

Recurrent idiopathic tumoral calcinosis in a child postoperatively

Garrett S. Booth, MS; Rukmalee E. Vithana, MD; Daphne E. DeMello, MD; Gerald A. Mandell, MD

Tumoral calcinosis is an uncommon disease characterized by the deposition of calcium salts and crystals in the periarticular soft tissues. It is almost entirely a disease of adults. Histological and radiologically, however, features of this condition are identical regardless of age. Lesions in adults usually involve the hip joint and tend to recur following surgery, whereas in children surgery is often curative. We report a case of recurrent tumoral calcinosis of the sternum of a Hispanic identical-twin female infant.

Introduction

First described by Duret (1) in 1899, tumoral calcinosis is a rare disease characterized by a progressively enlarging mass of amorphous calcium salts and calcium hydroxyapatite crystals in periarticular soft tissues, especially in the hips, elbows, and extensor surfaces. Lesions tend to be firmly attached to the underlying fascia, muscle, or tendon and may have a predilection for infiltrating surrounding structures. Although this condition has been described in patients of all ages, it is more commonly observed in the second decade of life.

Several variants of tumoral calcinosis have been described. Patients receiving extensive renal dialysis are at an increased risk of disordered calcium homeostasis and may subsequently develop tumoral calcinosis in multiple sites (2). The second subtype of tumoral calcinosis is due to a rare autosomal recessive disorder caused by a mutation in the *GALNT3* gene, which encodes glycosyltransferase (3). Genetically linked tumoral calcinosis is thought to be characterized by elevated serum phosphate levels, due to an increase in proximal tubular phosphate transport, often co-

occurring with increased serum calcitriol levels (4). Unlike patients on chronic renal dialysis, those with familial variants tend to have normal serum calcium and PTH concentrations. Familial tumoral calcinosis is generally found in patients of African or African-American descent. The final variant of tumoral calcinosis is idiopathic. These sporadic variants have no known phosphorus or calcium irregularity, and surgical excision is often curative.

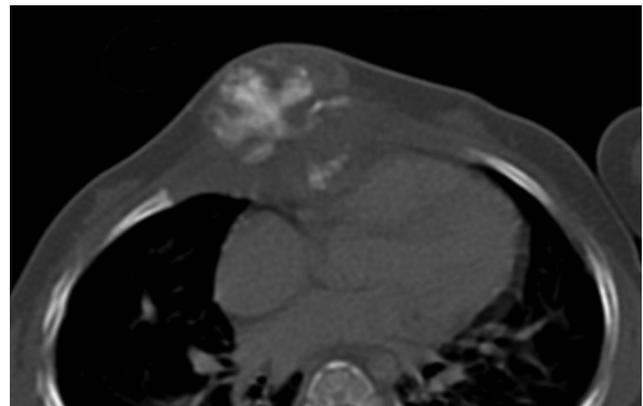


Figure 1. 34-month-old female with tumoral calcinosis. Representative CT slice through the mass showing collections of calcifications separated by radiolucent bands.

Tumoral calcinosis is typically a disorder of adulthood; children are rarely affected. Infants with tumoral calcinosis are extremely uncommon, having been described only in a handful of cases (5).

Citation: Booth GS, Vithana RE, DeMello DE, Mandell GA. Recurrent idiopathic tumoral calcinosis in a child postoperatively. *Radiology Case Reports*. (Online) 2012;7:735.

Copyright: © 2012 The Authors. This is an open-access article distributed under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs 2.5 License, which permits reproduction and distribution, provided the original work is properly cited. Commercial use and derivative works are not permitted.

Mr. Booth is a student at the University of Arizona College of Medicine, Tucson AZ. Dr. Vithana is affiliated with the Children's National Medical Center, Washington, DC. Dr. DeMello is a pediatric pathologist, and Dr. Mandell is a radiologist, both at Phoenix Children's Hospital, Phoenix AZ. Contact Mr. Booth at gbooth@email.arizona.edu.

Competing Interests: The authors have declared that no competing interests exist.

DOI: 10.2484/rcr.v7i4.735

Recurrent idiopathic tumoral calcinosis in a child postoperatively

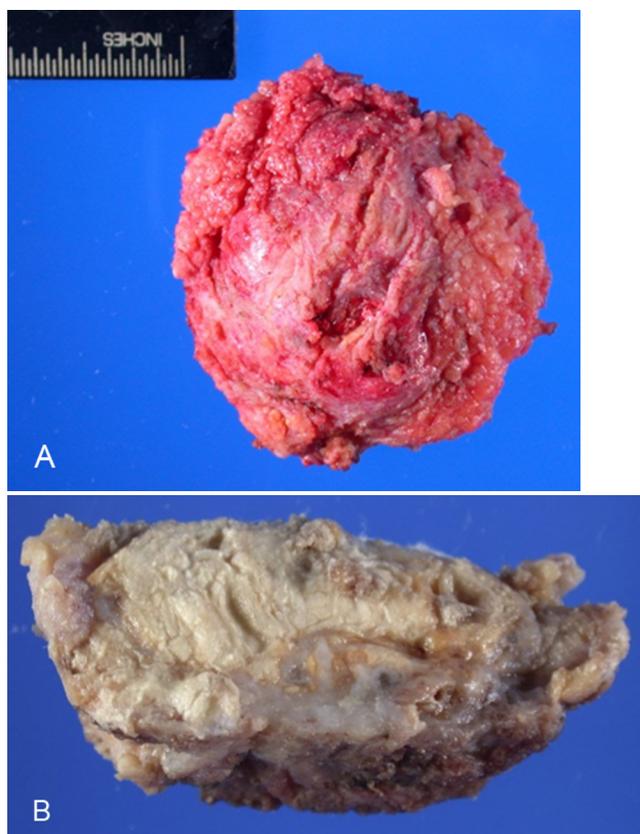


Figure 2. 34-month-old female with tumoral calcinosis. Resected specimen (A) and cross-section (B) showing chalky-white foci of calcification surrounded by fibrous tissue.

Case report

A 34-month-old Hispanic monozygotic twin-female patient, who was born 900 grams as twin A following a premature gestation of 26 weeks, presented with a history of recurrent sternal mass. In utero, an amniotic band was discovered, restricting the development of the patient's left hand. Subsequently, at birth the patient was diagnosed with a left-terminal-matrix hemorrhage and has since experienced developmental delay. At one month of age, the patient was bacteremic and underwent open-heart surgery via median sternotomy to remove an infected thrombus from her right atrium, as well as ligation of her ductus arteriosus. In addition, she was diagnosed with chronic lung disease of prematurity and retinopathy of prematurity. The twin sibling is without any medical issues or developmental delay. In addition, there is no known family history of tumoral calcinosis.

In followup, by 17 months of age the patient had developed an anterior chest-wall mass measuring up to 5 cm in the transverse dimension, 3.5 cm in the anterior-posterior dimension, and approximately 6.5 cm in the cranial-caudal dimension. At the time of her initial surgery, imaging studies demonstrated a mass appearing to be heavily calcified and consisting of small loculated compartments. Following

radical resection, the initial pathology report confirmed a nonmitotic lesion with dense fibrosis and dystrophic calcification with numerous giant cells.

By the age of 30 months, the patient's mother had begun to notice a recurrence of an enlarging sternal mass. Anterior-posterior and lateral chest radiographs four months later demonstrated a large cauliflower-like ossification on the anterior chest wall, extending anteriorly as well as retrosternally. Followup CT (Fig. 1) confirmed an anterior-chest-wall mass measuring 6.9 cm in the cephalad-to-caudad extent and 4.8 cm in the transverse diameter; the more anterior component measured 3.9 cm and the retrosternal component 1.8 cm in the anterior-posterior diameter. Surgical resection was again performed, excising the mass and portions of the sternum (Fig. 2). The specimen was a hard spherical mass measuring 4 cm in diameter (Fig. 3). The cut surface had scattered, chalky white foci of calcification. Microscopically, it was composed of dense fibrocollagenous tissue with extensive areas of calcification. The patient's lab values were serum phosphorus 4.3 mg/dL, vitamin D 25 ng/mL, and serum calcium 9.5 mg/dL, all within normal limits.

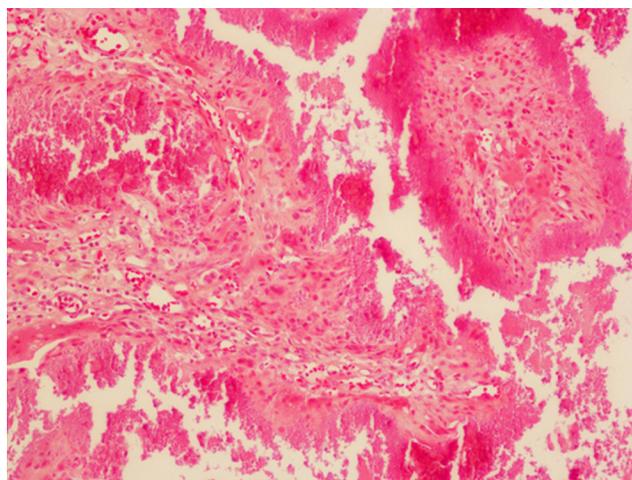


Figure 3. 34-month-old female with tumoral calcinosis. Microscopic section of the mass shows foci of calcification (granular pink material) surrounded by multinucleated foreign-body giant cells and a chronic inflammatory infiltrate. Mag x 100 H&E.

Discussion

Isolated sporadic variants of tumoral calcinosis present a diagnostic challenge to clinicians, having been reported in newborns to 88-year-olds (6). Without a family history, known genetic mutation, or identifiable risk factor like renal dialysis, often the first suspicion for tumoral calcinosis arises in the radiographic interpretation (7). Radiographically, tumoral calcinosis is described as well-demarcated, rounded collections of calcific material grouped in rounded or oval grape-like clusters near articulations. Radiolucent

Recurrent idiopathic tumoral calcinosis in a child postoperatively

fibrotic bands impart a “chicken-wire” appearance by separating the individual cystic collections.

Imaging studies cannot make the ultimate diagnosis, however, because isolated small calcified lesions may be caused by a variety of disorders: milk-alkali syndrome, hyperparathyroidism, hypervitaminosis D, chronic renal disease, osteochondroma, or chondrosarcomas. Often some of these conditions can be tentatively ruled out through laboratory analysis, but careful exclusion of malignant or tumor-like conditions requires a biopsy.

Macroscopically, specimens are firm to rubbery in consistency and lack a true capsule; this often allows the lesion to extend into adjacent structures. Microscopically, lesions reveal amorphous calcified material bordered by an elaborate proliferation of multinucleated giant cells and macrophages. Closer examination with electron microscopy demonstrates macrophages with needle-shaped hydroxyapatite crystals aggregated with a dense core (8).

The single largest difficulty in making the diagnosis of tumoral calcinosis in infants without a predisposing condition is the paucity of cases (9). Adult tumoral calcinosis is most commonly identified in patients with comorbid medical conditions, making the likelihood of recurrence higher in older populations. Continued surveillance and research into the underlying differences between adult and infantile forms of tumoral calcinosis may help to yield not only a greater understanding of human biology but also to clarify treatment and prognostic variables.

References

1. Duret, M.H.: Tumeurs multiples et singulières des bourses seruses. *Bull. Soc. Anat. Paris*, 74:725, 1899.
2. McGregor DH, Mowry M, Cherian R, McAnaw M, Poole E: Non-familial tumoral calcinosis associated with chronic renal failure and secondary hyperparathyroidism: Report of two cases with clinicopathological, immunohistochemical, and electron microscopic findings. *Human Pathology*, 26:607-613, 1995. [[PubMed](#)]
3. Topaz O, Shurman DL, Bergman R, et al. Mutations in GALNT3, encoding a protein involved in O-linked glycosylation, cause familial tumoral calcinosis. *Nature Genetics*, 36:579, 2004. [[PubMed](#)]
4. Steinherz R, Chesney RW, Einstein B, Metzker A, DeLuca HF, Phelps M.: Elevated serum calcitriol concentrations do not fall in response to hyperphosphatemia in familial tumoral calcinosis. *American Journal of Children*, 139:816-819, 1985. [[PubMed](#)]
5. Hammound S, McCarthy EF, Weber K.: Tumoral calcinosis in infants. *Clinical Orthopaedics and Related Research*, 436: 261-264, 2005. [[PubMed](#)]
6. Aprin H, Sinha A: Tumoral calcinosis: Report of a case in a one-year old child. *Clinical Orthopaedics*, 185:83-86, 1984. [[PubMed](#)]
7. Steinbach LS, Johnston JO, Tepper EF, Honda GD, Martel W: Tumoral calcinosis: Radiologic-pathologic correlation. *Skeletal Radiology*, 24:573-578, 1995. [[PubMed](#)]
8. Balogh K, Ferencz T, Csikos A, Timar J: Tumoral calcinosis in infancy: A light and electron microscopic study with X-ray microanalysis. *Pathology and Oncology Research*, 1:80-84, 1995. [[PubMed](#)]
9. Bostrom B: Tumoral calcinosis in an infant. *American Journal of Diseases in Children*, 135:246-247, 1981. [[PubMed](#)]