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Case Report

Alveolar soft part sarcoma–presenting as an unusual scapular swelling- A rare case report

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ABSTRACT

Alveolar soft part sarcoma (ASPS) is a very rare soft tissue tumor of unknown histogenesis and has propensity for recurrence and distant metastasis. It commonly occurs in the head neck region and on extremities in adolescents and young adults. Less number of cases have been reported in the scapular region so far. Till now, no definite treatment regime is available for ASPS; early diagnosis plays a pivotal role in its management and patient outcome. Here, we are presenting a case of 23 years old male presenting with a recurrent swelling in right scapular region. It was diagnosed as ASPS on the basis of radiology, histomorphology and immunohistochemistry.

1. Introduction

Alveolar soft part sarcoma (ASPS) is a very rare soft tissue neoplasm of unknow histogenesis with poor prognosis and accounts for 0.5%-1% cases of all soft tissue sarcomas.[1,2] It shows a very peculiar histological appearance (alveolar pattern) and cell morphology. Its exact nature of origin is still uncertain though few immunohistochemistry analyses showed its skeletal muscle origin.[3] ASPS is more common in females and occurs in adolescents and young adults.[3] It arises in deep soft tissue of upper and lower extremities, head neck region, trunk, mediastinum, retroperitoneum, and genital tract. In children, favorite site is head neck region particularly orbit and tongue. While in adults it occurs extremities.[4–6] This tumor generally shows indolent behavior but can metastasize to distant places like lungs, bone and brain.[5,7] Radiographically; it is soft tissue mass in deep tissue with foci of calcification or necrosis in larger lesions.

On computed tomography (CT) it appears as hyperdense vascular soft tissue mass with heterogeneous enhancement on contrast. CT angiography shows a hyper vascular mass with multiple connecting feeding vessels in the tumor.[8,9]

Surgery is the treatment of choice in localized tumor and surgical resection free margins show good prognosis. It shows poor response to chemotherapy.[9] Being a rare entity, awareness about its histomorphology and clinical feature would help in early diagnosis and management of patient. Tissue biopsy and immunohistochemistry are required for confirmatory diagnosis.

2. Case Report

Here we are discussing a case of a 23 years old male who presented at our hospital; with complaint of recurrent swelling in right scapular region. He developed small swelling in scapular region 5 year ago which was painless and gradually progressive. He got operated 2 years back at a peripheral hospital and no histopathology record was available. Patient was asymptomatic for 6-7 months after

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surgery; thereafter he again developed a swelling at the same site. The swelling was slowly increasing in size measuring 9 x 8.5 cm, well defined, firm, non-tender with restricted mobility. His general condition was unremarkable.

The CT thorax (Figure 1A) revealed a heterogeneously enhancing soft tissue lesion in suprascapular and posterior scapular region on right side with cortical thickening and irregularities of underlying scapula. Bilateral lung fields and mediastinum was unremarkable. No lymphadenopathy was noted. Visceral organs; liver kidney and urogenital tract were also unremarkable.

Fine needle aspiration cytology smears showed ill-defined cellular clusters of pleomorphic atypical cells having hyperchromatic nuclei with cytoplasmic microvacuoles along with few multinucleated giant cells and mitotic figures. It was suggestive of soft tissue tumor; our differentials were rhabdomyosarcoma, clear cell sarcoma and ASPS.

Later we received wide local excision (WLE) specimen of lesion with attached skin flap. (Figure 1B) The outer surface was rough and congested, cut surface showed solid greyish white growth with infiltrative borders. Growth was multinodular, largest measuring was 8.0 x7.0 x3.5cm with focal hemorrhagic spots. Histology sections showed malignant soft tissue neoplasm displaying alveolar and nodular growth pattern as dyscohesive tumor cell nests separated by thin fibro vascular septa. The tumor cells were uniform in size, polygonal or round, with well- defined borders and had centrally located bland looking nucleus with prominent nucleoli. Binucleation, nuclear atypia, and atypical mitotic figures were occasional. The cytoplasm was abundant eosinophilic and granular. In some area of tumor cells also have cytoplasmic clearing. Lympho-vascular tumor emboli were frequent and focal areas of necrosis along with hemorrhage were also evident (Figure 2A-D). All surgical resection margins were clear.

Our histological differentials were, ASPS, alveolar rhabdomyosarcoma, clear cell sarcoma, melanoma, paraganglioma, granular cell tumor and metastasis from renal cell carcinoma. Periodic acid Schiff's (PAS) stain after diastase digestion was done, which displayed magenta pink colored intracytoplasmic granules. Immunohistochemistry of tissue sections displayed diffuse cytoplasmic positivity for vimentin and focal positivity for Desmin in tumor cells. Myogenin was negative as it showed cytoplasmic positivity. CK, S100, Myogenin Melan-A HMB45 chromogranin and synaptophysin were negative in tumor cells (Figure 3A-D). On the basis of presenting age, clinic-radiological characteristic, histomorphology and immunohistochemical features, it was diagnosis as Alveolar soft part sarcoma - right scapular region.

1.

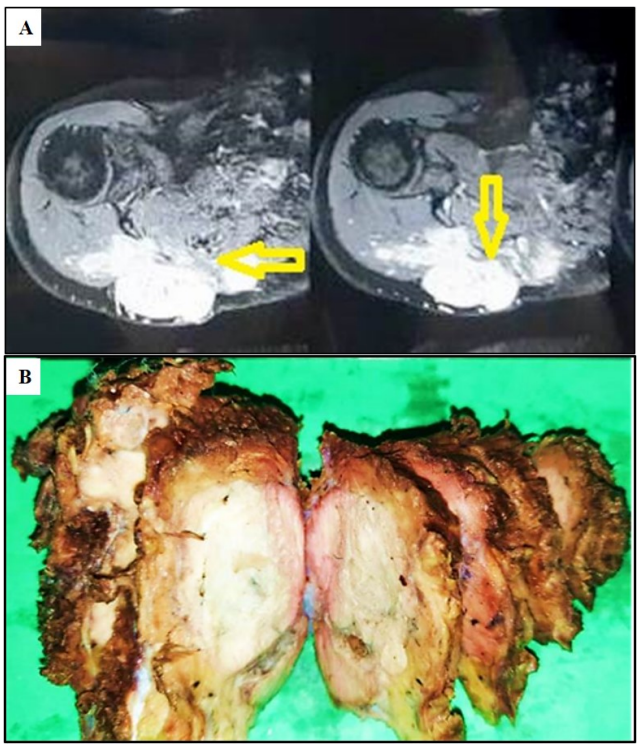

Fig. 1: Alveolar soft part sarcoma: **A:** CT Scan showing heterogeneously enhancing soft tissue lesion in the right scapular region; **B:** Grey white, firm and multi nodular growth.

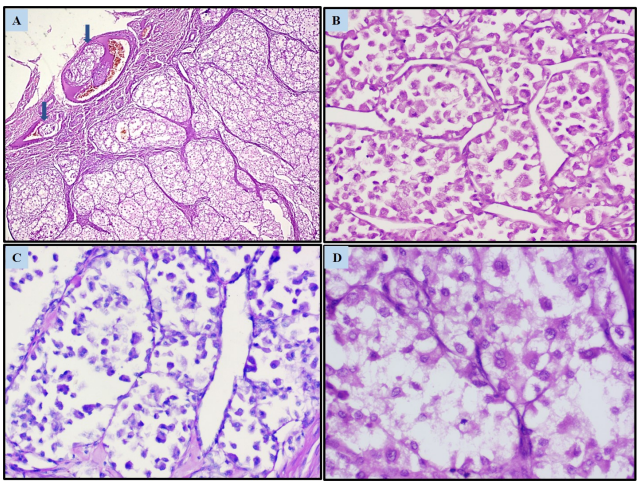

Fig. 2: Alveolar soft part sarcoma; **A:** tumor cells arranged in alveolar pattern and peripheral area with multiple vascular tumor emboli (arrow) H&E x 40X; **B:** Large oval to polygonal cells separated by thin vascular septa H&E x 200X; **C:** Tumor cells showing pas positive cytoplasmic granules. PAS x 200X; **D:** Mildly pleomorphic cells with binucleation and vesicular nucleus, prominent nucleoli and granular to clear cytoplasm. H&E x 400X.

Table 1: Review of literature of ASPS in scapula region.

Author	Year	Salient features Age (years) /Sex	Radiological features(CT,MRI, Tc 99)	Histological features
Demir H. et al.[10]	2002	46yrs/M	CT-Distruction of right scapula MRI-Contrast enhancing mass invading infraspinatus muscle TC99 Scan- A hypoactive area with an irregular hyperactive rim in the inferolateral portion of the right scapula.	Large and polygonal cells with distinct cell borders, and abundant eosinophilic cytoplasm. IHC-Vimentin positive S100, CK, Desmin, Actin (Negative)
Ohshika S et al.[11]	2012	33yrs/M	Heterogeneous mass with intensive invasion to the scapula with multiple bilateral lung metastases.	Large tumor cells with abundant eosinophilic and granular cytoplasm proliferated in an alveolar pattern, separated by thin sinusoidal vessels.
Yavuz, A. et al.[12]	2013	32 yrs/M	Expanded and disrupted bony cortex of scapula	Polygonal cells with granular cytoplasm, prominent nucleoli and alveolar structure, PAS positivity
Sahraoui, G et al.[13]	2017	25yrs/M	Heterogeneous mass within the left scapula with destruction of the bone cortex and local expansion to adjacent muscle, bone and joint structures.	Tumor cells arranged in a nesting or pseudo-alveolar pattern of large epithelioid cells with abundant eosinophilic granular cytoplasm and hyperchromatic vesicular nuclei having prominent nucleoli. IHC- Desmin, TFE3 (Positive) and Myogenin and CK (Negative)
Present case	2021	23yrs/M	Heterogeneously enhancing soft tissue lesion in right suprascapular and posterior scapular region with cortical thickening and irregularities of underlying scapula bone.	Tumor cells displaying alveolar and nodular growth pattern & separated by thin fibro vascular septae having granular cytoplasm and hyperchromatic nuceli. IHC-Vimentin (Diffuse Positive), Desmin (Focal Positive) Myogenin, S100, HMB45, CK (Negative)

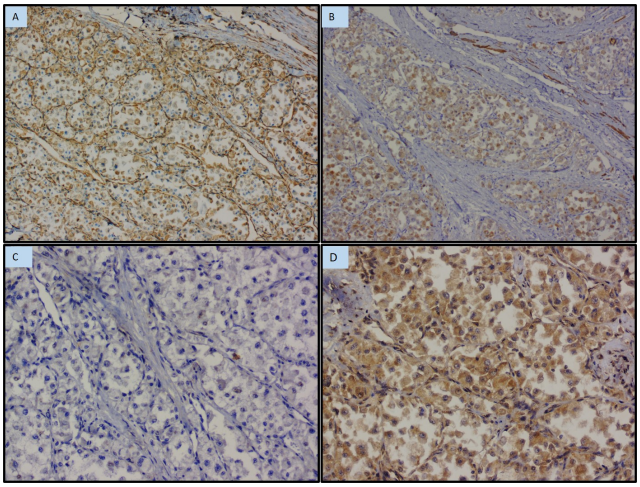

Fig. 3: Immunohistochemistry images of Alveolar soft part sarcoma; **A:** vimentin positive in tumour cells x 100X; **B:** Desmin faint positive in tumor cells x 100X; **C:** Cytokeratin negative in tumor cells x 200X; **D:** Myogenin cytoplasmic positivity in tumor cells x200X.

3. Discussion

ASPS is a very rare soft tissue neoplasm arising from deep connective tissue. It was first described by Christopherson et al.in 1952.[14] Thereafter it is described as case report or case series in the literature. It has characteristic alveolar pattern on histology hence also known as alveolar soft tissue sarcoma. Theses tumors are very vascular and have propensity to metastasis to distance places. It has uncertain line of differentiation. Some cases of ASPS showed myogenic differentiation and researchers consider as it has myogenic origin. But only cytoplasmic positivity with myogenin or myo-D1 has been detected so, this idea has been also discarded.[3,7]

According to genetic theory it is caused by a specific unbalanced translocation der(17) t(x:17)(p11; q25) and produces ASPL-TFE3 fusion gene. It can be detected by either molecular genetics or immunohistochemistry analysis using an antibody directed against the C-terminus of the transcription factor E3 (TFE3) gene, it gives strong nuclear staining and now, it is used as a specific marker for ASPS.[9,11]

Clinically it presents as slow growing painless mass in deep soft tissue of extremities and head-neck region. Our case was presented as recurrent painless swelling in scapular region which was an unusual site for ASPS. Very few cases have been reported in scapular region. Authors found that tumor presented with bony erosion and poorly defined tumor margins [10–13] and their relevant findings were summarized in Table 1.

Clinical differential of ASPS are schwannoma, hemangioma and soft tissue sarcoma. Grossly it is well demarcated lesion with size ranging from 3-14 cm. Cut surface is pale grey to yellowish in color and soft to firm in consistency. Some tumor shows focal area of necrosis and hemorrhage. [4,5,7]

Microscopic feature is very characteristic displaying alveolar pattern in a which nest of tumor cells is separated by thin fibrovascular septa. The tumor cells are line the wall of septa and relatively monomorphic round to polygonal in shape, with distinct cell borders, centrally placed nuclei with prominent nucleoli and moderate amount of eosinophilic granular or clear cytoplasm. Sometimes it resembles renal cell carcinoma. Tumor cell shows strong PAS (Periodic Acid Schiff's) positive diastase resistant cytoplasmic granules. Vascular invasion is very common findings in these tumors. Pathological differential of ASPS is alveolar rhabdomyosarcoma, clear cell sarcoma, melanoma, paraganglioma, granular cell tumor, metastasis from renal cell carcinoma and adrenal cortical carcinoma. Immunohistochemistry helps to separate ASPS from these differentials. It shows positive expression for vimentin, TFE3 and negative for S-100, HMB-45, Melan-A, synaptophysin, chromogranin, desmin myogenin and cytokeratin. [3,4,9]

Strong cytokeratin expression with expression of site-associated markers like renal cell carcinoma antigen in renal cell carcinoma, Melan-A positivity in adrenal cortical carcinoma and clinical correlation as to the presence of a renal mass or adrenal mass will help in this differential diagnosis. Malignant melanoma can mimic ASPS; but can be easily differentiated by positive staining for S100, HMB-45 and Melan -A. Paraganglioma, unlike ASPS, show strong expression with neural marker, chromogranin A and synaptophysin. Granular cell tumor can express TFE3 and simulates ASPS but lacks cytological atypia and show strong S100 protein expression. Alveolar rhabdomyosarcoma, despite its somewhat similar name and have alveolar pattern but it shows strong expression of desmin and myogenin nuclear regulatory proteins and negative for TFE3.

ASPS is a malignant soft tissue tumor with indolent course the high frequency of metastasis and have overall poor survival. [8,9,11] Portera et al[5] reported better outcome in localize disease; 5-year disease-free survival of 71% in patients presenting localized disease as compared with only 20% in patients presenting with metastases. For ASPS prognostic risk factors are age >10 years, tumor size >5cm and presence of distant metastases. [7,9,12,13]

Surgical excision remains the primary modality of treatment for ASPS. In advance disease, it may be followed by adjuvant therapy. [7,9]

4. Conclusion

ASPS is a rare slow growing soft tissue tumor and has propensity to distant metastasis with high mortality. It has a peculiar histological and immunohistochemical features which help in diagnosis. We should keep it as differentials in lesions arising in deep soft tissue of extremities and head-neck region in adolescents and adults.

5. Source of Funding

None.

6. Conflicts of Interest

None.

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