

CASE REPORT

Diagnostic and therapeutic approaches of para-testicular SCRMS in an adolescent patient: A case report

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Abstract

Background: Para-testicular rhabdomyosarcoma (RMS) is a rare and aggressive malignancy primarily affecting children and adolescents, with peak incidence between ages 2 and 5 years. Of the histological variants of RMS, the spindle cell type of RMS is particularly uncommon, accounting for approximately 5% of cases. Effective management typically involves a combination of surgery, chemotherapy, and sometimes radiation therapy, with prognosis influenced by factors such as tumor stage and genetic profile. Early diagnosis is essential for improving survival rates and reducing recurrence risk.

Case Presentation: We present the case of a 17-year-old male with scrotal swelling following trauma. Ultrasound revealed a large hypoechoic mass in the left hemi-scrotum. A left radical orchiectomy was performed. Histopathology showed a spindle cell tumor with a mutation in NRAS mutation (Q61L) and positive for skeletal muscle markers including Myogenin, MyoD1 (myogenic differentiation 1), Desmin, Vimentin, and Actin. Staging imaging showed an enlarged left para-aortic lymph node and a small pleural nodule. The patient was scheduled for three cycles of chemotherapy as initial management and regular follow-up with imaging.

Conclusion: Spindle cell RMS necessitates prompt diagnosis and intervention to reduce morbidity and mortality. This case highlights the importance of accurate assessment, diagnosis and management to minimize recurrence risk.

KEYWORDS

rare malignancy, sarcoma, spindle cell rhabdomyosarcoma, testicular rhabdomyosarcoma, testicular tumor, urological oncology

1 | INTRODUCTION

Para-testicular rhabdomyosarcoma (RMS) is a rare and aggressive malignancy that originates from the mesenchymal tissues around the testis. It is mostly seen in children and adolescents, but some cases have been reported in young adults^[1].

Histologically, RMS has various types, including embryonal, alveolar, pleomorphic, spindle, and mixed-

type, with embryonal RMS being the most frequently observed, while spindle cell RMS accounts only for approximately 5% of testicular mass cases^[2]. Effective management of para-testicular RMS generally involves a combination of surgery, chemotherapy, and occasionally radiation therapy^[3]. The prognosis depends on several factors, including tumor stage, tumor size, genetic signature, and response to treatment. Early diagnosis is crucial for improving survival rates and

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reducing the risk of recurrence^[3]. Herein, we present a case of para-testicular RMS spindle cell type in an adolescent, and discuss the presentation, treatment, diagnostic challenges, and outcome.

2 | CASE PRESENTATION

A 17-year-old young male, presented to the hospital immediately after sustaining trauma to his scrotum while playing football. On clinical examination, left scrotal swelling was noted. Scrotal Doppler ultrasound showed a large, mixed echogenic, predominantly hypoechoic lobulated mass lesion located in the inferior aspect of the left testis, displacing the interscrotal membrane and pushing the left testis superiorly and anteriorly. The mass also extends to the medial aspect of the spermatic cord. Apart from the displacement, the left testis appears normal in size, shape, and echotexture (Figure 1). A contrasted staging CT scan showed an enlarged left para-aortic lymph node measuring 23 mm × 16 mm with heterogeneous enhancement and no definite calcifications, small external iliac and ilio-colic lymph nodes (Figure 2), and a small pleural-based nodule at the superior segment of left upper lobe measuring 3 mm. Blood tests showed normal Alpha-fetoprotein and normal B-HCG. A left radical orchiectomy was performed. Postoperative course was uneventful. Histopathology showed a spindle cell tumor in which the cells are arranged in fascicles (Figure 3) and showed reactivity for skeletal muscle markers, including

NRAS mutation (Q61L) (Figure 4). Next-generation sequencing was performed on the tumor and showed recurrent genetic alterations. Table 1 summarizes the immunohistochemical profile of our patient's tumor, demonstrating diffuse and strong positivity for myogenic markers (Myogenin, MyoD1, Desmin, Vimentin, Actin) and negativity for markers associated with other tumor types, confirming the diagnosis of para-testicular spindle cell RMS. A plan was made for three cycles of chemotherapy, with a follow-up chest CT scheduled after those cycles to monitor the lung lesion and the left paraaortic lymph node.



FIGURE 2 Computed tomography with contrast demonstrating an enlarged left para-aortic lymph node (23 mm x 16 mm) with heterogeneous enhancement on arterial phase (red arrow).

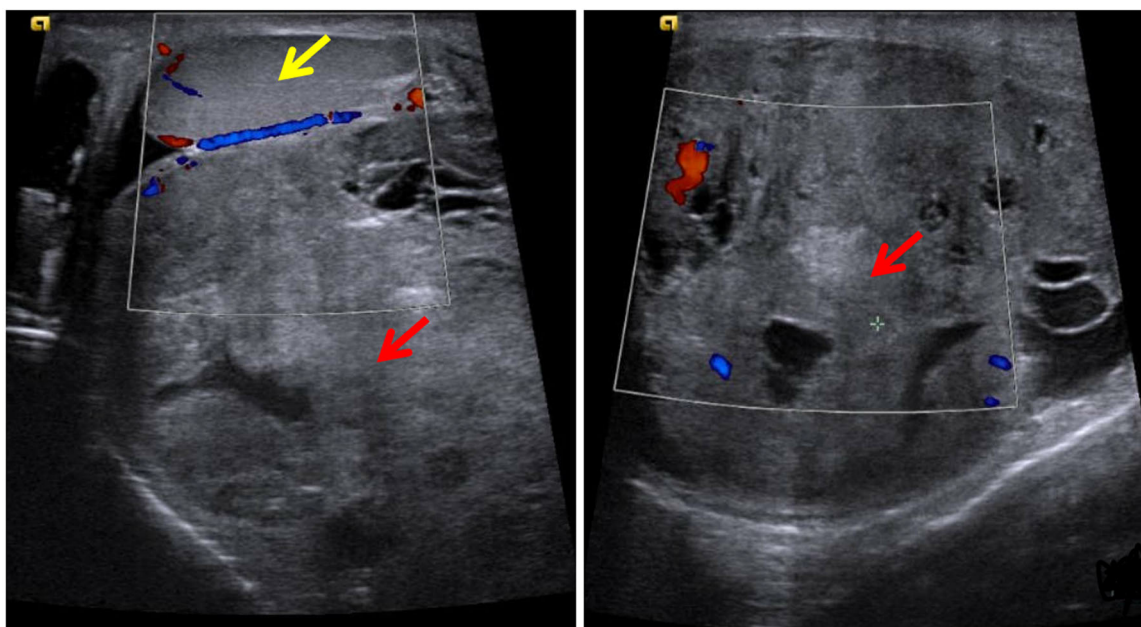


FIGURE 1 Scrotal ultrasound image showing the left testis (yellow arrow) displaced superiorly and anteriorly by a large, mixed echogenic, predominantly hypoechoic lobulated mass (red arrows).

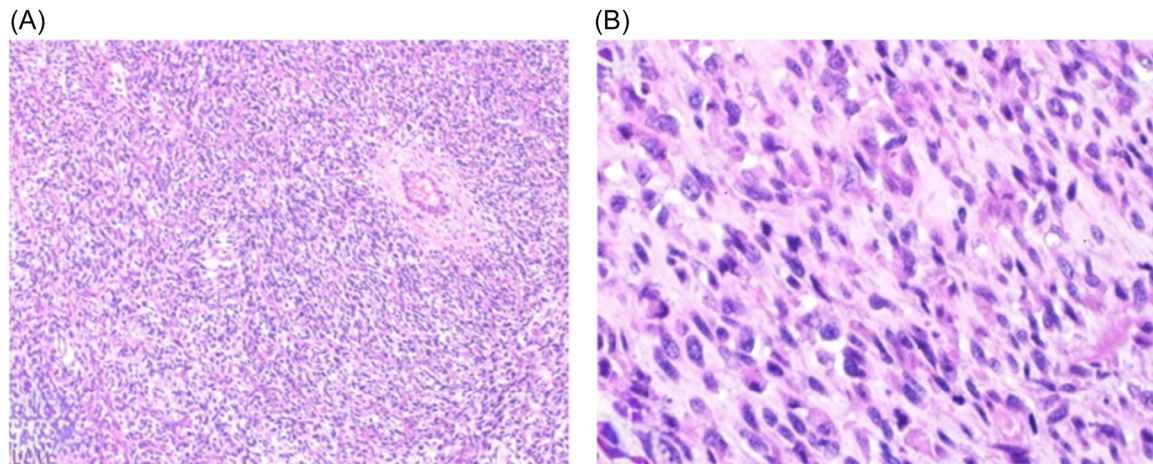


FIGURE 3 Microscopic features of the tumor. (A) Cellular spindle cell tumor in which the cells have fascicular arrangement (×40). (B) The tumor cells exhibit nuclear atypia with mitotic activity, some of the cells have deeply eosinophilic cytoplasm (×200).

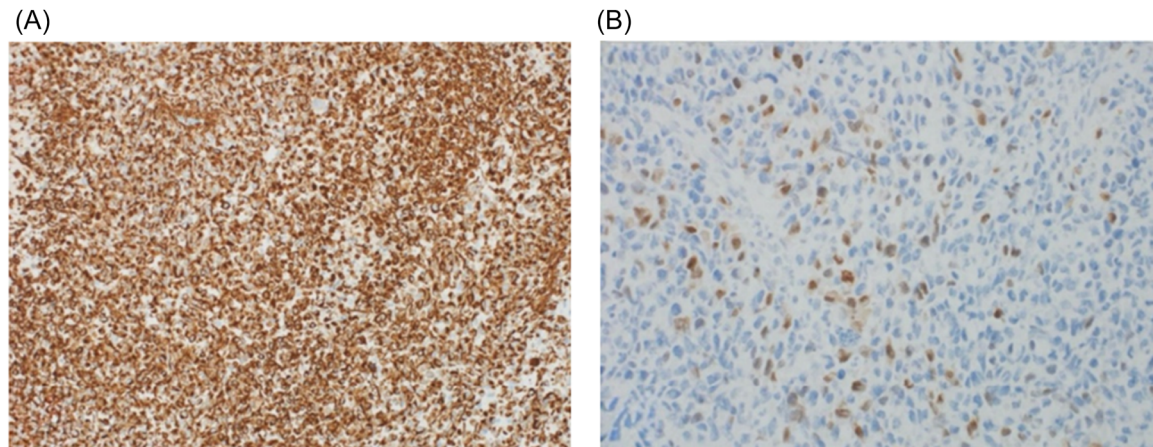


FIGURE 4 Immunohistochemical features of the tumor. (A) The tumor cells demonstrate diffuse strong reactivity for desmin (×100). (B) Focal moderate to strong nuclear staining for myogenin (×100).

3 | DISCUSSION

Para-testicular RMS is a rare tumor that predominantly affects adolescent^[4]. Adult testicular RMS is a rare and more aggressive entity^[1]. We present the case of an adolescent patient diagnosed with para-testicular spindle cell RMS, one of the rarest subtypes of RMS.

Histologically, spindle cell RMS can be particularly challenging to diagnose due to its resemblance to other spindle cell neoplasms. It can present with atypical features that may lead to initial misdiagnosis. The differential diagnosis for spindle cell RMS at the histological point includes a range of other spindle cell tumors such as fibrosarcoma and leiomyosarcoma^[5]. This makes precise histological evaluation and immunohistochemical staining crucial for accurate diagnosis even though the treatment of spindle cell RMS does not differ significantly in principle from other RMS types^[1]. However, the rarity of the spindle cell variant

necessitates a careful review of histological findings to ensure that the treatment regimen is appropriate^[6].

Spindle cell RMS is characterized histologically by spindle cell tumoral proliferation that are arranged in fascicles. The tumor may show focally evidence of skeletal muscle differentiation in the form of rhabdomyoblasts. These cells are characterized by having eccentric nuclei and deeply eosinophilic cytoplasm. By immunohistochemistry, the tumor cells are typically immunoreactive for Myogenin, MyoD1 (myogenic differentiation 1), Desmin, Vimentin, and Actin (markers of skeletal muscle differentiation)^[7].

Recently, it has been shown that spindle cell RMS shows various genetic alterations. The genotype of the tumor has been found to have a prognostic significance. For example, tumors with VGLL2/NCOA2 rearrangements have been found to carry favorable prognosis, while tumors that carry MYOD1 mutation behave more aggressively. A subset of tumors lacks recurrent genetic

TABLE 1 Immunohistochemistry results.

Marker	Result
Myogenin	Diffuse, strong positive
MyoD1	Diffuse, strong positive
Desmin	Diffuse, strong positive
Vimentin	Diffuse, strong positive
Actin	Diffuse, strong positive
CD99	Negative
FLI1	Negative
NKX2.2	Negative
EMA	Negative
MUC4	Negative
SMA	Negative
CK AE1/AE3	Negative
S100	Negative
SALL4	Negative
MDM2	Negative
Glypican	Negative
OCT3/4	Negative
AFP	Negative
CK7	Negative

abnormalities, those tumors have been found to have an overall better prognosis, as in our case^[7].

Patients usually present with palpable mass in the scrotum or inguinal region, often without significant pain. Scrotal swelling or discomfort may also be noted. Also, in some cases, systemic symptoms may present, which can indicate advanced disease^[1,8].

Early detection of para-testicular RMS is crucial due to its impact on treatment outcomes and overall prognosis and increases the likelihood of favorable outcomes. Early intervention is also important to prevent the tumor from advancing to a more aggressive stage^[1,6]. Imaging studies, including ultrasound and MRI, are essential for diagnosis, while histopathological analysis confirms the presence of the disease^[6,9].

The standard treatment of para-testicular spindle cell RMS involves surgery, ensuring complete resection to minimize the risk of recurrence^[9]. Surgery is typically followed by chemotherapy and radiation therapy, although specific chemotherapeutic agents and dosages may be adjusted based on tumor behavior and patient response^[6,8]. As demonstrated in our case, complete surgical resection was followed by chemotherapy.

4 | CONCLUSION

Para-testicular spindle cell RMS poses distinct diagnostic and therapeutic challenges due to its unique histological and genetic characteristics. Early detection and timely intervention are crucial for reducing morbidity and mortality. Our case emphasizes the importance of accurate histological assessment and prompt management. Regular follow-up remains essential for achieving favorable long-term outcomes and prompt detection of recurrence.

AUTHOR CONTRIBUTIONS

The case report, clinical documentation, literature review, and manuscript drafting were primarily carried out by Dr. Kholoud Alabassi and Dr. Yaser M. Ata. Dr. Ibrahim A. Khalil critically reviewed and revised the manuscript. The histopathological evaluation was conducted by Dr. Khaled Murshed and reviewed by Dr. Nohier M. Taha. Dr. Khalid Al Rumaihi supervised the project and provided overall guidance. All authors reviewed and approved the final version of the manuscript for publication.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

ETHICS STATEMENT

This case has been reviewed and approved by the Institutional Review Board (IRB) under approval number MRC-04-24-857, ensuring adherence to ethical guidelines and participant safety protocols.

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