ORIGINAL RESEARCH PAPER

Surgery

EMBRYONAL RHABDOMYOSARCOMA OF THE TESTIS IN A 19-YEAR- OLD MALE: A RARE CASE REPORT AND LITERATURE REVIEW

KEY WORDS: Embryonal rhabdomyosarcoma, testicular cancer, young adult, case report

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ABSTRACT

Background: Primary testicular embryonal rhabdomyosarcoma (ERMS) is an exceptionally rare malignancy, accounting for less than 1% of all rhabdomyosarcoma cases and less than 3% of all childhood cancers. Only 25 cases of primary testicular ERMS have been reported in the literature as of March 2025. **Case Presentation:** We present a case of a 19-year-old male who initially presented with right testicular swelling that was misdiagnosed as varicocele. Following multiple surgical interventions and a delayed diagnosis, histopathological examination confirmed embryonal rhabdomyosarcoma of the testis. The patient underwent radical inguinal orchiectomy followed by adjuvant chemotherapy with the VAC protocol. **Conclusion:** This case highlights the importance of maintaining a high index of suspicion for rare testicular malignancies in young adults presenting with atypical scrotal masses, even when conventional tumor markers are normal.

INTRODUCTION

Rhabdomyosarcoma (RMS) is a malignant soft tissue sarcoma arising from mesenchymal cells committed to skeletal muscle differentiation. Embryonal rhabdomyosarcoma (ERMS) represents the most common histological subtype, accounting for approximately 60% of all RMS cases and 15-20% of all soft tissue sarcomas. While RMS predominantly affects children and adolescents, primary testicular involvement is exceedingly rare, representing less than 1% of all RMS cases and less than 3% of all childhood cancers.

The rarity of testicular ERMS, combined with its non-specific clinical presentation and normal conventional tumor markers, often leads to diagnostic delays and potential mismanagement. We report a case of primary testicular ERMS in a 19-year-old male, discussing the clinical challenges, diagnostic approach, and management strategy.

Case Presentation

Patient Demographics and Initial Presentation

A 19-year-old male presented with a one-year history of right testicular swelling that had been rapidly increasing in size over the preceding three months. The patient also complained of mild, dull ache in the right scrotum for one month duration.

Medical History

- October 2024: Initial presentation with similar complaints led to a diagnosis of right varicocele, for which he underwent right varicocelectomy at a government hospital.
- February 2025: Persistent swelling prompted ultrasonography, which revealed a right paratesticular lesion and left varicocele.
- March 2025: The patient underwent left varicocelectomy with right paratesticular lesion biopsy via scrotal incision.
- March 29, 2025: Patient was referred with a histopathological report suggestive of embryonal rhabdomyosarcoma of the testis.

Post-Biopsy Clinical Course

Following the biopsy procedure, the patient experienced progressive enlargement of the right scrotal swelling from an initial size of 2×2 cm to 5×4 cm at the time of presentation, accompanied by increasing hardness and mild pain.

Physical Examination

- Hard, non-tender, irregular 5×4 cm mass in the right hemiscrotum, distinct from the testis
- No cord thickening
- Normal external genitalia
- Previous bilateral inguinal and right varicocelectomy scars over the right scrotum
- Normal contralateral scrotum

Laboratory Investigations

AFP: 1.29 ng/mL (normal <15 ng/mL)
-hCG: <0.2 ng/mL (normal 0 ng/mL)
LDH: 191 U/L (normal <250 U/L)

All other hematological and biochemical investigations were within normal limits.

Imaging Studies

Scrotal Ultrasonography: Heterogeneous, solid, hypoechoic mass clearly separate from the right testis.

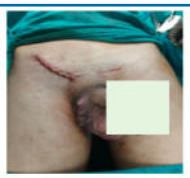
Chest X-ray: Normal findings with no evidence of pulmonary metastases.

PET-CT: Hypermetabolic soft tissue density mass lesion in the right testis measuring 5.2×4.5 cm with SUV max of 5.37. No significant hypermetabolic pelvic or retroperitoneal lymph nodes were identified, IRS Stage I, Group I.

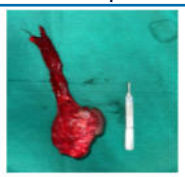
Treatment Approach

1. Radical inguinal orchiectomy with clear margins.
2. RPLND considered but deferred due to no radiographic evidence of nodal involvement.
3. Adjuvant chemotherapy (VAC protocol):
   - Vincristine: 1.5 mg/m² IV weekly
   - Dactinomycin: 0.045 mg/kg IV q3weeks
   - Cyclophosphamide: 2200 mg/m² IV q3weeks with Mesna
   - Duration: 42 weeks (6 cycles)
4. Regular follow-up for recurrence/metastases.

High inguinal incision and separate scrotal incision



Skin closure



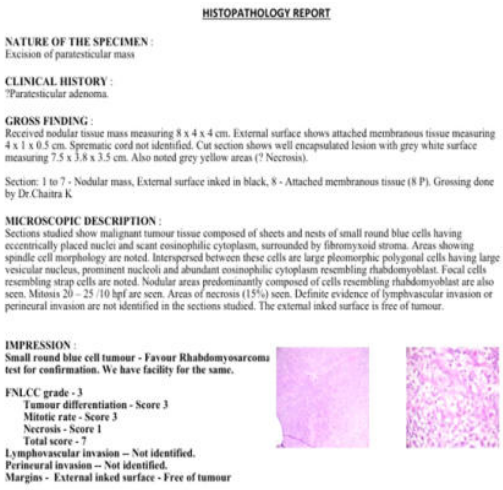
Resected Specimen- Testis with cord structures

Histopathological Analysis

Biopsy: Spindle cells, primitive round cells, and rhabdomyoblasts in myxoid stroma.

Immunohistochemistry: Positive for Desmin, MyoD1, Myogenin confirming rhabdomyoblastic differentiation.

Post-Orchiectomy specimen reconfirmed ERMS diagnosis.



HISTOPATHOLOGY REPORT

NATURE OF THE SPECIMEN :
Excision of paratesticular mass

CLINICAL HISTORY :
?Paratesticular adenoma.

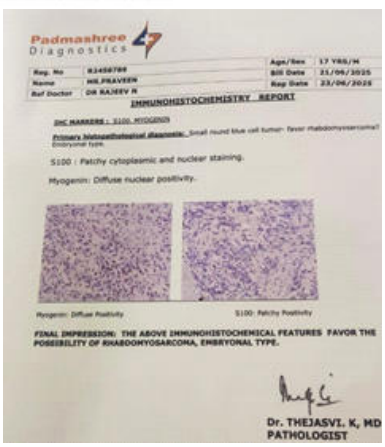
GROSS FINDING :
Received nodular tissue mass measuring 8 x 4 x 4 cm. External surface shows attached membranous tissue measuring 4 x 1 x 0.5 cm. Spermatic cord not identified. Cut section shows well encapsulated lesion with grey white surface measuring 7.5 x 3.8 x 3.5 cm. Also noted grey yellow areas (? Necrosis).

Section: 1 to 7 - Nodular mass, External surface inked in black, 8 - Attached membranous tissue (8 P). Grossing done by Dr.Chaitra K

MICROSCOPIC DESCRIPTION :
Sections studied show malignant tumour tissue composed of sheets and nests of small round blue cells having eccentrically placed nuclei and scant eosinophilic cytoplasm, surrounded by fibromyxoid stroma. Areas showing spindle cell morphology are noted. Interspersed between these cells are large pleomorphic polygonal cells having large vesicular nucleus, prominent nucleoli and abundant eosinophilic cytoplasm resembling rhabdomyoblast. Focal cells resembling strap cells are noted. Nodular areas predominantly composed of cells resembling rhabdomyoblast are also seen. Mitosis 20 - 25 /10 hpf are seen. Areas of necrosis (15%) seen. Definite evidence of lymphvascular invasion or perineural invasion are not identified in the sections studied. The external inked surface is free of tumour.

IMPRESSION :
Small round blue cell tumour - Favour Rhabdomyosarcoma test for confirmation. We have facility for the same.

FNLCC grade - 3
Tumour differentiation - Score 3
Mitotic rate - Score 3
Necrosis - Score 1
Total score - 7
Lymphovascular invasion -- Not identified.
Perineural invasion -- Not identified.
Margins - External inked surface - Free of tumour



Padmashree Diagnostics

IMMUNOHISTOCHEMISTRY REPORT

S100 : Patchy cytoplasmic and nuclear staining.

Myogenin: Diffuse nuclear positivity.

FINAL IMPRESSION: THE ABOVE IMMUNOHISTOCHEMICAL FEATURES FAVOR THE POSSIBILITY OF RHABDOMYOSARCOMA, EMBRYONAL TYPE.

Dr. THEJASVI. K, MD
PATHOLOGIST

Differential Diagnosis

- Right-sided epididymal cyst
- Right-sided testicular tumor
- Varicocele (initial misdiagnosis)

Discussion

Epidemiology and Rarity

Primary testicular ERMS is extremely rare, with only 25 cases reported as of March 2025. RMS peaks at ages 1-5 and 15-20 years, with adult onset >20 years being uncommon.

Risk Factors

Reported risk factors include:

- Cryptorchidism
- Trauma
- Advanced maternal age
- Paternal cigarette smoking
- Exposure to maternal estrogen
- X-ray exposure

Clinical Challenges

1. Non-specific presentation mimicking benign lesions.
2. Normal AFP and -hCG causing confusion.
3. Misdiagnosis led to scrotal violation and potential tumor seeding.

Diagnostic Approach

Diagnosis requires imaging, biopsy with IHC, and staging studies. Avoid scrotal approaches.

Treatment Considerations

Radical orchiectomy is standard. RPLND in select high-risk cases. All patients require chemotherapy (VAC).

Prognosis

5-year survival ~95% in paratesticular RMS. Favorable factors: age <10, embryonal histology, complete resection, no nodal disease.

Learning Points

1. Maintain suspicion in atypical scrotal masses.
2. Avoid scrotal surgery prior to diagnosis.
3. Multidisciplinary management is key.
4. Long-term follow-up essential.

CONCLUSION

Primary testicular embryonal rhabdomyosarcoma is rare and diagnostically challenging. Early recognition, correct surgical approach, and multimodal treatment are critical for favorable outcomes.

Declarations

Ethical Approval: Prepared in accordance with institutional guidelines.

Consent: Written informed consent obtained.

Competing Interests: None.
Funding: None.

REFERENCES

1. Intergroup Rhabdomyosarcoma Study Group guidelines and protocols
2. DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology, 11th edition
3. Recent literature from IRS-V and SIOP-MMT studies (Journal of Clinical Oncology)
4. WHO Classification of Soft Tissue Tumors
5. EpSSG guidelines