# A RARE CASE OF ALVEOLAR RHABDOMYOSARCOMA OF PERINEAL REGION

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**Abstract:**

The term "rhabdomyosarcoma" (RMS) dates back to Weber's description in 1854 and is characterized by two primary subtypes: Alveolar Rhabdomyosarcoma (ARMS) and Embryonal Rhabdomyosarcoma (ERMS). A 23-year-old male with complaints of mass and bleeding per rectum since a month was referred for Contrast Enhanced Computerized Tomography (CECT) Abdomen and Pelvis. The CECT revealed a mildly lobulated heterogeneously enhancing soft tissue mass arising from the perianal region with infiltration of bilateral ischioanal, ischiorectal fossae extending to anterior perineum and infiltrating to right side of prostate. Histopathological findings revealed small round cells showing round nucleus, coarse chromatin having high N:C ratio and scant cytoplasm. The tumor cells were immune positive for vimentin, desmin, myogenin and myoD1 with a leading differential diagnosis of Rhabdomyosarcoma. Imaging is required for both the initial diagnosis and the assessment of the tumor's response to treatment. Ultrasonography (US) of the mass is frequently the first imaging modality used in the diagnostic work-up whereas Magnetic resonance imaging (MRI) is the preferred modality for assessing primary rhabdomyosarcoma. Advanced multiparametric MRI may be useful in predicting prognosis. Both immunohistochemistry and histology are always used to confirm the diagnosis.

## Introduction:

The term known as "rhabdomyosarcoma" (RMS) dates back to Weber's description in 1854 [1]. Rhabdomyosarcoma malignancy develops from mesenchymal cells and usually targets the head and neck, then the urogenital tract, the limbs, and very rarely the perianal/perineal region. Alveolar Rhabdomyosarcoma (ARMS) and Embryonal Rhabdomyosarcoma (ERMS) are the two primary subtypes of RMS. Typically, Perianal Rhabdomyosarcoma (PRMS) presents as ARMS [2]. Compared to embryonic rhabdomyosarcoma, alveolar rhabdomyosarcoma is more common at younger age (10–25 years) [3]. Depending on the location of the main tumor mass and the existence of metastases, the clinical appearance and symptoms can take many different forms. Throughout the entire patient journey, imaging is essential: from the initial clinical suspicion to the definitive diagnosis of rhabdomyosarcoma, staging the disease for the best possible risk stratification, assessing the patient's response to treatment, organizing local therapy, and, at the end, monitoring and relapse detection [1].

The present report describes a case of PRMS in an adult patient and reviews the accurate diagnosis of rhabdomyosarcoma through CECT of Abdomen and Pelvis later confirmed by histopathology and immunohistochemistry.

**Case Presentation:**

A 23-year-old male with complaints of mass and bleeding per rectum since a month was referred for Contrast Enhanced Computerized Tomography (CECT) Abdomen with Pelvis. The Non Contrast Computed Tomography (NCCT) of the abdomen and pelvis showed a mass in the perianal region (Fig 1) and a malrotated kidney (Fig 2).

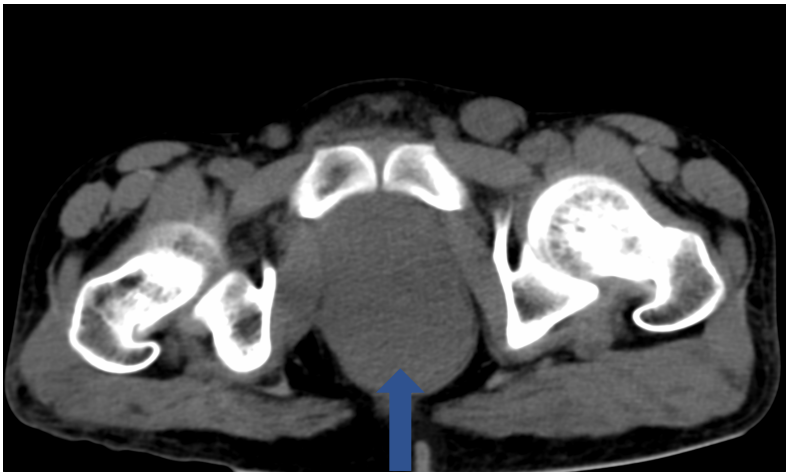

Fig 1: Mass in the perianal region

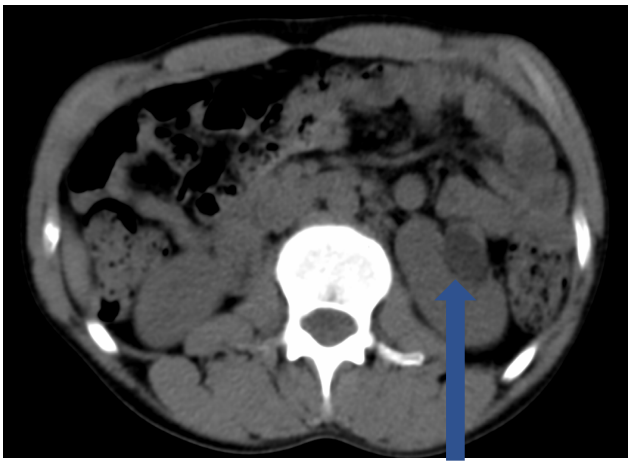

Fig 2: Malrotated Kidney

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The CECT of Abdomen and Pelvis revealed a mildly lobulated heterogeneously enhancing soft tissue mass arising from the perianal region (R>L) with infiltration of bilateral ischioanal, ischiorectal fossae extending to anterior perineum and infiltrating to right side of prostate. Medially, suspicious infiltration to right lateral wall of rectum and anal canal. Anteriorly, invading bilateral bulbospongious muscles were observed. Right laterally infiltrates to right obturator internus muscle. Multiple discrete heterogenenously enhancing bilateral perirectal, left obturator, bilateral external iliac, inguinal and presacral lymph nodes were enlarged (Fig 3).

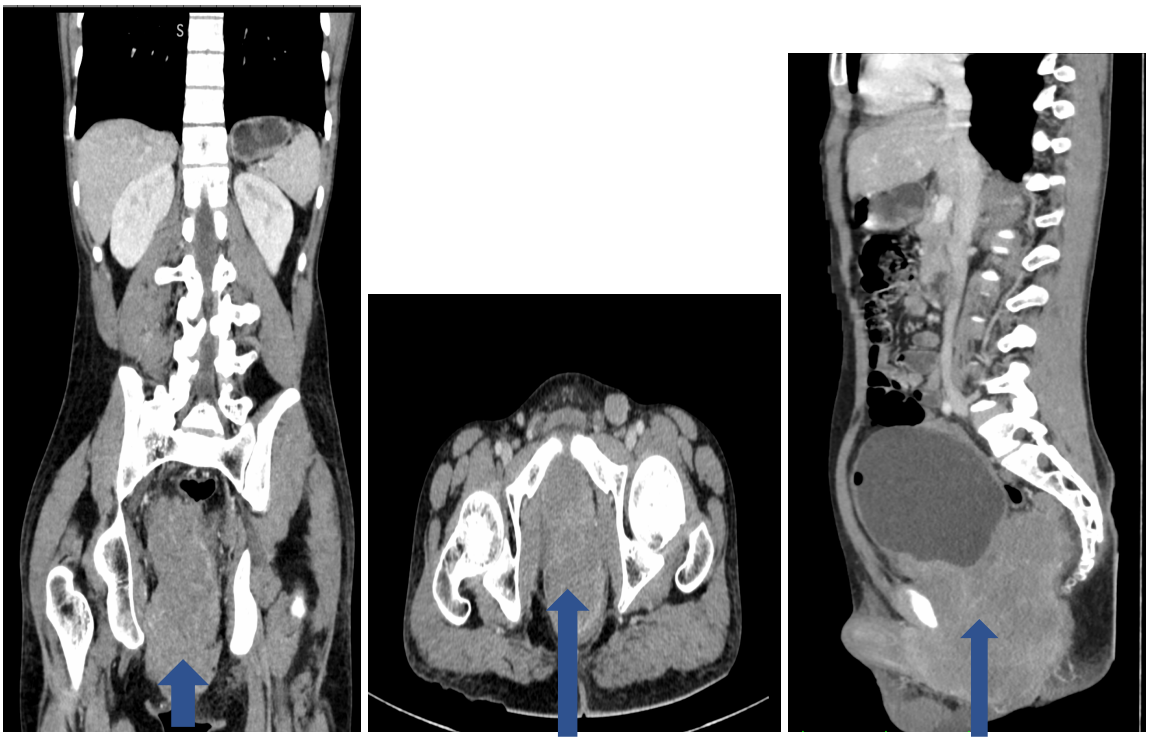

Fig 3: CECT revealing Heterogeneously enhancing soft tissue mass involving the perianal region with involvement of lymph nodes.

A differential diagnosis included Gastrointestinal Stromal Tumor or Rhabdomyosarcoma or Lymphoma. Following the CECT, biopsy was done.

The Histopathological findings revealed small round cells showing round nucleus, coarse chromatin having high N:C ratio and scant cytoplasm. Few scatted mitosis were seen. The tumor cells were separated by fibrocartilaginous tissue (Fig 4). A differential diagnosis included Non-Hodgkins Lymphoma/ Desmoplastic Small Round Cell Tumor.

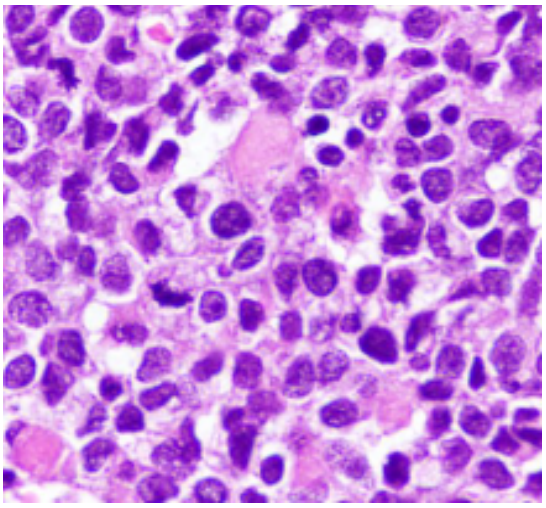

Fig 4: Small round blue cells in Alveolar Rhabdomyosarcoma

Further, immunohistochemistry revealed small round cell tumors arranged in the compact nests and alveoli. The cells showed uniform, mild hyper chromatic nuclei with evenly dispersed chromatin. The scant cytoplasm was clear to lightly eosinophilic. The tumor cells were immune positive for vimentin, desmin, myogenin and myoD1 with a leading differential diagnosis of Rhabdomyosarcoma based on immunohistochemistry and histological findings (Fig 5). The patient was further treated in a tertiary care centre.

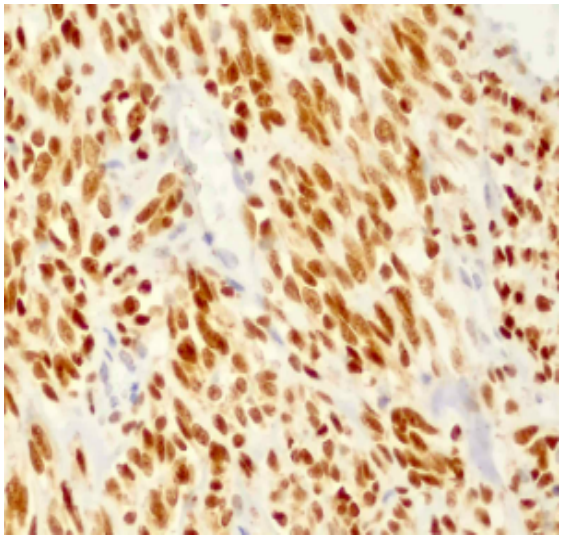

Fig 5: The lesional cells stained positive for myoD1.

**Discussion:**

PRMS is extremely uncommon, making up only 2% of RMS [2] with unknown etiology [3]. When perianal abscesses are mistakenly classified as PRMS, treatment is delayed, which might result in local or distant metastases, since PRMS is more strongly linked to the external anal sphincter, according to magnetic resonance imaging [2,7].

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The clinical and imaging manifestations of soft-tissue sarcomas vary according on the patient's age, the tumor's location, and its contents. Imaging is required for both the initial diagnosis and the assessment of the tumor's response to treatment [6,7]. A full work-up needs to be carried out as soon as an RMS is detected in order to correctly stage the tumour and obtain a histological diagnosis. At the beginning of the diagnostic work-up, all patients must have imaging of the main location using US, MRI, or CT [8].

Certain characteristics that set rhabdomyosarcomas apart from other soft tissue tumors and among these, the subtypes can be identified on MRI and CT scans, even if the final diagnosis is made pathologically [4].

Combining traditional and sophisticated multiparametric MRI allows for the examination and characterisation of soft-tissue sarcomas, for which MRI is the preferred modality due to its contrast resolution. For precise treatment planning prior to surgery, conventional MRI techniques can show the tumor's composition, extent, compartmental involvement, and connection with other structures. However, due to overlap in the signal intensity characteristics of these tumors, traditional approaches are still unable to distinguish between malignant and benign lesions. They are also limited in their ability to assess residual or recurrent disease following treatment [5].

Ultrasonography (US) of the mass is frequently the first imaging modality used in the diagnostic work-up. Generally speaking, US is readily available, can provide important details regarding the tumor, and can help with an initial differential diagnosis. Rhabdomyosarcoma typically manifests on US as a distinct, slightly hypoechoic inhomogeneous mass with the potential to exhibit enhanced flow. Magnetic resonance imaging (MRI) is the preferred modality for assessing the features of the tumor and its relationship to surrounding tissue if there is a suspicion of a soft tissue sarcoma. The original tumor, along with any loco regional disease extension, and any loco regional lymph nodes, should be seen within the MRI's field of view [1].

The tumor's radiological features are not particular. A CT scan can be used to evaluate bone deterioration. A more accurate description of the bulk and its invasion of nearby structures can be obtained using magnetic resonance imaging [3].

**Conclusion:**

Adults with rhabdomyosarcoma have a catastrophic prognosis because of this uncommon, extremely aggressive soft-tissue tumor. The preferred imaging method for assessing primary rhabdomyosarcoma is magnetic resonance imaging (MRI). When assessing residual disease following surgery and anticipating a patient's reaction to preoperative neoadjuvant treatment, advanced multiparametric MRI may be useful in predicting prognosis. Both immunohistochemistry and histology are always used to confirm the diagnosis.

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