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Case Report

Pathology Section

# An Anterior Abdominal Wall Alveolar Soft Part Sarcoma in a Child: A Case Report

ARTHI ANANDHAN[1], K CHANDRAMOULEESWARI[2], UMADEVI SRINIVASAN[3]



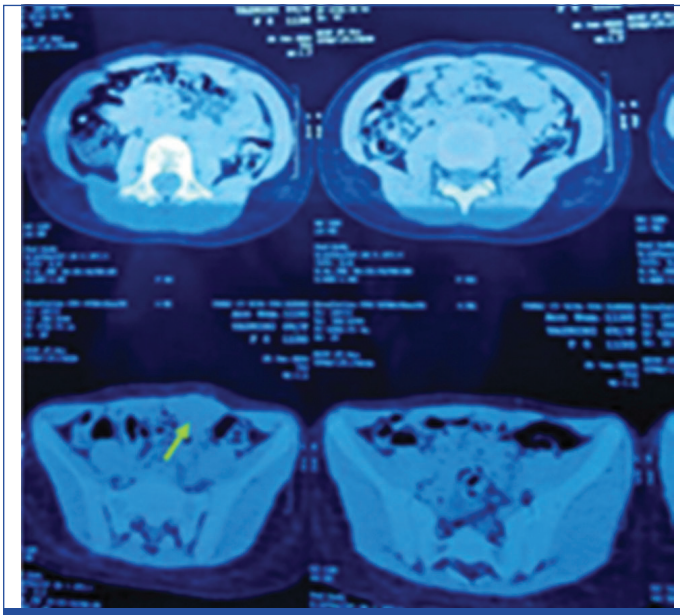

## ABSTRACT

Alveolar Soft Part Sarcoma (ASPS) is a rare and aggressive soft-tissue sarcoma, constituting less than 1% of all sarcomas, with a predilection for young adults and occasional paediatric presentations. This case report highlights an eight-year-old girl who presented with a one-month history of a painless swelling in the left anterior abdominal wall. Clinical evaluation and imaging revealed a 4×3.5×2.5 cm enhancing soft-tissue lesion in the left rectus abdominis muscle, raising suspicion of malignancy. Excisional biopsy revealed histological features typical of ASPS, including solid nests of polygonal cells with eosinophilic granular cytoplasm, prominent nucleoli, and pseudo-alveolar architecture. Tumour cells exhibited mild atypia, necrosis, and vascular invasion. Periodic Acid-Schiff (PAS) staining demonstrated intracytoplasmic crystals, and Immunohistochemistry (IHC) confirmed TFE3 nuclear positivity and CD34 expression, confirming the diagnosis of ASPS. ASPS is associated with a specific translocation, der(17) t(x;17)(p11;q25), resulting in ASPSCR1-TFE3 gene fusion, which activates oncogenic pathways such as c-Mesenchymal Epithelial Transition (MET) signalling. The tumour's propensity for vascular invasion and metastasis contributes to its poor prognosis, with the lungs, brain, and skeleton being the common metastatic sites. Standard chemotherapy is often ineffective, underscoring the importance of early diagnosis and complete surgical resection. This case underscores the diagnostic challenges and emphasises the need for molecular and immunohistochemical studies to distinguish ASPS from mimickers. Furthermore, it highlights the emerging role of targeted therapies and anti-angiogenic agents in improving outcomes for advanced or metastatic disease.


**Keywords:** Abdominal wall sarcoma, Children, Periodic Acid-Schiff staining

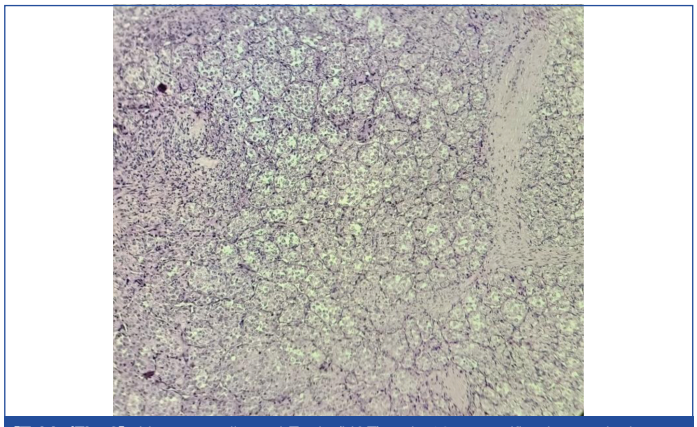
## CASE REPORT

An eight-year-old girl presented to the outpatient department in July 2024 with a one-month history of swelling on the left side of the anterior abdominal wall. Examination revealed a firm, approximately 4×3 cm swelling in the left anterior abdominal wall, below the level of the umbilicus. The swelling was non-tender, with no changes in the overlying skin. Her blood sample values were normal. A provisional diagnosis of a desmoid tumour was initially considered. However, Contrast-Enhanced Computed Tomography (CECT) of the abdomen showed a 4×3.5×2.5 cm intensely enhancing soft-tissue lesion with a central necrotic area in the left rectus abdominis muscle, suggesting a malignant soft-tissue neoplasm [Table/Fig-1]. The patient subsequently underwent an excision biopsy for further evaluation.



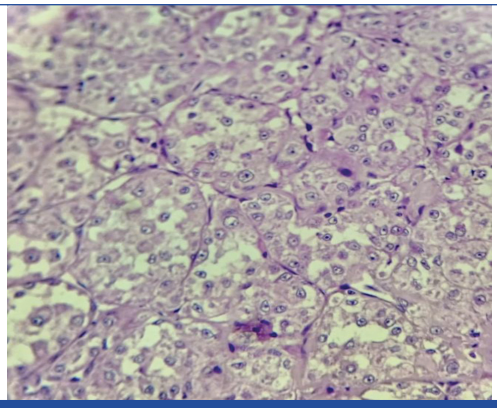
**[Table/Fig-1]:** Soft-tissue density lesion showing intense enhancement with necrotic area.

The specimen consisted of a single globular, grey-white to grey-brown soft-tissue mass measuring 3.2×2.5×1.5 cm, with a nodular, grey-white, and firm cut surface. Histological examination revealed a well-circumscribed tumour arranged in solid nests with organoid and pseudo-alveolar patterns. The tumour cells were polygonal, with eosinophilic granular cytoplasm, round nuclei, and prominent nucleoli [Table/Fig-2,3]. Dyscohesive cells within the nests created a pseudo-alveolar appearance, with thin-walled vascular spaces separating them. Mild atypia and atypical mitosis was observed, with mitosis occurring at a rate of 1-2 per 10 High-Power Fields (HPF), and necrosis was present in about 5% of the tumour. Tumour cells invaded adjacent skeletal muscle fibres, and vascular emboli were noted [Table/Fig-4].

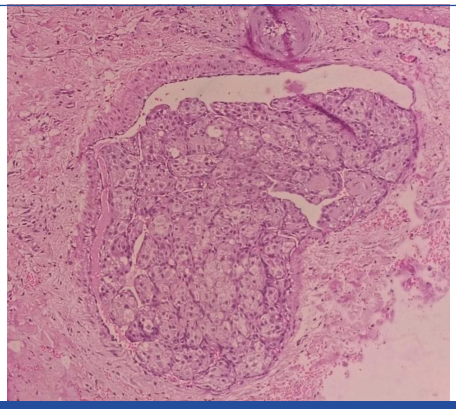


**[Table/Fig-2]:** Haematoxylin and Eosin (H&E) stain 10x magnification: typical organoid arrangement of tumor cell.

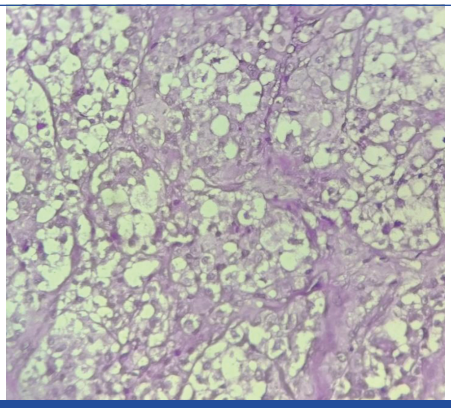
Based on the histomorphology, the differential diagnosis included paraganglioma, granular cell tumour, alveolar rhabdomyosarcoma, ASPS, and Perivascular Epithelioid Cell tumour (PEComa). PAS staining revealed positivity for intracytoplasmic crystals [Table/Fig-5]. IHC showed diffuse strong nuclear positivity for TFE3 in tumour cells [Table/Fig-6] and CD34 positivity in the vascular lining surrounding the tumour cells [Table/Fig-7]. S100 showed focal positivity in



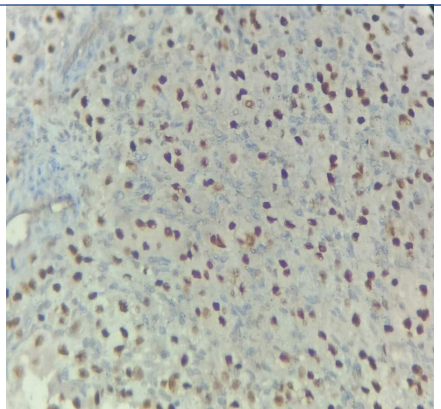
**[Table/Fig-3]:** 40x magnification- Solid nests are separated by thin walled vascular spaces central dyscohesion, resulting in pseudoalveolar pattern.



**[Table/Fig-4]:** 10x magnification shows vascular tumour emboli.



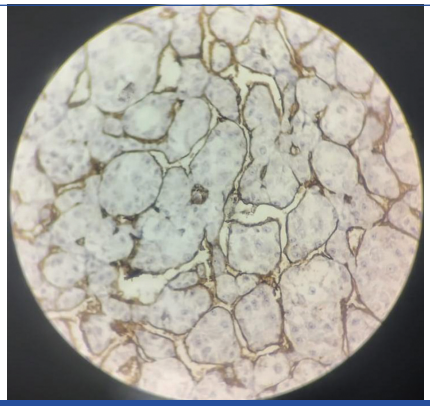
**[Table/Fig-5]:** 40x magnification, PAS special stain- Positive in intracytoplasmic crystals.



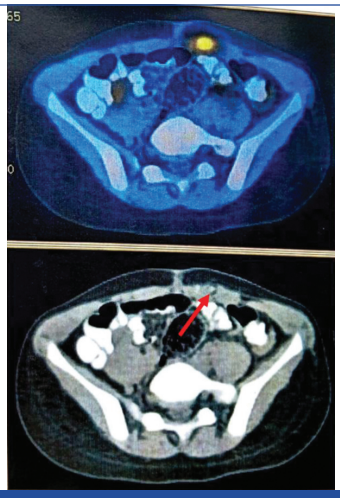
**[Table/Fig-6]:** 10x magnification -IHC TFE3 immunoreactivity in the nucleus of tumour cells.

tumour cells, while HMB45 and Desmin showed negative reactivity. The final diagnosis was ASPS.

After surgical resection, an Fluorodeoxyglucose (FDG) Positron Emission Tomography–Computed Tomography (PET-CT) was performed, showing subcutaneous post-procedural soft-tissue thickening in the left lower abdominal wall muscle at the level of the umbilicus [Table/Fig-8].



**[Table/Fig-7]:** 4x magnification-IHC CD34 positivity in endothelial cells around tumour nests.



**[Table/Fig-8]:** Non-FDG avid subcutaneous thickening in the left lower abdominal wall at the level of umbilicus.

## DISCUSSION

ASPS is a rare and distinct type of soft-tissue sarcoma with uncertain differentiation, accounting for less than 1% of all soft-tissue sarcomas [1]. It is associated with skeletal muscle and the musculofascial plane [2]. The most common sites include the thigh and buttock (39.5%), with other locations being the leg/popliteal region (16.6%), chest wall/trunk (12.9%), forearm (18.4%), back/neck (6.4%), tongue (3.2%), and retroperitoneum (3.2%) [3]. In children, cases have also been reported in the head and neck [4], with the tongue and orbit being the most common sites [5]. Primary pulmonary ASPS has been noted in a 25-year-old male without evidence of a soft-tissue tumour [6]. A diagnosis of primary pulmonary ASPS should exclude secondary ASPS [7].

ASPS is characterised by a specific translocation, der(17)t(x;17)(p11;q25), resulting in the ASPSCR1-TFE3 gene fusion [1]. Most studies have found a female preponderance in adult patients, but no such predilection has been noted in children [8]. Clinically, it presents as a soft, non-ulcerated, indolent, slow-growing mass [9], similar to the presentation in our case. It has characteristic Magnetic Resonance Imaging (MRI) findings: T1-weighted images show equal or slightly higher signals, whereas T2-weighted images show uneven high signals. A large number of thick and tortuous blood vessels are observed around the tumour. Due to rapid blood flow through these vessels, it presents as a flow void effect on MRI, a relatively characteristic feature of ASPS [9].

Essential diagnostic criteria include eosinophilic polygonal cells arranged in a nested or organoid pattern with rich sinusoidal capillaries and intracytoplasmic crystals, along with TFE3 nuclear expression detected by IHC [1]. Sometimes, ASPS may show a solid/compact pattern, especially in the head and neck of paediatric

patients [10]. The organoid appearance may sometimes be lost, and the tumour may instead comprise sheets of epithelioid cells. A minority of tumours show unusual features, such as myxoid changes, cystic changes, haemorrhage, and a prominent lymphocytic infiltrate [11]. While necrosis and haemorrhage may be notable, nuclear pleomorphism is typically minimal and focal, as is the level of mitotic activity. Significant vascular invasion, particularly involving dilated veins at the tumour's periphery, is frequently observed [12].

The location of ASPS significantly impacts diagnosis. Tumours occurring in the tongue should be distinguished from other conditions such as granular cell tumours, which primarily occur in middle-aged and elderly people. Granular cell tumours lack capillary networks and cytoplasmic glycogen in fibrous connective tissue. PAS staining is negative, while markers such as S100, SOX10, inhibin, and nestin are positive [13].

IHC is crucial for distinguishing ASPS from other tumours. Immunoreactivity for PAX8 is helpful in confirming renal cell carcinoma, as this antigen is absent in ASPS. In granular cell tumours, cells are less well-defined and exhibit distinct granular cytoplasm. Immunoreactivity for S100 protein and SOX10 aids in distinguishing granular cell tumours from ASPS [12]. ASPS is negative for epithelial markers, neuronal markers, and melanocytic markers [14].

ASPS generally has a poor prognosis due to its tendency to metastasise early, even when the primary tumour is asymptomatic or small. Its close differential diagnoses include metastatic renal cell carcinoma, paraganglioma, and granular cell tumour [12]. The ASPSCR1-TFE3 fusion protein localises to the nucleus, where it functions as an aberrant transcription factor, leading to c-MET overexpression and activation of the c-MET signalling pathway [1]. A similar translocation has been detected in a type of renal neoplasm, likely epithelial, which bears some morphological resemblance to ASPS [15].

The principal metastatic sites for ASPS are the lungs, followed by the brain and skeleton. Once diagnosed, these tumours are often indolent but frequently fatal, with survival rates decreasing from 87% at two years to only 18% at a 20-year follow-up [16]. The 5-year survival rate is around 60%, varying based on the stage and extent of the disease at diagnosis. Older age at presentation and tumour size greater than 10 cm are indicators of poor prognosis, whereas paediatric cases generally have a better outcome [16]. Paediatric follow-up is mandatory to detect metastasis [17].

The best treatment option for ASPS to prevent local recurrence is surgical excision with a tumour-free margin of 1.0-1.5 cm [14]. In the present case, the patient underwent surgical excision of the primary tumour with margin clearance and is on periodic follow-up.

For metastatic or advanced ASPS, novel treatment options are necessary, as the tumour is resistant to standard chemotherapy. Molecular-targeted therapies have emerged as promising strategies for ASPS [18]. Ongoing clinical trials are focussing on the overactivity of the MET receptor kinase gene induced by the ASPSCR1-TFE3 fusion protein. Additionally, the vascular nature of ASPS suggests a potential role for anti-angiogenic agents in its treatment [19].

## CONCLUSION(S)

This case report highlights the unique presentation and challenges associated with ASPS. Despite its indolent growth, ASPS has a propensity for late metastasis and resistance to conventional therapies. Early diagnosis, through a combination of imaging and histopathological evaluation, remains crucial for managing this rare malignancy. This case underscores the importance of considering ASPS in differential diagnoses, particularly in young patients with atypical soft-tissue masses. Ongoing research is needed to explore more effective treatment strategies and improve patient outcomes.

## REFERENCES


[1] World Health Organization. WHO classification of tumours editorial board. Soft-tissue and bone tumours. 5th ed. Lyon: International Agency for Research on Cancer; 2020. WHO Classification of Tumours Series, No 3.
[2] Sidi V, Fragandrea I, Hatzipantelis E, Kyriakopoulos C, Papanikolaou A, Bandouraki M, et al. Alveolar soft part sarcoma of the extremity: A case report. Hippokratia. 2008;12(4):251-53.
[3] Lieberman PH, Brennan MF, Kimmel M, Erlandson RA, Garin-Chesa P, Flehinger BY. Alveolar soft-part sarcoma: A clinicopathologic study of half a century. Cancer. 1989;63(1):01-13.
[4] Yam MK, Chan KK. Alveolar soft part sarcoma in a young woman: A case report from Hong Kong. Radiol Case Rep. 2022;17(6):1938-41.
[5] Oda T, Kikuchi K, Togao O, Baba S, Mizoguchi M, Tanabe M, et al. Alveolar soft part sarcoma of the orbit: A case report. Radiol Case Rep. 2021;16(12):3766-71. Doi: 10.1016/j.radcr.2021.09.005.
[6] Kaur K, Gami A, Shah A, Gandhi J, Trivedi P. Clinico-pathological spectrum of alveolar soft part sarcoma. Turk J Pathol. 2024;40(2):89-100.
[7] Xu Z, Wang J, Mao J, Xu D, Wu L, Zhou Y, et al. Primary alveolar soft-part sarcoma of the lung in a child. Front Surg. 2023;10:927597. Doi: 10.3389/fsurg.2023.927597.
[8] Singh A, Gupta S, Ghosh S, Yuwanati MB. Alveolar soft part sarcoma. BMJ Case Rep. 2014;2014:bcr2013203386. Doi: 10.1136/bcr-2013-203386.
[9] Wang B, Wang H, Wei J, Qu L, Meng L, Xin Y, et al. Alveolar soft part sarcoma of the right calf: A case report. Medicine. 2020;99(5):e18952. Doi: 10.1097/MD.0000000000018952.
[10] Mishra S, Dey AK. Understanding and identifying 'themes' in qualitative case study research. South Asian J Bus Manag Cases. 2022;101(5):187-92. Doi: 10.1177/22779779221134659.
[11] Folpe AL, Deyrup AT. Alveolar soft-part sarcoma: A review and update. J Clin Pathol. 2006;59(11):1127-32. Doi: 10.1136/jcp.2005.031120.
[12] Enzinger FM, Weiss SW, editors. Enzinger and Weiss's soft-tissue tumours. 7th ed. Philadelphia: Elsevier; 2020.
[13] Gong Y, Liu M, Wu Q, Liu Y, Zhang J, Yao L, et al. Alveolar soft part sarcoma of the tongue: A case report and review of the literature. Int J Clin Exp Pathol. 2020;13(5):1275-82.
[14] Rajan PS, Chauhan S, Singaram M, Subramaniyann A, Ahmad S. Alveolar soft part sarcoma of the tongue: A case report of a rare entity. Adv Oral Maxillofac Surg. 2021;4:100121. Doi: 10.1016/j.adoms.2021.100121.
[15] Rosai J. Ackerman's Surgical Pathology. 11th ed. Philadelphia: Elsevier; 2018.
[16] Fletcher CDM, Bridge JA, Hogendoorn PCW, Mertens F, editors. WHO classification of tumours of soft-tissue and bone. 4th ed. Lyon: IARC Press; 2013. WHO Classification of Tumours Series, Volume 5.
[17] Ata K, Farsakh H, Rjoop A, Matalka I, Rousan L. Alveolar soft part sarcoma of the extremity: Case report and literature review. World J Oncol. 2014;5(5):01-05. Available from: https://wjon.org/index.php/wjon/article/view/777/549.
[18] Soheilifar MH, Taheri RA, Emameh RZ, Moshtaghian A, Kooshki H, Motie MR. Molecular landscape in alveolar soft part sarcoma: Implications for molecular targeted therapy. Biomed Pharmacother. 2018;103:889-96.
[19] Mitton B, Federman N. Alveolar soft part sarcomas: Molecular pathogenesis and implications for novel targeted therapies. Sarcoma. 2012;2012:428789. Doi: 10.1155/2012/428789.



**PARTICULARS OF CONTRIBUTORS:**
1. Postgraduate Student, Department of Pathology, Institute of Child Health and Hospital for Children, Chennai, Tamil Nadu, India.
2. Professor and Head, Department of Pathology, Institute of Child Health and Hospital for Children, Chennai, Tamil Nadu, India.
3. Assistant Professor, Department of Pathology, Institute of Child Health and Hospital for Children, Chennai, Tamil Nadu, India.

**NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:**
Dr. Umadevi Srinivasan,
No. 42/47, First Floor, Dewan Raman Avenue Road, Purasaiwakam,
Chennai-600084, Tamil Nadu, India.
E-mail: drumadevisrinivasan@gmail.com



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