DOI: 10.7860/JCDR/2021/47753.15319

Case Report

Pathology Section

Primary Extraskeletal Myxoid Chondrosarcoma: A Case Report

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ABSTRACT

Extraskeletal Myxoid Chondrosarcoma (EMC) is a rare malignant soft tissue sarcoma with uncertain differentiation, most often seen in males. The incidence is 3% of all soft tissue tumours with limited literature available on its cytological features. EMC may arise from lower extremities, upper extremities, retroperitoneum, pelvis, and buttocks. This case report was an effort to understand the role of Fine Needle Aspiration Cytology (FNAC), histopathology and immunochemistry in the diagnosis of EMC. Authors hereby report a case of a 70-year-old male patient with slow-growing soft tissue swelling on the back just behind the right shoulder who was referred for FNAC. Patient complete history and clinical findings were recorded. Radiological images were suggestive of malignant soft tissue neoplasm with no involvement of underlining bone. FNAC reveled tumour cells which appeared monotonous and they were seen in a myxoid stroma background. Subsequently, the excised lesion was sent for histopathological examination and the report revealed the presence of abundant chondromyxoid matrix material within which were found numerous elongated spindly shaped cells. These cells had moderately pleomorphic elongated nuclei with focal solid fibrocollagenous areas along intersecting fascicles of the moderately pleomorphic spindly cell. Few of these cells had multilobulated bizarre nuclei with nuclear inclusions. Immunohistochemical stains showed diffuse positivity for S-100, vimentin, and focally positive for Epithelial Membrane Antigen (EMA). The FNAC, histopathology and immunohistochemical features confirm the diagnosis of EMC on right shoulder. It's a rare tumour whose diagnosis is made depending on history, clinical location, growth pattern, histopathology, and immunohistochemistry.

Keywords: Fine needle aspiration cytology, Pleomorphic myxoid sarcoma, Pleomorphic spindle cell

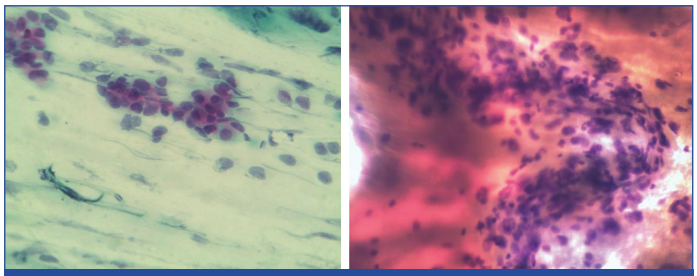
CASE REPORT

A 70-year-old male patient with the mass on the back near right shoulder was referred for FNAC. On evaluation, the patient gave the history of painless, slow-growing mass on the back for the past six months. His only symptom was that, he was not able to sleep on the back. Local examination revealed a solitary firm to hard mass measuring 20×20 cm on the back near right shoulder which had a smooth surface and well-defined border. The X-ray showed no involvement of the bone.

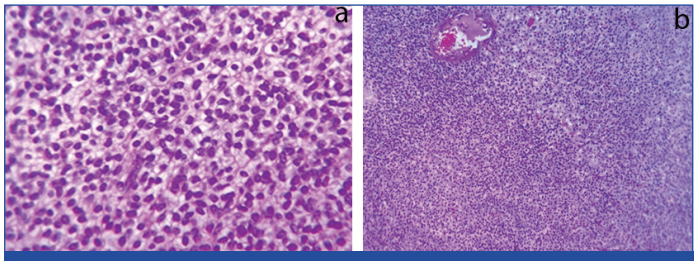
Fine Needle Aspiration (FNA) was carried out under all aseptic precaution by using 24 gauge needle and aspirate were stained with Papanicolaou and May Grunwald-Giemsa (MGG) stain to confirm the lesion. The smear showed good cellularity with cells which had a highly pleomorphic nucleus. Some of the cells seen showed binucleation and multinucleation and had well-defined cell border with abundant cytoplasm. Myxoid material was abundant on the background [Table/Fig-1].

The clinical and cytological features favoured the diagnosis of myxoid pleomorphic sarcoma. Surgical excision of the lesion with an extended margin was performed. Grossly, the lesion had extensive myxoid material altogether measuring 23×25×8 cm and was removed in parts. The largest one measured 5×5×4 cm. The cut surface was gelatinous and showed a grey-white tumour with an area of necrosis and haemorrhage. Imprint smear was done which showed bizarre cells [Table/Fig-2].

The excised lesion was sent for histopathological examination for further evaluation. The histopathological report revealed the presence of abundant chondromyxoid matrix material within which were found numerous elongated spindly shaped cells. These cells had moderately pleomorphic elongated nuclei with focal solid fibro collagenous areas along intersecting fascicles of the moderately pleomorphic spindly cell. Few of these cells had multilobulated bizarre nuclei with nuclear inclusions. Mitotic figures upto 4-6/High Power Field (HPF) were seen. [Table/Fig-3] showing lesion with the chondromyxoid area with haemorrhage (H&E 10X). Other myxoid tumours of soft tissue that should be considered in the differential diagnoses on histopathology include soft tissue chondroma, myxoma, a myxoid variant of malignant fibrous histiocytoma, myxoid liposarcoma, and chondrosarcoma was thought of.



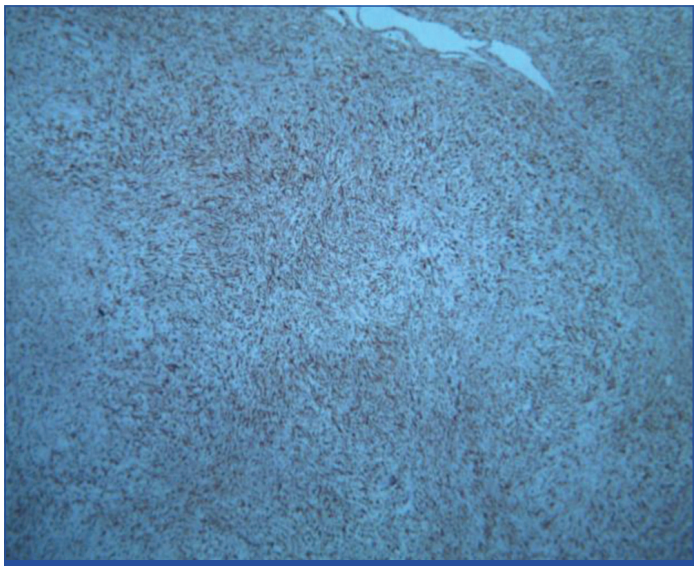
[Table/Fig-1]: Bizarre tumour cells in groups, papinicalou stain ((40X)).
[Table/Fig-2]: Dysplastic tumour cells with large bizarre nucleus, H&E ((40X)).
(Images from left to right)



[Table/Fig-3]: a) Tumour cells with bizarre nucleus H&E stain (40X); b) Tumour cells in myxoid background,H&E stain (10X).

Immunohistochemical stains were performed that showed the lesion was diffusely positive for S100 [Table/Fig-4], vimentin and focally positive for Epithelial Membrane Antigen (EMA). Based on clinical, histopathological and immunohistochemistry findings diagnosis of

extraskeletal chondromyxoid sarcoma was made and the patient was referred for the higher centre for further management.



[Table/Fig-4]: Immunohistochemical stain showing positivity for S-100 (10X).

DISCUSSION

Extraskeletol chondrosarcoma has uncertain differentiation and is a very rare sarcoma [1]. Men are commonly affected in their 5th decade and account for 3% of all the soft tissue sarcomas [2]. The diagnostic workup of this group of tumours is challenging due to its rarity and conspicuous biologic diversity [3]. Extraskeletal Myxoid Chondrosarcoma is a slow-growing tumour with a prolonged survival that is prone to local recurrences and occasionally, metastasis. However, a significant risk of death has been reported as a result of these tumours. This unique tumour usually appears as an expanding mass in adults extremities, although it can also appear in other places [4]. Histologically, EMC and other mesenchymal malignancy have overlapping phenotypic features and vaguely resemble human cartilage and shows uncertain histogenesis. Most EMC are positive for vimentin and focally positive for S100 protein [2].

EMC is a rare soft tissue sarcoma of the extremities that Stout and Verner identified as a separate pathologic entity. It is most commonly seen in the soft tissue of a pulmonary [2], lower extremity [4-6], labium majus [7], waist [8], intradural, spinal mass [9], and neck [10] has been reported. Metastasis to the lungs is also reported [11]. A study had shown that the average age of occurrence is 50 years with male preponderance and 62% of the presentation in the upper extremities, 17% of cases in lower extremities and 21% in other location [2]. EMC has a prolonged clinical course with 10-year survival among 65-85% and 15-year survival in 58%. Risk of metastasis is documented in 40% of cases for 10 years.

In the present case report, the patient had come back with local recurrence with multiple large lesions and he was later referred to higher centre. EMC has shown to have “the recurrent balanced chromosomal translocation t(9;22) (q22;q12.2), which leads to the oncogenic fusion geneEWSR1-NR4A3 [6]. This chimeric gene activates the transcription of target genes involved in cell proliferation and has been detected in approximately 65% of the cases [2]. This is seen more commonly in men and often asymptomatic. Prognostic factors like tumours having a diameter of more than 10 cm, high cellularity, anaplasia, and mitotic activity in more than two out of every ten high-power fields are linked to a decrease chance of survival [7]. Radiologic features also help in the diagnosis of Extraskeletal Myxoid Chondrosarcoma [11].

Microscopically, the distinct cytological features that were present in this case is shown in [Table/Fig-1]. However, the presence of typical chondrocyte-like cells embedded within lacunae, as seen in some case reports, was not observed in this case. EMC does not have any specific immunohistochemical markers. Nearly all the cases are vimentin positive and synaptophysin positive. S100 and EMA show focal and weak immunoreactivity in 20% cases. Other previous studies also confirmed the diagnosis of EMC with immunohistochemical analysis [7,10]. This sarcoma is classified as a tumour of intermediate grade, with a prolonged clinical history and the potential for local recurrence and distant metastasis. Non-surgical treatment is usually reserved for distant metastasis/ recurrent disease [2]. Clinical features shared by all carcinoma of the unknown primary should be focused and primary metastatic cancer should be considered as an archetype of metastatic disease [12]. It was observed that Neoadjuvant Chemotherapy (NAC) can be considered to avoid axillary surgery. Chemotherapy for the ipsilateral breast had a better outcome, which prevented mastectomy [13].

Other differential diagnoses on cytology include myxoid liposarcoma, soft tissue chondroma, a myxoid variant of pleomorphic sarcoma, myxoma and chondrosarcoma [7]. The distinct cytological features of EMC help us to diagnose the case with ease on FNAC. This diagnosis is supported by histopathology and immunohistochemical profile. In this case, the histopathological features and immunohistochemistry were helpful to confirm the final diagnosis.

Complete surgical resection is the standard treatment of primary EMC followed by radiation in high-risk cases. In advanced disease, the patient usually receives medical treatment. Unfortunately, response rates to conventional chemotherapy are low [5]. In the above case, the close differential diagnosis was pleomorphic sarcoma as a lesion was positive for S-100 and vimentin, negative for synaptophysin along with other cytological feature pleomorphic sarcoma was ruled out.

CONCLUSION(S)

The EMC is a rare tumour. For this unique tumour, diagnosis depends on accurate histology, immunohistochemistry, location, and growth pattern. It should remain in the differential diagnosis for patients presenting with soft tissue mass on back. FNAC plays a vital role in the diagnosis of EMC and is a valuable tool for cytologist to make a presumptive diagnosis.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Nov 13, 2020
- Manual Googling: Jun 16, 2021
- iThenticate Software: Jul 24, 2021 (18%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Nov 12, 2020**
Date of Peer Review: **Dec 18, 2020**
Date of Acceptance: **Jun 19, 2021**
Date of Publishing: **Sep 01, 2021**