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Massive Retrorectal Extraosseous Ewing Sarcoma/Primitive Neuroectodermal Tumor in an Adult: A Case Report

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ABSTRACT

Ewing sarcoma is a rare, aggressive small round-cell sarcoma that predominantly affects children and young adults. Extraosseous pelvic presentations are uncommon, particularly in older adults. We report the case of a 59-year-old man who presented with progressive asthenia, constipation, and intermittent rectal bleeding. Pelvic ultrasound and magnetic resonance imaging demonstrated a large multiloculated presacral mass with cystic and enhancing solid components, initially raising the possibility of a tailgut cyst. Contrast-enhanced computed tomography showed a heterogeneous pelvic mass with marked upstream colonic dilatation; no distant metastases were identified on CT of the chest, abdomen, and pelvis. A core biopsy was reported as an extraosseous Ewing sarcoma/primitive neuroectodermal tumor. The lesion was considered unresectable at multidisciplinary review because of its extent and local relationships. Systemic chemotherapy was initiated, but the patient's clinical condition deteriorated after two cycles. This case highlights the nonspecific and potentially cystic-appearing imaging presentation of extraosseous Ewing sarcoma in the retrorectal space and the diagnostic challenges posed by this rare presentation in an older adult.

KEYWORDS

Extraosseous Ewing sarcoma, Primitive neuroectodermal tumor, Retrorectal space, Presacral mass, Tailgut cyst, Magnetic resonance imaging

MAIN ARTICLE

INTRODUCTION

Primitive neuroectodermal tumors (PNETs) are highly malignant tumors characterized by undifferentiated small round cells of neuroectodermal origin [1]. They belong to the Ewing sarcoma family of tumors and typically have a characteristic balanced translocation t(11;22)(q24;q12) involving the EWSR1 gene, which most of the time makes an EWS-FLI1 fusion oncoprotein [2]. PNET/Ewing sarcoma predominantly affects children and adolescents, its incidence in older adults is exceedingly rare [3]. The historical terms peripheral primitive neuroectodermal tumor and extraosseous Ewing sarcoma have been used for closely related tumors within the Ewing spectrum; current WHO classification places Ewing sarcoma among molecularly defined undifferentiated small round-cell sarcomas [2]. Pelvic soft-tissue PNETs are extremely rare with limited cases reported in the literature. In particular, PNETs that develop in the presacral or retrorectal space of adult patients are exceptional.

Patients with pelvic PNET often present with non-specific symptoms related to the mass effect on adjacent organs. Common presentations include pain, neurological symptoms from nerve compression, or visceral symptoms such as constipation or trouble urinating, in rare cases, rectal bleeding can occur when the tumor invades the rectum. Diagnosing PNET based on imaging alone is challenging because the radiologic features are not pathognomonic. Typically, these tumors appear as large heterogeneous masses with necrotic or cystic areas and variable enhancement, these signs are similar with other pelvic neoplasms. Because these imaging findings overlap with those of other pelvic neoplasms, tissue sampling is required for definitive diagnosis." Multimodal therapy is the standard of care for PNET/Ewing sarcoma, consisting of intensive systemic chemotherapy combined with local control measures (surgical resection and/or radiotherapy). Despite aggressive treatment, the prognosis for pelvic PNET remains poor compared to extremity or bone lesions. Tumor volume, location and presence of metastases at diagnosis are key prognostic factors [4]. We present a rare case of a large retrorectal extraosseous PNET in a 59-year-old man, with an emphasis on the imaging findings, histopathology, and a review of the relevant literature.

CASE PRESENTATION

A 59-year-old male patient with no notable medical history presented with a several-month history of generalized weakness (asthenia), lower abdominal discomfort, constipation and intermittent rectal bleeding. On physical examination, he appeared pale but had stable vital signs. Abdominal examination revealed distension without an obvious palpable mass. Digital rectal examination found a firm extrinsic bulge compressing the anterior rectal wall, but no mucosal lesions were felt. Laboratory testing showed mild normocytic anemia (hemoglobin, 10.8 g/dL). Serum tumor markers (including CEA and PSA) were within normal limits.
Given the rectal bleeding, colonoscopy evaluation demonstrated an extrinsic compression of the mid-rectum with intact overlying mucosa. The endoscopic lumen was narrowed by an external mass effect, but no intraluminal tumor was seen.
An initial pelvic ultrasound revealed the presence of a large complex mass in the pelvis (Figure 1). For better characterization, pelvic magnetic resonance imaging (MRI) was obtained, which showed a voluminous mixed mass with solid and multiloculated cystic components in the presacral (retrorectal) space. The lesion was fairly well-circumscribed, measuring approximately 15 cm in maximum dimension. It exhibited predominantly high signal on T2-weighted sequences and low signal on T1-weighted sequences, consistent with fluid components, with multiple internal septations and nodules. Some of the fluid compartments demonstrated T1 hyperintense contents on fat-suppressed images, suggesting proteinaceous or hemorrhagic material. The septa and peripheral solid components enhanced after gadolinium administration. On this initial MRI, there were no definitive signs of invasion into the rectal wall or sacrum; the interface with adjacent organs appeared mostly preserved, and the sacral neural foramina were intact (Figure 2,3,4). Given the multicystic appearance, a benign retrorectal cystic hamartoma (tailgut cyst) was considered in the differential at that time.
A contrast-enhanced CT scan of the chest, abdomen, and pelvis was later performed revealing a massive pelvic tumor occupying nearly the entire pelvic cavity. The mass was heterogeneous, with soft-tissue density solid components and multiple low-attenuation nonenhancing areas compatible with cystic or necrotic change. The tumor caused marked upstream colic distension up to 7 cm in diameter (Figure 5,6). No distant metastases were identified on CT of the chest, abdomen, and pelvis.

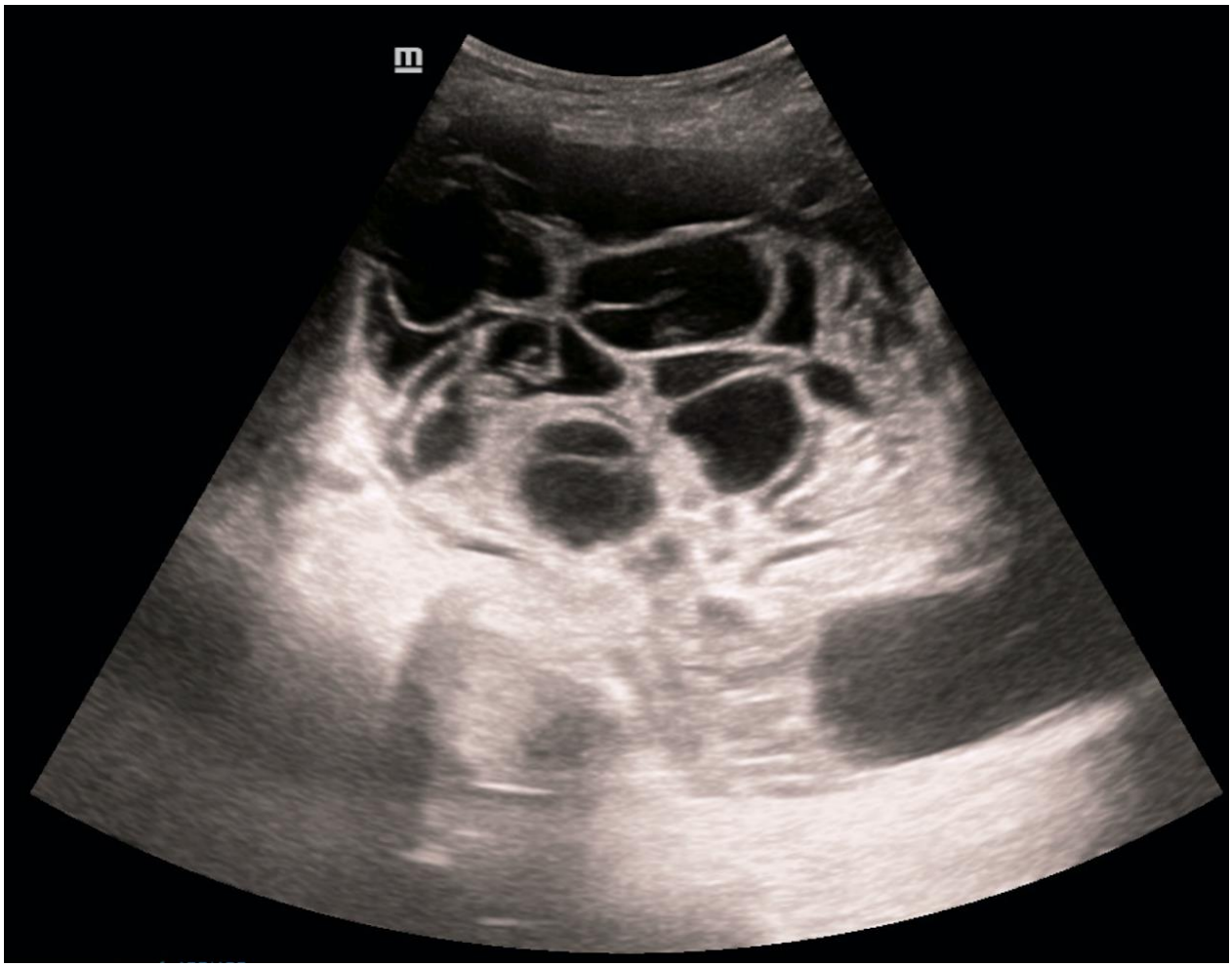


Figure 1: Pelvic ultrasound image showing a multicystic pelvic mass with thick septations and solid-appearing components.

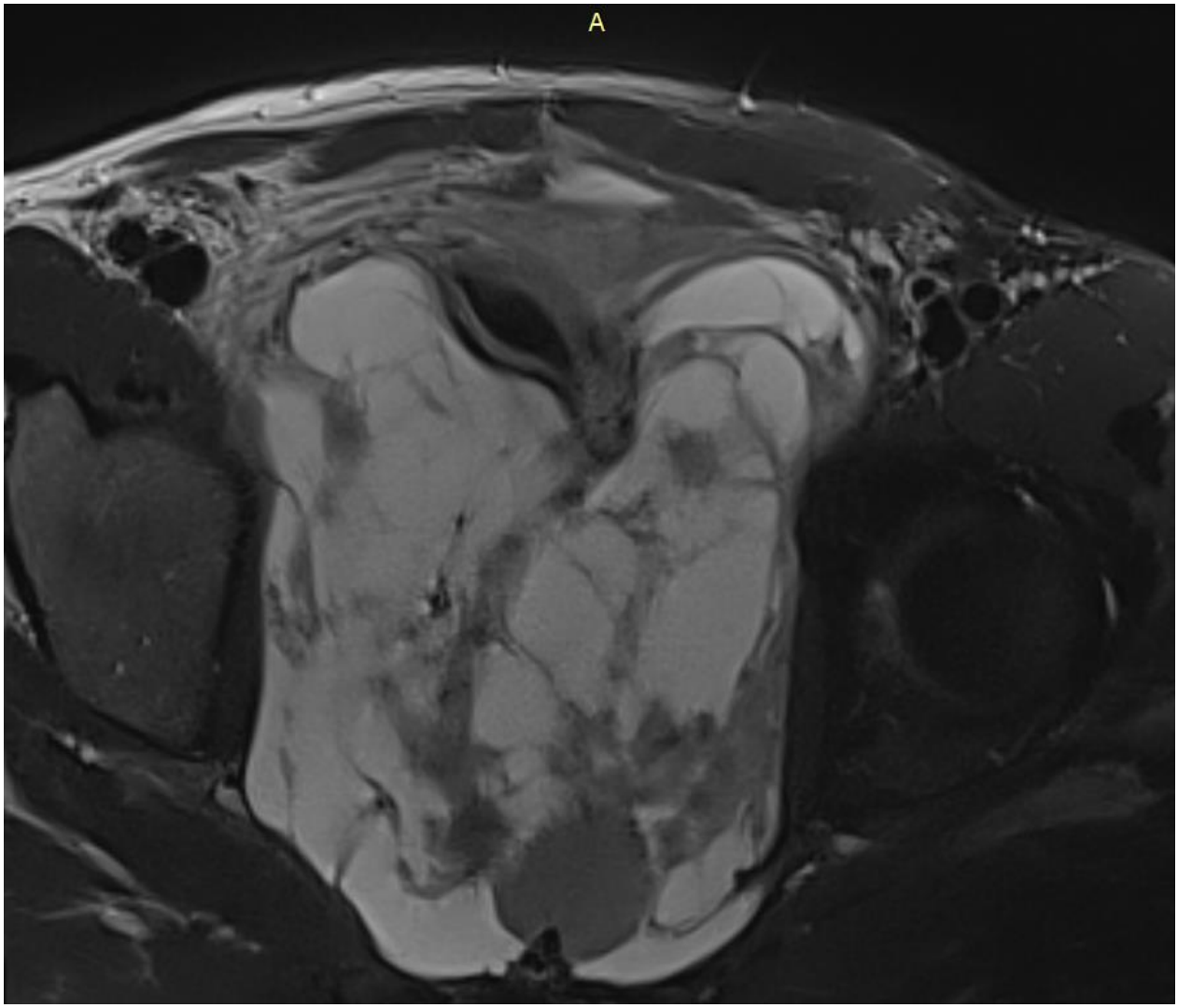


Figure 2: Axial T2-weighted MRI of the pelvis showing a large lobulated high-signal mass occupying the retrorectal space. The lesion exhibits internal septations dividing it into multiple lobules, corresponding to the multicystic components observed on ultrasound. Surrounding organs are displaced: the rectum is compressed and shifted anteriorly by the mass.

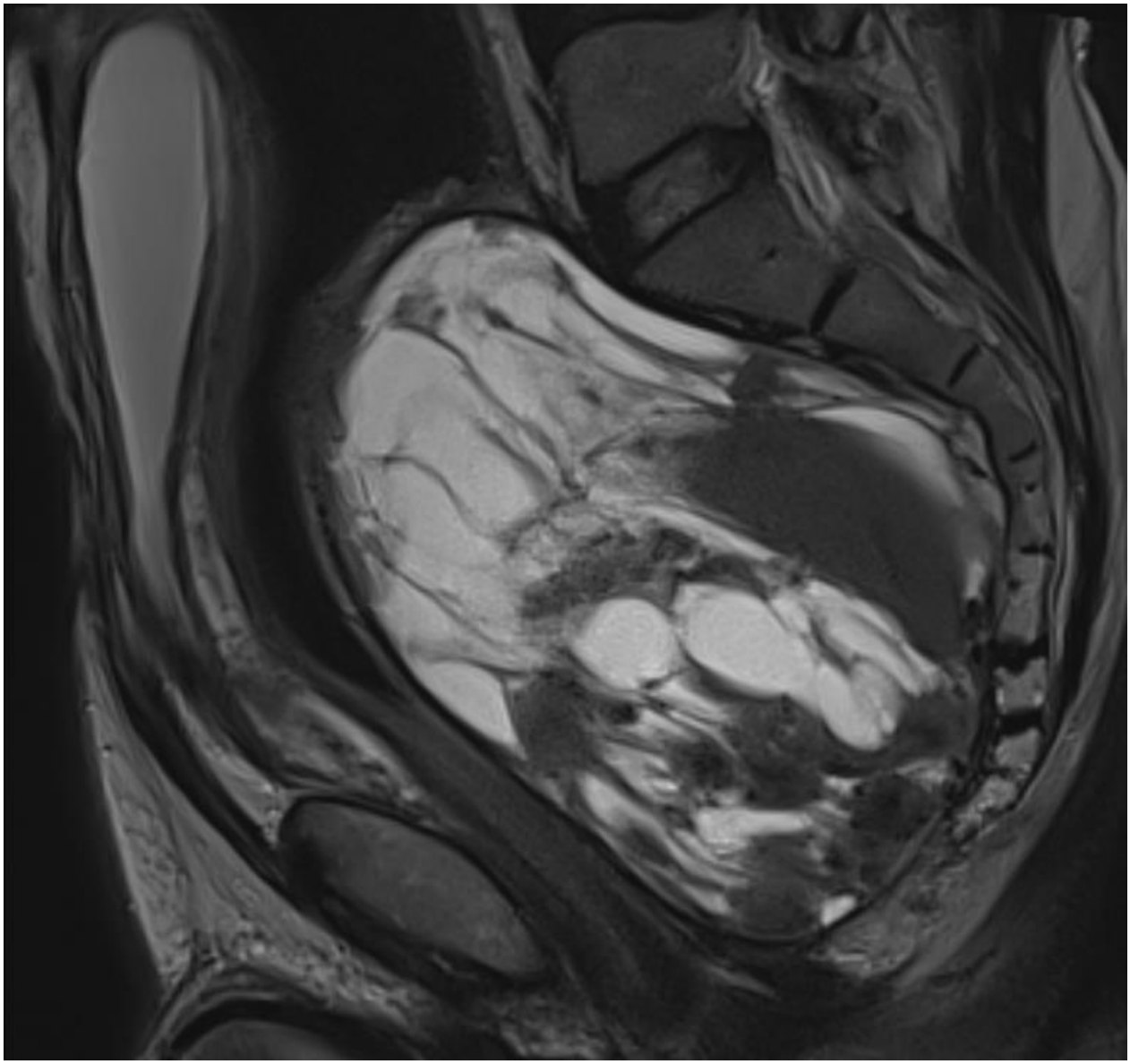


Figure 3: Sagittal T2-weighted MRI demonstrating the craniocaudal extent of the lobulated presacral tumor. The mass lies posterior to the rectum and anterior to the sacrum, occupying the entire retrorectal space.

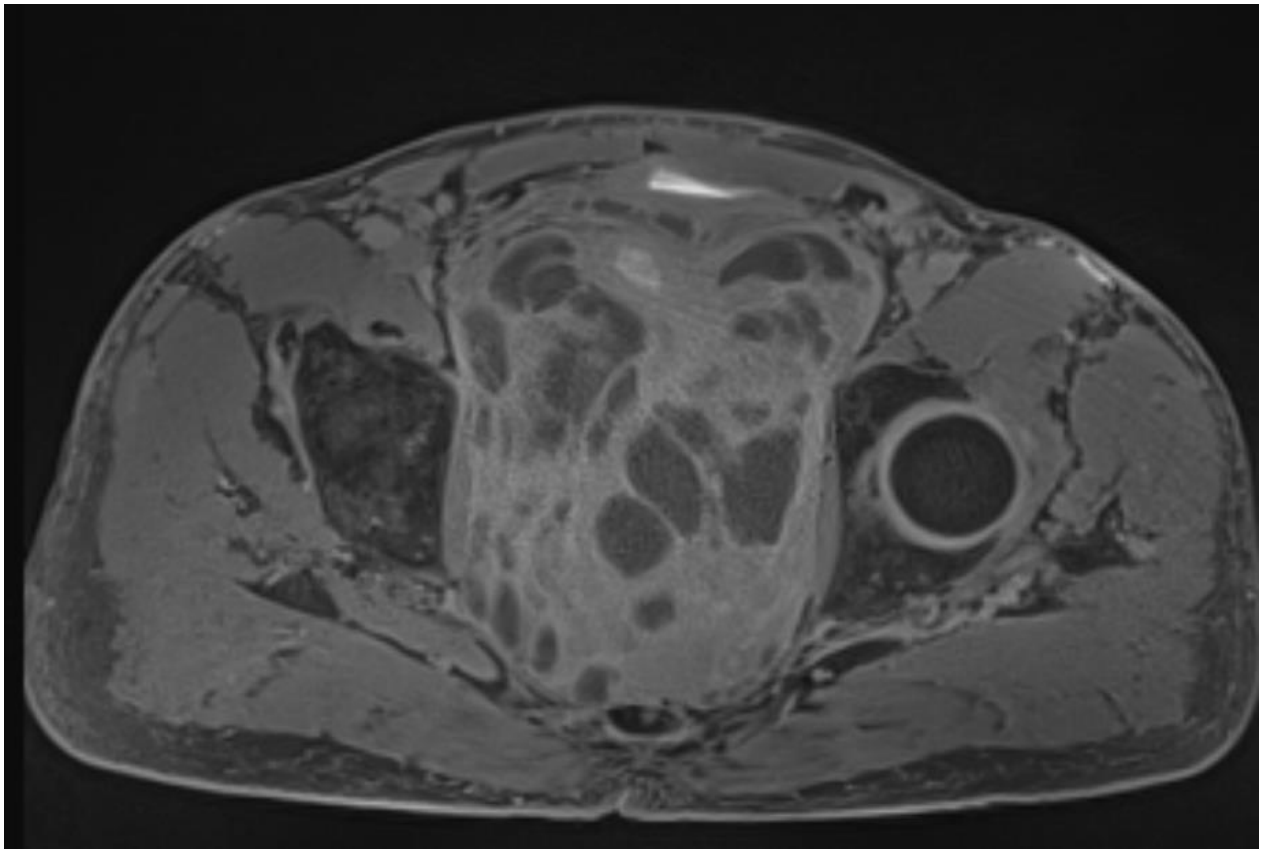


Figure 4: Axial T1-weighted MRI with contrast (fat-suppressed sequence) showing heterogeneous enhancement of the presacral tumor. Enhancing solid portions of the mass demonstrate contrast uptake, whereas non-enhancing areas correspond to necrotic or cystic components.



Figure 5: Axial contrast-enhanced CT image revealing a large heterogeneous mass in the retrorectal region. The tumor consists of both soft-tissue attenuation solid portions and low-attenuation fluid-filled areas, consistent with solid tumor components and cystic degeneration. The mass compresses the rectum anteriorly and abuts the sacrum posteriorly.



Figure 6: Axial contrast-enhanced CT at a higher abdominal level showing marked upstream colonic dilatation.

A transrectal aspiration and core biopsy was realized. A core biopsy was obtained, and the pathology report concluded that the lesion was an extraosseous Ewing sarcoma/primitive neuroectodermal tumor.

The case was discussed in a multi-disciplinary tumor board. Given the tumor's unrespectability at presentation, the patient was started on systemic chemotherapy according to an Ewing sarcoma multi-agent protocol. Unfortunately, after two cycles of VAC/IE chemotherapy, the patient's clinical condition deteriorated and further intensification of therapy was not possible. Supportive and palliative measures were initiated thereafter.

DISCUSSION

Extraosseous PNETs are rare aggressive neoplasms that are histogenetically related to Ewing sarcoma. They typically affect adolescents and young adults, and occurrences in the pelvis of middle-aged or older patients are exceedingly uncommon [1]. In the current case, a 59-year-old man developed a massive retrorectal PNET, which is a very rare presentation. Published pelvic cases predominantly involve children, adolescents, or younger adults, making presentation at 59 years of age unusual [5]. This case expands the clinical spectrum of PNET by demonstrating that, although infrequent, these tumors may manifest in late adulthood and in unusual locations such as the retrorectal space.

The differential diagnosis for a large retrorectal mass in an adult is broad and includes both benign and malignant lesions. Common considerations include congenital cystic lesions such as tailgut cysts (retrorectal cystic hamartomas), which classically appear as multilocular cystic masses in the presacral space on MRI [6]. Indeed, our patient's initial MRI showed a multiloculated cystic lesion, resulting in a presumptive diagnosis of a tailgut cyst. Other differential diagnosis for a presacral mass include chordoma (a malignant bone tumors of the sacrum), which typically shows bone destruction on imaging (absent in this case), schwannomas or neurofibromas arising from sacral nerve roots, extragonadal germ cell tumors, and various soft tissue sarcomas (e.g., leiomyosarcoma or liposarcoma) that can occur in the pelvic retroperitoneum. The heterogeneous appearance of the mass with necrotic areas on CT in this case was suggestive of a high-grade malignancy. However, there is no distinct radiological characteristic that distinguishes PNET from other sarcomas or tumors in this region. As demonstrated here, extraosseous Ewing sarcoma may occasionally display prominent cystic-appearing components and mimic a developmental harmless cystic lesion, which underlines the importance of histological diagnosis for any indeterminate presacral mass.

Current classification of undifferentiated small round-cell sarcomas relies increasingly on molecular features. Because molecular confirmation was not available in this patient, we retained the diagnostic terminology documented in the biopsy report, namely extraosseous Ewing sarcoma/primitive neuroectodermal tumor.

In general, PNET is an aggressive neoplasm with a high tendency for metastasis and recurrence. Around 20–25% of patients have metastatic disease at presentation most frenquently to the lungs, bone, or bone marrow [4]. No distant metastases were identified on

the available CT staging examination in our patient; however, the very large pelvic tumor and its local extent complicated management.

The primary therapy for extraosseous PNET/Ewing sarcoma is systemic chemotherapy, using protocols similar to those for skeletal Ewing sarcoma [7]. The best way to control the disease is with multimodal treatment that includes surgery, chemotherapy, and radiotherapy [7]. Complete surgical resection of the tumor, when possible, is an important predictor of a long-term survival. In some reported cases, neoadjuvant chemotherapy to shrink the tumor followed by surgical resection and adjuvant chemoradiotherapy has resulted in prolonged remission. However, in many cases (especially with large unresectable tumors or metastatic disease), the prognosis remains very poor despite aggressive treatment.

In our patient, surgery was not initially possible. Chemotherapy was initiated in hopes of cytoreduction, but the lack of significant response and the patient's deterioration showed the aggressive biology of PNET in this context. New treatments, including targeted molecular treatments, are being explored for refractory Ewing/PNET, but the outcomes are still not hopeful.

CONCLUSION

We report an unusual case of a very large retrorectal extraosseous Ewing sarcoma/PNET in a 59-year-old man. The case illustrates that Ewing-spectrum tumors can rarely present in older adults and that a presacral tumor with prominent cystic components may mimic a developmental cystic lesion. Imaging is central to defining tumor extent and relationships but is not specific enough to establish the diagnosis. Tissue-based diagnostic confirmation should therefore be retained even when histopathology images and detailed microscopic descriptions are unavailable. Multidisciplinary management with systemic therapy and local control when feasible remains the therapeutic principle.

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