



## Paratesticular Embryonal Rhabdomyosarcoma with Lung Metastasis – A Rare Case Report

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### ABSTRACT

Paratesticular rhabdomyosarcomas (RMS) are very rare malignant tumors arising from the mesenchymal tissues of tunica, epididymis or spermatic cord. They present as painless hard masses in inguinoscrotal region, and large tumors can be mistaken for testicular tumors. They can spread to retroperitoneal lymph nodes or hematogenously metastasize to lung, bones and bone marrow. Here, we report a case of Embryonal RMS with spindle cell component presenting with painless scrotal mass and lung metastasis at initial diagnosis.

**Keywords:** Paratesticular RMS, Embryonal, Lung Metastasis, Myogenin

### Introduction

Rhabdomyosarcomas (RMS) of the paratesticular region are very rare and comprise only 7% of all RMS and 17% of all intrascrotal malignant tumors (children < 15 years old).<sup>(1)</sup> They are aggressive tumors and present as painless hard scrotal masses.<sup>(2)</sup> Embryonal subtype is the most common histomorphology encountered.<sup>(3)</sup> Diagnosis is based on a high degree of suspicion, radio-imaging for staging the tumor and histological examination & immunohistochemical confirmation. Multi-modality treatment comprising of high inguinal orchidectomy, systemic chemotherapy and radiotherapy is helpful in attaining maximum tumor control.<sup>(4)</sup>

### Case Report

A 5-year-old male child was brought by his mother with the complaints of right testicular swelling for 8 months duration. The swelling was gradually progressive and it was not associated with pain or fever. On palpation of right scrotal swelling, there was no warmth or tenderness and the swelling was firm in consistency. Left testis was unremarkable upon examination.

Ultrasound of scrotum showed suspiciously elongated right testis with a large solid isoechoic heterogenous mass with internal vascularity. The provisional diagnosis was right testicular tumor, following which a PET-CT scan was performed which revealed a heterogenous lesion (134 x 95 x 92 mm) in the right scrotum replacing the testis, with <sup>18</sup>F-fluoro-2-deoxy-D-glucose (FDG) avid solid and cystic components noted within. Also, 2 nodules of size 4 x 4 mm and 2 x 2 mm were noted in the postero-basal segment and

apical segment of right lower and upper lobes of the lung respectively.

Right orchidectomy was done. On gross examination, the external surface was grey-white and irregular. Cut surface showed a solid grey-white mass measuring 12.8 x 7.5 x 7 cm replacing the entire sac wall. Compressed testicular tissue was also identified. Histopathological examination of orchidectomy specimen showed sheets of small round to oval cells with scant cytoplasm, hyperchromatic nuclei with focal anaplasia and areas of spindled out cells. Extensive rhabdomyoblastic differentiation was made out in some areas. Circumferential margin was focally involved. By IHC, the tumor cells were positive for Vimentin, Desmin, Myogenin and TLE1 (10%) focally in spindle cells. The diagnosis was Paratesticular Embryonal rhabdomyosarcoma with extensive rhabdomyoblastic differentiation, focal spindle cell component and diffuse anaplasia.

Post surgically, patient was placed on systemic chemotherapy VAC regimen (Vincristine, Dactinomycin and Cyclophosphamide). After 6 cycles of chemotherapy, PET-CT scan was repeated to assess metastasis. The lung nodules were showing no increase in size to the previous scan and there was no active metabolic residual disease in scrotal sac. Following this, Right hemiscrotectomy along with biopsy of lung nodule was done. The right hemiscrotectomy tissue specimen showed granulation tissue formation, while the lung biopsy confirmed the presence of metastatic tumor deposits. Patient is currently on chemotherapy with routine follow-up.



## Discussion

Rhabdomyosarcoma (RMS) is one of the commonest soft tissue sarcomas of childhood. However, paratesticular

location of RMS is very rare and constitutes only 7% of all rhabdomyosarcomas. They develop from the mesenchymal tissues of tunica, epididymis or spermatic cord. <sup>(5)</sup>The

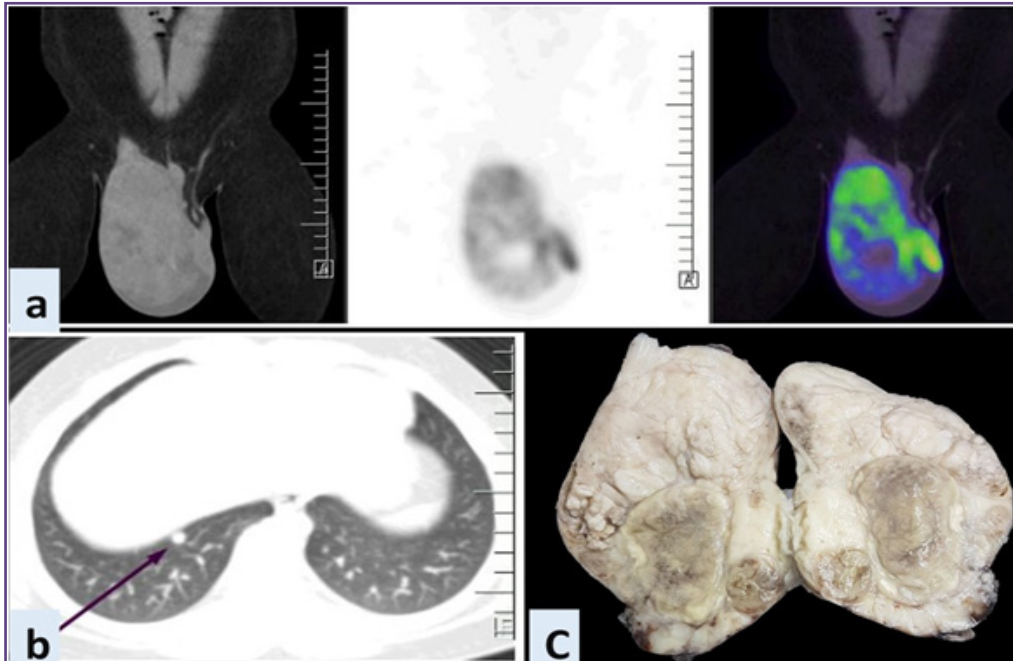


Fig. 1a: PET CT scan showing intrascrotal tumor with FDG uptake. b.) Lung nodule seen in the right lower lobe. C.) Gross specimen of right orchidectomy showing grey-white solid tumor with compressed testicular tissue.

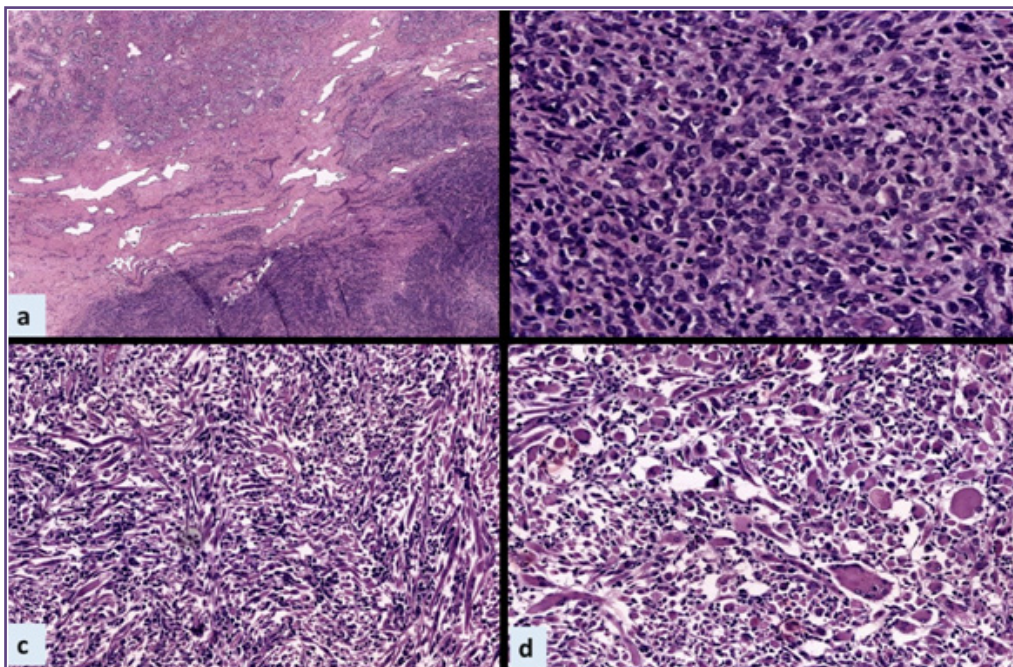


Fig. 2a: Tumor with adjacent compressed testicular parenchyma b.) Embryonal RMS showing sheets of small round cells c.) Spindle cell component d.) Rhabdomyoblasts (strap cells).

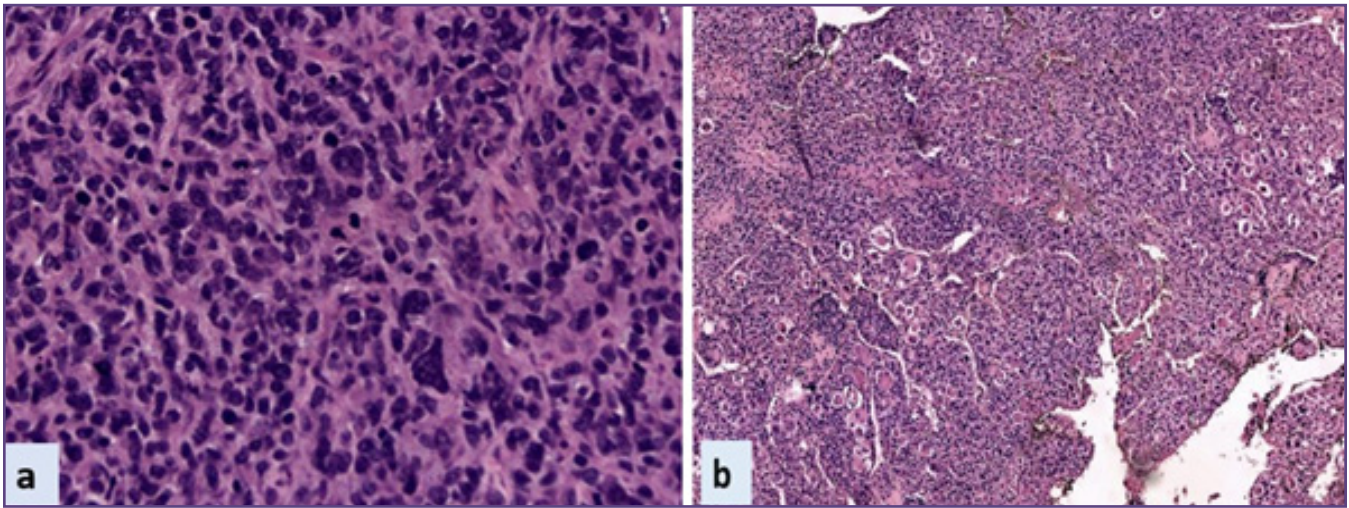


Fig. 3a: Scattered large anaplastic cells b.) Lung nodule showing tumor deposit.

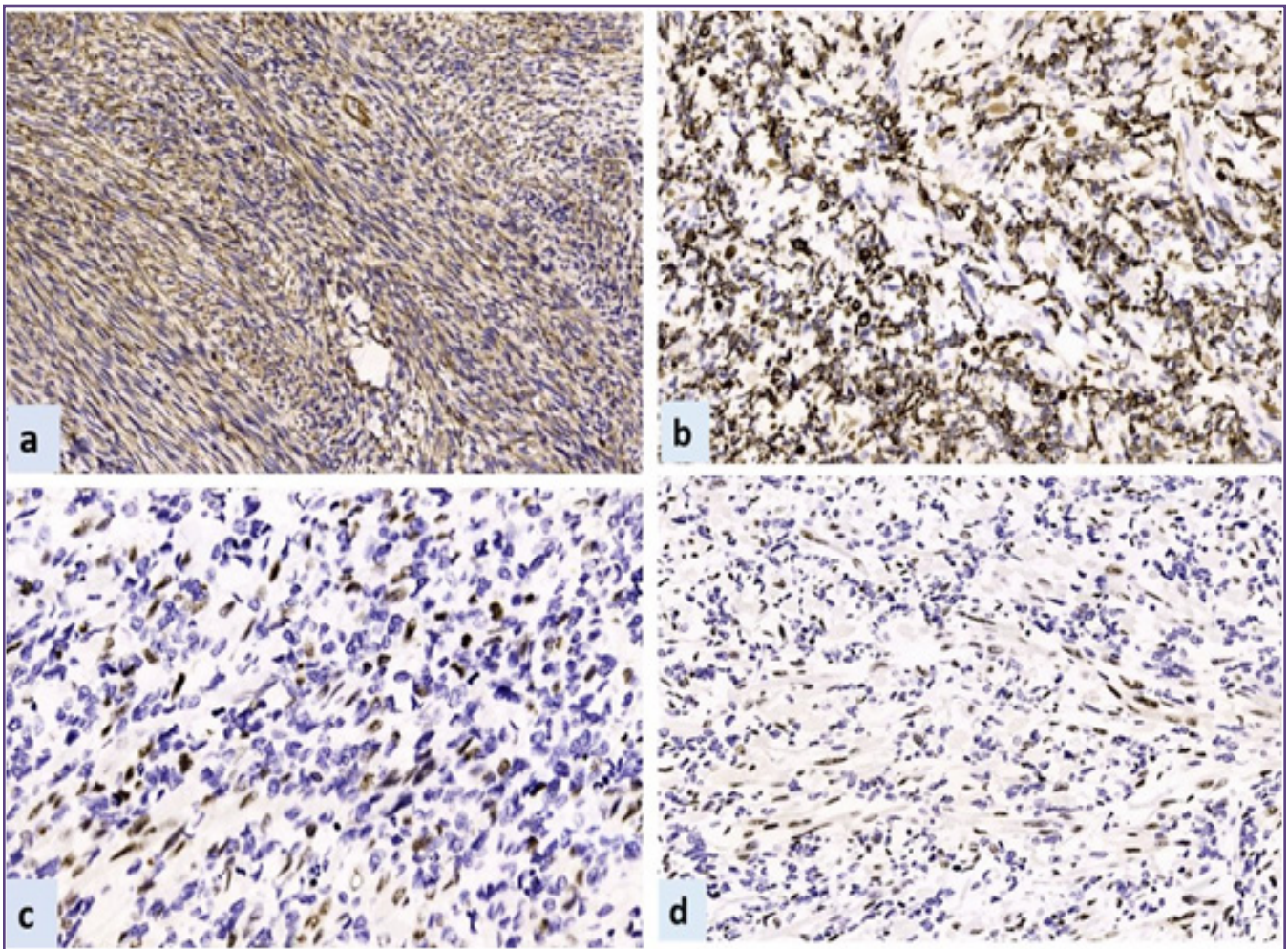


Fig. 4a.: Tumor cells showing positivity for Vimentin b.) Desmin c.) Myogenin d.) Focally for TLE1.

other common sarcomas in the paratesticular region are leiomyosarcoma (32%), liposarcoma (20%), and malignant fibrous histiocytoma (13%).<sup>(6)</sup> There are two peaks of incidence, with the first peak more commonly in 1-5 years followed by a second peak in adolescence.<sup>(2)(5)(7)</sup>

The tumor presents as a painless hard mass in the inguinoscrotal region which rarely invades the scrotal skin. The tumor can be confused with conditions like epididymo-orchitis, testicular torsion, scrotal abscess, hydrocoele, strangulated hernia and rarely testicular tuberculosis.<sup>(2)(5)</sup>

However, painful lesions are seen in only 7% of cases.<sup>(6)</sup> Large tumors can be mistaken for tumors of testicular origin as in our case. Spread of tumor can occur via lymphatics preferentially to retroperitoneal lymph nodes and by hematogenous spread to lung, bone and bone marrow.<sup>(4)(5)(8)</sup>

For initial workup, scrotal ultrasonography is the imaging modality of choice. A frequency of 7.5-10 MHz is required to distinguish testicular and paratesticular lesions. USG helps to demonstrate the solid component, and eliminate differential diagnosis like hydrocoele and varicocele. CT scan of thoraco-abdomino-pelvic regions is performed to look for retroperitoneal and mediastinal lymphadenopathy and distant metastasis. <sup>18</sup>F-fluoro-2-deoxy-D-glucose (FDG) PET-CT scan has been shown to be a sensitive tool in staging and re-staging of RMS and also in assessment of chemotherapy response.<sup>(7)</sup>

Based on histomorphology, three types of RMS have been identified, namely embryonal type, alveolar type and pleomorphic type, of which embryonal type is the most common subtype, making up approximately 80% of the cases. Additional variants have more recently been identified, such as spindle cell RMS and sclerosing RMS that comprise 5% to 10% of RMS.<sup>(7)</sup> Rhabdomyoblast is the characteristic cell but is not essential for diagnosis.<sup>(2)(4)</sup> In embryonal RMS, small round cells with scant rim of eosinophilic cytoplasm and small oval nuclei as well as spindled out cells with cross striations are seen in a loose myxoid stroma. Occasional tumor cells with abundant eosinophilic cytoplasm (strap cells) are a feature of rhabdomyoblastic differentiation. On Immunohistochemistry, the tumour cells are immunoreactive for desmin, actin and myogenin and negative for Pan CK, S100 and CD99. Immunoreactivity for myogenin clinches the diagnosis.<sup>(4)</sup>

The differential diagnosis (DD) for rhabdomyosarcoma include leiomyosarcoma, fibrosarcoma, dedifferentiated

liposarcoma, malignant fibrous histiocytoma, malignant triton tumor and malignant mesothelioma.<sup>(4)(9)(10)</sup> However, these tumors occur more commonly in adults. Small round cell tumors should be included in the differential diagnosis of Embryonal RMS. Non Hodgkin lymphoma (NHL) of B cell type involving the testis can mimic Embryonal RMS. NHL (B cell type) will usually be positive for CD45 and CD20 and negative for desmin and myogenin.<sup>(11)</sup>

Other DD include desmoplastic small round cell tumor (DSRCT) and extraskeletal Ewing sarcoma. DSRCT mimics Embryonal RMS as it frequently shows desmin positivity and an alveolar pattern imparted by dense sclerosis that separates tumor nests cells. However, Myogenin will be negative in DSRCT and helps in differentiating the two. CD99, often used as a marker in Ewing sarcoma may be positive in rhabdomyosarcoma, whereas FLI-1 will be negative.<sup>(9)</sup> Myogenin positivity helps to clinch the diagnosis of RMS in such cases.

Localized tumors are associated with good prognosis whereas metastatic disease is associated with poor prognosis. Multimodal therapy using systemic chemotherapy in conjunction with either surgery, radiotherapy is used to maximize tumor control. For localized tumor, high inguinal orchidectomy is the treatment of choice. Hemiscrotectomy is performed if there is scrotal violation.<sup>(12)</sup>

The use of retroperitoneal lymph node dissection (RPLND) is controversial. The protocol for Children's Oncology Group (COG) however recommends that staging ipsilateral RPLND be done for all boys aged >10 years, and in those < 10 years it is reserved for those with suspicious lymph nodes on CT scan. RMS is chemo-sensitive and the most commonly used chemotherapeutic regimen is Vincristine, Dactinomycin and Cyclophosphamide (VAC).<sup>(7)</sup>

## Conclusion

Paratesticular RMS are rare aggressive tumors. Due to varied presentation and histomorphology, IHC plays a crucial role in diagnosis of this tumor. Positive myogenin clinches the diagnosis. Radio-imaging plays a crucial role in diagnosis and staging.

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## Competing Interests

Authors have no competing interests to declare.

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