Received: 23 May 2025 | Accepted: 18 August 2025

DOI: 10.1002/msp2.70017

Malignancy Spectrum

CASE REPORT

Primary mediastinal synovial sarcoma in an infant: A case report

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Abstract

Background: Primary mediastinal synovial sarcoma (PMSS) is an uncommon and aggressive soft tissue tumor, particularly rare in infants. Its diagnosis is challenging due to overlapping features with other thoracic neoplasms and requires integration of histopathology, immunohistochemistry, and molecular genetics.

Case presentation: We present a case of a 4-month-old girl admitted with respiratory distress and found to have a large posterior mediastinal mass. Histopathology and immunohistochemistry were compatible with biphasic synovial sarcoma. However, molecular confirmation via detection of *SS18-SSX* fusion was not performed. Despite multimodal chemotherapy, the tumor progressed, and complete surgical resection was unachievable. The patient died during follow-up in the pediatric intensive care unit.

Conclusion: Accurate diagnosis is critically dependent on *SS18-SSX* molecular testing, particularly in infants. Incomplete resection and lack of genetic confirmation may contribute to poor prognosis. Literature supports that complete resection is the only consistent predictor of survival. This case illustrates the limitations and consequences of managing PMSS without molecular confirmation or complete resection. A multidisciplinary approach with current diagnostic standards is essential in these high-risk pediatric tumors.

KEYWORDS

synovial sarcoma, mediastinal tumor, pediatric sarcoma

INTRODUCTION

Synovial sarcoma (SS) is a malignant mesenchymal neoplasm with partial epithelial differentiation, comprising 5%–10% of all soft tissue sarcomas. Though most commonly found in the extremities of adolescents and young adults, SS has been identified in virtually any anatomical location, including the mediastinum [1].

Although once considered exceedingly rare, primary mediastinal synovial sarcoma (PMSS) is now better characterized through accumulating case reports. Terra et al. documented 21 thoracic SS cases [2], and Kato et al. conducted a systematic review of thoracic presentations of SS [3]. However, PMSS in infancy remains extremely rare, and its clinical and radiological similarity to other mediastinal tumors continues to delay diagnosis.

We present an infantile case of PMSS and review the critical diagnostic and therapeutic challenges, particularly emphasizing the need for molecular diagnostics and complete surgical excision.

CASE PRESENTATION

A 4-month-old girl was admitted to our hospital with progressive respiratory distress and significant abdominal distension. On physical examination, breath

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sounds were markedly diminished on the right side, and signs of respiratory compromise were evident. Initial laboratory investigations were unremarkable, but due to worsening symptoms, an urgent thoracic computed tomography (CT) scan was performed.

Thoracic CT revealed a large posterior mediastinal mass measuring approximately $53 \times 57\ mm^2$, extending into the right hemithorax and compressing the adjacent lung parenchyma and airways (Figure 1a). Coronal reconstruction demonstrated marked displacement of the mediastinum and compression of the right upper lung (Figure 1b). These findings contributed to severe airway obstruction and atelectasis.

The patient was intubated and transferred to the pediatric intensive care unit for respiratory stabilization. Based on the initial radiological impression and patient age, neuroblastoma was suspected, and chemotherapy with vincristine, etoposide, and carboplatin was initiated. Subsequently, a partial surgical resection of the mass was performed in September 2022 to obtain tissue for definitive diagnosis and to relieve airway compression.

Highlights

- Primary mediastinal synovial sarcoma (PMSS) is extremely rare in infants and poses significant diagnostic challenges.
- Accurate diagnosis requires molecular confirmation of *SS18-SSX* fusion, particularly in atypical age groups and locations.
- Radiological and histopathological documentation must be complemented with molecular studies to avoid misdiagnosis.
- Complete surgical resection is the most critical factor influencing survival.
- This case underscores the consequences of incomplete resection and the lack of molecular confirmation in PMSS.

HISTOPATHOLOGICAL FINDINGS

Microscopy revealed a biphasic neoplasm with spindle-cell and epithelial components. Immunohistochemistry demonstrated positivity for vimentin, CD99, and pan-cytokeratin, and focal positivity for Wilms' tumor 1 (WT1), transducin-like enhancer of split 1 (TLE-1), and neuron-specific enolase (NSE). CD34, desmin, chromogranin, and calretinin were negative. Ki-67 proliferation index was 60%. Despite these findings, molecular testing for the *SS18-SSX* translocation was not performed, which is a critical limitation in this case. Recent advances have introduced *SS18-SSX* fusion-specific antibodies that may offer valuable diagnostic utility in such settings, as described by Baranov et al. [4, 5].

TREATMENT AND CLINICAL COURSE

The patient underwent multiple chemotherapy regimens: three cycles of ifosfamide and doxorubicin, two cycles of cisplatin-doxorubicin-ifosfamide, and the ifosfamide-carboplatin-etoposide (ICE) protocol. No clinical or radiological improvement was observed. Complete surgical excision was not feasible due to anatomical constraints. The patient deteriorated and died during follow-up in the pediatric intensive care unit.

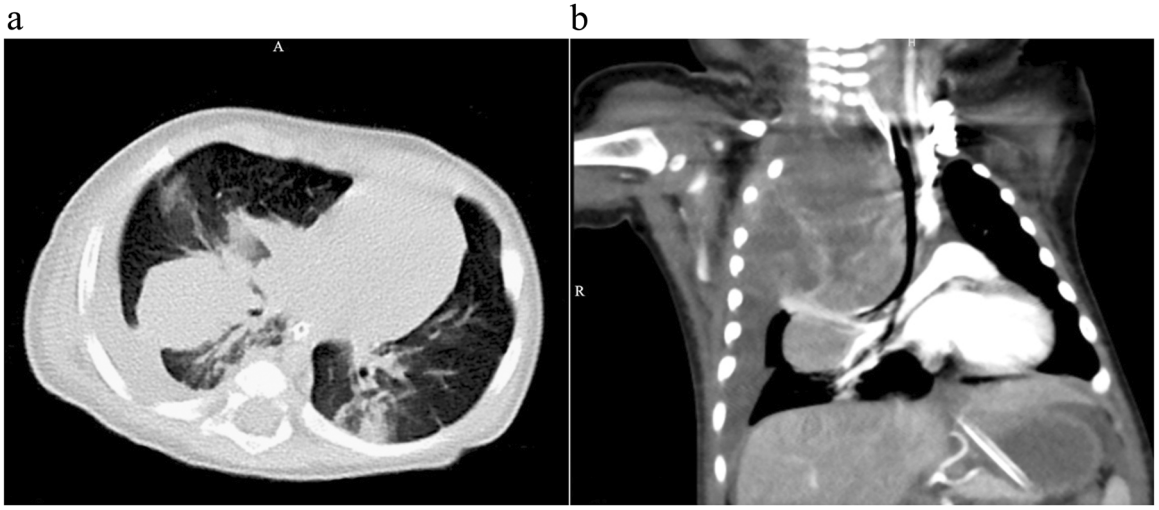

FIGURE 1 (a) Axial contrast-enhanced thoracic computed tomography (CT) image showing a large posterior mediastinal mass extending into the right hemithorax with significant airway compression and associated lung collapse. (b) Coronal CT reconstruction demonstrating the mass displacing the mediastinum and compressing the adjacent lung parenchyma.

DISCUSSION

Although once considered exceedingly rare, PMSS is now better characterized through an increasing number of reported cases in the literature [2, 3]. Nonetheless, accurate diagnosis remains challenging, especially in atypical age groups such as infants and at uncommon anatomical sites like the mediastinum. SS shares radiological and histopathological features with other spindle-cell and epithelial tumors of the thoracic cavity, which can lead to diagnostic delay or misdiagnosis.

None of the currently used immunohistochemical markers are pathognomonic for SS, and their interpretation must be combined with molecular studies for definitive diagnosis [6, 7]. A major limitation in this case is the absence of molecular confirmation of the *SS18-SSX* fusion, which is recognized as the defining genetic hallmark of SS. While the histopathological and immunohistochemical features were consistent with biphasic SS, the lack of genetic confirmation introduces diagnostic uncertainty, especially in atypical locations and age groups such as infancy. This may affect the accuracy of the diagnosis, the prognostic assessment, and treatment decisions [8]. European Society for Medical Oncology guidelines and recent literature strongly recommend molecular or cytogenetic testing to establish a definitive diagnosis, guide appropriate therapeutic strategies, and reduce the risk of misdiagnosis. Therefore, the inability to demonstrate the fusion transcript in this case underscores the importance of comprehensive diagnostic workup, including fusion-specific antibodies or molecular assays when standard methods are unavailable.

The use of *SS18-SSX* fusion-specific antibodies, as described by Baranov et al., can be a valuable diagnostic tool, particularly in limited resource settings or when tissue availability for molecular testing is insufficient [5]. This case emphasizes the need to integrate all available diagnostic modalities to ensure an accurate diagnosis and appropriate management plan.

From a therapeutic standpoint, a systematic review by Syred et al. demonstrated that completeness of surgical resection is the only consistent prognostic factor associated with improved survival in thoracic SS [3]. Our patient's tumor location within the posterior mediastinum and its proximity to vital structures rendered complete surgical excision infeasible, despite multimodal chemotherapy. This experience aligns with the conclusions of Espinoza et al., who emphasized that achieving negative margins or re-resection when possible is vital for improving outcomes in pediatric SS [9].

In summary, this case illustrates the complex diagnostic and therapeutic challenges posed by PMSS in infancy. It highlights the critical role of comprehensive molecular diagnostics to ensure diagnostic accuracy and appropriate clinical decision-making. It also reinforces the well-established finding that complete surgical resection is the most significant factor influencing survival, underscoring the need for a multidisciplinary approach and adherence to current clinical practice guidelines to optimize outcomes in children with SS.

CONCLUSION

This case exemplifies the diagnostic and therapeutic challenges of PMSS in infancy. It emphasizes the crucial importance of molecular confirmation, appropriate radiological documentation, and complete surgical resection. It underscores the necessity of molecular confirmation, the importance of appropriate radiological documentation, and the vital importance of complete surgical resection. Adoption of a standardized, multidisciplinary approach incorporating current guidelines is essential for improving outcomes in pediatric SS.

AUTHOR CONTRIBUTIONS

Şule Çalışkan Kamış: Conception; writing; data collection and/or processing. **Begül Yağcı:** Supervision; design; analysis and/or interpretation.

ACKNOWLEDGEMENTS

Not applicable.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

The datasets generated and/or analyzed during our study are not publicly available due to ethical and participant confidentiality reasons, but they are available from the corresponding author, Dr. Şule Çalışkan Kamış, upon reasonable request.

ETHICS STATEMENT

This case report was conducted in accordance with ethical standards. As it is a case report, ethics approval was not required. However, informed consent for publication was obtained from the patient's family. Informed consent for publication was obtained from the patient's family.

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How to cite this article: Kamış ŞÇ, Yağcı B. Primary mediastinal synovial sarcoma in an infant: a case report. *Malig Spect*. 2025;2(3):163-166. doi:10.1002/msp2.70017