Embryonal rhabdomyosarcoma within abdomen and pelvis in an adult

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Abstract

Rhabdomyosarcoma arising in abdomen and pelvis is an uncommon but important type of soft tissue sarcoma, posing a great challenge for clinicians. Sporadic cases of intra-abdominal rhabdomyosarcoma were reported, but mostly in pediatrics. We demonstrated a rare case of primary abdominopelvic rhabdomyosarcoma in an elderly woman who presented with a notable increase in abdominal circumference and constipation. Abdominal magnetic resonance imaging showed a huge mass throughout the abdomen and pelvis. Cytoreductive surgery was performed by gynecologists due to the suspicious diagnosis of disseminated leiomyosarcoma. However, the final pathological analysis revealed embryonal rhabdomyosarcoma. Although adjuvant chemotherapy was administered, localized recurrence was identified 6 months after the initial operation. Gynecologists and radiologists should be aware of it so it can be listed in the differential diagnosis of masses that primarily arise in the abdomen and pelvis.

Keywords

Introduction

Rhabdomyosarcoma (RMS) is one of the most common soft tissue sarcomas (STSs) in pediatrics. However, adult RMS is a rare occurrence, representing only 3% of STSs and less than 1% of all malignancies.[1] Similar to other soft-tissue sarcomas, RMS originates most commonly in the extremities, followed by the trunk wall, retroperitoneum, and head and neck. Nevertheless, RMS is now considered to develop from early rhabdomyoblasts that can be identified in a wide range of locations throughout the body with a different scale of differentiation.[2]

Primary abdominopelvic RMS is extremely rare than other soft-tissue sarcomas such as liposarcoma, leiomyosarcoma and gastrointestinal stromal tumor. At the time of diagnosis, abdominopelvic RMS is usually a large mass due to its clinically silent characteristics until it compresses or invades vital organs. Therefore, this kind of tumor in female adults is sometimes misdiagnosed as female malignancies due to the similarity in terms of clinical presentations. Magnetic resonance imaging (MRI) is the most appropriate modality for evaluation of tumors in the pelvis and abdomen. Cross-sectional imaging data with regard to abdominopelvic RMS in adults is limited, reflecting the rarity of these tumors.[3] Multi-institutional trials have not been performed, and only case reports have been published. Our case is probably the third well-documented case of primary abdominopelvic RMS reported in literature.

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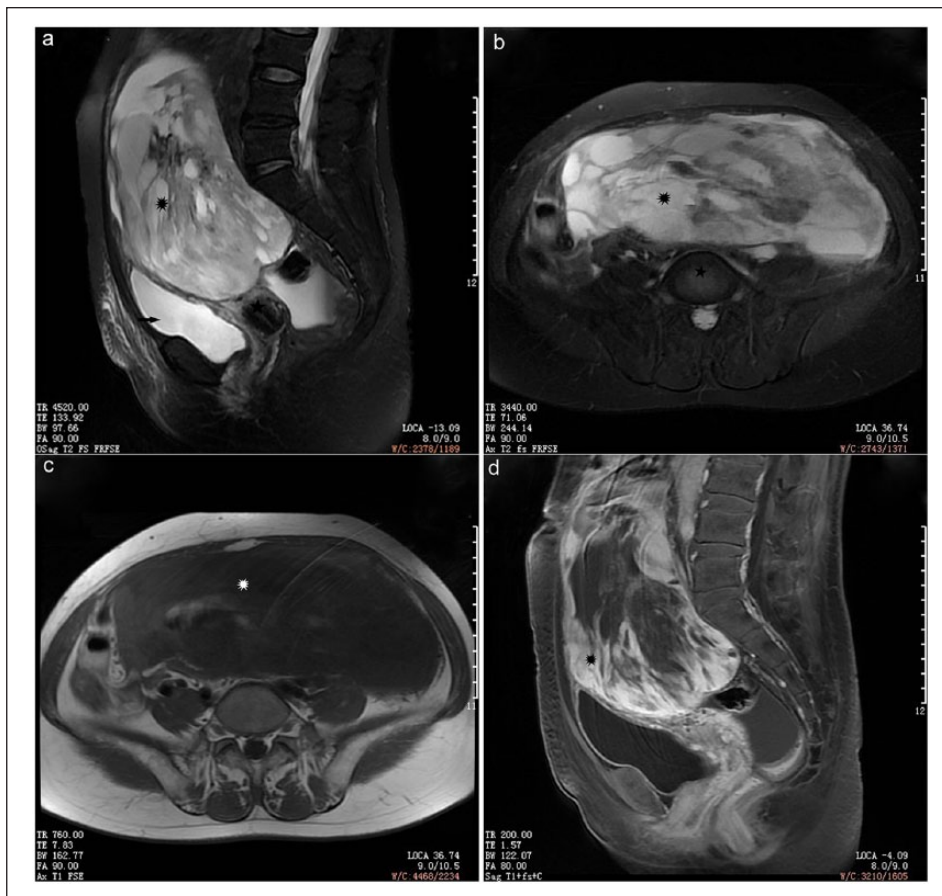Email: yangsufenedu@sina.com

Figure 1. (a) Sagittal T2-weighted MR demonstrates a heterogenous high-signal mass (asterisk) located posterior to the urinary bladder (black arrow). The dominant mass is separated from the cervix (five-pointed star) by a small amount of fluid. (b) Axial T2-weighted image demonstrates most portions of the mass (asterisk) to be primarily high signal with relatively small areas of fluid. (c) Axial T1-weighted precontrast image demonstrates predominantly low signal intensity throughout the mass (asterisk). (d) 3 min delayed post-contrast T1-weighted image demonstrates persistent heterogeneously moderate enhancement in portions of the dominant mass (asterisk).

Case presentation

A 52-year-old postmenopausal woman was referred to our gynecologic clinic, with the chief complaint of a notable increase in abdominal circumference and constipation caused by abdominal pressure. Six years previously, she underwent subtotal hysterectomy through laparoscopic morcellation for uterine leiomyoma. That operating records did not refer any peritoneal disease in that procedure, and the final pathological analysis demonstrated benign leiomyoma with hyaline degeneration, no evidence of malignancy.

Informed consent, after an explanation of the potential benefits and risks of surgical alterations, was obtained from the patient. The patient's written consent was also obtained for the tumor histological analysis and the use of photographs in terms of clinical findings. On physical examination, the abdomen was excessively enlarged similar to full term and tender. MRI revealed a cystic and solid mass throughout the abdomen and pelvis, measuring $18 \times 15 \times 12$ cm. The ill-defined mass did not appear to be originating from the cervix and ovaries (Figure 1(a)). Axial T2-weighted image demonstrates most portions of the mass (asterisk) to be primarily high signal with relatively small areas of fluid (Figure 1(b)). T1-weighted image demonstrates homogeneous low-signal intensity through the whole mass (Figure 1(c)), while portions of the tumor presented heterogeneously moderate enhancement on contrast-enhanced T1-weighted imaging (Figure 1(d)). Except an elevated cancer antigen 125 (CA125) level of 125.7 U/L, all other blood counts were within normal ranges.

With suspicious diagnosis of parasitic leiomyoma or leiomyosarcoma, gynecologists performed abdominal exploration. On laparotomy, the mass
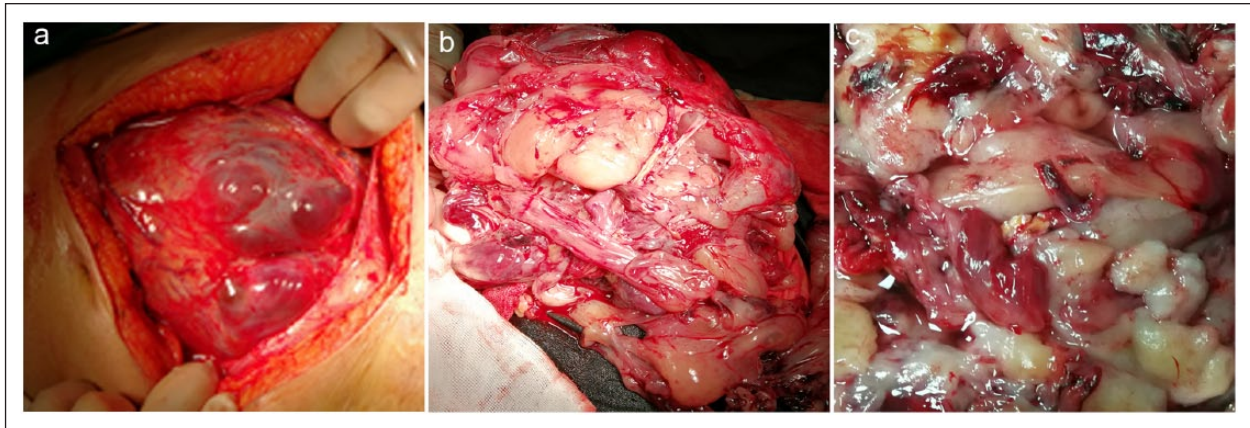
Figure 2. (a) On laparotomy, the abdomen and pelvis was found to be completely occupied by a huge multilobate mass that were surrounded by omentum and colon. (b) Large amounts of amorphous, gelatinous tissue were invested within all compartments of the intraperitoneal spaces. (c) Pseudocapsule around the tumor was ruptured and tissue with whitish-/yellowish-colored appearance flowed out.

surrounded by omentum and colon extended from the pelvic cul-de-sac to a portion of hemidiaphragm (Figure 2(a)). The cervix, ovaries, liver, kidneys and spleen appeared to be normal. Large amounts of amorphous, gelatinous tissue were invested within all compartments of the intraperitoneal spaces (Figure 2(b)), but no clear site of origin was recognized. Cytoreductive surgery with resection of all seen lesions and omentectomy was performed. During the procedure, pseudocapsule around the tumor was ruptured and tissue with whitish-/yellowish-colored appearance flowed out (Figure 2(c)).

A biopsy of tissue of approximately 0.4 cm × 0.4 cm thickness was obtained from non-necrotic abdominal tumor, washed thoroughly with phosphate buffered saline (PBS), placed in 10% paraformaldehyde for 48 h and then kept in 70% ethanol solution until embedding for immunohistochemistry. The tumor specimen was evaluated with respect to cellular morphology, necrosis, nuclear pleomorphism and mitotic count. Immunohistochemistry was performed on 4-mm-thick deparaffinized sections after antigen retrieval by pressure cooking in 1 mm ethylenediaminetetraacetic acid (EDTA) solution at pH 6.0 for 30 min. Freshly prepared 3, 3'-diamino-benzidine tetrahydrochloride (DAB) was applied as a chromogen and sections were counter-stained with haematoxylin. The following antibodies were used: smooth muscle actin (Abcam, Cambridge, UK), desmin (Dako), MyoD1 (Dako), estrogen receptor (ER; Dako) and Ki67 (Dako).

Microscopical findings revealed a mesenchymal tumor containing neoplastic cells of haphazard arrangement. Tumor cells were undifferentiated,

demonstrating polygonal morphology with cells short spindled shape to more rounded cells in myxoid stroma and varying amounts of eosinophilic cytoplasm. Furthermore, polynuclear cells and cells with bizarrely enlarged nuclei could be detected (Figure 3(a)). The overall histologic pattern of the resected tumor was largely suspicious of embryonal RMS. Immunohistochemical (IHC) analysis demonstrated strongly positive staining for smooth muscle actin (Figure 3(b)) and Desmin (Figure 3c), moderate staining for MyoD1 in morphologically appearing rhabdomyoblasts (Figure 3(d)), and focal positive staining for ER (Figure 3(e)), consistent with a diagnosis of RMS. The Ki67 proliferation index reached as high as 70% (Figure 3(f)). There were no postoperative complications, and the patient was administered with combined chemotherapy with six courses of doxorubicin and ifosfamide. However, 6 months later, abdominal MR imaging revealed the recurrence of the tumor. Because of poor performance status and extent of the disease, the patient was not considered a candidate for any surgical intervention and chemotherapy. Palliative therapy was administered, and then the patient died within 1 year after the initial surgery.

Discussion

Sporadic cases of primary RMS arising in the abdominopelvic cavity have been reported. This rarity makes our case a meaningful presentation to help in understanding the clinicopathological features of primary abdominopelvic RMS. Through extensive search of English language literature, we
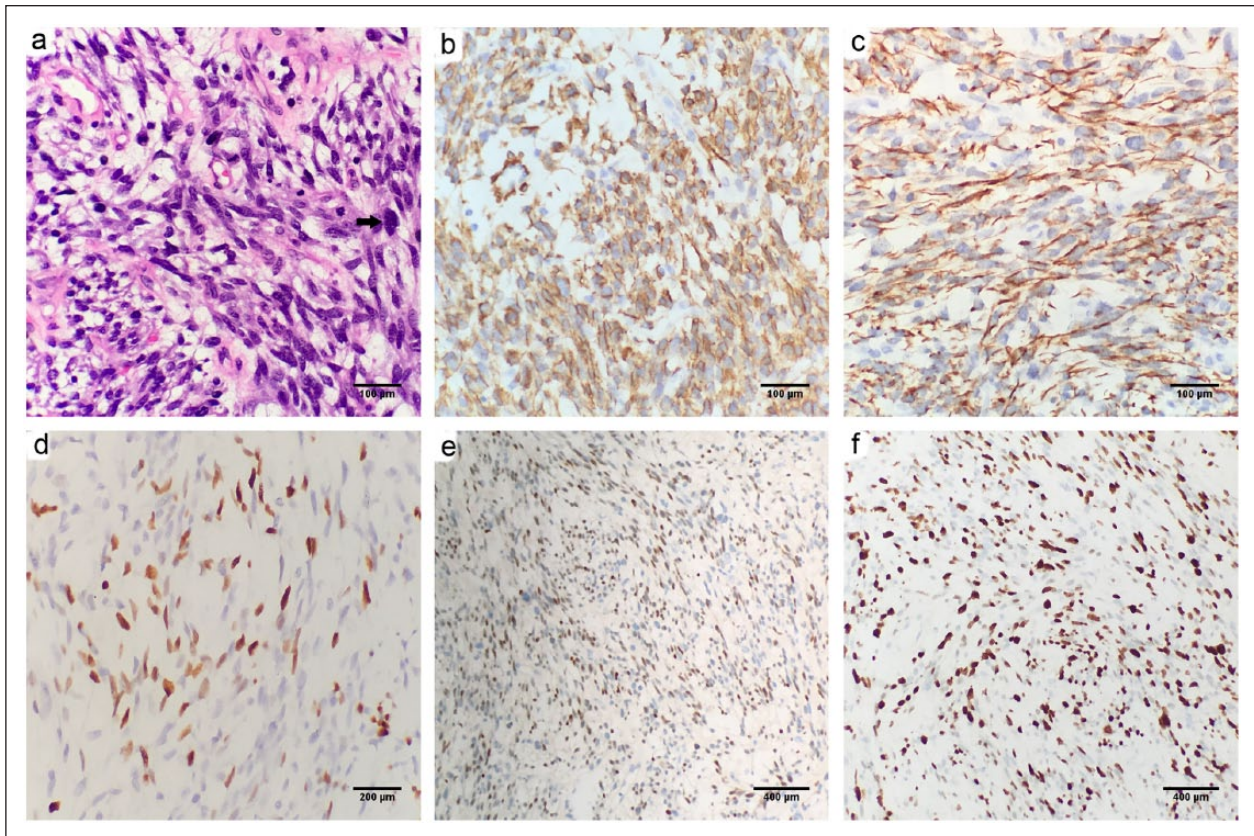
Figure 3. (a) Most of the neoplastic cells demonstrated polygonal morphology with short spindled shape to more rounded cells in myxoid stroma and varying amounts of eosinophilic cytoplasm. Furthermore, polynuclear cells and cells with bizarrely enlarged nuclei (arrow indicated) could be detected (HE staining; magnification ×20). (b) IHC demonstrated a malignant spindle cell neoplasm positive for smooth muscle actin (Envision: magnification ×20). (c) IHC showed strong positive staining for desmin (Envision: magnification×20). (d) IHC showed moderate cytoplasmic staining for MyoD1 in morphologically-appearing rhabdomyoblasts (Envision: magnification ×10). (e) IHC presented focal positive staining for ER (Envision: magnification ×5). (f) The Ki67 proliferation index was 60%–70% (Envision: magnification 5×).

found two case reports describing primary intra-abdominal RMS in adults. In 1999, Andrew M. Kaplan reported the first well-documented case of intra-abdominal RMS in a female adult.[4] In that case, abdominal exploration revealed similar appearance to that of our case. Also, no origin of the ill-defined tumor was recognized. The second case described that a 65–year-old male patient was diagnosed as intra-abdominal RMS with a clear margin and solid appearance.[5] Both cases were classified as embryonal RMS.

The World Health Organization (WHO) divided RMS into four distinct subtypes: embryonal, alveolar, pleomorphic, and spindle cell/sclerosing. The incidence of RMS subtype in adults still remains uncertain. Pediatric and adult RMS differ in terms of histopathological subtype that embryonal types arise in childhood, pleomorphic types occur in older adults with a mean age of 51 years at diagnosis,[6] and alveolar tumors affect all age groups. Histologically, this age distribution corresponds with the histologic maturity of the RMS subtype, because embryonal types resemble embryonic tissues and pleomorphic groups appear as aggressive adult carcinomas with malignant fibrous histiocytoma like characteristics. The patient in our case was confirmed as embryonal RMS based on the microscopic findings with undifferentiated neoplastic cells as well as IHC results. RMS cells could express desmin, as well as myogenic transcription factors MyoD1 or myogenin, or both.[6] Due to the high sensitivity and specificity of MyoD1 in the diagnosis of RMS, it is a very useful tumor marker to identify RMS from other non-RMS.[7]

So far, limited numbers of studies exist regarding the practicability of whole-body MRI application for patients with RMS. In our case, MRI demonstrated high signal intensity on T2-weighted MR images and low signal intensity on T1-weighted

MR images, which was consistent with one previous report.[8] Due to the past surgical history of subtotal hysterectomy through laparoscopic morcellation, the patient was initially suspicious of disseminated leiomyosarcoma. However, one previous report described the MR imaging features of disseminated uterine leiomyosarcoma, with a low-signal intensity on T2-weighted MR and intermediate intensity on T1-weighted MR.[9] Therefore, MR can also be applied in the differentiation between leiomyosarcoma and RMS.

It is difficult to determine the optimal management therapy due to the relative rarity of RMS in adults. The current guidelines for treating adult RMS are in line with the multimodal therapy (MMT: resection, chemotherapy, and radiation) suggested by the Intergroup Rhabdomyosarcoma Studies (IRS) which achieved prominent improvements in long-term survival for children with RMS.[10] However, the same treatment protocols have been less effective for adult RMS, with 5-year overall survival as low as 31%–44% from the limited retrospective case series.[11] Small studies have found that in adults with RMS, chemotherapy responders had improved metastasis-free survival compared to nonresponders. Adjuvant chemotherapy with doxorubicin-based regimes and ifosfamide showed reduction in recurrence rate and improved prognosis in patients with localized resectable soft-tissue sarcoma.[12] In our case, we administered six courses of chemotherapy without combined radiotherapy, due to the particular location of intra-abdominal RMS as well as no organ origin responsible for the tumor. The disease recurred shortly after termination of chemotherapy probably due to the subtype and the difficulty of complete resection.

In conclusion, we present a rare case of primary abdominopelvic RMS mimicking a gynecologic malignancy. Literature regarding RMS clinicopathological features and management option is very limited. It is important for gynecologists and radiologists to recognize RMS so it can be listed in the differential diagnosis of masses arising in the abdomen and pelvis.

Declaration of conflicting interests

Funding

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