# Soft Tissue Aneurysmal Bone Cyst in the Sartorius Muscle of a 13-Year-Old Boy Mimicking Myositis Ossificans: Case Report

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## Abstract

**Introduction:** A soft tissue aneurysmal bone cyst is an extremely rare tumor. The objective of the article is to present the clinical, radiological, and histopathological features of a very unusual neoplasm of soft tissues. **Case report:** A 13-year-old male patient presented a painful, mobile, and rapidly growing mass on the posteromedial aspect of his left knee. Imaging studies revealed a mass that arose from the medial surface of the distal sartorius muscle, with extension to the subcutaneous fat tissue. It was a well-circumscribed solid tumor with a peripheral rim calcification on plain film, computerized tomography, and ultrasound (zonal phenomenon). On magnetic resonance imaging, a heterogenous mass on T1-weighted images (WI) and T2-WI was seen, with a peripheral hypointense rim in both sequences. An outstanding edema on T2-WI extending to the soft tissue and muscles of the medial compartment of the knee was detected. The mass was resected, and the "tumoral mimickers" histopathological and molecular (next-generation sequencing) diagnoses confirmed a soft tissue aneurysmal bone cyst. A follow-up showed that the patient was free of disease 12 months after surgery. **Conclusion:** Soft tissue aneurysmal bone cyst is a rare tumor. Appropriate clinical and radiological correlation should be performed to differentiate it from other tumor mimickers.

#### Keywords

aneurysmal bone cyst, soft tissue aneurysmal bone cyst, myositis ossificans

## Introduction

A soft tissue aneurysmal bone cyst is an extremely rare osteoclastic giant cell-rich benign tumor that was first described by Salm and Sissons.<sup>1</sup> It is histologically indistinguishable from its bone counterpart (aneurysmal bone cyst).<sup>2</sup> By definition, soft tissue aneurysmal bone cyst has no attachment to the adjacent bone surface.<sup>3</sup> Only 39 soft tissue aneurysmal bone cyst cases have been reported to date in English literature.<sup>4–7</sup> Secondary cystic changes in benign and malignant tumors of bones resembling aneurysmal bone cyst are known as secondary aneurysmal bone cyst.<sup>8</sup>

The radiological appearance of soft tissue aneurysmal bone cyst consisted of multicameral appearances, with multiple septations with low signal on T1 and T2 that demarcate spaces that display fluid–fluid levels and demonstrate enhancement after the administration of contrast, resulting in a "honeycomb appearance".<sup>9–12</sup>

Histologically, an aneurysmal bone cyst is a wellcircumscribed multilocular tumor composed of blood-filled cystic spaces without endothelial lining and separated by thin septa.<sup>2</sup> The fibrous septa contain a dense proliferation of fibroblasts, scattered multinucleated osteoclast-type giant cells, and strands of woven bone rimmed by osteoblasts.<sup>2</sup> No evidence of cytologic atypia is seen in an aneurysmal bone cyst.<sup>2</sup> Most

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primary aneurysmal bone cysts present t(16; 17)(q22; p13) translocation involving ubiquitin-specific protease 6 (USP6).<sup>13</sup>

The objective of this article is to present clinical, radiological, and histopathological features, and surgical management of this very rare tumor of soft tissues.

# **Case Report**

## **Clinical Findings**

A 13-year-old boy was referred to our hospital with a painful enlarging mass on his left knee, which had developed over a month. The patient denied recent local trauma but was an active social soccer player and had suffered repeat trauma to his limbs over time. He could not remember any trauma related to the appearance of the mass. No personal or family medical antecedents were relevant. A physical examination revealed a painful mobile mass on his left knee with a full range of motion (Figure 1).

## Radiological and Pathological Findings

Plain film, computerized tomography (CT), and magnetic resonance imaging (MRI) were performed in his original medical center due to a soft-tissue sarcoma suspicion.

The initial radiographs (Figure 2A and B) and CT (Figure 2C) identified a  $3 \times 3.5$  cm mass in the medial aspect of the knee, with coarse calcifications and a central hypodense area. The lesion did not contact any bone structure. An MRI evaluation (Figure 2D-F) showed a high-signal periphery due to edema, an intermediate zone of low signal that corresponded to calcifications, and a central area of low signal that appeared to be slightly hyperintense in T1 sequences, which could be attributable



Figure 1. (A and B) Soft tissue mass on the medial aspect of the left knee.

to areas of fat or bleeding. Myositis ossificans, synovial sarcoma, soft-tissue osteosarcoma, and a solid aneurysmal bone cyst were included in the differential diagnosis.

In our "high-resolution" hospital team approach, bone scintigraphy was performed as a staging imaging protocol in bone-forming tumors. Bone scintigraphy images after the intravenous administration of 99mTc-HMDP showed the presence of a hypervascularized area on the vascular phase (Figure 2G) and on the delayed phase (Figure 2H) on the inner side of the left knee, with an increase in osteogenic activity with high uptake levels of 99mTc in the tumor. In the SPECT-CT study, these changes corresponded to a rounded lesion with calcified edges (Figure 2I and J).

After analyzing the image studies carried out by the patient and the ones performed in our hospital, radiologists were confident with a myositis ossificans diagnosis in the sartorius muscle. A fine needle aspiration was made to avoid general sedation and to exclude malignancy. With the first results from cytological exams, which proved nonmalignancy and cellularity in accordance with the myositis ossificans suspicion, a "wait and see" surveillance technique was proposed.

The lesion continued growing and after 3 months the mass showed a thin peripheral mineralized bone shell, and a central solid component with calcified thin septations and hypodense areas, which suggested blood-filled cystic spaces (Figure 2K and L). An evolution from the zonal phenomenon to a bubble soap appearance was very uncommon. The only lesion that fulfilled all the imaging criteria was probably a soft tissue aneurysmal bone cyst.

A core needle biopsy was carried out to exclude alternative diagnoses of a solid soft tissue aneurysmal bone cyst, soft-tissue osteosarcoma, and synovial sarcoma, and to confirm an atypical evolution of myositis ossificans. The CT-guided biopsy showed a fibroblastic cell neoplasm without cytological atypia in a myxoid stroma compatible with myositis ossificans (Figure 3A).

During the following 4 weeks, the mass continued growing and the pain increased. After the case was presented to the multidisciplinary tumor committee and due to the clinical–radiological–pathological discordance, a marginal tumor resection of the mass with the surrounding soft tissues was indicated.

The surgical specimen was grossly conformed by an ovoid well-circumscribed mass that measured  $5.5 \times 4 \times 3.5$  cms. Upon sectioning, it was predominantly solid in the center, with patchy blood-filled cystic spaces mainly at the periphery (Figure 3B).

Histologically, the main pattern was a storiform bland spindle cell proliferation without cytological atypia embedded in a fibromyxoid stroma with scattered multinucleated, osteoclast-type giant cells, and woven bone rimmed by osteoblast (Figure 3C). The woven bone was arranged parallel to the cystic cavities (Figure 3C). This bone was focally spiculated, basophilic, and with a chondroid-like appearance ("blue bone") (Figure 3D and E). No mitotic figures were observed. Peripherally blood-filled cysts and septa were observed



**Figure 2.** (A and B) Conventional radiography: Posteroanterior and lateral radiographs demonstrated a calcified well-circumscribed solitary lesion, with peripheral rim appearance in the lateral view. (C) Axial images from noncontrast computerized tomography (CT; soft tissue window): a mass that arose in the sartorius muscle (arrow), peripheral calcification pattern, and central calcium with lower density. (D) Noncontrast T2-weighted fat-suppressed coronal image: the lesion presented a central high signal with tiny hypointense septa, and a hypointense periphery with a radiate pattern that corresponded to calcifications. An extensive peripheral edema pattern was also seen. (E) Noncontrast T1-weighted coronal image: the lesion showed patchy areas of slight hyperintensity that did not cancel in fat-suppressed sequences, which could be attributable to foci of bleeding. (F) Contrast T1-weighted fat-suppressed axial image: after contrast administration, the lesion showed a zonal phenomenon with central and peripheral enhancement. Enhancement was also seen in the soft tissue around the lesion, probably associated with an inflammatory/edema response. (G) Scintigraphy (vascular phase) and (H) Scintigraphy (delayed phase): the lesion showed a hypervascularized area. (I) SPECT-CT axial image and (J) 3D SPECT-CT: rounded lesion with calcified edges. (K) Posteroanterior radiography and (L) noncontrast axial CT 3 months later (post-biopsy): it showed growth, a thin peripheral bone shell, and a central component with calcified thin septations and areas of hypodensity with a level, which some fluid–fluid levels (arrow).



**Figure 3.** (A) Computerized tomography (CT)-guided biopsy: fibroblastic cell neoplasm with myxoid stroma (200×, H&E). (B) Middle section of surgical specimen: predominantly solid mass with patchy blood-filled cystic spaces. (C) Surgical specimen: storiform bland spindle cell proliferation without cytological atypia embedded in a fibromyxoid stroma with scattered multinucleated osteoclasts and woven bone rimmed by osteoblast (100×, H&E). (D) Spiculated blue bone (200×, H&E). (E) Spiculated, blue bone (400×, H&E). (F) Peripherally blood-filled cysts and thin septa with several giant multinucleated cells (200×, H&E).

(Figure 3F). The border of the tumor was well-circumscribed and composed by an eggshell layer of lamellar bone.

Next-generation sequencing showed a *COL1A1::USP6* gene fusion with chr17:48278772 (exon 1-*COL1A1*) and chr17:5033231 (exon 1-*USP6*) as chromosome coordinates and transcript ID, Genbank: NM\_000088.3 (*COL1A1*) and NM\_004505.3 (*USP6*). Genomic RNA was extracted and purified from a paraffin-embedded

tissue sample. Two panels of genes were studied by amplicon-based enrichment (Archer FusionPlex Sarcoma Panel v2) and capture enrichment (Custom solid tumor solution, Sophia Genetics) with subsequent sequencing (MiSeq, Illumina). The alignment was done against the reference genome GRCh37/hg19. The bioinformatic analysis was performed with the software and analysis algorithms developed by Sophia Genetics (Sophia DDM) as well as

Table 1. Mai	n Characterist	ics of the Repor	rted Cases of S	TABC in 13 Years	-Old Patients or	Younger.			
	Petrik et al. (1993)	Dalet al. (2000)	Nielsen et al. (2002)	Ajilogba et al. (2005)	Ellison et al. (2007)	Sahu et al. (2008)	Hao et al. (2012)	Olshinka et al. (2020)	Present case (2023)
Age Sex	7 Male	8 Male	8 Male	12 Female	10 Female	12 Female	10 Female	12 Female	13 Male
Site	Common carotid artery	Shoulder region	Shoulder region	Thigh	Thigh	Palm	Shoulder region	Knee (intra-articular)	Knee (sartorius muscle)
Size (cm)	З	Not mentioned	8	7	8.5	6	8.8	2.8	3.5
Conventional radiography	Not mentioned	Not mentioned	Not specified	Normal	Peripheral shell of bone	Lytic lesion with sharply circumscribed bony outline	Round soft tissue lesion with a well-defined calcification margin	Peripheral Calcification > ossified mass with well-defined sclerotic margins	Calcified well-circumscribed solitary lesion, with peripheral rim appearance
Ultrasound	Aneurysmal mass	Not mentioned	Not specified	Hypoechoic mass with a feeding vessel and intralesional vascularity	Not mentioned	Not mentioned	Not mentioned	Not mentioned	Not performed
b	Aneurysmal mass	Not mentioned	Not specified	Not mentioned	Not mentioned	Lytic lesion with bony outline	Arcuated thin rim with an ambiguous density, suggesting calcification	Not mentioned	Peripheral zone calcification pattern and central calcium with lower density
R	Not mentioned	Not mentioned	Not specified	Soft-tissue signal intensity with intense enhancement following administration of IV contrast	Partially hemorrhagic, cystic mass with peripheral rim of low signal intensity and central fluid– fluid collection	Not mentioned	Overall signal intensity similar to surrounding normal muscles on T1-WI, well-defined margin on T2-WI, fluid-fluid levels, peripheral and intralesional	High T2 center, low T1 signal, heterogenic enhancement with a rim of low intensity consistent with calcified boarders surrounded by severe soft tissue edema	Heterogenous mass on T1-WI and T2-WI was seen, with a peripheral hypointense rim in both sequences. An outstanding edema on T2-WI extending to soft tissue and muscles of the medial compartment of the knee was detected

(continued)

Table 1. (co	ntinued)								
	Petrik et al. (1993)	Dalet al. (2000)	Nielsen et al. (2002)	Ajilogba et al. (2005)	Ellison et al. (2007)	Sahu et al. (2008)	Hao et al. (2012)	Olshinka et al. (2020)	Present case (2023)
Bone scintigraphy	Not mentioned	Not mentioned	Not mentioned	Not mentioned	Not mentioned	Not mentioned	septa enhancement Not mentioned	Not mentioned	Hypervascularized area on vascular phase and on delayed phase, with an increase in
Molecular tests	Not mentioned	Cytogenetic analysis: t(17;17)(q12; p13)	Not mentioned	Not mentioned	Cytogenetic analysis: t(5;17)(q33; p13)	Not mentioned	Not mentioned	Not mentioned	osteogenic activity NGS: <i>COL1A1::USP6</i> gene fusion
Treatment	Not mentioned	Not mentioned	Not mentioned	Total excision	Total excision	Not mentioned	Not mentioned	Total excision	Marginal resection
Follow-up	Not mentioned	Not mentioned	16 months, no	Not mentioned	Not mentioned	Not mentioned	Not mentioned	18 months, no recurrences	12 months, no recurrences
			recurrences						

with the IGV sequence viewer. A variant analysis was performed on the Archer platform.

### Clinical Evolution

Twelve months after tumoral excision the patient was free of disease and asymptomatic, and returned to his normal sports activities. Consent for publication has been obtained from the family.

## Discussion

Abbreviations: CT, computerized tomography; MRI, magnetic resonance imaging; NGS, next-generation sequencing, WI, weighted images.

Soft tissue aneurysmal bone cyst is an extremely rare tumor. We have found 162 aneurysmal bone cyst cases in the Pathology Department files of our University Hospital, with only three of them being located in soft tissues. Two of these cases had been previously reported by Rodríguez-Peralto et al<sup>14</sup> and López-Barea et al.<sup>15</sup>

We report a case of a 13-year-old boy who presented with an extraosseous aneurysmal bone cyst. The average age at diagnosis of soft tissue aneurysmal bone cyst is 29 years old (range 7-60 years old) with a male-to-female ratio of 1:1.2.<sup>6,7,16</sup> Soft tissue aneurysmal bone cyst most commonly affects skeletal muscles or the subcutaneous tissues of lower limbs; followed by the arms, the shoulders, and other rarer sites that include the abdominal wall, pelvis, breasts, or carotid artery.<sup>16–18</sup> The main characteristics of previously soft tissue aneurysmal bone cysts that arose in patients younger than 13 years old are summarized in Table 1. In these patients, the most frequent sites are the shoulder region<sup>10,19</sup> and the thigh.<sup>20,21</sup> This is the first reported case in the sartorius muscle.

The specific radiological diagnostic features of soft tissue aneurysmal bone cyst include a peripherally calcified soft tissue mass with a hypodense center on CT, hemorrhagic spaces with fluid-fluid levels, and enhancing septations on MRI. These features were not all visible in the initial radiological studies. This disparity has been previously described and may reflect the different phases of the evolution of this entity.<sup>21</sup> The present case initially showed a mass with radiated peripheral coarse calcification, a predominantly central solid component, and was surrounded by perilesional edema. These features are characteristic of myositis ossificans. However, later the mass grew, and the peripheral calcification became thinner. The central solid component showed scattered hypodense areas, suggestive of necrotic and/or cystic changes, which is not the usual evolution of myositis ossificans. The classical diffuse ossification pattern on CT of the mature phase of myositis ossificans was not seen. Perilesional edema in adjacent soft tissues can lead to the impression of an aggressive or even malignant lesion. This feature is speculated to be only present in the active phase of the current case, and disappear later on in a more quiescent stage.<sup>22</sup> The main radiological differential diagnosis of soft tissue aneurysmal bone cyst includes

myositis ossificans, giant cell tumor of soft tissue, giant cell tumor of the tendon sheath, and extraskeletal telangiectatic osteosarcoma.<sup>23,24</sup>

Blood-filled cavernous spaces without endothelial lining separated by fibrous septa that contain spindleshaped fibroblasts, osteoclast-type giant cells, and osteoblastic-rimmed woven bone are characteristic histological features of soft tissue aneurysmal bone cyst. Some tumors can histologically mimic it. Myositis ossificans, nodular fasciitis, giant cell tumor of soft tissue, ossifying fibromyxoid tumor, and extraskeletal osteosarcoma should be ruled out. Giant cell-rich tumors of soft tissue exhibit a multinodular architecture that includes bland, round-to-oval mononuclear cells, unlike soft tissue aneurysmal bone cyst.<sup>2</sup> Both feature giant multinucleated cells, but giant cell-rich tumors usually do not present a peripheral calcified rim.<sup>2</sup> Myositis ossificans is also wellcircumscribed with a peripheral lamellar bone rim that may also harbor COL1A1::USP6 gene fusion.<sup>13</sup> It is usually related to a traumatic antecedent and are self-limited lesions, unlike our case. Myositis ossificans does not show blood-filled cystic cavities, and no blue bone has ever been described.<sup>2</sup> Multinucleated giant cells may be occasionally observed in myositis ossificans, while they are frequently seen in aneurysmal bone cyst, as in our case. The classical zonal pattern of myositis ossificans, hyaline matrix, or osteoma-like areas was not observed in our case. Amir et al<sup>24</sup> suggested that a subset of myositis ossificans-like lesions is an early phase of an evolving soft tissue aneurysmal bone cyst. The morphology in the initial biopsy performed in our case may overlap to that early myositis ossificans-like phase. Ossifying fibromyxoid tumors could also be partially surrounded by a shell of metaplastic lamellar bone. They are composed of uniform round to epithelioid cells arranged in cords with a characteristic S-100 protein and desmin-positive immunophenotype.<sup>2</sup> Other malignant soft tissue tumors like extraskeletal osteosarcoma enter in differential diagnosis. Unlike soft tissue aneurysmal bone cyst, extraosseous osteosarcoma shows a dense atypical peripheral and infiltrative cellularity with centrally located osteoid matrix.<sup>2</sup> Finally, secondary aneurysmal bone cyst to these benign and malignant lesions needs to be discarded. Neither secondary aneurysmal bone cyst nor none of the abovementioned entities, except myositis ossificans, present COL1A1::USP6 gene fusion.13

# Conclusion

We present a case of a 13-year-old boy soft tissue aneurysmal bone cyst and highlight the rarity of this tumoral entity that affects a wide age range. It is essential to recognize its clinical, radiographic, and histological features to exclude other tumoral mimickers. A cytogenetic analysis can be a helpful tool to confirm the diagnosis in selected cases.

#### **Authors' Note**

All data generated or analyzed during this study are included in this article. Further enquiries can be directed to the corresponding author.

#### **Declaration of Conflicting Interests**

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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#### **Ethical Approval**

Not applicable. Consent for publication has been obtained from the family.

#### **Informed Consent**

Informed consent for publication was obtained from the family.

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