Section: Pathology

Case Report

MALIGNANT PERIVASCULAR EPITHELIOID CELL TUMOR: A RARE CASE REPORT WITH REVIEW OF LITERATURE

Archana Shinde[1], Srushti Patel [2], Pramod Shinde[3], Ruchita Patel[4] Manisha Raut[4]

[1]*Associate Professor, Department of Pathology, NAMO Medical Education and Research Institute, Silvassa, Dadra and Nagar Haveli, India*
[2] *Tutor, Department of Pathology, NAMO Medical Education and Research Institute, Silvassa, Dadra and Nagar Haveli, India*
[3]*Professor and Head, Department of Pathology, NAMO Medical Education and Research Institute, Silvassa, Dadra and Nagar Haveli, India*
[4]*Assistant Professor, Department of Pathology, NAMO Medical Education and Research Institute, Silvassa, Dadra and Nagar Haveli, India*

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Corresponding Author:
Dr. Shrusti Patel,
Tutor, Department of Pathology, NAMO Medical Education and Research Institute, Silvassa, Dadra and Nagar Haveli, India.
Email: docsrushti@gmail.com

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ABSTRACT

Background: Perivascular epithelioid cell neoplasms (PEComa) are a family of mesenchymal tumors characterized by perivascular epithelioid cells with co-expression of melanocytic and muscle markers and occur most common in the abdominal pelvic location. Malignant PEComa are exceptionally uncommon in the pediatric population, and widespread metastatic disease with bone marrow involvement in exceedingly rare.
Case Report: A 9 years old girl presented with left axillary mass since 1 year. On examination, left axilla showed a mass measuring 5x4 cm. USG revealed heterogeneously enhancing hyper-hypoechoic nodal mass. FNAC examination showed High Grade Malignant Tumor.Core biopsy with immunohistochemistry was done for definite diagnosis.
Results: Histopathological findings showed sheets and solid nests of atypical cells having abundant eosinophilic cytoplasm, round central nucleus, inconspicuous nucleoli. Mitosis are seen (8/10 HPF). Immunohistochemistry revealed tumor cells are positive for CD68, S100, HMB45, H-Caldesmon, SMA. Ki67 is 80%.The combined morphological and immunophenotypic findings supported a diagnosis of malignant PEComa. **Conclusion:** This case highlights an exceptionally rare occurrence of malignant PEComa in a child, presenting as an axillary soft tissue mass with aggressive clinical behaviour, extensive multisystem metastases and bone marrow involvement. Awareness of this entity and recognition of it's characteristic dual melanocytic and smooth muscle immunophenotype are crucial for accurate diagnosis and distinction from other pediatric high grade epithelioid malignancies.
Keywords: PEComa,Malignant mesenchymal tumor,Bone marrow Metastasis, HMB45.

INTRODUCTION

Perivascular epithelioid cell tumors (PEComa) are a group of mesenchymal tumors that classically composed of distinctive cells that show a focal association with blood vessel walls and usually express melanocytic and smooth muscle markers.These tumors exhibit an epithelioid appearance, with clear or granular acidophilic cytoplasm and perivascular distribution, had an immunoreaction with melanocytic markers and were the most sensitive to human melanoma black 45(HMB45). [1,2]

They are rare mesenchymal tumors, presenting in numerous anatomic sites including lung,kidney,liver, genitourinary tract, soft tissue and skin. [3] PEComa are more frequent in females than males (M:F ratio: -0.2:1), with a wide age range and a peak in young to middle-aged adults (mean age: 45 years). [1]

Their biological behaviour ranges from benign or indolent lesions with relatively good prognosis, but a minority of them have malignant potential with few criteria, such as tumor size >5cm, mitotic activity

1/50 hpf, cell necrosis, nuclear pleomorphism and an infiltrative growth pattern.

We report an unusual case of malignant PEComa arising in the axillary region of a 9 year old girl with extensive metastatic disease involving the lung and pleura, skeleton, liver, muscle, cervical lymph nodes and bone marrow. The rarity of this tumor in childhood, unusual primary site and widespread systemic and bone marrow involvement make this case diagnostically and clinically noteworthy.

CASE REPORT

A 9 years old girl presented with no significant medical history, complaining with left axillary mass since 1 year. No additional relevant medical, family, genetic or psychological history was available at the time of reporting. On examination, left axilla showed a mass measuring 5x4 cm. Overlying skin was stretched and shows focal ulceration. [Figure 1]

Fine needle aspiration cytology(FNAC) of the lesion revealed singly scattered and loose clusters of large round to polygonal cells with large prominent nucleoli and pale / clear cytoplasm with many mitotic figures. Based on these cytomorphological features, a diagnosis of High Grade Malignant Tumor was rendered.[Figure 2].

Computed tomography (CT) showed heterogeneously enhancing soft tissue density lesion with loss of fat plane / infiltration in adjacent pectoralis major muscle.The radiological differential diagnoses included a primary soft-tissue sarcoma, particularly rhabdomyosarcoma, and a metastatic malignant neoplasm.

A core biopsy of a lesion showed sheets and solid nests of atypical cells having abundant eosinophilic cytoplasm, round central nucleus, inconspicuous nucleoli.Mitosis are seen (8/10 HPF). [Figure 3]

Immunohistochemical examination: tumor cells coexpress smooth muscle and melanocytic markers i.e. positive for CD68, S100, HMB45, H-Caldesmon, SMA and Negative for CK, CD20, CD30, CD34, ALK, SALL4, EBV, TFE3. Ki67 is 80%. [Figure 4]

In view of the characteristic epithelioid morphology and immunohistochemical coexpression of melanocytic and smooth-muscle markers, the findings were consistent with a diagnosis of **malignant perivascular epithelioid cell tumor (malignant PEComa).**

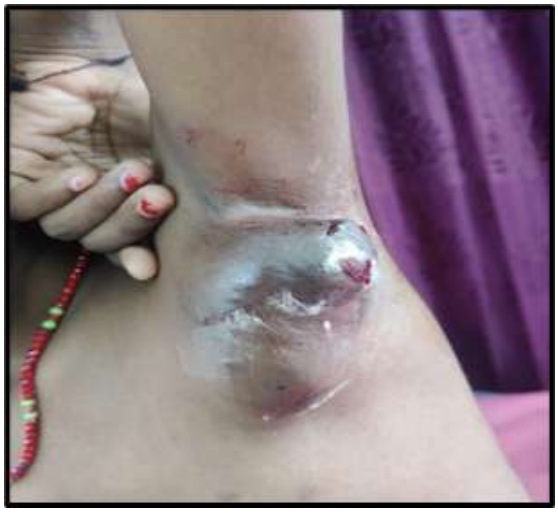

Figure 1: left axilla having large fungting mass

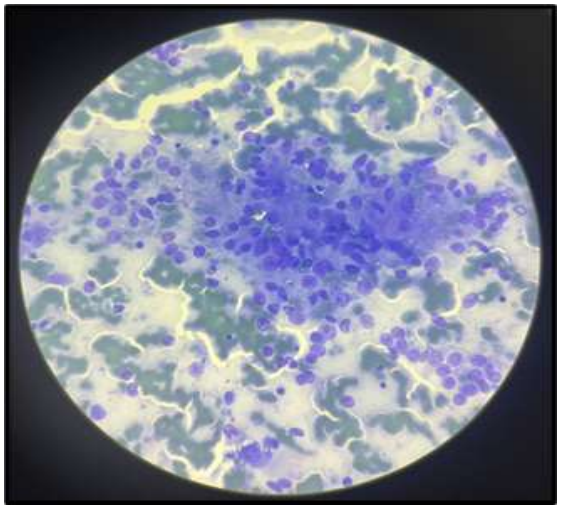

Figure 2: large round to polygonal cells with large prominent nucleoli and pale / clear cytoplasm

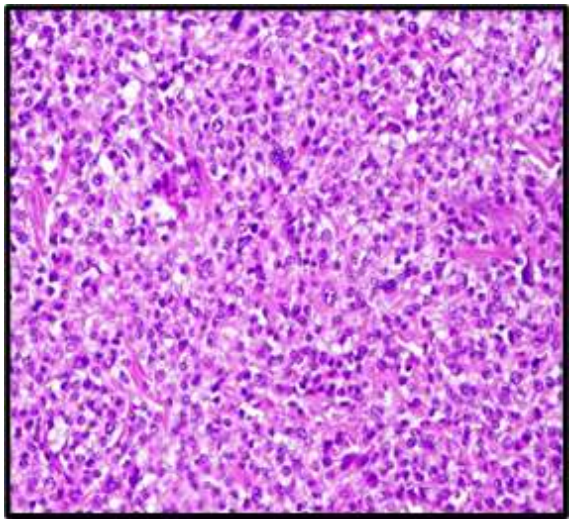

Figure 3: sheets of atypical cells having abundant eosinophilic cytoplasm, round central nucleus, inconspicuous nucleoli

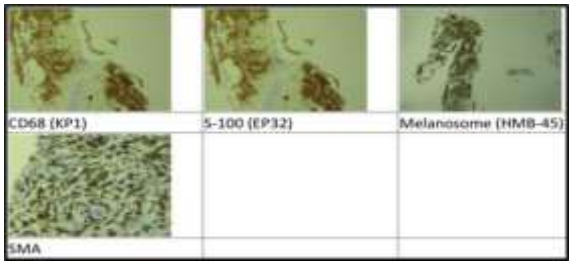

Figure 4: immunohistochemistry shows tumour cells are positive for smooth muscle and melanocytic markers, I.e. HMB45, SMA.

Whole-body positron emission tomography–computed tomography (PET-CT), performed for disease staging, demonstrated extensive metastatic disease involving multiple organ systems. The scan revealed multiple discrete and coalescent bilateral pleuro-pulmonary lesions, numerous lytic sclerotic skeletal lesions, hepatic lesion,few muscle plane deposits and few left lower neck nodes. **Bone marrow aspiration and trephine biopsy** shows metastatic deposits of morphologically similar to the primary axillary tumor, confirming bone marrow involving by malignant PEComa.

Overall, the clinical, radiological, histomorphological, and immunohistochemical findings were consistent with **malignant PEComa of the left axillary region with widespread systemic metastases.**

Differential Diagnosis

The major differential diagnoses considered in this case included rhabdomyosarcoma, clear cell sarcoma, epithelioid sarcoma, anaplastic large cell lymphoma, metastatic carcinoma, and other high-grade epithelioid sarcomas.

DISCUSSION

Although perivascular epithelioid cell neoplasms were first described in 1943 by Apitz,[5] (as an "abnormal myoblast" in renal angiomyolipoma; it was in 1996 that Zamboni et al. in their first case of pancreatic clear-cell "sugar" tumor (CCST) suggested the name PEComa for neoplasms composed of a pure proliferation of perivascular epithelioid cells.[6]

As per the World Health Organization classificationy of tumors of soft tissues and bones, perivascular epithelioid cell neoplasms are classified as "Tumor of uncertain differentiation" with a make-up of perivascular epithelioid cells.[7]

PEComas occasionally associated with tuberous sclerosis complex, are defined by the presence of perivascular epithelioid cells that co-express muscle and melanocytic markers. This family of tumors includes angiomyolipoma (AML), clear cell sugar tumor of the lung (CCST), lymphangioleiomyomatosis (LAM), and very rare tumors in other locations,[8]

Histologically, PEComas typically show a nested architecture and are composed of epithelioid cells with abundant granular eosinophilic or clear cytoplasm and round nuclei with small nucleoli. The nests or trabeculae are typically surrounded by thin-walled capillary vessels. [1] Immunohistochemically angiomyolipomas, lymphangiomyomas, and related tumors are distinctive and co-express markers of smooth muscle (consistently SMA-positive, variably desmin-positive) and melanocytes (HMB-45, Melan-A, MART). Therefore, such a constellation of markers is useful in the diagnosis of angiomyolipoma-related mesenchymal tumors, especially when they occur in an unusual clinical setting.[9,10]

The molecular pathogenesis of PEComa commonly involves alteration of tuberous sclerosis complex genes TSC1 or TSC2 gene. small subset of PEComas harbour TFE3 gene fusions, which correlate with strong nuclear immunoreactivity for TFE3.[1]

The present study being a clinopathology based case report study.It provides a primary baseline tool to work up for future case based targeted studies on PEComas. It also served the purpose of providing basic clinopathological data which can be compared with available studies in the literature.

Table 1: comparison of clinopathological features of PEComa of present study with other studies

	J. Zhong, Y. Hu, L. Si et al. (2020),[11]	R.Freeman and E. Kelley et al. (2022),[12]	S.Mah, L.Hoang, S. Lee et at.(2023),[13]	Tahiri Elousrouti et al (2023),[14]	Present Study
Age/ Sex	33 years/Female	35 years/ Male	37years/Female	92 years/ female	9 years/ female
site	Right distal humerus	Renal	cervix	right thigh	left axillary mass
size	11.8 cm	12 cm	8 mm	7 cm	5 cm
presentation	Pain	left upper abdominal and flanky pain	Intermittent postcoital bledding since 1 year	Exophytic, ulcerated, hemorrhagic nodular tumor rapidly growing for 8 months over the right thigh	left axillary mass since 1 year
radiological findings	expansile osteolytic lesion with destruction of the cortex and soft tissue expansion	complex fatty lesion involving upper pole of left kidney posterolaterally measuring 5x4xcm	No any preoperative investigations done.	heterogeneously enhancing, extra-aponeurotic, budding cutaneous mass	heterogenously hyper-hypoechoic lesion with minimal internal vascularity

		consistent with angiomyolipoma.			
histological features	Epithelioid cells morphology	Epithelioid cells morphology	Epithelioid cells morphology	Epithelioid cells morphology	Epithelioid cells morphology
IHC markers	HMB45, SMA, desmin and caldesmon positive and were weakly positive for Melan-A	HMB45, Melan A, Desmin Positive	Vimentin, Desmin, ER, HMB45, melan-A, caldesmon	melan A, SMA	CD68,S100,HMB45,H-Caldesmon, SMA
Prognosis	Malignant	Malignant	Benign	Malignant	Malignant

CONCLUSION

Malignant PEComa is an exceptionally rare mesenchymal neoplasm in children and may pose considerable diagnostic difficulty because of its morphological overlap with other pediatric high grade malignant tumor. Awareness of this entity and recognition of it's characteristic dual melanocytic and smooth muscle immunophenotype are crucial for accurate diagnosis and distinction from other pediatric high grade epithelioid malignancies.

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