

Pleomorphic Rhabdomyosarcoma in the Deltoid Muscle with Pulmonary Spread: Radiologic and Histopathologic Insights

NILLOHIT SAHA¹, JENIKAR PAULRAJ², SAI SHANKAR MANKUZY GOPALAKRISHNAN³, HARSHAVARDHAN BALAGANESAN⁴, R REMYA⁵



ABSTRACT

Rhabdomyosarcoma (RMS) is a malignant soft-tissue tumour primarily affecting children, but it is comparatively rare in adults. This case study outlines the clinical presentation, imaging characteristics, and histopathological findings of pleomorphic RMS in a 56-year-old male patient with a painless, progressively enlarging mass in the left deltoid region. Imaging techniques, including ultrasound and Contrast-Enhanced Magnetic Resonance Imaging (CEMRI), revealed a large, lobulated intramuscular lesion with significant vascularity, diffusion restriction, and features suggestive of haemorrhage. Histopathological Examination (HPE) confirmed the diagnosis of undifferentiated pleomorphic RMS, supported by positive immunohistochemical staining for myogenin. Following imaging for metastatic evaluation, multiple cannonball metastases were identified in the lungs. These findings underscore the necessity for early diagnosis and the importance of imaging in evaluating the extent of the disease and guiding therapeutic decisions in this aggressive tumour.

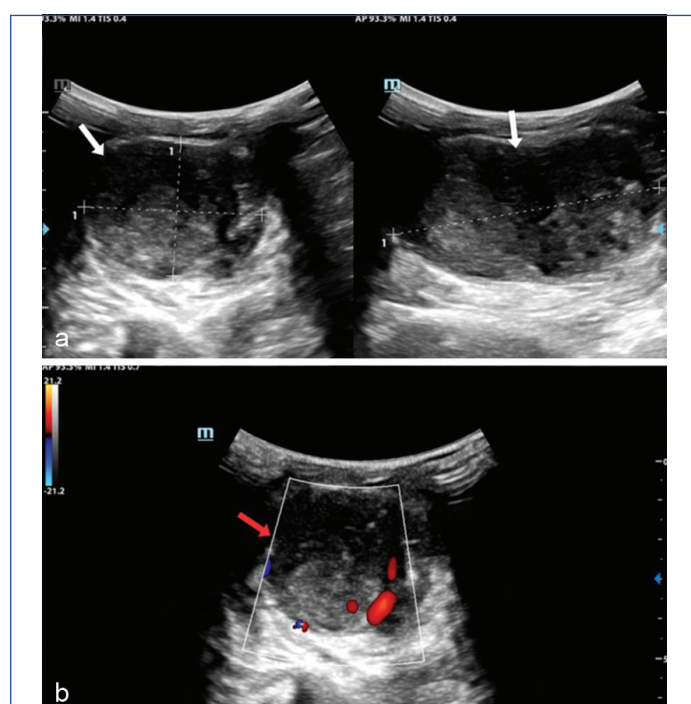
Keywords: Adult, Imaging, Oncology, Radiology, Rare, Soft tissue tumour

CASE REPORT

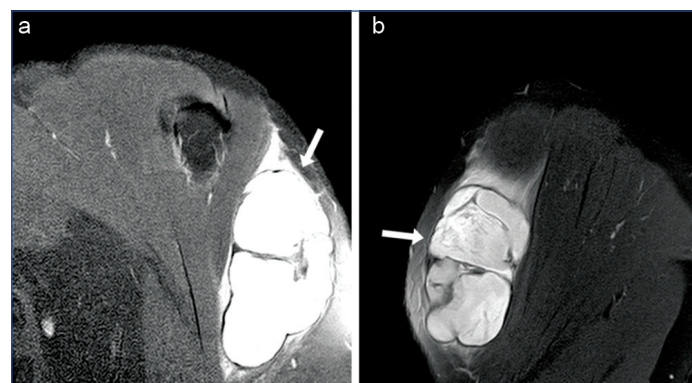
A 56-year-old male presented to the Department of General Surgery with an intramuscular swelling in the left deltoid region that had been progressively enlarging over the last month. The swelling was painless and did not reduce the range of movement of the left shoulder. Palpation revealed a mass in the left deltoid region measuring about 4×4 cm. The patient was then referred to the Radiology department after a clinical examination. On ultrasound of the left deltoid region, a large, lobulated, intramuscular, heteroechoic lesion was noted, which involved the left deltoid muscle, measuring 4.5 cm in length and 4.3 cm in width, with increased vascularity observed on colour Doppler [Table/Fig-1a,b]. CEMRI showed an

encapsulated, multilobulated lesion involving the left deltoid muscle that appeared predominantly hypointense to skeletal muscle on T1-weighted sequences and hyperintense on T2/PD-weighted sequences [Table/Fig-2a,b]. The lesion also showed a “split fat” sign on the T1 coronal sequence [Table/Fig-3] [1]. Short T1 Inversion Recovery (STIR) sequences show the septations within the lesion, and gradient sequences show blooming in the more superior aspect of the lesion, which suggested haemorrhage [Table/Fig-4a,b]. The lesion demonstrated areas of significant diffusion restriction, as observed with Diffusion-Weighted Imaging (DWI) and the Apparent Diffusion Coefficient (ADC) [Table/Fig-5a,b], representing thin rinds of fat around the lesion on T1 coronal sequences. After the administration of contrast, the lesion showed peripheral and septal enhancement [Table/Fig-6a,b].

RMS typically appears as an ill-defined, heterogeneously enhancing soft-tissue mass; however, several conditions can mimic it. Pleomorphic undifferentiated sarcoma, leiomyosarcoma, and liposarcoma often show similar heterogeneity with necrosis, while intramuscular myxoma is usually well-circumscribed with low T1 and high T2 signal. Haemangiomas demonstrate serpiginous flow



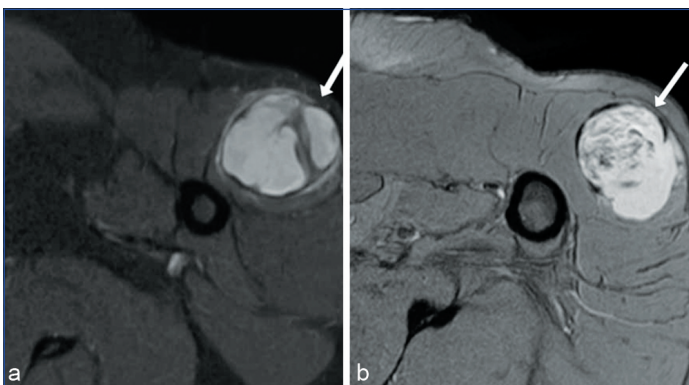
[Table/Fig-1]: a) Ultrasound of the local area showed a well-defined lobulated heteroechoic lesion involving left deltoid muscle (white arrows); b) Colour doppler showed increased vascularity within the lesion (red arrow marking the lesion).



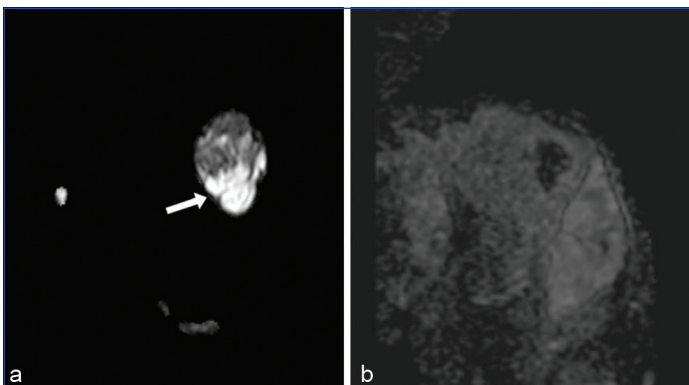
[Table/Fig-2]: a) PD SPIR coronal sequence showed an encapsulated lobular hyperintense lesion (white arrow) involving the left deltoid muscle with surrounding subcutaneous edematous changes; b) showed the same lesion (white arrow) in the PD SPIR sagittal sequence with multiple hypointense septations and surrounding oedema.
PD-SPIR - Proton density spectral pre-saturation with inversion recovery



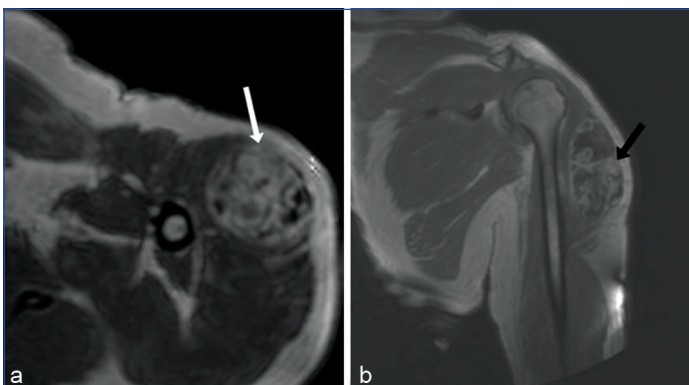
[Table/Fig-3]: MRI shoulder coronal T1 images showed a 'split fat' sign representing rinds of fat along the margins of the lesion (red arrows).



[Table/Fig-4]: a) STIR axial images show a hyperintense septated lesion within the deltoid muscle (white arrow); b) Gradient axial images show the lesion with blooming, suggesting haemorrhage (white arrow).



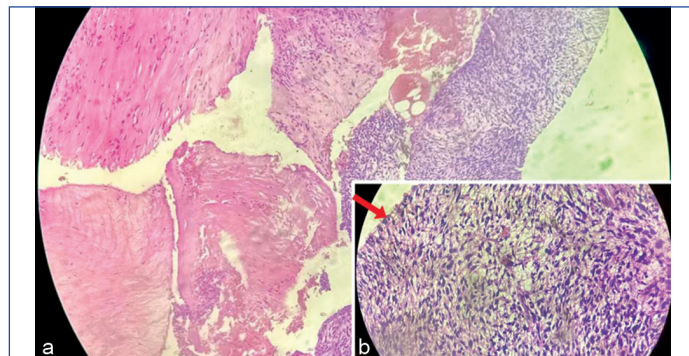
[Table/Fig-5]: The lesion showed areas of diffusion restriction on DWI and ADC. (area of diffusion restriction marked by white arrow).



[Table/Fig-6]: Postcontrast T1 axial and coronal images show peripheral and septal contrast enhancement (lesion marked by black and white arrows).

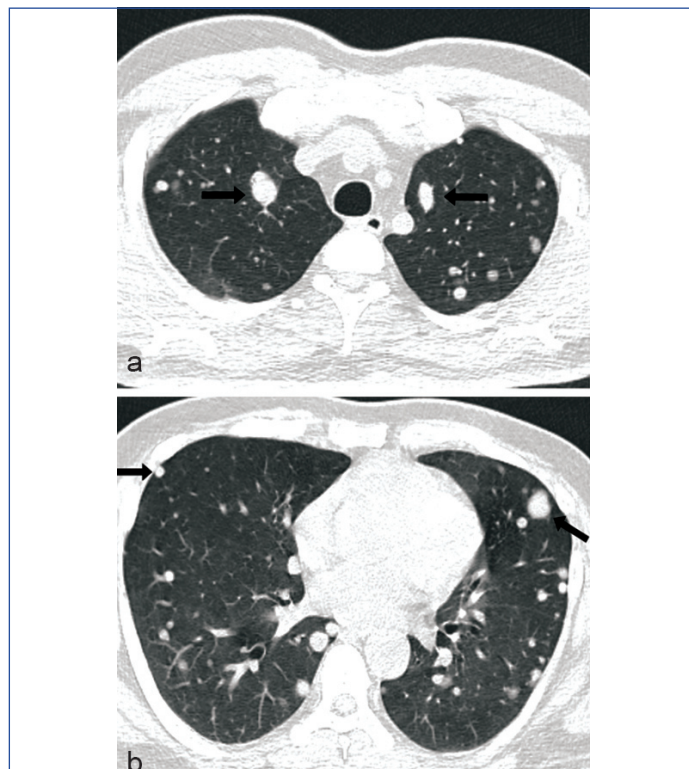
voids or phleboliths, unlike sarcomas. Metastases typically present as solid, enhancing nodules, sometimes multiple, while pyomyositis shows central fluid with rim enhancement accompanied by clinical signs of infection. Lymphoma within muscle tends to be homogeneous on MRI with less necrosis. Thus, integration of signal pattern, enhancement features, and clinical context is key before histological confirmation.

A HPE was recommended, and the patient underwent an ultrasound-guided core needle biopsy for diagnosis. The HPE revealed tumour cells arranged in sheets and fascicles, showing hypercellular and hypocellular areas in a fibromyxoid stroma. The individual tumour cells are highly pleomorphic, appearing spindled to round, with hyperchromatic nuclei and sparse eosinophilic to clear cytoplasm, as well as multinucleated giant cells. This confirmed the diagnosis of undifferentiated pleomorphic RMS [Table/Fig-7a,b].



[Table/Fig-7]: a) HPE showed tumour cells arranged in sheets and fascicles with hyper and hypocellular areas in fibromyxoid stroma; b) Individual tumour cells with hyperchromatic nuclei with spindled to round appearance (red arrow).

After HPE, immunohistochemistry was done to confirm pleomorphic RMS, with immunostaining for Myogenic Differentiation 1 (myoD1) or myogenin turned out to be positive. High-Resolution Computed Tomography (HRCT) chest and Contrast-Enhanced Computed Tomography (CECT) abdomen were undertaken to screen for any distant metastases. HRCT chest showed multiple cannonball metastases involving bilateral lungs [Table/Fig-8a,b], with unremarkable findings on CECT abdomen. The patient was later referred to a tertiary oncology institute for palliative management,



[Table/Fig-8]: HRCT chest: showing multiple cannonball metastases diffusely involving bilateral lungs (black arrows).

as surgical treatment was not feasible with multiple metastases involving bilateral lung fields. The patient had undergone radiation therapy for tumour size and pain reduction, with the treatment aimed at palliative care.

DISCUSSION

The RMS is a malignant tumour originating from primitive mesenchymal cells, characterised by its tendency to differentiate into striated muscle tissue [2]. RMS is a frequently occurring soft-tissue sarcoma in the paediatric age group but is a rarity among adults [3]. Over half of soft-tissue tumours in children under 16 years of age are caused by these tumours, and they occur at a rate of 4 cases per million per year [1]. In contrast, these tumours are rare in non-paediatric populations, comprising 11% of cancers in adolescents and only 1-2% in adults [4].

RMS can develop in almost any anatomical location; however, in adults, it most commonly occurs in the extremities, followed by the trunk, the genitourinary tract, and the head and neck regions [5]. Very few cases of deltoid RMS in adults have been reported in the literature to date, as discussed in the [Table/Fig-9] below [6-8].

Imaging techniques, such as Magnetic Resonance Imaging (MRI) and Computed Tomography (CT) scans, are essential for diagnosing RMS. MRI scans with contrast agents help determine the local extent of the tumour, while CT scans are used for distant staging. MRI is the preferred imaging method for determining the local extent of RMS because it can clearly distinguish between different types of soft tissues [14]. In the present case, the contrast MRI highlighted the lesion's characteristics, including haemorrhage, cellular components, and septations, as well as its local extent.

The RMS differs from other soft-tissue sarcomas because it often involves nearby lymph nodes and can even spread to distant lymph nodes. These enlarged lymph nodes are typically first identified through ultrasound scans, followed by confirmation with CT or MRI scans, which reveal thickened outer layers, internal blood vessel growth, and abnormal structures [3]. More than 20% of cases exhibit distant metastases at the time of diagnosis, predominantly affecting the lungs [15]. Within this framework, a maximum diameter that surpasses 5 mm is commonly used as the threshold for identifying a pulmonary nodule as a possible metastasis in cases of high-grade sarcomas [16].

Study	Age/sex	Site and presentation	Imaging	HPE & IHC	Treatment	Outcome
Furlong MA et al., 2001 [6]	21-81 yrs (mean 51); M:F = 28:10	Predominantly extremities (18 lower, 3 upper limbs); usually painful, rapidly growing masses	Tumour size 1.5-15 cm; lobulated, firm, with necrosis; grossly circumscribed	Classic features of pleomorphic rhabdomyoblasts; 3 morphologic variants (classic, round-cell, spindle-cell). IHC positive for ≥1 skeletal muscle marker (myoglobin 97%, desmin 95%, myogenin ~56%, MyoD1 ~53%)	Surgery; some had adjuvant therapy (not standardised)	Poor prognosis: 70% died of disease (mean survival 20 mo); ~27% disease-free at follow-up
Xi S and Tong W, 2018 [7]	36-yr-old male	Right shoulder; mass infiltrating deltoid, subscapularis, infraspinatus with oozing into adjacent tissues	MRI: ill-defined heterogeneous lesion with infiltration; later abdominal CT showed small bowel intussusception due to metastasis	Biopsy confirmed PRMS. IHC: vimentin+, desmin+, SMA+, partial myogenin+, focal MyoD1+, Ki-67 high; negative for AE1/AE3, CD34, S100	Neoadjuvant ifosfamide + epirubicin → wide excision → adjuvant chemotherapy + radiotherapy → resection of intestinal metastasis	Alive without recurrence 6 months after surgery
Azam M et al., 2016 [8]	45-year-old male	Left shoulder; painless swelling for 1 year, 12×10 cm mass involving deltoid and triceps	MRI: large, well-defined heterogeneously enhancing lesion; supplied by axillary/subclavian branches; intact humeral cortex	HPE: spindle and bizarre pleomorphic cells; IHC: vimentin+, desmin+, negative for SMA, S-100, CD34	9 cycles neoadjuvant ifosfamide + epirubicin → wide local excision → 4 cycles adjuvant chemo (ifosfamide, epirubicin, etoposide) → adjuvant radiotherapy (66 Gy, 33 fractions)	Complete response; disease-free at 2-year follow-up

[Table/Fig-9]: Few cases of pleomorphic RMS in extremities/shoulder muscles presented in the literature [6-8].
HPE: Histopathological examination; IHC: Immunohistochemistry
CD34-Cluster of Differentiation 34 AE1 / AE3 - clones of anticytokeratin monoclonal antibodies

Imaging is crucial in managing RMS, by provides a comprehensive diagnostic evaluation that including determining the extent of the primary tumour, assessing nodal involvement, guiding biopsies, evaluating treatment response, and monitoring during follow-up [3]. It is generally a swiftly advancing tumour in apparently healthy patients with no presenting complaints. Clinical symptoms vary depending on the location of the primary tumour and the presence of metastases [9]. In the present case, the patient did not complain of any symptoms. Although RMS can occur anywhere, adult cases are most commonly found in the limbs (40%), cervical region (30%), and urogenital system (20%) [4]. In the present case, the RMS was located in the upper extremity, specifically in the deltoid region.

Histologically, RMS is generally subclassified into embryonal (which includes rare variants like spindle cell and botryoid), pleomorphic, and alveolar types, while some cases remain unclassified (NOS: Not Otherwise Specified) [10]. Typically, the pleomorphic subtype of RMS manifests in the extremities in patients aged 45 and older. In contrast, the embryonal subtype does not have a specific site preference in adults [11,12].

Unlike paediatric RMS, adult RMS is rarely, if ever, associated with syndromes or hereditary diseases like Li-Fraumeni syndrome, Costello syndrome, Neurofibromatosis type 1 (NF1), and hereditary retinoblastoma [13]. Although the patient of the present case did not have any associated syndromes or familial conditions.

High accuracy in evaluating lytic bone lesions is a notable advantage of CT is its high accuracy in evaluating lytic bone lesions, effectively complementing MRI assessment of intramedullary bone [17]. In the present case, HRCT of the chest and CECT of the abdomen were done to find distant metastases, and HRCT chest showed multiple cannonball metastases involving the upper and lower lobes of bilateral lungs. Microscopically, pleomorphic RMS is characterised by sheets and clusters of highly variable cells with cytoplasm that displays intense eosinophilia [11,18]. Immunohistochemical analysis reveals that RMS is positive for desmin and myogenin, a transcription factor in skeletal muscle [19]. The diagnosis of pleomorphic RMS in this case was confirmed by HPE and myoD1 positive immunohistochemistry.

Surgery is the mainstay of treatment for patients with pleomorphic RMS [2]. Within the scope of radiotherapy, this approach has proven beneficial for individuals with advanced stages of the disease. Radiotherapy is commonly utilised to manage microscopic or residual tumours, particularly in unresectable sites. Additionally, notable advancements in diagnostic technologies alongside cutting-edge radiotherapy techniques, including conformal radiotherapy, proton therapy, and Intensity-Modulated Radiotherapy (IMRT), help to minimise adverse effects associated with radiation exposure while achieving effective local control in the treatment of RMS in children [20].

Factors influencing prognosis include the tumour's location and size, the patient's age, the potential for surgical resection, the surgical margins achieved, and the presence of metastases [21]. Regarding clinical outcomes, the five-year survival rate in adults is significantly lower than that in children (27% compared to 61%) [5].

CONCLUSION(S)

Magnetic resonance imaging is preferred for evaluating RMS across various body locations, with additional benefits through Diffusion Weighted Imaging (DWI). The combination of imaging characteristics-such as a large, irregular primary tumour, extensive lymphadenopathy, and metastases in both the lungs and bones-paired with clinical insights, the patient's age, and the tumour's location, can significantly improve the precision of the differential diagnosis. Histopathological assessment and immunohistochemistry provide consistent confirmation of the diagnosis.

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