



A RARE CASE OF MALIGNANT RHABDOID TUMOR OF THE ANORECTUM: IMAGING FINDINGS

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ABSTRACT

The term '*Malignant Extrarenal Rhabdoid Tumor (MERT)*' is used for those tumors occurring in extrarenal locations. Since the initial description, only a few cases of MERT involving the anorectum have been reported in the literature and none has been reported in the radiology literature to our knowledge. Here, we report on the computed tomography, magnetic resonance imaging and ultrasonographic features of a rare case of malignant rhabdoid tumor of the anorectum in a 53 year old female patient. To our understanding, this is the first case report of MERT involving the anorectum that includes imaging features. MERTs present as very aggressive neoplasms at the time of diagnosis thus have poor prognosis. Radiologists should be aware of the imaging findings of the MERT of the anorectum and include it as a differential diagnosis when there is as a large, heterogeneous tumor involving the anorectum.

KEYWORDS:

MERT, heterogeneous tumor, iso-hypodensity, coarse calcification

INTRODUCTION

In 1978, Beckwith and Palmer first reported the Malignant Rhabdoid tumor the kidneys of children as a rare variant of Wilm's tumor with rhabdomyosarcomatoid pattern [1]. But even before this report, carcinomas with rhabdoid morphologic features were first described in 1954 as a "*pleomorphic giant cell carcinoma*" of the pancreas [2]. The term "*Malignant Extrarenal Rhabdoid Tumor (MERT)*" is used for those tumors occurring in extrarenal locations [3,4,11]. Colorectal MERTs are very rare neoplasms with only very few cases have been reported with even fewer cases involving the anorectum in the literature.

CASE REPORT

A 53 year old female patient was admitted to our hospital with two months history of pain abdomen on and off and bleeding per rectum. There was no significant past medical history. On general examination the patient was obese with pallor and icterus noted. Her abdomen was soft and non-tender with no palpable mass and active bowel sounds noted.

Initial laboratory examination revealed hemoglobin 8.0 g%; total leukocyte count 5300 cells/mm³; differential count - neutrophil 74%, lymphocyte 20%, monocytes 4%, eosinophils 2%; platelet count 1.9 lakhs/mm³; and erythrocyte sedimentation rate 20 mm/1st hour. Serum electrolytes, renal and liver functions tests were within the normal limits.

Colonoscopic examination revealed a smooth growth compromising the lumen just above the anal verge. Histopathological examination revealed sheets of loosely cohesive epitheloid cells with abundant eosinophilic cytoplasm and eccentrically placed pleomorphic vesicular to hyperchromatic nuclei with occasional macro-nucleoli with many abnormal mitotic figures. Immunohistochemistry also was conclusively confirming the histopathological examination.

Initial imaging with computed tomography (CT) revealed a well-defined, large, iso-hypodense mass lesion involving the left lateral wall of the anal canal and distal rectum measuring 8 x 7 x 6 cm³ (craniocaudal x mediolateral x anteroposterior) with area of coarse calcification measuring 1.6 x 1.3 cm². Post-contrast study showed heterogeneous enhancement of the lesion (Fig. 1). Isolated enlarged necrotic right external iliac and left presacral lymph nodes were also detected (Fig. 2). No evidence of invasion to the adjacent pelvic organs noted. Ascites and bilateral pleural effusion were also noted. On subsequent magnetic resonance imaging (MRI), the lesion appeared as isointense on T1W images and heterogeneously hyperintense on T2W images with irregular signal void suggestive of coarse calcification

(Fig. 3). ultrasonography (USG) revealed a heterogeneously hypoechoic mass lesion involving the anorectal region with central and peripheral vascularity showing low resistance, low systolic velocity flow (Fig. 4).

Figure 1

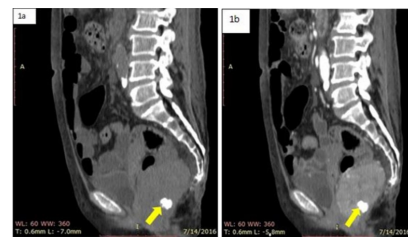


Fig. 1 Plain (1a) and post-contrast (1b) sagittal CT images of the pelvis show a iso-hypodense mass lesion involving the anorectum with heterogeneous enhancement and areas of coarse calcification (arrows).

Figure 2

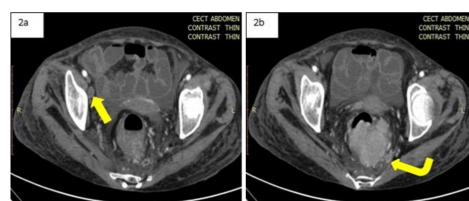


Fig. 2 Axial CECT images (2a & 2b) of the pelvis show enlarged, necrotic right external iliac (straight arrow) and left presacral (curved arrow) lymph nodes.

Figure 3

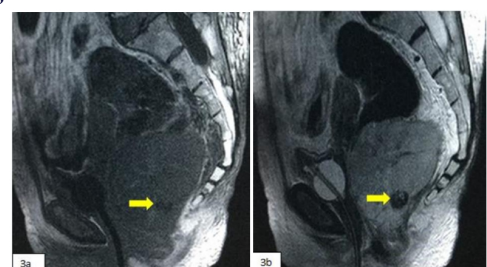


Fig. 3 Sagittal T1W image (3a) of the pelvis shows a isointense mass lesion involving the anorectal region with heterogeneous hyperintense signal on T2W image (3b). A well-defined rounded irregular signal void (arrows) noted on both the images suggestive of coarse calcification. Magnetic field 1 Tesla, TR – 608, TE – 14, slice thickness – 5 mm, FOV – 230*230.

Figure 4

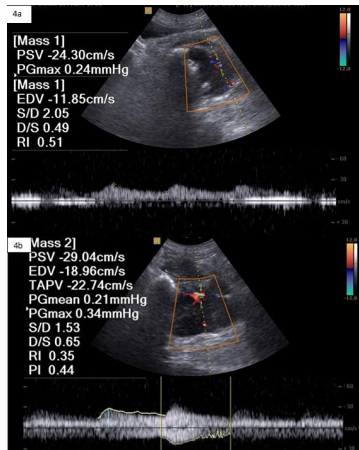


Fig. 4 USG shows a heterogeneously hypoechoic mass lesion involving the anorectal region with peripheral (4a) and central (4b) vascularity showing low resistance, low systolic velocity flow.

In view of poor performance status of the patient and unwillingness of the patient's relatives, surgical resection of the lesion was not attempted and the patient died due to circulatory shock after one cycle of chemotherapy with actinomycin D.

DISCUSSION

Malignant rhabdoid tumors were originally described as rhabdomyosarcomatoid variant of Wilm's tumor in the kidneys of children in 1978 by Beckwith and Palmer, have been recently reported in extrarenal locations, termed as 'Malignant extrarenal rhabdoid tumors' [1,3,4,11]. The median age of presentation in MERT is 12 months, but it can range from 3 months to 70 years. There is no gender predilection. MERTs involving the gastrointestinal tract have been reported in older patients in comparison to the MERTs of other locations [4]. MERT are aggressive neoplasms with large size at presentation usually although cases have been reported with size ranging from 3 cm to 15 cm [1,4].

Only a mere handful cases of colorectal MERTs have been reported in the literature. Most of the MERTs usually occur in children while the colorectal MERTs have a predilection for the elderly population [3]. MERT involving the rectum was first reported by Macak J et al in 1995 [5]. Since then only very few cases have been reported in the literature and none in the radiology literature to our understanding. The age of the patients in whom MERT involving the anorectum were reported varied from 23 years to 83 years [2,3,6].

MERTs can occur either as a pure rhabdoid phenotype alone or as a focal "rhabdoid phenotype" admixed with other malignancies such as mesenchymal and epithelial origin. The latter form also called as "Composite rhabdoid tumors". Histopathologically pure malignant rhabdoid tumors are characterized by loosely cohesive solid sheets of pleomorphic epithelioid cells with abundant eosinophilic cytoplasm with large eccentric nucleus (hence the name 'rhabdoid') containing prominent nucleolus and paranuclear inclusions of intermediate filaments. Worse prognosis has been reported in pure rhabdoid phenotype [1,4,7]. The percentage of rhabdoid tumor cell in the composite rhabdoid tumors is more than 60% in the previously reported cases [3].

Though MERTs morphologically resemble skeletal muscle tumors, they are usually negative for skeletal muscle markers such as desmin and myogenin and usually show positivity for vimentin and pancytokeratin hence the name 'rhabdoid' was given to differentiate them from rhabdomyosarcomas [2,13]. The histogenesis of the MERTs is still unclear. Most studies have shown that these tumors arise due to

biallelic inactivation of the SMARCB1(INI1) / hSNF5 / INI1 gene located in long arm of chromosome 22, with lack of normal nuclear INI1 expression on immunostaining which is pathognomonic for malignant rhabdoid tumors, though in one case report of rhabdoid carcinoma of anal canal by Shetuni et al did not show inactivation of this gene. But the aforementioned histomorphology and immunohistochemistry alone essential to make the diagnosis [2,8].

There are no pathognomonic imaging findings for MERT have been described to date. They usually present as a large heterogeneous tumors on CT and MRI. MERTs involving the brain typically present as hyperdense, enhancing mass on CT with isointense or hypointense on T1W and isointense on T2W images [9]. Imaging characteristics of the uterine and hepatic MERTs in two case reports presented by Abdulla A et al in 2010 were large, solid, heterogeneously echogenic mass with internal vascularity on USG with hypodensity as a consistent feature on CT with MR images showed isointensity on T1W images and heterogeneously iso- to hyperintensity on T2W images with post-contrast study in both CT and MR images showed heterogeneous enhancement with areas of non-enhancement suggestive of necrosis [10]. In 2014, Shim E et al presented a MERT involving the adrenal gland which presented as a low attenuating mass with poor enhancement on contrast-enhanced CT [4]. In our case, histopathologically a pure MERT involving the anorectum in a postmenopausal female patient presented as predominantly a large, heterogeneously hypoechoic lesion with central and peripheral vascularity showing low resistance, low systolic velocity flow on USG, iso-hypodensity on CT with heterogeneous contrast enhancement. On MRI, isointensity on T1W images with heterogeneous hyperintense signal on T2W images noted. Mathew D et al presented imaging features of an MERT involving the sacral canal with neural foraminal involvement mimicking a peritheral nerve sheath tumor which showed isointensity on T1W images with areas of hyperintensity, and heterogeneous yet predominantly T2 high signal with avid contrast enhancement [12]. In a retrospective study conducted by Garces-Inigo EF et al during the period 1989 to 2007 with 9 infants, which is the largest radiological descriptive case series of the extrarenal rhabdoid tumors outside the central nervous system involving the thorax/mediastinum, liver, neck, shoulder and axilla, hypointensity on T1W images and heterogeneous hyperintensity on T2W images was the consistent feature with cystic component with fluid-fluid levels on T2W images noted in two patients [8]. Areas of calcification on CT have been reported in MERTs involving the thorax, neck, liver and uterus [8,9,10]. In our case, MERT involving the anorectum also showed areas of irregular signal void on T1 and T2W images suggestive of coarse calcification. The iso-hypodense nature of the lesion may be due to the discohesive arrangement of the tumor cells. Local invasion to ureter, urinary bladder and mesosalpinx with regional and distal metastatic lymphadenopathy have been reported at the time of presentation in the previous MERTs involving the rectum [3,7]. Our case showed regional and distal lymphadenopathy with no evidence of local invasion to adjacent pelvic organs.

The prognosis of the MERTs is very poor especially those involving the GIT with median rate of survival is approximately 5.5 months with death usually occurs an average of 6 months after the diagnosis [4,10]. Complete surgical resection at an early stage is the best treatment modality for the MERTs.

TEACHING POINTS

MERT involving the anorectum is a very rare tumor with poor prognosis. Though the imaging features are non-specific for the MERT involving the rectum and the diagnosis is mainly based on the histopathology and immunohistochemistry, the radiologists should be aware of the imaging findings of the MERT of the anorectum and include it as a differential diagnosis when there is a large, iso-hypodense, heterogeneous tumor involving the anorectum.

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ABBREVIATIONS

MERT - Malignant Extrarenal Rhabdoid Tumor
CT - Computed Tomography

CECT - Contrast Enhanced Computed Tomography

MRI - Magnetic Resonance Imaging

USG - Ultrasonography

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