitamin D toxicity, milk-alkali syndrome, and significant bone resorption.

In cases of secondary tumoral calcinosis, author Smack observed that both complete and partial surgical excision yielded poor outcomes, with a recurrence rate of 79%. Dietary changes provided little benefit. In some cases, effectively treating the underlying conditions promoting calcification could lead to successful outcomes (e.g., treating hyperparathyroidism resulting in the resolution of calcifications)..

In individuals with end-stage renal disease, insufficient glomerular filtration leads to reduced phosphate excretion and impaired vitamin D activation, resulting in secondary hyperparathyroidism. This condition promotes calcium release from bones, known as renal osteodystrophy, leading to increased calcium-phosphate product levels and their precipitation in soft tissues [6].

3.2. Tumoral Calcinosis Imaging

Diagnostic tests commonly employed include imaging studies and histopathology. Conventional X-rays in cases of tumoral calcinosis typically reveal amorphous, lobulated, cystic calcifications located around the joints. A notable feature of this condition is the absence of bone destruction or infiltration into the surrounding soft tissues. Another characteristic finding is the "sedimentation sign," where mineral deposits settle, forming a calcium-fluid level [7]. Computed tomography scans are valuable for assessing the lesion's size, its proximity to surrounding structures, and for aiding in surgical planning, as illustrated in Figure 2. Histologically, tumoral calcinosis is defined by irregular calcified deposits encircled by multinucleated giant cells and inflammatory cells, resembling a granulomatous reaction. [8].

3.3. Surgical Intervention

Currently, surgery is often considered in cases where large calcified masses cause pain, restricted movement, nerve compression, or other complications [6], surgical removal of the calcified masses can provide pain relief and improve the patient's motor function. However, the risk of tumor recurrence following surgery is also commonly observed [9].

3.4. Non-Surgical Treatment

The primary goal of this approach is to correct metabolic disorders that promote calcification, manage pain, and reduce inflammation. Several reported solutions have been found effective, including:

Dietary phosphate restriction: Reducing dietary phosphate intake is a crucial first step in managing tumoral calcinosis associated with hyperphosphatemia. Patients are advised to follow a low-phosphate diet, limiting high-phosphate foods such as dairy products, nuts, and meat. Nutritional counseling can assist patients in creating balanced meal plans that ensure adequate intake of essential nutrients [10].

Use of phosphate binders: Medications like sevelamer carbonate, sevelamer hydrochloride, lanthanum carbonate, and aluminum hydroxide act as phosphate binders in the gastrointestinal tract, reducing phosphate absorption. These drugs are usually administered with meals and can effectively lower serum phosphate levels. However, they may lead to gastrointestinal side effects, including constipation, nausea, and abdominal discomfort [10].

3.5. Limitations and Lessons Learned

In this case, based on the knowledge acquired from medical literature, we identified the following: The pathogenesis was identified as secondary tumoral calcinosis. There were limitations in the initial preoperative assessments, such as the absence of serum calcium and phosphate measurements and parathyroid hormone assays, due to a lack of experience with this condition. Surgical intervention is indicated when the tumor causes symptoms such as deformity, nerve pain, ulceration, or impaired joint function, making the decision to operate in this case appropriate. However, the cornerstone of treatment is maintaining serum phosphate levels within the normal range..

IV. CONCLUSION

Tumoral calcinosis is an uncommon condition, and addressing its root cause is essential for effective symptom control and preventing recurrence. This case highlights the clinical, imaging, and pathological features of a challenging soft tissue mass in a patient with a history of chronic kidney disease, contributing to the understanding and management of similar cases.

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