DOI: https://doi.org/10.4038/slju.v15i1.4113

Sri Lanka Journal of Urology.2024;**15**(1):52-56

Case Report

From diagnosis to remission: A rare case of para testicular embryonal rhabdomyosarcoma in a young male.

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Abstract

We present the case of a 24-year-old male diagnosed with paratesticular embryonal rhabdomyosarcoma, who presented with a painless left inguinal swelling. Initial investigations, including ultrasound and biopsy, indicated a poorly differentiated malignancy, later confirmed as embryonal rhabdomyosarcoma by immunohistochemistry. Following multidisciplinary team discussions, the patient underwent radical inguinal orchidectomy, and adjuvant chemotherapy. Post-treatment PET-CT scans revealed complete resolution of metastatic lymph nodes. Nine months after the initial surgery, the patient is doing well, with no evidence of active disease. This case underscores the importance of a comprehensive, multidisciplinary approach in the management of rare and aggressive paratesticular rhabdomyosarcoma.

Introduction

Paratesticular rhabdomyosarcoma is a rare malignant tumors arising from mesenchymal tissue. It predominantly affects children and adolescents, accounting for a small percentage of paediatric malignancies (1). This tumour type is known for its aggressive nature and potential for metastasis, often presenting as a painless inguinal or scrotal mass. Early diagnosis and prompt treatment, including surgery, chemotherapy, and radiation therapy, are crucial for improving outcomes. This case report details the presentation, diagnosis, and multidisciplinary management of a 24-year-old male with metastatic Paratesticular embryonal rhabdomyosarcoma, highlighting the challenges and strategies in treating this rare condition.

Case Presentation

A 24-year-old male presented to the ward with a painless left inguinal swelling that had persisted for a few weeks. He had no history of trauma or previous inguinal surgery. He also denied any symptoms indicative of metastasis or cancer cachexia, and there was no family history of malignancy. On examination, he was a thin, normally built young man. Abdominal examination revealed a large, hard inguinal mass extending into the left testis along the cord structures. An initial ultrasound scan suggested the presence of a vascular mass in the inguinal and scrotal region and confirmed by computed tomogram images (Figure 1a, b). An inguinal lymph node biopsy indicated a poorly differentiated malignancy, with the differential diagnosis including rhabdomyosarcoma or a poorly

differentiated germ cell tumour. Immunohistochemistry later confirmed the diagnosis of rhabdomyosarcoma.

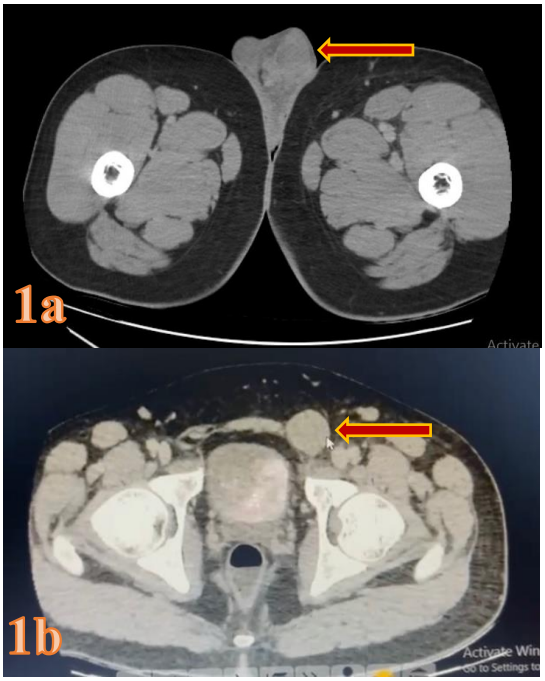

Fig 1a. *CT image- the left testicular mass*, and **1b**.*extension of the mass into the left inguinal canal.*

The case was discussed at a multidisciplinary team meeting, and it was decided that the patient would undergo a left radical inguinal orchidectomy. The biopsy revealed a 140 x 45 x 40 mm Para testicular soft tissue mass with an atrophic testis. The epididymis and spermatic cord were incorporated into the tumour mass. The para-testicular tumour is composed of polygonal malignant cells with pleomorphic hyperchromatic nuclei. The cytoplasm is moderate but abundant in some cells. Frequent mitotic figures are noted; the background is myxoid in many areas (Figure 2a). Occasional tad pole-like cells are also present. The tumour cells show strong and focal positivity for desmin and myo-D1, respectively, with negativity for cytokeratin, PLAP, LCA and CD30 (Figure 2a, 2b). The immunomorphological features, in conjunction with the location and the age of the patient, a diagnosis of an embryonal rhabdomyosarcoma was made.

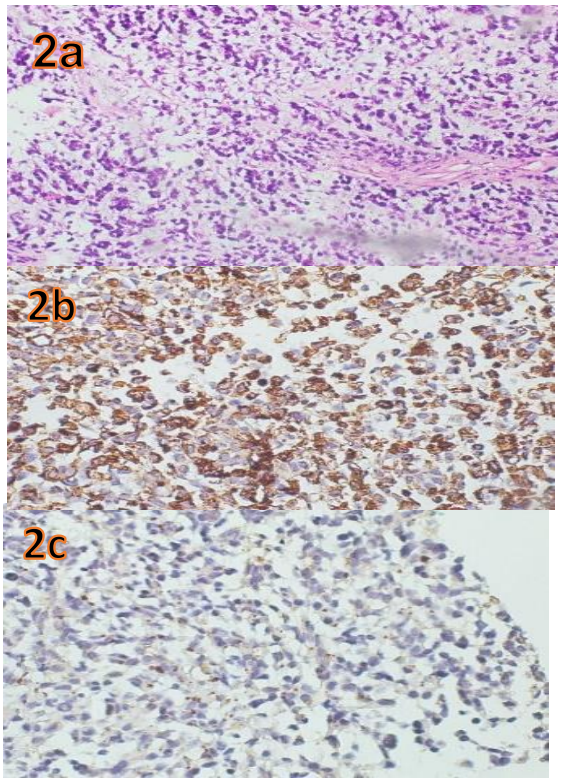

Fig 2a. *H & E x 20 image shows malignant rhabdoid cells in a myxoid background*, **2b.***Desmin immunostaining shows cytoplasmic and occasional perinuclear dot-like positivity in the tumour cells, and* **2c)** *Myo-D1 immunostaining shows positive nuclear staining in occasional tumour cells.*

Contrast-enhanced computed tomography (CT) scan of the chest, abdomen, and pelvis revealed multiple enlarged, enhancing lymph nodes along the left external and internal iliac vessels, left common iliac vessels (Figure 3a), and left infrarenal para-aortic, precaval, and retrocaval lymph nodes in the retroperitoneal space (Figure 3b), indicating metastatic involvement.

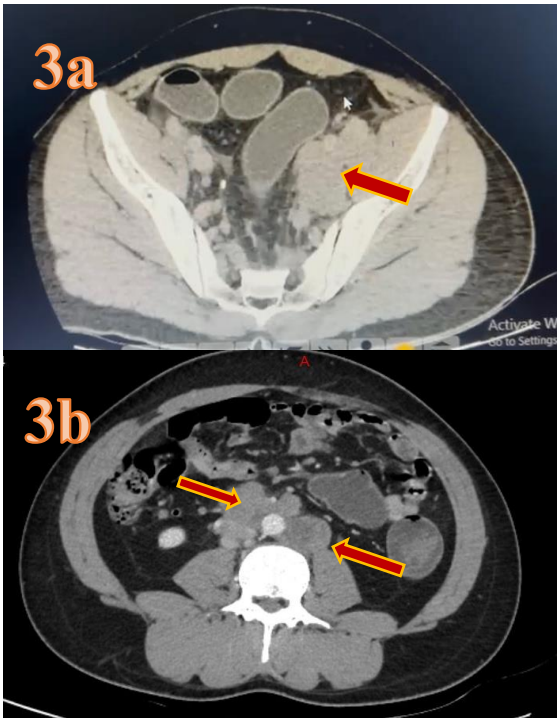

Fig 3a. *CT pelvis images - the enlarged lymph nodes along the iliac vessels*, **3b** *the enlarged left infra renal para-aortic and precaval lymph nodes.*

The case was again discussed at the multidisciplinary team meeting, and adjuvant chemotherapy was planned. The patient underwent several cycles of chemotherapy (Infosphamide, vincristine, actinomycin, and Doxorubacin), resulting in significant reduction and resolution of the lymph nodes after six to eight weeks, as evidenced by PET-CT scans (Figure 4a). Subsequently, he received radiation therapy to the residual lymph nodes in the pelvic and retroperitoneal region. Follow-up PET-CT scans of the chest, abdomen, and pelvis showed complete resolution of lymph node metastases and no evidence of active disease (Figure 4b). The patient is doing well nine months after the initial surgery.

Discussion

Paratesticular embryonal rhabdomyosarcoma (PT-RMS) is a rare and aggressive malignancy, accounting for approximately 7% of all rhabdomyosarcomas and typically affecting the paediatric population, with a bimodal age distribution, with peaks at 1 to 5 years and then at 16 years of age (1). However, its occurrence in young adults, as illustrated by this case, is exceedingly uncommon, posing unique diagnostic and therapeutic challenges.

PT-RMS can arise from the spermatic cord, epididymis, or testicular tunics, typically presenting as a painless scrotal mass (2). In this case, the patient had a painless inguinal mass, a common symptom of PT-RMS. Occasionally, it may present as bruising, hernia, or hydrocele. Ultrasound, a key diagnostic tool, often reveals a highly reflective mass with increased vascularity on colour Doppler (2). Serum tumour markers, such as β-HCG and alpha-fetoprotein, remain normal in PT-RMS, distinguishing it from testicular tumours (3). CT scans of the chest, abdomen, and pelvis are essential for staging and detecting metastases.

Para testicular Rhabdomyosarcoma is traditionally classified into three main histologic subtypes: embryonal, alveolar, and pleomorphic. Embryonal RMS is the most common, accounting for approximately 80% of PT-RMS cases. A study by Ferrari and colleagues reviewed outcomes of 216 patients with paratesticular RMS. The histologic subtypes were embryonal RMS in 181 cases (84%), alveolar RMS in 18 (8%), spindle cell RMS in 10 (5%), and “not

otherwise specified" in 7 (3%). Our case falls under the common embryonal histology subtype (4).

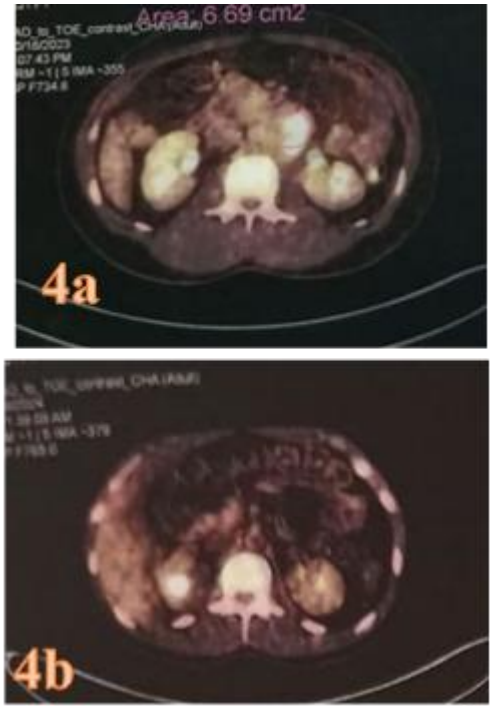

Fig 4a. *PET scan shows retroperitoneal lymph nodes with reduction in size after six cycles of chemotherapy*, **4b**. *The complete resolution of retroperitoneal lymph nodes after completion of chemoradiation.*

Management of paratesticular rhabdomyosarcoma generally requires a multimodal approach, combining surgery, chemotherapy, and radiation therapy. Before chemotherapy became standard, surgical resection of localized disease alone resulted in approximately a 50% two-year relapse-free survival rate (5). PT RMS patients typically undergo radical orchiectomy through an inguinal incision, with high ligation and dissection of the spermatic cord. Re-resection is necessary when there is inadequate initial resection or if histology reveals positive margins for localized disease. Approximately 25% of patients present with retroperitoneal lymph node metastasis. All patients aged 10 years or older should receive ipsilateral infrarenal lymph node evaluation, regardless of imaging findings (6).

Chemotherapy plays a crucial role in adjuvant treatment, improving disease-free survival. Actinomycin, vincristine, and cyclophosphamide are commonly combined, with 85.7% of patients achieving disease-free status at over two years of follow-up in one study (7). Adjuvant radiation therapy is indicated for incomplete surgical resection, gross residual disease, or positive retroperitoneal lymph nodes (8). Our patient presented with metastatic disease, received adjuvant chemotherapy, and later achieved complete resolution with radiation therapy after completing chemotherapy.

In conclusion, paratesticular embryonal rhabdomyosarcoma is a rare but aggressive tumour in adults. This case highlights the importance of early diagnosis, precise histopathological classification, and a multidisciplinary approach combining surgery, chemotherapy, and radiation therapy. Complete resolution of the disease was achieved, adding to the evidence supporting the effectiveness of comprehensive treatment strategies in managing PT-RMS.

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