e-ISSN: 0975-5160, p-ISSN: 2820-2651

Available online on www.ijtpr.com

International Journal of Toxicological and Pharmacological Research 2024; 14(9); 07-11

Original Research Article

Labial Tumescence: A Rare Diagnostic Encounter with Epithelioid Sarcoma

Pratiksha Mishra[1], Meenakshi Mohapatro[2], Kalyani Prava Gouda[3], Chinmayee Lenka[4], Sanghamitra Patra[5], Sampada Mohanty[6], Kambala Divya Teja[7], Lity Mohanty[8]

[1]Junior Resident, Department of Pathology, SCB Medical College and Hospital
[2]Assistant Professor Department of Pathology, SCB Medical College and Hospital
[3]Professor Department of Pathology, SCB Medical College and Hospital
[4]Assistant Professor, Department of Pathology, SCB Medical College and Hospital
[5]Senior Resident, Department of Pathology, SCB Medical College and Hospital
[6]Senior Resident, Department Of Pathology, SCB Medical College and Hospital
[7]Junior Resident, Department of Pathology, SCB Medical College and Hospital
[8]Professor and HOD, Department of Pathology, SCB Medical College and Hospital

Received: 18-06-2024 / Revised: 21-07-2024 / Accepted: 26-08-2024
Corresponding author:Dr. Meenakshi Mohapatro
Conflict of interest: Nil

Abstract:
Epithelioid sarcoma is a very rare aggressive soft tissue neoplasm comprising less than 1% of all vulval neoplasms. Median age of presentation is usually 34 years almost similar to our case. It is characterized tumor cells having typical epithelioid morphology. Histopathology and immunohistochemistry (IHC) are essential in making a final diagnosis. SMARCB1 is presently helpful for making an accurate diagnosis for adequate management. Herein we present a very rare case of epithelioid sarcoma of vulva in a 27-year-old patient.
Keywords: Labial Tumescence, Epithelioid Sarcoma.

Introduction

Epithelioid sarcoma (ES) is a very rare, extremely aggressive soft tissue malignancy first introduced by Enzinger in 1970 [1]. Proximal type ES tends to involve the deeper tissues soft tissues of the external genital tract. However distal (conventional) types involves the extremeties, especially hand. “Hybrid subtype” is referred when both the proximal and distal involvement is seen [2]. Herein we present a rare case report of a 27 year female diagnosed as epithelioid sarcoma, proximal type.

Case Report

A 27-year-old female presented with a gradually increasing right labial swelling of 3cm in diameter associated with mild itching since 6 months. She had no other comorbidities. There was no family history of any malignancy. Her past surgical history was also insignificant. On clinical examination, a firm, non-tender, non-ulcerative swelling measuring 3cm in diameter was present over the right labia major. No significant skin redness or signs of discharge was seen. Comprehensive blood examinations, laboratory tests were unremarkable. With the clinical diagnosis of malignancy, excisional biopsy was planned and the tissue was sent for histopathological examination. Grossly, skin attached tissue measuring 3cm in diameter was received. Cut section showed solid, yellowish white homogenous areas. Microsection showed normal keratinized stratified squamous epithelium with underlying tumor tissue (Figure 1a). Tumor cells were arranged in diffuse pattern separated by fibrovascular septa. Individual tumor cells were round to polygonal in shape having moderate amount of eosinophilic cytoplasm, round to oval to spindeloid nucleus having vesicular chromatin and prominent nucleoli (Figure 1b). Both typical and atypical mitosis were found, 8/10HPF. Plenty of myxoid areas in between the tumor cells and necrosis was seen. There were presence of numerous multinucleated tumor giant cells. With the suspicion of malignant mesenchymal tumor, immunohistochemistry (IHC) was done. On IHC, tumor cells showed positivity for vimentin (Figure 2a), epithelial membrane antigen (EMA) (Figure 2b) and negative for cytokeratin-7, cytokeratin-20(CK20), carcinoembryonic antigen (CEA) and S100. IHC and combined negative clinical examination findings and imaging studies helped us

rule out other mucinous neoplasm of labia. So the final diagnosis of epithelioid sarcoma was made. Further the patient was advised for cytogenetic study, for the loss of INI1/SMARCB1 correlation. The analysed cells presented with heterozygous deletion of 22q11.2 gene locus (Figure 3) and hence the specimen was reported as positive for SMARCB1 (INI1) gene deletion.

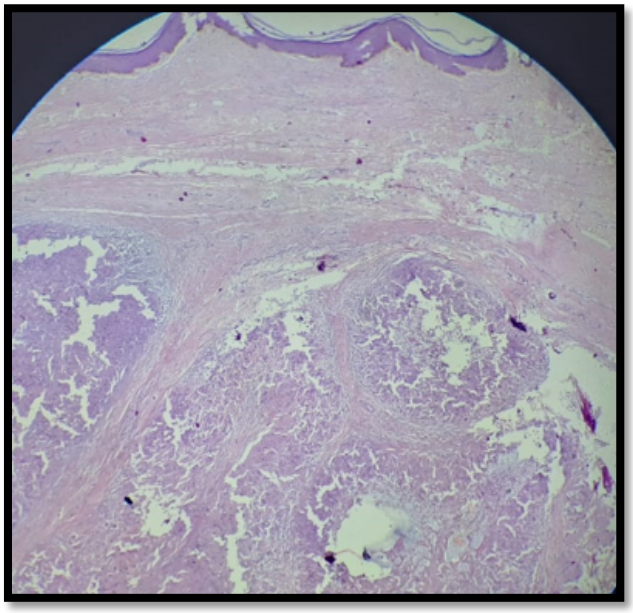

Figure 1a: Microsection shows keratinized stratified squamous epithelium, with underlying tumor tissue admixed with myxoid areas. H&E (40X)

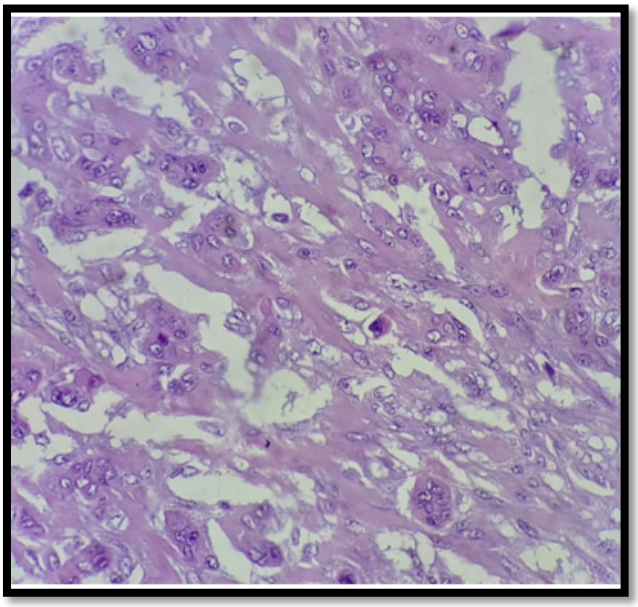

Figure 1b: Individual cells are round to polygonal with moderate eosinophilic cytoplasm, vesicular chromatin and prominent nucleoli. H&E (400X)

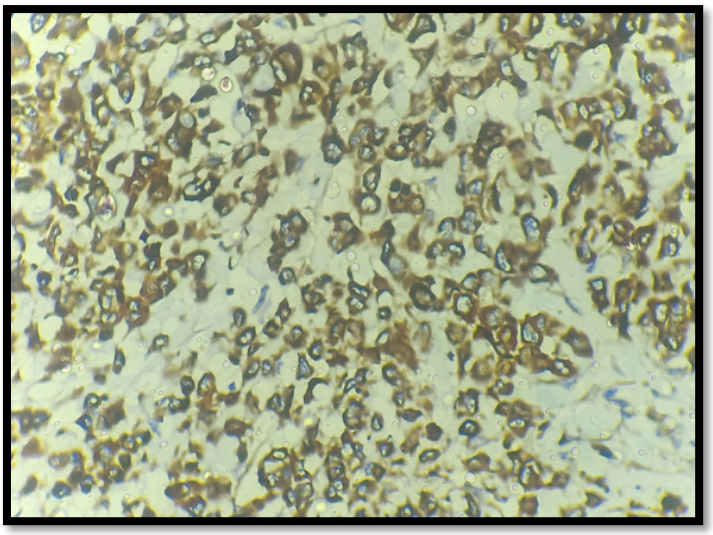

Figure 2a: Vimentin shows strong cytoplasmic positivity in tumor cells. IHC (400X)

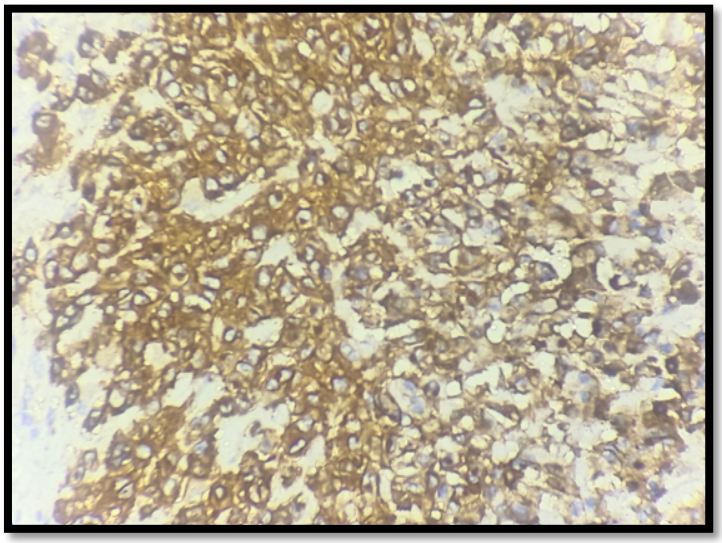

Figure 2b: EMA shows strong cytoplasmic positivity in tumor cells. IHC (400X)

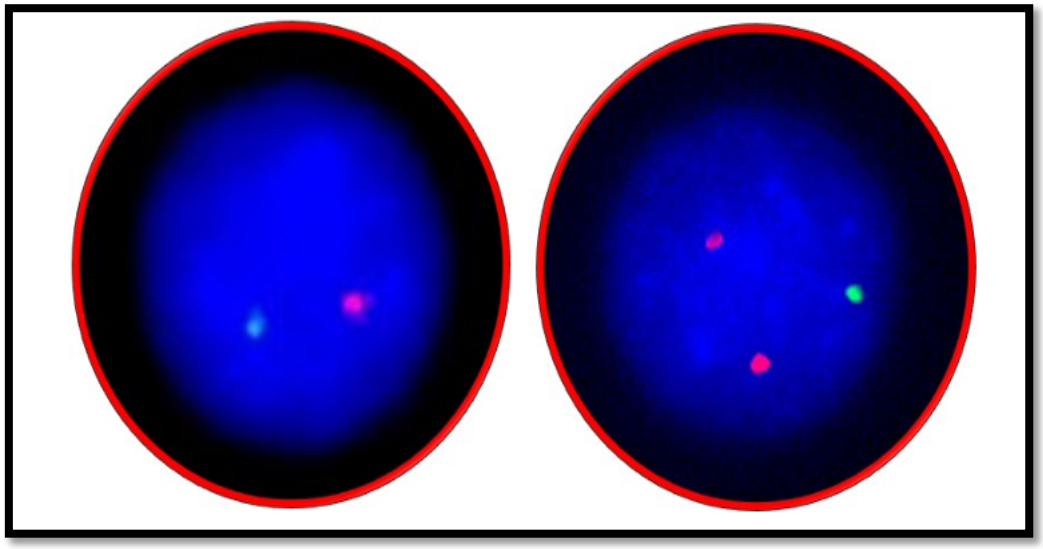

Figure 3: Fluorescent microscopy imaging shows 40% positive cells for 22q11.2 deletion

Discussion

Epithelioid sarcoma (ES), a rare aggressive soft tissue malignancy similar to other sarcomas presents with median age of 35 years [3]. ES constitutes approximately 1% of all vulvar malignancies [1, 2]. Clinically, patient usually presents with a gradually progressive painless mass in the external genitalia, hence misdiagnosed as an infectious of benign lesion due to its rarity. Distal type ES commonly seen in young presents less aggressively in contrast to proximal type ES [4].

Deep seated proximal tumors are usually multinodular, on cut section may reveal solid to myxoid areas. Histopathological examination is essential in making a final diagnosis. Biopsy usually reveals tumor cells arranged in multinodular or diffuse sheets of polygonal/epithelioid cells. These large, atypical malignant cells show moderate to abundant amount of eosinophilic cytoplasm, centrally placed highly pleomorphic nucleus, vesicular chromatin and prominent nucleoli. Binucleation or multinucleation is common. Numerous tumor giant cells and abundant necrosis also seen in some areas. Variable amount of myxoid stroma along with rhabdoid intracytoplasmic inclusions also seen. Both typical and atypical mitosis may be seen. The presence of central areas of hemorrhage and necrosis mimics that of infectious granulomas, however they are negative for IHCs typical of ES and show strong cytoplasmic or membranous positivity for CD68 due to the presence of abundant histiocytes. The presence of partial or complete epithelioid morphology mimics squamous cell carcinoma and adenocarcinoma. However lack of glandular architecture and squamoid differentiation rules out both histopathologically. Further IHC can be done to confirm the same. IHC shows strong cytoplasmic staining for cytokeratin (CK), Vimentin and membranous positivity for epithelial membrane antigen (EMA). CD34 may show cytoplasmic positivity in 50% of the cases. Negative p63, p40 rules out squamous cell carcinoma. Epithelioid malignant peripheral nerve sheath tumor (MPNST) also presents with epithelioid morphology however a pre-existing benign nerve sheath neoplasm has to be present.

SMARCB1 (INI-1) is a tumor-suppressor gene located on chromosome 22q11.2. Its gene product is ubiquitously expressed in nuclei of all normal tissues also. SMARCB1 gene inactivation has been implicated in the pathogenesis of a various group of malignant neoplasms that tend to share "rhabdoid" cytomorphology. SMARCB1 acts as a tumor suppressor gene, and loss of function of both alleles gives rise to SMARCB1-deficient tumors. Epithelioid sarcoma is characterized by the loss of SMARCB1/INI1 (integrase interactor 1) or other proteins of the SWI/SNF complex which can be assessed by fluorescence in situ hybridization (FISH) based on the scoring of signal pattern as seen in our case [5,6,7].

Currently, surgical excision is the standard management protocol for ES. The role of medical management including chemotherapy and radiation therapy is still unclear. Local lymph nodes should be dissected if they are enlarged due to metastases [8]. Lungs apart from lymph nodes are the most common sites of metastases. Prognostic factors depends on the tumor size, depth of invasion, hemorrhage, necrosis, high rate of atypical mitosis and vascular invasion [9].

Conclusion

Proximal type of ES most commonly seen in middle aged and elderly females presents with an aggressive clinical course and worse prognosis than the distal counterparts. Proper histopathological examination, IHC studies and loss of molecular marker SMARCB1 gives a final diagnosis. Surgical excision is the mainstay of the treatment.

References

1. Enzinger, F.M., 1970. Epithelioid sarcoma. A sarcoma simulating a granuloma or a carcinoma. Cancer 26(5), 1029–1041.
2. Oda, Y., Dal Cin, P., Le Loarer, F. and Nielsen, T.O., 2020. Epithelioid Sarcoma. Tumors of uncertain differentiation/Ewing sarcomaSoft tissue and bone tumours. World Heal Organ. Classif. Tumors Editor board, Ed, pp.294-296.
3. Noh, J.J., Jeon, J.-E., Jung, H., Kim, H.-S., Lee, Y.-Y., Choi, C.H., Lee, J.-W., 2021. Vulvar epithelioid sarcoma: A case report with literature review. Taiwan J. Obstet. Gynecol. 60 (1), 132–135.
4. Rekhi, B., Gorad, B.D., Chinoy, R.F., 2008. Clinicopathological features with outcomes of a series of conventional and proximal-type epithelioid sarcomas, diagnosed over a period of 10 years at a tertiary cancer hospital in India. Virchows Arch. 453 (2), 141–153.
5. International Standing Committee on Human Cytogenomic Nomenclature. ISCN 2020: an international system for human cytogenomic nomenclature (2020). Karger; 2020.
6. Pawel BR. SMARCB1-deficient tumors of childhood: a practical guide. Pediatric and Developmental Pathology. 2018 Jan;21(1):6-28.
7. Bishop JA, Antonescu CR, Westra WH. SMARCB1 (INI-1)-deficient carcinomas of the sinonasal tract. The American journal of surgical pathology. 2014 Sep 1;38(9):1282-9.
8. Czarnecka, A.M., Sobczuk, P., Kostrzanowski, M., Spalek, M., Chojnacka, M., Szumera-Cieckiewicz, A., Rutkowski, P., 2020. Epithelioid Sarcoma—From Genetics to

Clinical Practice. Cancers (Basel). 12 (8), 2112. https://doi.org/10.3390/ cancers12082112.

9. Johnson, S., Renz, M., Wheeler, L., Diver, E., Dorigo, O., Litkouhi, B., Behbakht, K., Howitt, B., Karam, A., 2020. Vulvar sarcoma outcomes by histologic subtype: A Surveillance, Epidemiology, and End Results (SEER) database review. Int. J. Gynecol. Cancer 30 (8), 1118–1123.