Retrorectal Mass: An Atypical Case Presentation of Metastatic Myxoid Liposarcoma

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Background	Retrorectal masses often pose a diagnostic and management challenge due to their rarity and heterogeneous etiologies. Myxoid liposarcoma (MLS) is a lipomatous malignancy that can have atypical, extra-pulmonary metastatic sites.
Summary	A 36-year-old male patient who initially underwent a radical resection of a left posterior thigh MLS was found to have an asymptomatic 9 cm solitary retrorectal mass during a surveillance MRI a year later. Given the clinical history and imaging characteristics, the mass was suspected to be a solitary metastatic disease from his MLS. He underwent chemotherapy followed by radiation therapy, after which his tumor demonstrated response without evidence of further metastatic disease. The patient then underwent a low anterior resection, and the final pathology confirmed metastatic MLS.
Conclusion	Metastatic MLS to the retrorectal space has not previously been reported and should be part of the differential diagnosis when evaluating a retrorectal mass, especially in the context of prior known primary disease. This case report also highlights the efficacy of a radical resection with a total mesorectal excision in the surgical management of retrorectal metastatic MLS.
Key Words	metastatic myxoid liposarcoma; mesorectal; presacral; retrorectal; pelvic mass

DISCLOSURE:
The authors have no conflicts of interest to disclose.

FUNDING/SUPPORT:
The authors have no relevant financial relationships or in-kind support to disclose.

RECEIVED: August 26, 2021
REVISION RECEIVED: September 13, 2021
ACCEPTED FOR PUBLICATION: November 1, 2021

To Cite: Cyprich J, Yoon D, Shin J, Patel DB, Thomas J, Tseng WW. Retrorectal Mass: An Atypical Case Presentation of Metastatic Myxoid Liposarcoma. *ACS Case Reviews in Surgery.* 2021;3(5):6–10.

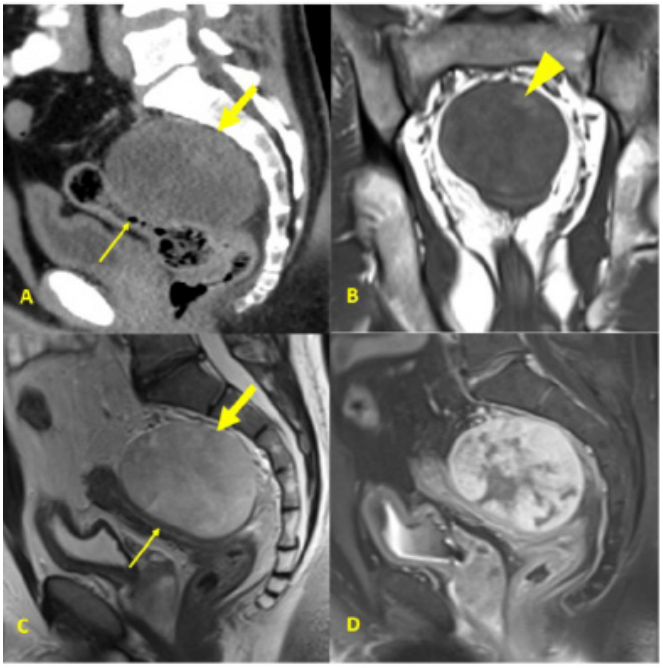
Case Description

Retrorectal masses are uncommon lesions found in the potential space confined by the sacrum posteriorly, rectum anteriorly, and peritoneal reflection superiorly. They often pose a diagnostic and management challenge due to their rarity and heterogeneous etiologies.[1] Based on the classification proposed by Uhlig and Johnson,[2] etiologies of retrorectal masses are categorized into congenital, neurogenic, osseous, inflammatory, and miscellaneous, with benign and malignant lesions in each category. We describe a unique case of a patient with a history of surgically treated primary myxoid liposarcoma (MLS) of the left posterior thigh who presented with a solitary retrorectal mass.

The patient is a 36-year-old healthy male who presented to another hospital three years ago with a left posterior thigh mass. After evaluation of local extent with MRI, the biopsy revealed high-grade lipomatous malignancy with the presence of round cells. DNA damage-inducible transcript 3 (DDIT3) gene translocation was detected on fluorescence in situ hybridization, confirming the diagnosis of MLS. CT chest showed no pulmonary metastasis. The patient underwent neoadjuvant radiation therapy followed by radical resection. The final pathology revealed a 10 cm tumor with negative margins.

Although initial surveillance imaging was negative, pelvic MRI at one-year follow-up revealed interval development of a 9 cm well-circumscribed, heterogeneous retrorectal mass without regional lymphadenopathy (Figure 1). Staging CT of the chest, abdomen, pelvis did not show any other sites of disease. At this time, the patient was referred to us. Metastatic MLS was highly suspected given his history and subtle fat components in the tumor seen on a careful review of the imaging. The patient was treated with three cycles of trabectedin 1.5 mg/m^2 intravenously every three weeks. Upon completion, restaging showed overall stable disease for the retrorectal tumor and, again, no other metastase.

Figure 1. Cross-sectional Images of Retrorectal Mass. Published with Permission



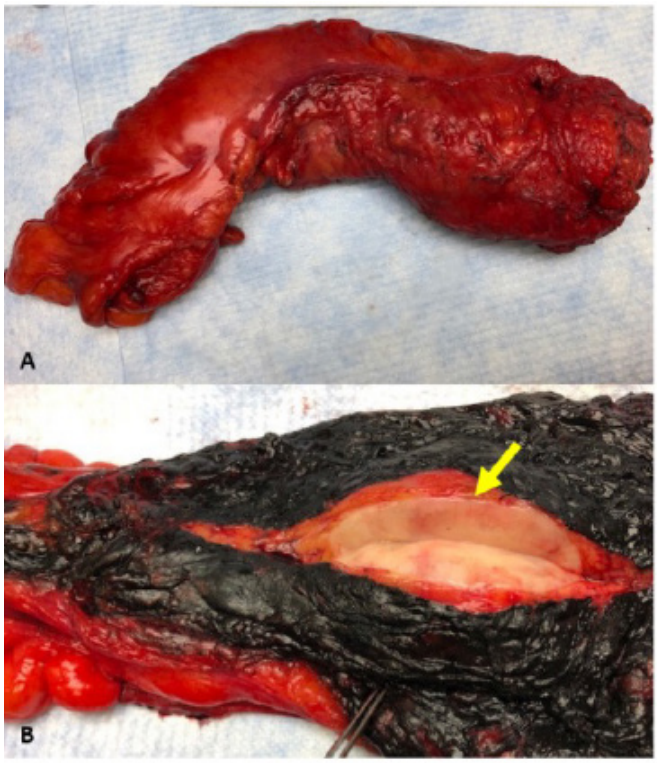
A) Sagittal reformat CT image shows retrorectal mass (thick arrow) with broad area of contact with rectum (thin arrow); B) coronal T1 MRI image shows isointense mesorectal mass. Focal areas of T1 hyperintense signal in mass are consistent with fat (arrowhead); C) sagittal T2 MRI image shows heterogeneously hyperintense mass (thick arrow) relative to muscle. Mass abuts rectum (thin arrow) without gross invasion; D) sagittal post-contrast T1 with fat saturation MRI image shows heterogeneous enhancement of mass

The patient then underwent radiation therapy with 5000cGy delivered over 25 fractions. Repeat CT now showed partial response to therapy, with decreased tumor size (>20%) and heterogeneous internal characteristics with less enhancing components. He remained asymptomatic and notably, without evidence of metastatic disease elsewhere. After consultations with both a surgical oncologist and a colorectal surgeon, the patient consented to surgical resection.

At surgery, the tumor was found to be confined within the posterior mesorectal envelope. Given the extent of involvement, we proceeded with low anterior resection and diverting loop ileostomy to achieve complete en bloc resection (Figure 2). In the immediate postoperative period, the patient had high ileostomy output that was managed conservatively but otherwise had an uneventful recovery. He was discharged on postoperative day 5.

The final pathology revealed a 6.2 cm metastatic MLS with negative margins. The tumor abutted the rectum without invasion into the muscularis propria.

Figure 2. Low Anterior Resection Surgical Specimen. Published with Permission



A) Specimen with intact mesorectal envelope encasing retrorectal mass; B) posterior margin inked encasing lipomatous solid mass with negative gross margin of excised mesorectum (arrow)

A CT of the chest, abdomen, pelvis performed six weeks postoperatively was negative for residual or metastatic disease and served as a new baseline imaging study. The patient then underwent an uneventful ileostomy reversal three months later. At his most recent follow-up six months postoperatively, he remains disease-free.

Discussion

The differential diagnosis of a retrorectal mass is wide given a variety of tissue types in close proximity.[1] A review by Baek et al[3] including 1,708 patients reported an extensive list of differential diagnoses for primary retrorectal masses. Most lesions were benign (70%) and the most common category overall was congenital (60.4%). Primary liposarcoma had a reported prevalence of 0.8% of retrorectal masses.

Liposarcomas are malignant mesenchymal tumors of adipocytic differentiation with three main subtypes: well-differentiated/dedifferentiated, myxoid/round cell, and pleomorphic.[4] MLS is the second most common subtype, representing 20 to 30% of all liposarcomas. Its incidence peaks in the fourth and fifth decades of life,[5,6] but MLS is also the predominant subtype of liposarcoma affecting the pediatric population.[7] It is characterized by pathognomonic chromosomal translocation resulting in an oncogenic FUS-DDIT3 fusion protein.[6] Presence of round cells is characteristic of high-grade disease.[5]

The most common sites for primary presentation of MLS are the extremities and trunk. After complete resection, local recurrence or distant metastatic disease can be seen in up to 40% of cases, typically in high-grade (=round cell) tumors.[5,8,9] Other factors associated with increased risk of local or distant recurrence include older age, male gender, large tumor size, and depth of the primary tumor.[5] Timing of recurrence of MLS is highly variable amongst published reports. Most cases of recurrence disease occur within the 2 to 3 years after primary resection, although the time interval ranges from 0 (synchronous) to almost 12 years (very late).[8,10–12]

Unlike other soft tissue sarcomas, potential metastatic sites for MLS are uniquely extra-pulmonary to fat-bearing areas of the body, including the retroperitoneum and spine.[5,9,10,13] This pattern of metastasis as opposed to second primary sites is supported by detailed molecular tumor characterization.[14,15] Other reported atypical metastatic sites include the heart and small bowel mesentery. To our knowledge, metastatic disease to the retrorectal space, as in this case, has not been previously reported.

It is important to mention that our patient did not undergo a preoperative biopsy of his retrorectal mass. Based on history and imaging characteristics, we determined that the retrorectal mass was likely metastatic MLS from his primary tumor site (thigh) and proceeded with the treatment accordingly with neoadjuvant therapy and resection. To our knowledge, literature is scarce on the risk of needle tract seeding after a percutaneous biopsy. Two recent papers discuss this in the setting of sarcoma (all body sites), including MLS, that found seeding to be a very rare complication (0.37% to 2%).[16,17] The caveat is that these studies looked at primary and not metastatic disease, as was the case for our patient.

The treatment of locally advanced and metastatic MLS involves a multidisciplinary approach. MLS is uniquely sensitive to radiation and systemic therapy. MLS is particularly sensitive to trabectedin, but radiographic responses have been seen with traditional chemotherapy agents, including anthracyclines and ifosfamide.[18–20] A phase II study of trabectedin in patients with locally advanced MLS resulted in pathologic complete response in three and partial pathologic responses in 12 out of 23 patients.[21] Another large trial, in which neoadjuvant trabectedin was compared to neoadjuvant anthracycline/ifosfamide for patients with primary MLS, showed that trabectedin might be at least as effective but with significantly less toxicity.[22]

The indication for surgical management of metastatic disease is less well defined. In this case, after a multidisciplinary discussion, we felt that surgical resection was indicated given tumor response to systemic therapy and confirmed solitary metastasis. Various surgical approaches, including anterior (transabdominal), posterior (perineal, trans-sacral, and paracoccygeal), and combined, have been described. The choice of approach depends on the sacral level of the tumor, size, involvement of sacral nerves, or other major structures.[3] Given the cephalad position of the tumor above the level of S4, we were able to achieve en bloc resection with negative margins via an anterior approach with a low anterior resection. Overall, published data regarding outcomes following resection of metastatic MLS are limited. Some recent evidence suggests that MLS may be one subtype of soft tissue sarcoma in which select patients with metastatic disease may benefit from surgery.[11,12,23]

Conclusion

A wide differential diagnosis for a retrorectal mass is essential to identify the etiology and guide treatment correctly. This case report describes a previously unreported presentation of a metastatic MLS as a solitary retrorectal mass. After a multidisciplinary discussion and multimodality therapy, complete resection was achieved via a low anterior resection adhering to the principles of total mesorectal excision.

Lessons Learned

Metastatic MLS may present in the retrorectal space and should be considered as part of the differential diagnosis, especially in the clinical context of an existing or prior primary disease elsewhere. A retrorectal MLS without invasion to the presacral fascia may be safely excised by total mesorectal excision.

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