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Case report

Cardiac Synovial Sarcoma in Children: A Case Report and Literature Review

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Abstract

Background: Synovial sarcomas are rare malignant mesenchymal tumors, primarily observed in adolescents and young adults. These tumors are predominantly located in the extremities. Cardiac involvement is exceptionally rare, **is** infrequently reported in the literature, and presents diagnostic, therapeutic, and prognostic challenges.

Case Presentation: We report a case of cardiac synovial sarcoma in a 12-year-old child, located in the right ventricle and measuring 67 mm at its largest dimension. Histological and immunohistochemical analyses confirmed the diagnosis. The extension workup was negative. The tumor was resected by morcellation, and chemotherapy with an ifosfamide–doxorubicin regimen was initiated, combined with adjuvant radiotherapy. The patient is currently alive but presents with a recurrence at the initial site after 23 months.

Conclusion: Cardiac synovial sarcomas are rarely documented in the literature. Although rare, it is crucial to consider this diagnosis in the presence of a malignant cardiac tumor with biphasic or spindle cell morphology.

Keywords: Pediatric cardiac tumor, Synovial sarcoma, Primary cardiac sarcoma, Right ventricle, Biphasic sarcoma

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1. Introduction

Synovial sarcomas (SS) are rare malignant mesenchymal tumors, primarily observed in adolescents and young adults. They represent the second most common type of malignant mesenchymal tumor after rhabdomyosarcomas and are often associated with a poor prognosis due to significant diagnostic delays, which can extend up to 98 weeks [1,2]. The diagnosis relies on histological analysis following a tumor biopsy. Contrary to what their name suggests, synovial sarcomas do not originate from synovial membrane cells, and their exact cell of origin remains uncertain [3]. In most cases, these tumors are located in the extremities, near large joints [3]. Cardiac involvement is exceptionally rare, with very few cases reported in the literature. This rarity poses significant diagnostic, therapeutic, and prognostic challenges.

2. Case Presentation

Patient A.Z., a 12-year-old Tunisian, prepubescent child, was born after a well-monitored pregnancy and delivered full term by vaginal delivery, with vaccinations up to date. He had normal psychomotor development. His medical history included a urinary tract infection at 2 months of age, complicated by dehydration.

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There was no family history of congenital heart disease or neoplasms. He presented with a deterioration in general condition, including an estimated weight loss of approximately 2 kg and dyspnea that initially occurred with exertion and later at rest, in the absence of fever.

At admission, the patient exhibited signs of moderate right-sided heart failure, including mild lower-limb edema, tachypnea with a respiratory rate of 26 breaths/min, and perioral cyanosis. Oxygen saturation was 88%. A cardiac ultrasound revealed a mobile echogenic mass in the right ventricle measuring 35×49 mm, with probable calcifications in the right atrium.

Due to the unavailability of cardiac magnetic resonance imaging (MRI), a thoracic angiocomputed tomography (CT) scan was performed as the first-line investigation and confirmed the presence of a lobulated, tissue-dense mass in the right ventricle, with its base implanted on the apicolateral wall of the right ventricle (RV), occupying the entire ventricular cavity and measuring 67 × 36 mm (Figure 1). It extended into the RV outflow tract, with its upper pole located 13 mm below the pulmonary valve. The right atrium was dilated. An extension workup, including bone scintigraphy, thoracoabdominopelvic CT, and brain MRI, was negative. The patient underwent surgery 10 days after admission. Intraoperatively, the mass was polylobulated and encapsulated, infiltrating the septal leaflet of the tricuspid valve, the interventricular septum, and the free wall of the RV.

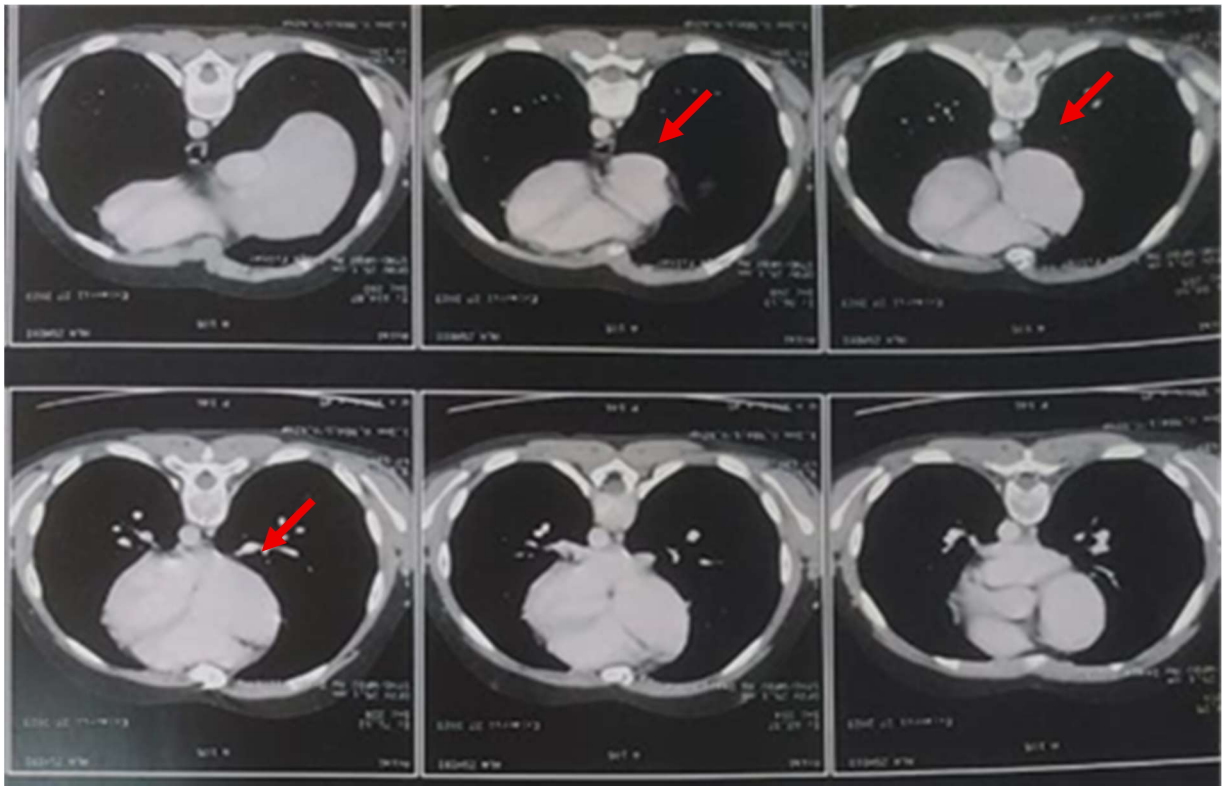

Fig. 1. Thoracic CT scan showing a highly enhanced tissue mass arising from the right ventricle of the heart.

The tumor was completely resected by morcellation, with microscopic tumor remnants (R0). Histological analysis revealed a hypercellular tumor proliferation infiltrating the ventricular muscle. It was organized in fascicles composed of slightly atypical, relatively monomorphic ovoid cells, exhibiting numerous mitotic figures, with a mitotic index exceeding 15 mitoses per 10 high-power fields. The tumor was richly vascularized, displaying a hemangiopericytoma-like pattern (Figure 2).

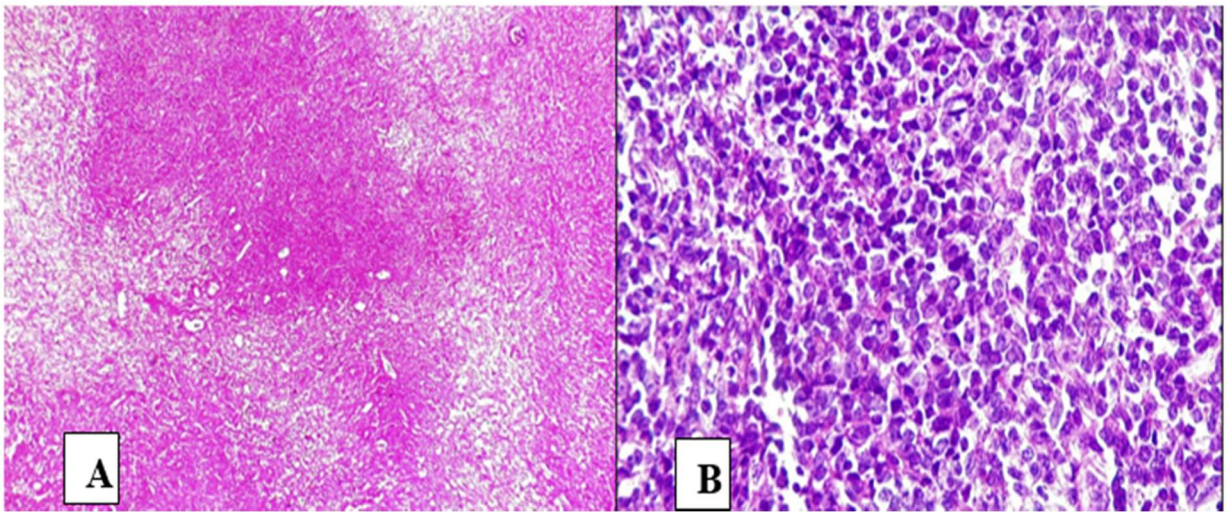

Fig. 2. Histological sections of a cardiac synovial sarcoma A: Tumor proliferation with storiform architecture (H&E, X100) B: Monomorphic atypical tumor cells (H&E, X400).

Immunohistochemical analysis showed diffuse positive staining of tumor cells for transducin-like enhancer of split 1 (TLE1), with focal positivity for ERG. The cells were negative for MyoD1, desmin, and smooth muscle actin (SMA) (Figure 3).

After surgery, the patient received adjuvant chemotherapy with ifosfamide–doxorubicin, administered over six cycles at 21-day intervals: four cycles of ifosfamide–doxorubicin followed by two cycles of ifosfamide alone.

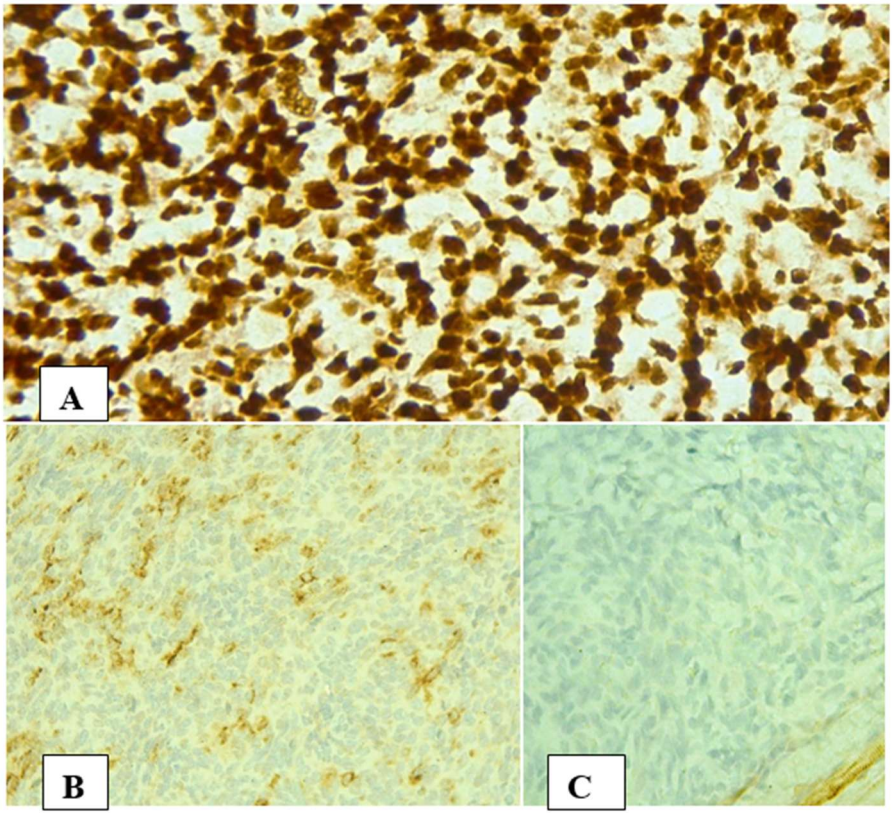

Fig. 3. Immunohistochemical staining of the tumor. A: Intense and diffuse staining with TLE1 B: Moderate and focal staining with EMA C: Negative staining for MyoD1

Adjuvant radiotherapy was discussed at the multidisciplinary team meeting due to the morcellation technique used during surgery. It was indicated because of the risk of residual microscopic disease. The administered dose was 50.4 Gy delivered in 28 fractions, with good clinical tolerance. The patient underwent sperm cryopreservation for fertility preservation. Twenty-three months later, the patient remains alive but presents with a recurrence at the initial site.

3. Discussion

The primary cardiac localization of synovial sarcoma is extremely rare. Reported cases in the general population number fewer than one hundred to date and are primarily described in the literature as individual case reports. This tumor accounts for only approximately 5% of malignant primary cardiac tumors [4]. To our knowledge, pediatric cases represent a small minority of reported instances. The age range reported is between 12 and 23 years, with a median age of 17 years, and a marked male predominance (3:1), as observed in our case (Table 1).

Clinically, as with synovial sarcomas of the extremities, cardiac synovial sarcoma is characterized by nonspecific functional symptoms, leading to delays in diagnosis and treatment. The symptoms are generally caused by tumor extension into the cardiac tissue and adjacent structures, with hemodynamic manifestations being predominant. Dyspnea is the most frequently reported symptom, as observed in our patient, followed by syncope, fatigue, chest pain, and fever (Table 1) [9-18].

According to the literature and the cases reviewed, blood tests and chest radiography are nonspecific for this condition and may even appear completely normal. Radiological examinations are also nonspecific with regard to cardiac localization, and typical findings such as calcifications, commonly described in extracardiac locations, are rarely reported.

The diagnostic tool most often detecting the presence of the mass is cardiac ultrasound, which has good sensitivity for tumor size and location. In our case, the diagnosis was initially suggested by cardiac ultrasound findings. Thoracic CT and cardiac MRI also provide valuable anatomical and tissue characterization support.

In our review, tumor size ranged from 2.5 to 11 cm, with a median size of 8 cm. Cardiac tumors are typically larger than extracardiac ones. Histological subtypes observed in the heart are comparable to those described in extracardiac locations. According to the World Health Organization (WHO) Classification of Tumors of Soft Tissue and Bone (5th edition, 2020), two classic histological subtypes are recognized: the monophasic subtype, which is the most common and consists exclusively of spindle cells or cells with an epithelial-like appearance, and the biphasic subtype, which is composed of both spindle and epithelial-like cells with glandular differentiation, present in varying proportions. Children show a higher frequency of monophasic forms, similar to the adult population.
Immunohistochemical findings are similar to those in other locations. Synovial sarcomas may express epithelial markers through the epithelial component, such as epithelial membrane antigen [9, 15–17] or cytokeratin [9–11,17], as

Table 1. Clinical and histological features of the 10 primary cardiac synovial sarcoma cases identified in the literature along with the present case.

Author & Year	Age	Sex	Symptoms	Location	Size (cm)	Histologic Type	IHC	Treatment	Follow-up
Nicholson et al. (1997)	13	M	Syncope; Vomiting; Headache	RA	5	Biphasic	Cytokeratin (+); EMA (+); CAM5-2 (+)	R0 surgery; CT	Progression-free survival at 10 months
Oizumi et al. (1999)	19	F	Dyspnea; General asthenia	Pericardium	11	Biphasic	Cytokeratin (+); Vimentin (+); EMA (–); S100 (–)	R1 surgery	Recurrence at 7 months
Al-Rajhi et al. (1999)	19	M	Dyspnea	Pericardium	10	Biphasic	Cytokeratin (+); Vimentin (+)	R2 surgery; CT + RT	–
Provenzano et al. (2006)	14	M	Poor general condition	RA & RV	RA: 8 / RV: 4	Biphasic	–	R2 surgery; CT	Progression-free survival at 18 months
Boulmay et al. (2007)	19	F	Dyspnea	RV	–	–	CD99 (+); BCL2 (+)	R2 surgery	Pulmonary metastasis
Yu et al. (2011)	20	M	Dyspnea	RV & RA	RV: 11 / RA: 2.5	Monophasic	–	R0 surgery	Recurrence at 14 months
Chekrine et al. (2013)	13	M	Dyspnea	Pericardium	7.3	Monophasic	Vimentin (+); EMA (+); CD34 (–)	R2 surgery; CT + RT	Recurrence at 21 months
Yoshino et al. (2013)	13	M	Fever; Chest pain; Asthenia	Pericardium	–	Monophasic	Vimentin (+); EMA (+); BCL2 (+); CD99 (+)	R2 surgery; CT	Progression-free survival at 3 months
Khan et al. (2014)	17	M	Fever; Chest pain; Asthenia	RA & RV	8 (resected tumor)	Monophasic	Cytokeratin (+); EMA (+); CD34 (+)	R2 surgery; CT	Progression-free survival at 11 months
Teng et al. (2021)	23	F	Dyspnea; Chest tightness	–	10	Monophasic	–	–	Died after 3 months
Present case (2025)	12	M	Dyspnea; Poor general condition	RV	4.5	Monophasic	TLE (+), MyoD1 (-); AML & Desmin (–)	R0 surgery; CT; RT	Progression-free survival at 23 months

R0 surgery indicates negative surgical margins; R1 indicates microscopically involved margins; R2 indicates macroscopically involved margins; CT: chemotherapy; RT: radiotherapy; RA: right atrium; RV: right ventricle.

as well as mesenchymal markers through the mesenchymal component, such as vimentin. The antibody specific to the SS18–SSX fusion protein is highly sensitive and specific for synovial sarcoma. Immunohistochemistry using this antibody may replace molecular genetic or cytogenetic testing in most cases. An antibody targeting the C-terminal region of SSX is also highly sensitive, though slightly less specific [5].

Transducin-like enhancer of split 1 (TLE1) is widely recognized as a highly sensitive and relatively specific immunohistochemical marker for synovial sarcoma. It is typically expressed in both the monophasic and biphasic subtypes of the tumor. TLE1 exhibits predominantly nuclear staining, and its diagnostic utility is most pronounced when moderate to strong nuclear positivity is observed. Although not entirely exclusive to synovial sarcoma, TLE1 helps to distinguish it from other spindle cell neoplasms, such as malignant peripheral nerve sheath tumors or fibrosarcomas, which usually do not express this marker at similar intensities [6].

Cluster of Differentiation 99 (CD99) is a transmembrane protein that shows strong membranous immunoreactivity in a majority of synovial sarcomas. However, CD99 is not specific to this entity; it is also characteristically and diffusely expressed in Ewing sarcoma, where it similarly displays intense membranous staining. This overlap may lead to diagnostic confusion, particularly in small round cell tumors, and highlights the necessity of interpreting CD99 expression in conjunction with other immunohistochemical markers and molecular testing for SS18–SSX gene fusion to reach an accurate diagnosis [7].

Synovial sarcomas are cytogenetically characterized by a recurrent chromosomal translocation, t(X;18)(p11.2; q11.2), in more than 95% of cases [6], in which the SS18 gene on chromosome 18 fuses with one of three SSX genes located on the X chromosome: SSX1, SSX2, or SSX4 [8].

Genetic testing can be crucial for diagnosis, although it is not mandatory in several national protocols, such as the Tunisian and Italian protocols [3].

Management of these tumors follows the same guidelines as for extracardiac locations. However, the larger tumor size and the technical difficulty of achieving complete surgical resection in cardiac localizations necessitate more frequent use of adjuvant therapies. The same chemotherapy

agents, namely ifosfamide and doxorubicin, are typically used.

Prognostic factors for cardiac localizations are comparable to those of other disease sites, with particular emphasis on the adverse role of the deep and axial location. Due to the rarity of this tumor, no studies specifically examining survival rates or metastatic patterns in pediatric cardiac cases have been conducted. In adults, the lungs are the most common metastatic site, a parameter that deserves further investigation in both pediatric and adult populations. In this review, only one of the 11 cases presented with pulmonary metastasis. Four of the 11 reported cases had tumor recurrence at the initial site. Six cases achieved progression-free survival ranging from 3 to 23 months.

Conclusion

In conclusion, pediatric cardiac synovial sarcomas represent an extremely rare and aggressive disease, posing significant diagnostic and therapeutic challenges. They are rarely reported in the literature, typically as isolated cases. Due to their rarity, unusual location, and symptom similarity with more common cardiac pathologies, diagnosis is often delayed, thereby limiting therapeutic options and adversely affecting prognosis. It is crucial to continue research and promote dedicated registries for pediatric synovial sarcomas to improve early diagnosis, refine treatment strategies, and increase survival rates of young patients affected by this complex condition.

Patient Consent

The patient provided written informed consent to participate in this study.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Conflict of Interest

The authors declare no conflicts of interest.

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None.

Authors' Contributions

All authors contributed to the manuscript and have read and approved the final manuscript.

References

[1] Ferrari A, Casanova M. New concepts for the treatment of pediatric nonrhabdomyosarcoma soft tissue sarcomas. Expert Rev Anticancer Ther. 2005;5(2):307-18. https://doi.org/10.1586/14737140.5.2.307

[2] Chotel F, Unnithan A, Chandrasekar CR, Parot R, Jeys L, Grimer RJ. Variability in the presentation of synovial sarcoma in children: a plea for greater awareness. J Bone Joint Surg Br. 2008;90(8):1090-6. https://doi.org/10.1302/0301-620X.90B8.19815

[3] Ferrari A, Bisogno G, Alaggio R, Cecchetto G, Collini P, Rosolen A, et al. Synovial sarcoma of children and adolescents: the prognostic role of axial sites. Eur J Cancer. 2008;44(9):1202-9. https://doi.org/10.1016/j.ejca.2008.03.016

[4] Zhang L, Qian J, Li Z, Jing H. Primary synovial sarcoma of the heart. Cardiol J. 2011;18(2):128-33. https://doi.org/10.1016/j.ejca.2008.03.016

[5] Baranov E, McBride MJ, Bellizzi AM, Ligon AH, Fletcher CDM, Kadoch C, et al. A Novel SS18-SSX Fusion-specific Antibody for the Diagnosis of Synovial Sarcoma. Am J Surg Pathol. 2020;44(7):922-33. https://doi.org/10.1097/PAS.0000000000001447

[6] Chastagner P. Facteurs pronostiques des synovialosarcomes chez l'enfant. Archives de Pédiatrie 2003;10(12):1096-1097. https://doi.org/10.1016/j.arcped.2003.09.023

[7] Blay JY, Von Mehren M, Jones RL, Martin-Broto J, Stacchiotti S, Bauer S, et al. Synovial sarcoma: characteristics, challenges, and evolving therapeutic strategies. ESMO Open. 2023;8(5):101618. https://doi.org/10.1016/j.esmoop.2023.101618

[8] Clark J, Rocques PJ, Crew AJ, Gill S, Shipley J, Chan AM, et al. Identification of novel genes, SYT and SSX, involved in the t(X;18)(p11.2;q11.2) translocation found in human synovial sarcoma. Nat Genet. 1994;7(4):502-8 https://doi.org/10.1038/ng0894-502

[9] Nicholson AG, Rigby M, Lincoln C, Meller S, Fisher C. Synovial sarcoma of the heart. 1997;30(4):349-52. https://doi.org/10.1046/j.1365-2559.1997.d01-616.x

[10] Oizumi S, Igarashi K, Takenaka T, Yamashiro K, Hiraga H, Fujino T, et al. Primary Pericardial Synovial Sarcoma With Detection of the Chimeric Transcript SYT-SSX. JpnCirc J. 1999;63(4):330-2. https://doi.org/10.1253/jcj.63.330

[11] Al-Rajhi N, Husain S, Coupland R, McNamee C, Jha N. Primary pericardial synovial sarcoma: A case report and literature review. J Surg Oncol. 1999;70(3):194-8. https://doi.org/10.1002/(sici)1096-9098(199903)70:3<194::aid-jso10>3.0.co;2-8

[12] Provenzano SC, Con R, Jones OD, Grant PW. Synovial Sarcoma of The Heart. Heart, Lung and Circulation. 2006;15(4):278-9. https://doi.org/10.1016/j.hlc.2006.01.009

[13] Boulmay B, Cooper G, Reith JD, Marsh R. Primary Cardiac Synovial Sarcoma: A Case Report and Brief Review of the Literature. Sarcoma. 2007;2007:1-4. https://doi.org/10.1155/2007/94797

[14] Yu L, Shi E, Gu T. A Giant Primary Cardiac Synovial Sarcoma. Journal of Cardiac Surgery. 2011;26(1):74-4. https://doi.org/10.1111/jocs.14254

[15] Chekrine T, Sahraoui S, Cherkaoui S, Eddakkaoui H, Labsaili H, Marouane S, et al. Primary pericardial synovial sarcoma: A case report and literature review. J Cardiol Cases. 2014;9(1):40-3. https://doi.org/10.1016/j.jccase.2013.09.005

[16] Yoshino M, Sekine Y, Koh E, Kume Y, Saito H, Kimura S, et al. Pericardial synovial sarcoma: a case report and review of the literature. Surg Today. 2014;44(11):2167-73. https://doi.org/10.1007/s00595-013-0720-4

[17] Khan I, Gul S, Tufail Z, Khan K, Sharma P, Waheed A. Primary cardiac synovial sarcoma. Asian Cardiovasc Thorac Ann. 2015;23(6):713-5. https://doi.org/10.1177/0218492314524753

[18] Teng F, Chen D, Li Y, Fang W, Yang S, Shang J, et al. Primary cardiac synovial sarcoma: a clinicopathological, immunohistochemical, and molecular genetics study of five

clinical cases. Cardiovasc Pathol. 2021;50:107286. https://doi.org/10.1016/j.carpath.2020.107286

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