Radiology Case Reports 20 (2025) 3295–3298

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journal homepage: www.elsevier.com/locate/radcr

Case Report

Transducin-like enhancer of split-1 (TLE-1)-positive primary pleuropulmonary synovial sarcoma in a 14-year-old female adolescent: A case report and literature review ☆,☆☆

Gashaw Arega[a], *Hewan Asfaw*[a], *Samuel Sisay*[b], *Fathia Omer Salah*[b], *Tihitena Negussie Mamo*[c], *Michael A. Negussie*[d,*], *Leul Adane*[b]

[a] *Department of Pediatrics and Child Health, College of Health Sciences, Addis Ababa University, Addis Ababa, Ethiopia*
[b] *Department of Radiology, College of Health Sciences, Addis Ababa University, Addis Ababa, Ethiopia*
[c] *Department of Surgery, College of Health Sciences, Addis Ababa University, Addis Ababa, Ethiopia*
[d] *School of Medicine, College of Health Sciences, Addis Ababa University, Addis Ababa, Ethiopia*

ARTICLE INFO

Article history:
Received 23 January 2025
Revised 16 March 2025
Accepted 17 March 2025

Keywords:
Pleuropulmonary synovial sarcoma
CT scan
Immunohistochemistry
TLE-1

ABSTRACT

Primary pleuropulmonary synovial sarcoma (PPSS) is a rare malignant soft tissue sarcoma primarily affecting adolescents and young adults. Diagnosis relies on clinical examination, radiological imaging, and confirmation through histopathological and immunohistochemical analyses. Due to nonspecific symptoms, diagnosis is often delayed. Treatment typically involves a multimodal approach, including systemic chemotherapy, surgical intervention, and radiotherapy. We present the case of a 14-year-old female with a 5-month history of cough, low-grade fever, and weight loss. A contrast-enhanced chest CT scan revealed a large left thoracic mass with lung infiltration, mediastinal invasion, and multiple enlarged lymph nodes. Histopathological and immunohistochemical analyses confirmed a primary pulmonary synovial sarcoma positive for transducer-like enhancer of split-1 (TLE-1). The patient underwent 2 cycles of neoadjuvant chemotherapy with ifosfamide and doxorubicin, but no significant improvement was observed. Local control options, including surgery and radiotherapy, were deemed infeasible, and palliative care was initiated.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported on this paper.

☆☆ Acknowledgments: No funding was received for this study.

* Corresponding author.
E-mail address: michielnegussie@gmail.com (M.A. Negussie).

https://doi.org/10.1016/j.radcr.2025.03.063

Introduction

Synovial sarcoma, a soft tissue tumor originating from pluripotent mesenchymal cells, accounts for 8% of all soft tissue tumors. While it typically arises near large joints in the extremities, it can also occur in the lungs, mediastinum, abdomen, head, neck, and heart [1,2]. Pleuropulmonary synovial sarcoma (PPSS), a rare subtype, originates from the lung or pleura, often presenting diagnostic challenges due to its nonspecific symptoms and radiological features [3].

Histologically, synovial sarcoma is classified into biphasic, monophasic spindle cells, and rare monophasic epithelial types [1,4,5]. Primary pulmonary synovial sarcoma is an aggressive malignancy, predominantly affecting adolescents and young adults. Common symptoms include chest pain, cough, shortness of breath, hemoptysis, and pleural effusion [2,6–9]. Radiologically, PPSS appears as a well-defined mass with heterogeneous enhancement on CT, often accompanied by pleural effusion [7].

Diagnosis is challenging due to morphological similarities with other neoplasms, necessitating immunohistochemical (IHC) markers such as transducer-like enhancer of split-1 (TLE-1) and molecular techniques like detection of the SS18-SSX fusion gene [10–12]. Treatment involves a multidisciplinary approach, including surgery, chemotherapy, and radiotherapy, though prognosis remains poor, with a 5-year survival rate of 50% [2].

Case presentation

A 14-year-old female presented with a 5-month history of dry cough, low-grade intermittent fever, and unintentional weight loss. Symptoms worsened one week prior to admission, with increased coughing and shortness of breath. On examination, her pulse rate was 120 beats per minute, respiratory rate 36 breaths per minute, temperature 36.4°C, and oxygen saturation 91% on 1 liter of intranasal oxygen. Chest examination revealed dullness and reduced air entry over the left lung fields.

Initial investigations, including complete blood count, organ function tests, serum electrolytes, and bone marrow biopsy, were normal. A chest X-ray showed complete opacification of the left hemithorax with a rightward mediastinal shift (Fig. 1). A contrast-enhanced chest CT scan revealed a large, heterogeneously enhancing left hemithoracic mass (20.4 × 17 cm) with coarse calcifications, infiltrating the left lung and extending into the left main bronchus. The mass caused a rightward mediastinal shift and obscured the fat plane with the aortic arch and pulmonary artery. Multiple enlarged lymph nodes were noted in the perivascular, supraclavicular, subcarinal, paratracheal, and para-aortic regions (Fig. 2).

After obtaining written consent, the patient was taken to the major operating room and positioned in the right lateral decubitus position. A left posterolateral thoracotomy was performed, revealing a large, fragile mass occupying the entire left hemithorax. The mass exhibited extensive vascular invasion and significant engorgement of collateral veins on the left side of the chest. Access to the left hemithorax was achieved through the fifth intercostal space, where profuse oozing was observed from both the tumor and its collateral veins. The tumor completely filled the left hemithorax, leaving no space between the mass and the chest wall. An incisional biopsy was taken from the mass, and further exploration was abandoned. The biopsy confirmed high-grade spindle cell sarcoma. Immunohistochemistry (IHC) showed diffuse positivity for TLE-1 and negativity for PanCK, BCL2, S100, and CD99, confirming TLE-1-positive primary pleuropulmonary synovial sarcoma.

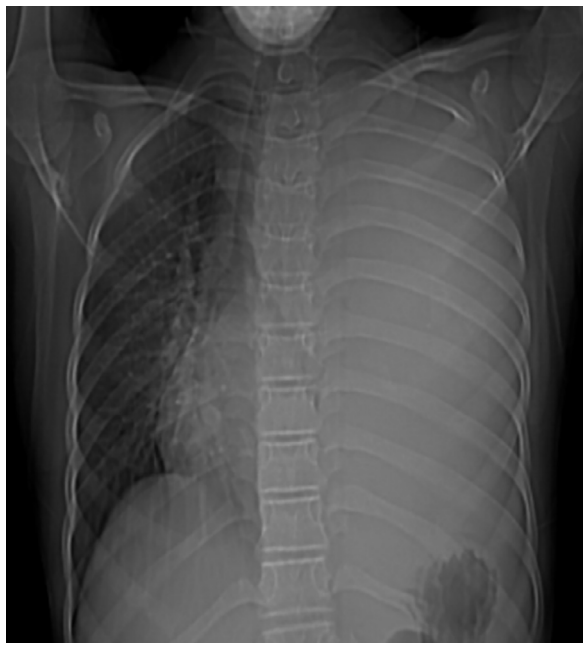

Fig. 1 – Anteroposterior (AP) chest X-ray demonstrating complete opacification of the left hemithorax accompanied by a rightward mediastinal shift.

Following the diagnosis, the patient underwent 2 cycles of neoadjuvant chemotherapy with ifosfamide and doxorubicin. Unfortunately, follow-up imaging showed no significant improvement in tumor size or extent. Given the lack of response and the extensive local invasion, options for local control, including surgery and radiotherapy, were deemed infeasible. The family was counseled on the poor prognosis, and palliative care was initiated to manage symptoms and improve quality of life.

Discussion

Synovial sarcoma is a rare soft tissue tumor, with pleuropulmonary synovial sarcoma (PPSS) representing an even rarer subset. PPSS predominantly affects adolescents and young adults, often presenting with nonspecific symptoms such as cough, chest pain, and weight loss [2,6–9]. Radiological findings, including well-defined masses with heterogeneous enhancement on CT, aid in diagnosis but are not pathognomonic [7–9].

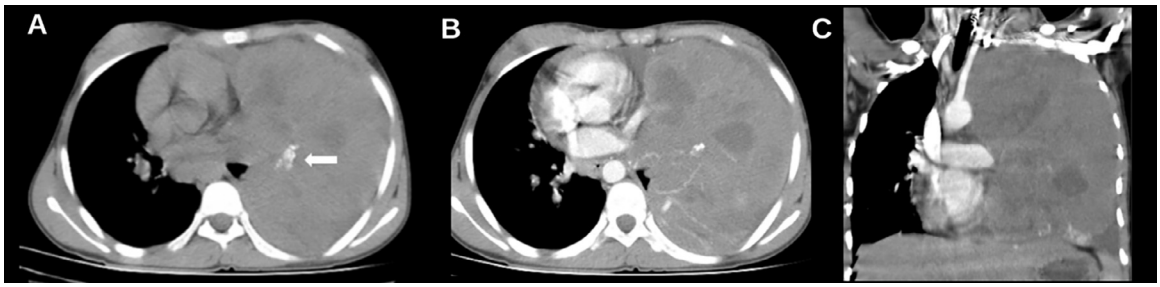

Fig. 2 – (A) Precontrast axial chest CT revealing a well-defined, predominantly soft tissue attenuating mass lesion in the left hemithorax with an irregular calcific focus (white arrow). (B, C) Postcontrast axial and coronal reformatted chest CT images demonstrating a well-marginated, heterogeneously enhancing left thoracic mass with cystic components.

The diagnosis of PPSS is challenging due to its morphological overlap with other neoplasms, such as fibrosarcoma, malignant mesothelioma, and metastatic carcinoma [13]. Accurate diagnosis relies on histopathological and immunohistochemical analyses. The SS18-SSX fusion gene, resulting from the t(X;18) translocation, is present in over 90% of cases and serves as the gold standard for diagnosis [10,12]. Immunohistochemical markers, including TLE-1, epithelial membrane antigen (EMA), cytokeratin, and BCL-2, are crucial in differentiating PPSS from other tumors [13,14]. TLE-1, a transcriptional corepressor involved in embryogenesis and hematopoiesis, has emerged as a sensitive and specific marker for synovial sarcoma [5,10,15,16]. A study done in 2020 reported a mean sensitivity of 94% and a mean specificity of 81% for TLE-1 in detecting synovial sarcoma [5]. Gene expression profiling studies have consistently demonstrated overexpression of TLE-1 in synovial sarcoma cells, making it a valuable diagnostic tool [5,16]. In our case, the tumor cells were diffusely positive for TLE-1 and negative for other markers, confirming the diagnosis of PPSS.

Radiologically, PPSS typically presents as a well-defined mass with heterogeneous enhancement on CT, often accompanied by pleural effusion [7–9]. In our patient, the chest CT revealed a large left hemithoracic mass with coarse calcifications, lung infiltration, and mediastinal invasion. These findings are consistent with previous reports of PPSS, highlighting the importance of advanced imaging in the diagnostic workup [7,8].

The prognosis for PPSS remains poor, with a 5-year survival rate of approximately 50% [2]. Poor prognostic factors include tumor size >5 cm, male gender, age >20 years, high mitotic count, and vascular invasion [2,4,8]. Although our patient is a young female, the large tumor size and vascular invasion place her in a high-risk category. There is no established gold standard treatment for PPSS, but a multimodal approach involving surgery, chemotherapy, and radiotherapy is recommended [7,8,11,17]. Radical resection remains the primary treatment method, with neoadjuvant chemotherapy often used to reduce tumor size and address micrometastasis [8]. In our case, the patient underwent 2 cycles of neoadjuvant chemotherapy with ifosfamide and doxorubicin. Unfortunately, follow-up imaging showed no significant improvement, and local control options, including surgery and radiotherapy, were deemed infeasible due to the extensive local invasion and poor response to chemotherapy. The family was counseled on the poor prognosis, and palliative care was initiated to manage symptoms and improve quality of life. This case demonstrates the aggressive nature of PPSS and the challenges in managing advanced disease, particularly when standard treatment modalities fail to achieve disease control.

Conclusion

Primary pleuropulmonary synovial sarcoma is a rare and aggressive malignancy requiring precise clinical, radiological, and pathological evaluation for diagnosis. TLE-1 is a valuable immunohistochemical marker for distinguishing PPSS from other mesenchymal tumors. A multidisciplinary treatment approach, including surgery, chemotherapy, and radiotherapy, is essential for managing this condition. However, as demonstrated in this case, advanced disease with poor response to treatment poses significant challenges, often necessitating a shift to palliative care. Further research into targeted therapies and improved treatment protocols is urgently needed to improve outcomes for patients with PPSS.

Availability of data and materials

Data supporting this case are available from the corresponding author upon request.

Patient consent

Written informed consent was obtained from the patient's parents for publication.

CRediT authorship contribution statement

Gashaw Arega: Writing – original draft, Resources, Data curation, Conceptualization. **Hewan Asfaw:** Writing – original draft, Resources, Data curation, Conceptualization. **Samuel Sisay:** Writing – original draft, Resources. **Fathia Omer Salah:**

Writing – original draft. **Tihitena Negussie Mamo:** Writing – review & editing. **Michael A. Negussie:** Writing – review & editing. **Leul Adane:** Writing – review & editing, Data curation.

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