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# Unusual Manifestation of Alveolar Rhabdomyosarcoma: A Case Report.

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**KEYWORDS**
Alveolar rhabdomyosarcoma, Metastasis, Immunohistochemistry.

**ABSTRACT:**

**Introduction**: Rhabdomyosarcomas (RMS) are rare soft-tissue tumours. It accounts for 5% for all childhood cancers whereas around 0.03% in adults.

**Case report**: A 23-year-old Indian male complained of a progressively enlarging perianal mass associated with rectal bleeding, accompanied by scrotal involvement and tenderness. Diagnostic workup revealed a heterogeneous pelvic mass with extensive lymph node involvement on CT and PET scans. Fine needle aspiration cytology (FNAC) and USG-guided Tru-cut biopsy confirmed the diagnosis of ARMS, supported by immunohistochemical analysis showing positivity for vimentin, Desmin, myogenin, and Myo-D1. This case underscores the diagnostic challenges and comprehensive management required for diagnosis of rare malignancies.

**Conclusions**: Unusual location, atypical presentation of rhabdomyosarcoma often confuses the clinical diagnosis. Hence minimally invasive techniques for tissue retrieval and ancillary diagnostic tests will confirm the diagnosis and pave the way for proper management.

## Introduction

Rhabdomyosarcomas (RMS) represent a unique and rare form of soft-tissue tumours. They are particularly noteworthy because they encompass a mere 5% of all childhood cancers, while in adults, their prevalence is even lower, approximately 0.03%.[1] These tumours are primarily characterized by their histological types, of which there are three distinct varieties: Embryonal, Alveolar, and Pleomorphic. Among these, alveolar RMS is predominantly seen in young adults, marking it as a significant health concern for this age group.[2]

The most common site for alveolar RMS is the deep soft tissue of the extremities. This localization preference contributes to the complexity of diagnosis and treating this condition, as the symptoms can often be subtle or confused with other soft tissue ailments. [3,4] The rarity of this condition in adults further complicates the diagnosis, as it may not be the first consideration in differential diagnoses. [5]

Rhabdomyosarcomas, in general, and alveolar rhabdomyosarcoma, in particular, are aggressive tumours. They are derived from primitive mesenchymal cells with a propensity to differentiate into skeletal muscle tissue.[6] This differentiation aspect is critical in understanding the biological behaviour of these tumours, as well as in guiding therapeutic approaches.

The clinical presentation of RMS can vary, but it usually manifests as a soft tissue mass. However, in rare instances, symptoms can be atypical, making the diagnosis challenging.

## Case report

A 23-year-old Indian male presented with complaints of mass per rectum which was insidious in onset, gradually progressive in nature, became prominent while defecation. He also complained of bleeding per rectum which was sudden in onset gradually progressive, bright red colour associated while passing stools.

Upon physical examination, the patient appeared moderately built, with signs of pallor. On per rectal examination, reveals bulge over rectal wall with congestion measuring 4x4 cm mass. On palpation –A hard nodule arising from anterior wall of rectum noted extending into the scrotal wall and a hard palpable nodule in the scrotal wall. Tenderness was present. Other Systemic examination was unremarkable. And the patient was a known case of epilepsy disorder and on medication since 15 years.

## Laboratory Investigations

Laboratory results showed a haemoglobin level of 11.8 g/dL, a platelet count of 4,89,000/cu mm, and neutrophilia.

## CT Abdomen and pelvis

CT scan showed heterogenous mass in perianal region and enlargement of pelvic lymph nodes. Mildly lobulated heterogeneously enhancing soft tissue mass in the perianal region (**Right**>Left) with infiltration of bilateral ischioanal, ischiorectal fossae extending to anterior perineal region

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infiltrating right side of prostate, bilateral bulbospongiosus, right obturator internus muscles. Multiple discrete heterogeneously enhancing bilateral perirectal, left obturator, left external iliac, right external iliac, presacral, bilateral inguinal lymph nodes.

PET scan showed increased FDG uptake with SUVmax 2.7 in the pelvic mass and pelvic lymph nodes also showed increased uptake SUVmax 1.9. Differential diagnosis of Lymphoma Vs Metastasis. was given. (Fig A)

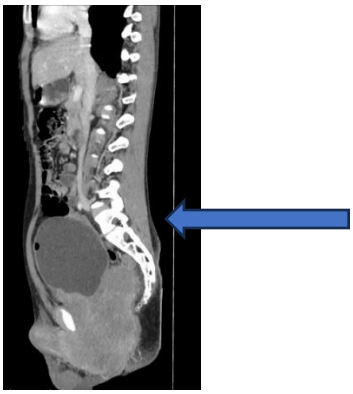
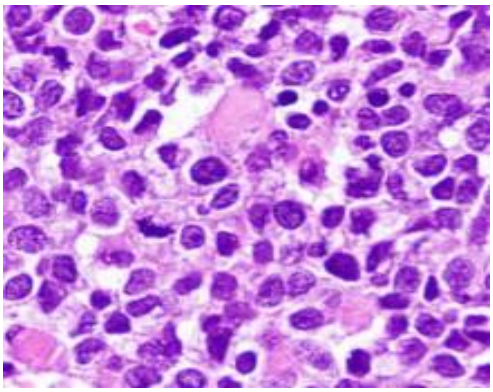


**Figure A: CT SCAN SHOWED HETEROGENOUS MASS IN PERIANAL REGION**

## FNAC

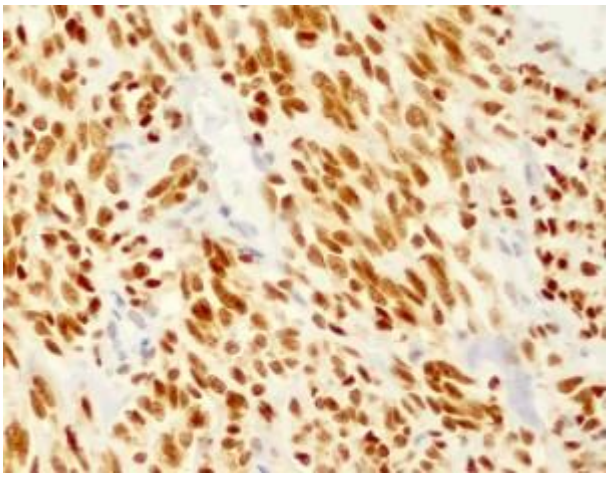
FNAC from bulge over scrotal region showed high cellularity, comprised of singly scattered pleomorphic cells as well few vague follicular/glandular patterns. Individual cells show coarse chromatin, irregular cell membrane with scant cytoplasm. Background shows RBCs and necrosis. The diagnosis of small round blue cell tumour was given. (Fig B)



**B: MICROPHOTOGRAPH SHOWING HYPERCHROMATIC TUMOR CELLS IN GLANDULAR PATTERN**

## USG guided Tru cut biopsy from scrotal bulge:

It showed small to medium sized cells with hyperchromatic nucleus, arranged in alveolar pattern**.** The diagnosis of Alveolar Rhabdomyosarcoma was given. On immunohistochemistry tumour cells are positive for vimentin, Desmin, myogenin, Myo-D1 and Negative for NKX2, S100, LCA, Synaptophysin, Pancytokeratin. It confirmed the diagnosis of Alveolar Rhabdomyosarcoma. (Fig. C)



**C: THIS FIGURE IS SHOWING TUMOR CELLS POSITIVE FOR MYOGENIN**

## Discussion

In recent years, organizations, such as the Intergroup Rhabdomyosarcoma Study (IRS), the Paediatric Oncology Group (POG), the Children's Cancer Study Group (CCSG), and the International Society for Paediatric Oncology (SIOP), play crucial roles in facilitating these collaborative studies. Rhabdomyosarcoma (RMS) is an early-stage tumor originating from primitive mesenchymal tissue with a tendency to differentiate into skeletal muscle. Among the various RMS subtypes, alveolar RMS stands as the second most prevalent.

Unfortunately, it carries the worst prognosis, attributed to its distinctive molecular characteristics driven by the PAX3-FOXO1 fusion gene. This subtype demonstrates poorly differentiated cells and a tendency for distant metastases, contributing to its unfavorable prognosis.

The primary locations where rhabdomyosarcoma (RMS) most frequently arises include the head/neck, extremities, and genitourinary tract. In contrast, the perianal and perineum areas are uncommon sites and are generally considered unfavourable for RMS occurrence. In adults, RMS is more prone to develop in these unfavourable sites compared to adolescents. [11,12]

Typically, perianal rhabdomyosarcoma is of the alveolar subtype (ARMS). This form of rhabdomyosarcoma, a rare tumour, manifests at a rate of 4 cases per 1 million in the

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general population annually, and the incidence is 4.5 cases per 1 million among children. In Europe, around 400 cases are diagnosed each year in individuals aged 0 to 19. The overall 5-year survival rate for rhabdomyosarcoma is approximately 70%, necessitating a comprehensive treatment approach involving surgical resection, radiotherapy, and chemotherapy. [8,9] Lymph node metastases were frequently observed in patients with RMS (46%), and alveolar PRMS had an exceptionally high risk of lymph node spread (78%).[10]

The outlook for primary perianal and perineal rhabdomyosarcoma is exceptionally bleak. Moreover, the diagnosis of RMS, particularly alveolar rhabdomyosarcoma, often necessitates a multidisciplinary approach. This includes imaging studies, histopathological examination, and increasingly, immunohistochemistry (IHC). IHC plays a pivotal role in distinguishing RMS from other small round blue cell tumours, especially in atypical cases. [13,7]

The present case has responded very well with chemotherapy and completed 5 cycles and disease free till follow up of 1 year after treatment.

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