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A RARE CASE OF GLUTEAL SOFT TISSUE SARCOMA PRESENTING AS MALIGNANT MYOEPITHELIOMA

General Surgery

Dr. Akshaya Gunasekar* Junior Resident, Department Of General Surgery, SBMCH *Corresponding Author
Dr. G. V. Praveen Junior Resident, Department Of General Surgery, SBMCH
Dr. S. Karthikeyan Assistant Professor, Department Of General Surgery, SBMCH
Dr. K. S. Ravishankar Professor, Department Of General Surgery, SBMCH

ABSTRACT

Sarcomas are heterogeneous group of neoplasms that arise predominantly from cells of embryonic mesoderm. Soft tissue malignant myoepithelioma is a rare tumour showing myoepithelial elements with lack of obvious ducts differentiation. Its rarity and similarity to other tumours makes them difficult to diagnose. Here we present a case of gluteal swelling which on further evaluation, surgical excision and pathological assessment came out as soft tissue myoepithelial carcinoma.

KEYWORDS

Soft tissue sarcoma, Myoepithelial tumors, Myoepithelial carcinoma

INTRODUCTION

Soft tissue sarcomas are diverse group of neoplasms arising virtually from any anatomical sites including skeletal muscles, adipose cells, blood, lymphatic vessels and connective tissues or those cells with a common mesoderm origin. Primary soft tissue sarcomas most commonly arise from extremity (50% - 60%); trunk (19%); retro peritoneum (15%) and head & neck (9%).Sarcomas account for 1% of all malignancies in adults and 15% of all malignancies in children. [1] Sarcomas arise from point mutations, translocations causing over expression of growth factors producing a cellular environment prone to malignant transformation. [2]

Soft tissue sarcomas are classified as benign, intermediate (locally aggressive), intermediate (rarely metastasising) and malignant. According to WHO classification of Tumors of Soft tissue and Bones, these neoplasms are classified into 12 categories. Assessing the histological type is important for prediction of behaviour and prognosis of the tumor. Malignant tumors have potential for local invasion, recurrence and risk of distant metastasis. [3]

Soft tissue sarcomas of trunk and extremities present as painless accidentally observed tumors which do not influence function or general health of the patient despite large tumor volume. The rarity of soft tissue sarcomas often lead to misinterpretation as benign conditions. Hence recognition of soft tissue sarcomas during the diagnostic evaluation of the patient is essential for further management.

Here we present a case of a 47 years old man who presented with a gluteal swelling was diagnosed as a benign swelling and on further work up turned out be soft tissue sarcoma of myoepithelial origin – myoepithelial carcinoma.

Soft tissue myoepithelial carcinoma are variegated tumors showing morphologic, immunohistochemical and genetic variation. They occur with equal gender distribution, predominantly occurring in 2nd to 4th decades with 20% occurring in paediatric age group. Most common sites are extremities, limb girdles, head, neck and trunk. These neoplasms have a local recurrence rate of 20-42% without metastasis and 40-52% with metastasis. Histologically these are lobulated tumors varying from epitheloid, spindles and clear to plasmacytoid cells with nuclear pleomorphism. Immunoprofile expresses S100 protein, EMA and shows variable positivity for GFAP, p63, SOX10. A subset exhibits nuclear loss of expression of INI1/ SMARCB. ESWR1 gene rearrangement is present in up to half of all myoepitheliomas and myoepithelial carcinomas. [4,7,9]

Myoepithelial tumors are insignificant due to their overlap with other neoplasms in all aspects and lack of awareness among physicians. Recognition and identification of these tumors is important for further management, refinement in genetic characterisation, extemporise literature and develop targeted therapeutic avenues in future.

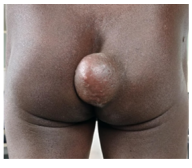
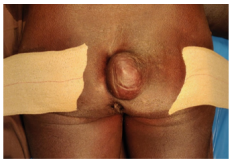
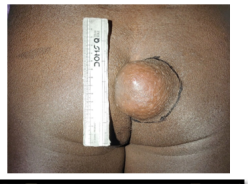
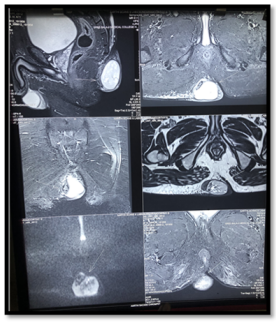
CASE REPORT

A 47 years old man presented with swelling in the right gluteal region for the past 10 years which developed spontaneously and was insidiously progressive to attain present size. It was associated with pricking type of pain on and off on sitting down.

On examination swelling present over the right gluteal region in the upper inner quadrant, oval in shape of size 6x4 cm, smooth surfaced, well defined edges with skin over it stretched.

On palpation there was no warmth or tenderness, firm to hard nodular in consistency and there were no regional or distant nodes.

Provisional diagnosis of a benign swelling was made and further evaluation was done.

MRI of the gluteal region showed- A lobulated T2 hyper intense mass 6x3.5 cm with eccentric hemosiderin deposit and surrounding soft tissue in the subcutaneous plane of right medial gluteal region.

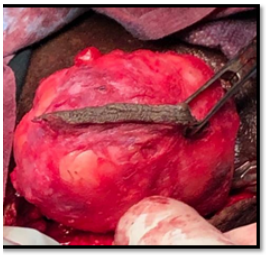
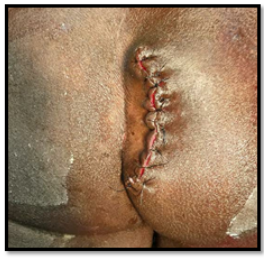
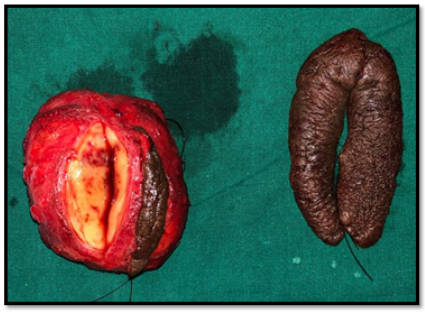
FNAC of the swelling – Aspirate shows monotonous sheets of round cells with vesicular nuclei and scanty cytoplasm, occasional organic arrangement – Round cell neoplasm.

Trucut biopsy – Fragments of neoplasms composed of polygonal cells and spindle shaped cells with dark staining nuclei arranged in sheets in a myxoid and fibrillation background. Vacuolated cells and focal areas of micro cystic degeneration are seen.

Picture that of myxopapillary ependymoma.

Neurosurgeon's opinion was obtained in view of the result from trucut biopsy as myxopapillary ependymoma which is a tumor arising from ependyma surrounding the spinal cord for spinal communication. As there was no spinal communication or any compression symptoms, neurosurgeon advised to proceed with the wide excision of the tumor.

Wide local excision of the swelling was done and sent for pathological evaluation.

Histopathology showed sections showing capsulated neoplasm with nests of epithelial cells infiltrating into stroma. Cells show enlarged vesicular nucleus, indistinct cell outline arranges in nests, cords and cribriform pattern. There were also clear cell components in a myxoid background, areas of cystic degeneration and necrosis and also areas of osseous and chondroid metaplasia. Margins were free of tumor. Final result was that of picture suggestive of malignant myoepithelioma. The diagnosis was confirmed with Immunohistochemistry which was positive for p63, vimentin, S100, High Ki67 index and focal Pan Cytokeratin

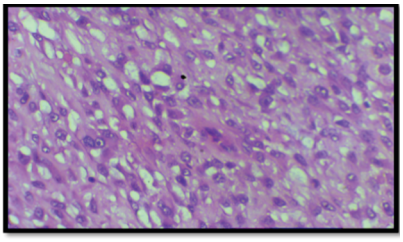


Fig. (A)

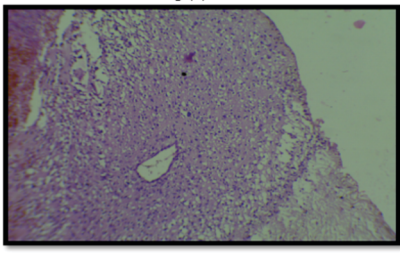


Fig. (B)

Figure (A) and (B) shows Round to oval cells with nuclear atypia

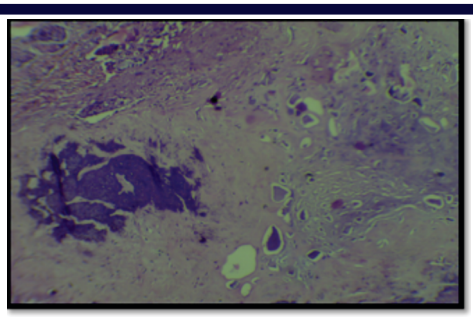


Fig. (C)

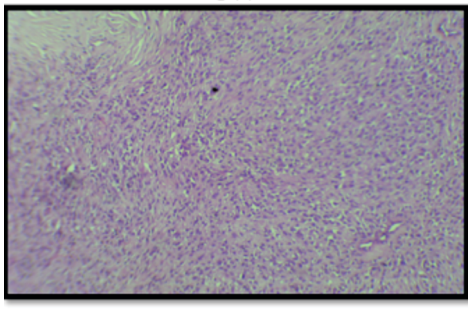


Fig. (D)

Figure (C) shows ossifications and calcifications, (D) shows spindle shaped cells arranged in fascicles.

Immunohistochemistry:

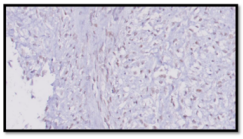


Fig. (E)

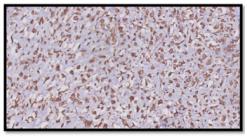


Fig. (F)

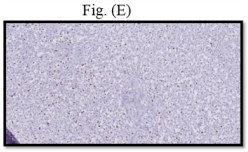


Fig. (G)

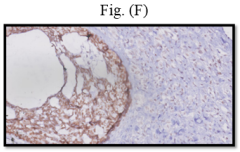


Fig. (H)

Figure (E) S100 postive; (F) Vimentin positive; (G) High Ki Index; (H) p63 positive

DISCUSSION

Soft tissue sarcomas are often managed with multimodality treatment. The imaging modality of choice in soft tissue sarcoma is MRI for diagnosing characterising and staging of the tumor, relation to adjacent neurovascular structures. Also used in guiding biopsy, planning surgery, evaluating response to chemotherapy/radiotherapy, restaging and in long term follow up for recurrences. Followed by imaging, the approach to diagnosis is multiple core needle biopsies. Biopsy is necessary to establish malignancy, assess histological type and grade of the tumor. CT scan of chest is performed in most of the cases to assess the distant metastasis which is an important factor in staging of the tumor. The pathological diagnosis is made according to 2013 WHO classification, grading of the tumor according to The Federation National des Centres de Lutte Contre le Cancer grading system (FNCLCC system) and staging according to AJCC TNM 2017 classification. [5, 6]

According to Soft Tissue Sarcoma AJCC TNM classification the case here falls under **Stage III A – T2N0M0** and according to 2013 WHO classification of soft tissue tumors, **Tumors of Uncertain Differentiation** under the sub-classification of **Intermediate (Rarely metastasising) tumors– Myoepithelial Carcinoma.**

Myoepithelial neoplasms of soft tissue are unrecognised tumors characterised by clinicopathologic and genetic means. In contrast to myoepithelial neoplasms in salivary gland, primary myoepithelial tumors in soft tissue lack ductal differentiation.

Myoepithelial tumors of soft tissue are rare tumors which exhibit a wide range of cytological and architectural features both within a given

lesion and between different tumors. The age of occurrence is between 3rd to 5th decades of life with the mean age being around 38 years and carcinomas are more common in children under 10 years of age. The common sites of tumors are the subcutaneous or deep subfascial soft tissues of limbs, trunk, head and neck region. Most patients present with a painless slow growing tumor, non-tender cutaneous/ subcutaneous mass. Larger masses may be associated with pain and local mass effect. These tumors are grossly well circumscribed, lobulated but frequently show infiltrative growth. These tumors are white to tan yellow, myxoid, gritty with areas of haemorrhage and necrosis. [7, 8]

Microscopically myoepithelial tumors are characterised by presence of spindled, ovoid or epitheloid cells arranged in reticular, trabecular or nested patterns. Occasionally there might be a dominant growth single type of cells or patterns. Other morphological features include plasmacytoid cytoplasm, clear vacuolated cytoplasm or rhabdoid features. Rarely 15% of the tumors show heterogenous differentiation with chondro-osseous, adipocytic and squamous differentiations. The characteristic feature of malignancy include nuclear pleomorphism, prominent nucleoli, atypical mitosis, mitotic figures more than 5 per high power field and necrosis. Cytologic atypia is the sole criterion for grading malignancy as it is the best predictor for malignant behaviour. [7-11]

Myoepithelial carcinomas show aggressive behaviour with a recurrence rate of 42% and a distant metastasis of 52% of affected patients. Commonly reported sites of metastasis are lungs, bones, lymph nodes and soft tissue.

Myoepithelial neoplasms have a varied immunoprofile. A panel of IHC markers are specific for recognition of myoepithelial differentiation. These include cytokeratin (93-100%), S -100 (72-100%), EMA (19-79%) and variable GFAP staining (50%). Myogenic markers calponin (86%) is the most sensitive and others which do not contribute much like SMA (36-64%) and Desmin (0-20%) are supportive for the diagnosis. ESWR1 gene rearrangement is seen in around 45% of cases which is specific. Gene fusion partners include POU5F1, PBX1, ZN444, ATF1 more commonly occurs in tumors which do not show ductal or glandular differentiation. Tumors with ESWR1- POU5F1 fusion gene show nests of epitheloid cells with clear cytoplasm and spindle cell proliferation with sclerotic stroma in ESWR1-PBX1 fusion. PLAG1 immunostaining is negative in myoepitheliomas without ductal differentiation. Diagnosis of myoepithelial origin is based on characteristic morphological features, Immunohistochemical panel and ESWR1 gene rearrangement as a supportive diagnosis. [4,7,8,9,11]

Differential diagnosis of myoepithelial tumors of soft tissue includes mixed tumors of skin which are histologically identical to myoepithelial tumors of soft tissue with location of the tumor being the differentiating point. Myxopapillary ependymoma is an important differential presenting more commonly in presacral and retro sacral region. Microscopically they show spindle cells in a myxoid or chondromyxoid background. The differentiating feature is myoepithlial tumors show mostly epitheloid cells and strong S100 positivity which is less in the latter. Ossifying fibromyxoid tumor shows a rim of metaplastic bone at the periphery and strong desmin positivity which is absent in myoepithelial tumors. Leiomyoma sans schwannomas show broad cigar shaped nuclei and alternating zones of nuclear palisading with hypo cellular myxoid zones respectively with Desmin positivity and less than 5% are S100 positive whereas myoepithelial tumors show tapering nuclei and has no alternating zones or nuclear palisading. Other tumors which are included in differential diagnosis of myoepithelial carcinoma are metastatic carcinoma and metastatic melanoma which most commonly occur in elderly patients and lack myxoid stroma, multinodular architecture and are not immunoreactive. Proximal epitheloid carcinoma shows rhabdoid morphology with EMA and keratin positivity but negative for other markers which is the differentiating feature of myoepthelial carcinoma. [8,9,11,12]

Surgery is the standard treatment of all patients with soft tissue sarcomas. The standard procedure is wide local excision with negative margins (absence of residual tumor R0). The margin of tumor on fixed tissue depends on histological subtype, preoperative therapies and presence of resistant anatomical barriers like muscular fascia, periosteum or epineurium. Sometimes marginal excision can be acceptable in selected cases. This is followed by radiotherapy as the treatment of high grade, deep, more than 5 cm lesions. Adjuvant chemotherapy is not a standard treatment in adult type soft tissue sarcoma. It can be given as an option to high risk individuals. Neoadjuvant chemotherapy with anthracyclines plus ifosfamide for 3 cycles is an option in high risk patients. [5, 13]

In this case with history, clinical examination and investigations, it was thought to be a benign swelling. Also the site of occurrence – gluteal region is very rare in case of soft tissue sarcoma. Initially FNAC was done to determine the cell type which came as Round cell neoplasm and hence proceeded with trucut biopsy which shoed features of Myxopapillary ependymoma which a tumor of ependyma surrounding the spinal cord. It is a benign but recurrent tumor and can arise in this region with or without spinal communication. Hence neurosurgeon's opinion was obtained, wide local excision of the tumor was performed and sent for histopathological evaluation. On histopathological evaluation it was found out to be mixed myoepithelial carcinoma with tumor free margins which was confirmed with immunohistochemistry.

In cases of deep, more than 5cm lesions or site specific lesions with R0 resection (tumor free margins) or R1 (microscopic margins) post-operative radiotherapy is highly essential to delay or prevent recurrences.

Although the cell type is diagnosed with histo pathology, the diagnosis is confirmed by immuno histochemistry in cases of soft tissue sarcoma for early recognition and treatment.

CONCLUSION:

Myoepithelial tumors of soft tissue are rare under recognized neoplasms. They show a wide variety of different morphological features and it is difficult to diagnose without immuno histochemistry. A precise diagnosis is essential for treatment as non-recognition leads to potentially fatal outcomes.

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