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Case Report

Pathology Section

Unveiling the Uncommon: A Case of Parietal Rhabdomyosarcoma in a 60-year-old Woman

JAYASHREE BHAWANI[1], SAMARTH SHUKLA[2], SOURYA ACHARYA[3]

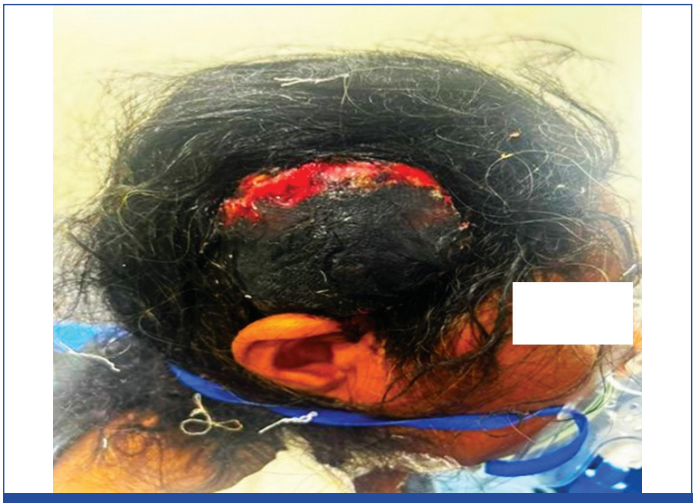
ABSTRACT

Malignant mesenchymal tumours are rare, aggressive neoplasms originating from mesenchymal tissues. These tumours are characterised by a high metastatic potential and pose significant diagnostic and therapeutic challenges. Mesenchymal tumours are of interest to oncologists because of their comparatively low occurrence, especially those with a high proliferation index. Authors report a case of a 60-year-old female who presented to the tertiary care rural hospital with persistent headaches, dizziness, and respiratory distress. Imaging studies revealed a heterogeneously enhancing soft-tissue density lesion in the right parietal region with extensive metastasis to both lungs. Histopathological analysis of the parietal lesion indicated features suspicious of a high-grade malignant tumour of muscular origin. Immunohistochemistry results were conclusive for Rhabdomyosarcoma. The patient was treated with radiation therapy for the primary tumour site and systemic chemotherapy, including doxorubicin and ifosfamide, for the metastasis. This case highlights the aggressive nature of malignant mesenchymal tumours and the complexities involved in their management. The high Ki-67 index and extensive metastasis indicate a poor prognosis. A multidisciplinary approach is crucial for symptom management and improving quality of life. Early detection, accurate diagnosis, and a comprehensive, multidisciplinary treatment strategy are essential for managing these challenging cases. Patients with such tumours typically have a bad prognosis since they have a significant chance of the tumour spreading to other organs, such as the liver, lungs, and bones. This case report aims to highlight the clinical presentation, diagnostic process, and therapeutic challenges associated with this aggressive tumour type, contributing to the limited but growing body of literature on rhabdomyosarcoma.

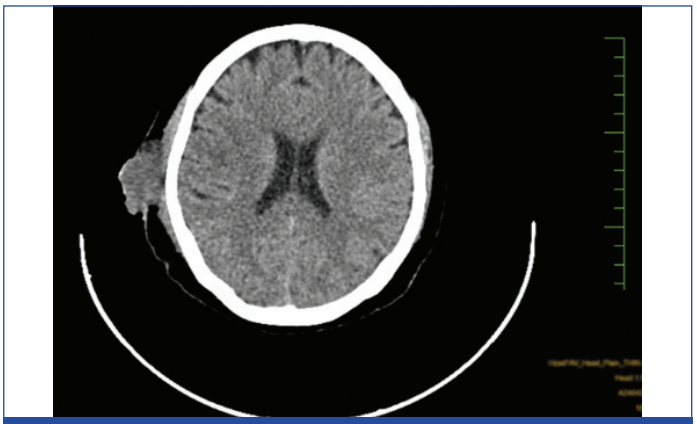
Keywords: Chemotherapy, Malignant mesenchymal tumour, Metastasis, Immunohistochemistry, Multidisciplinary approach, Radiation therapy

CASE REPORT

A 60-year-old female presented to the emergency department with neurological and respiratory symptoms. Her complaints included severe headache, dizziness, confusion, cough, occasional blood-tinged sputum, and shortness of breath. The patient was initially assessed and stabilised. A comprehensive physical examination was performed which revealed a hard swelling over the right side of the parietal region of the skull [Table/Fig-1] with decreased coordinative movements. Pulmonary examination showed diminished breath sounds in the left upper lung field and crackles on auscultation. The patient was admitted to the surgery department for further investigations. She underwent radiological investigations like Contrast Enhanced Computed Tomography (CECT) of the brain [Table/Fig-2], which discovered an extracalvarial heterogeneously enhancing soft-tissue density lesion with non enhancing necrotic areas in the right parietal region (approximately 4.8×2 cm) with the involvement of adjacent skull musculature, extending to the skin surface. It also revealed degenerative changes in the visualised spine. Radiological investigation suggested a neoplastic entity, probably melanoma.

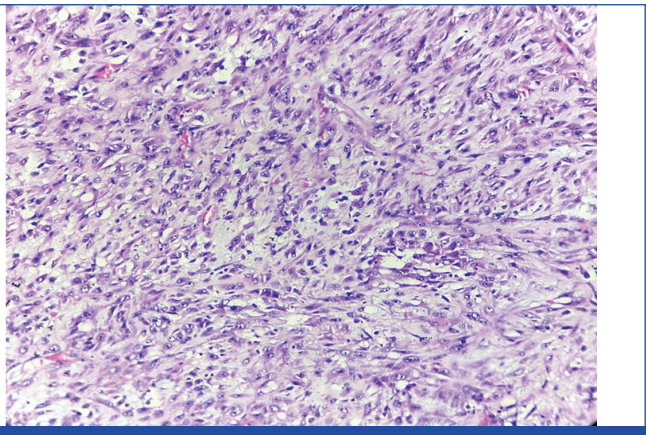


[Table/Fig-1]: Photograph showing a large right parietal region lesion.

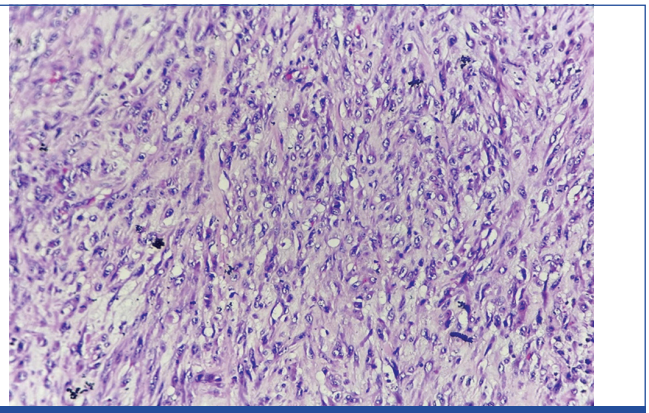


[Table/Fig-2]: CECT brain showing an extracalvarial heterogeneously enhancing soft-tissue lesion.

Surgical excision of the lesion was performed and sent for histopathological examination. The haematoxylin and eosin slides showed tumour cells arranged in a diffuse sheet-like pattern. These cells were cytologically undifferentiated, atypical, having large, pleomorphic nuclei with prominent nucleoli. Spindle-shaped cells were seen in some areas, and high mitotic counts were also observed [Table/Fig-3,4]. On microscopic evaluation, a diagnosis of high-grade malignant neoplasm arising from muscle was made. As radiological and histopathological analyses were contrary to each other, immunohistochemistry was performed, which resulted in

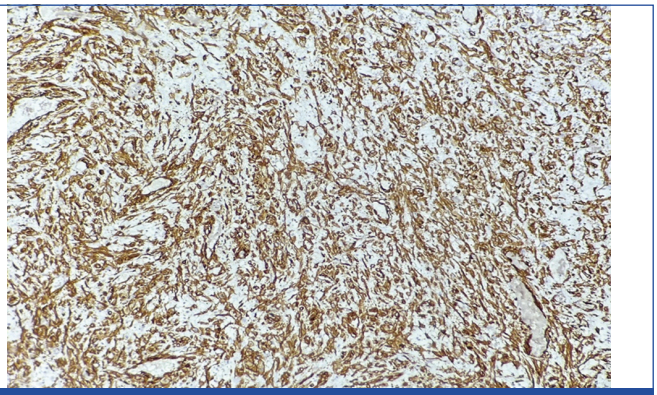


[Table/Fig-3]: Photomicrograph showing atypical, polygonal, spindle shaped tumour cells arranged in diffuse sheets (H&E,10x).



[Table/Fig-4]: Photomicrograph showing atypical rhabdomyoblasts. (H&E, 10x).

positive staining for Smooth Muscle Actin (SMA), Myogenin, Myo D-1, a high proliferation index for Ki-67, and negative staining for CD 34, HMB45, S100. SMA positivity ruled out the diagnosis of melanoma [Table/Fig-5]. Thus, a complete IHC panel resulted in the diagnosis of rhabdomyosarcoma.



[Table/Fig-5]: Photomicrograph showing positive immunostaining with SMA (smooth muscle actin) (IHC, 10x).

After the confirmation of the diagnosis, the tumour board meeting suggested the treatment plan. The patient was planned for radiation therapy for the parietal mass, and simultaneously chemotherapy sessions were started for the pulmonary metastasis. After two weeks of treatment, the patient showed partial relief of neurological symptoms and improved respiratory function. Regular follow-up was planned to monitor treatment response and manage any emerging complications. The patient was scheduled for repeat imaging and reassessment of the treatment plan after one month.

DISCUSSION

Malignant mesenchymal tumours are a diverse group of soft-tissue neoplasms that arise from mesenchymal tissues such as muscle, fat, and connective tissue [1]. These tumours are known for their aggressive behaviour and high metastatic potential. They account for a small percentage of all malignancies but pose significant diagnostic and therapeutic challenges due to their rarity and varied presentation [2].

The incidence of mesenchymal tumours, particularly those with a high proliferation index, is relatively low, making them a subject of interest in oncology [3]. The prognosis for patients diagnosed with such tumours is generally poor, with a high likelihood of metastasis to distant organs, including the lungs, liver, and bones [4]. Rhabdomyosarcoma, a subtype of malignant mesenchymal tumours, is a highly aggressive and malignant neoplasm originating from skeletal muscle progenitors [5]. While more commonly seen in paediatric populations, rhabdomyosarcoma in adults is rare and often presents with a poorer prognosis. The tumour's aggressive nature is characterised by rapid growth, high metastatic potential, and resistance to conventional therapies. The histological subtypes of rhabdomyosarcoma include embryonal, alveolar, and pleomorphic, with each subtype having distinct clinical and pathological features [6]. The incidence of rhabdomyosarcoma, particularly in adults with high proliferation indices, is relatively low, making it a subject of interest in oncology. The prognosis for patients diagnosed with such tumours is generally poor, with a high likelihood of metastasis to distant organs, including the lungs, liver, and bones. This case presents a 60-year-old female diagnosed with a malignant mesenchymal tumour rhabdomyosarcoma characterised by a highly aggressive and proliferative nature, as evidenced by a Ki-67 index greater than 90%. The tumour, originating in the right parietal region, demonstrated extensive metastasis to both lungs, causing significant clinical symptoms and posing substantial diagnostic and therapeutic challenges. Rhabdomyosarcoma, specifically, is a rare and aggressive form of soft-tissue sarcoma that arises from skeletal muscle progenitors [7]. While it predominantly affects children, adult cases like the one presented here are uncommon and often associated with a poorer prognosis. The histopathological analysis in this case revealed features consistent with rhabdomyosarcoma, ncluding the high proliferation index indicated by the Ki-67 staining. The immunohistochemical profile, showing SMA, Myogenin, Myo-D1 positivity and negativity for CD34, HMB45, and S100, helped confirm the diagnosis. Adult rhabdomyosarcoma is typically more resistant to treatment and has a higher rate of metastasis, which was evident in the extensive pulmonary involvement seen in this patient.

Diagnostic Challenges

Malignant mesenchymal tumours, although rare, encompass a wide range of soft-tissue sarcomas [8]. These tumours are known for their varied histological presentations and aggressive behaviour. In this case, the differential diagnosis included various soft-tissue sarcomas, such as leiomyosarcoma, malignant peripheral nerve sheath tumours, and undifferentiated pleomorphic sarcoma. Immunohistochemistry played a crucial role in narrowing down the diagnosis. The tumour was negative for CD34, HMB45, and S100 but positive for SMA, Myogenin, Myo-D1 indicating a smooth muscle origin, which aligns with the characteristics of rhabdomyosarcoma. Imaging studies provided essential insights into the extent of the disease. The CECT brain scan revealed a large, heterogeneously enhancing lesion with necrotic areas, suggesting a high-grade malignancy. The involvement of adjacent skull musculature and extension to the skin surface further underscored the aggressive nature of the tumour. The CECT thorax scan highlighted the extensive metastatic burden, with large lesions in the lungs and pleural effusion, complicating the patient's respiratory status.

Therapeutic Approach

The treatment of rhabdomyosarcoma tumours typically involves a multimodal approach, including surgery, radiation therapy, and chemotherapy. In this case, surgical resection of the primary tumour

was not feasible due to the lesion's location and extensive involvement of surrounding structures. Instead, the patient underwent a biopsy to confirm the diagnosis and guide further treatment. Radiation therapy was administered to the primary tumour site to reduce the tumour burden and alleviate neurological symptoms. This approach is supported by evidence indicating that radiation therapy can be effective in local control of soft-tissue sarcomas, particularly when surgical options are limited. Systemic chemotherapy was initiated to address the metastatic disease. The patient received a combination of doxorubicin and ifosfamide, which are commonly used in the treatment of high-grade soft-tissue sarcomas. Despite the aggressive treatment regimen, the prognosis for patients with metastatic soft-tissue sarcomas remains poor, with limited survival benefits observed in advanced stages.

Prognosis and Follow-up

The prognosis for rhabdomyosarcoma, particularly those with a high proliferation index and extensive metastasis, is generally unfavourable [9]. Factors influencing prognosis include tumour size, histological grade, and the presence of metastatic disease at diagnosis. In this case, the large size of the primary tumour, high Ki-67 index, and significant metastatic burden suggest a poor outcome [10]. Follow-up care focuses on palliative measures to manage symptoms and maintain the quality of life. Regular imaging studies are essential to monitor disease progression and assess the effectiveness of ongoing treatment. Palliative interventions, such as thoracentesis for pleural effusion and analgesia for pain management, are critical in addressing the patient's comfort and functional status.

Literature Context

The aggressive nature of rhabdomyosarcoma tumours and their propensity for early and widespread metastasis are well-documented in the literature [11]. Studies indicate that high-grade soft-tissue sarcomas, such as rhabdomyosarcoma, often present with advanced disease and have a high recurrence rate [12,13]. The immunohistochemical profile observed in this case (SMA, Myogenin and Myo-D1 positive, CD34 negative, HMB45 negative, S100 negative) aligns with characteristics described in other reports of rhabdomyosarcoma and related sarcomas [14]. Overall, this case underscores the importance of a multidisciplinary approach in managing complex and aggressive tumours. Collaboration among oncologists, radiologists, pathologists, and palliative care specialists is crucial in developing comprehensive treatment plans tailored to individual patient needs.

CONCLUSION(S)

This case report presents a 60-year-old female with a rhabdomyosarcoma in the right parietal region, demonstrating extensive metastasis to both lungs. Despite aggressive treatment, the prognosis for patients with such advanced and high-grade tumours remains poor. This case underscores the critical need for early detection, accurate diagnosis, and comprehensive treatment planning involving a multidisciplinary team to manage symptoms and improve the patient's quality of life.

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PARTICULARS OF CONTRIBUTORS:
1. Junior Resident, Department of Pathology, Jawaharlal Nehru Medical College, DMIHER, Sawangi (Meghe), Wardha, Maharashtra, India.
2. Professor, Department of Pathology, Jawaharlal Nehru Medical College, DMIHER, Sawangi (Meghe), Wardha, Maharashtra, India.
3. Professor, Department of General Medicine, Jawaharlal Nehru Medical College, DMIHER, Sawangi (Meghe), Wardha, Maharashtra, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Jayashree Bhawani,
Jawaharlal Nehru Medical College, Sawangi (Meghe),
Wardha-442107, Maharashtra, India.
E-mail: jayashree1993rey@gmail.com

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