

# Pleomorphic rhabdomyosarcoma metastasis to small intestine causing intussusception

## A case report

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### Abstract

**Rationale:** Pleomorphic rhabdomyosarcoma (PRMS) is a rare soft tissue malignancy which is frequently misdiagnosed and associated with metastasis to the lungs, lymph nodes, and bone marrow. Case studies are needed to improve the awareness of the disease and our understanding of it.

**Patient concerns:** In this study, we present a case of a 36-year-old man with a lesion on the right back shoulder. Lesion was confirmed by magnetic resonance imaging (MRI) around the right armpit, subscapularis, deltoid, and infraspinatus muscle, with oozing surrounding soft tissues.

**Diagnosis:** The tumor was diagnosed as PRMS which metastasized to the intestine, where it caused intussusception.

**Interventions:** The patient was treated by complete surgery in combination with neo-adjuvant chemotherapy including ifosfamide and epirubicin.

**Outcome:** The patient remained alive 6 months after the treatment with no recurrence and metastasis.

**Lesson:** PRMS can be aggressive, and surgical treatment in combination with multidrug chemotherapy can be used in the management.

**Abbreviations:** CT = computed tomography, PRMS = pleomorphic rhabdomyosarcoma, RMS = Rhabdomyosarcoma.

**Keywords:** pleomorphic rhabdomyosarcoma, rhabdomyosarcoma, sarcoma, soft tissue tumor

## 1. Introduction

Rhabdomyosarcoma (RMS) is a malignant mesenchymal tumor that originates from skeletal muscle cells. It mostly occurs in children and adolescents,<sup>[1]</sup> accounting for about 5% of all pediatric cancers and 50% of all soft tissue sarcomas.<sup>[2]</sup> Among the 3 major histotypes of RMS: embryonal, alveolar, and pleomorphic subtypes, embryonal RMS is mostly diagnosed in children under 10 years old,<sup>[2]</sup> alveolar RMS predominantly in adolescents and young adults<sup>[2]</sup> while pleomorphic RMS (PRMS) almost exclusively affects adults aged over 45.<sup>[1]</sup> The histotypical variants of RMS are associated with differential prognosis with embryonal RMS having the most favorable outcome.<sup>[2]</sup> In addition, older age predicts disease metastasis and poor survival,<sup>[2]</sup> likely due to the increased incidence of unfavorable

histotype, poor chemosensitivity, and decreased tolerance of patients towards chemotherapies.<sup>[1]</sup>

Pleomorphic RMS represents the most aggressive subtype of RMS, with a 5-year overall survival of 27% to 44%,<sup>[1]</sup> however, the diagnostic criteria for it has been controversial,<sup>[1,3]</sup> leading to frequent misdiagnosis of this lethal malignancy. Metastasis is often found at diagnosis, commonly to lungs, lymph nodes, and bone marrow. In the present study, we report a rare case of metastasis of PRMS, which originated from the right back shoulder, to the small intestine causing intussusception and obstruction. The patient was treated by chemotherapy and surgical resections of primary and metastatic tumors and remained alive half a year later.

## 2. Case description

This case study was approved by the Ethics Committee of the First Affiliated Hospital of Jilin University, and written informed consent was obtained from the patient.

A 36-year-old man presented with a mass on the right back shoulder in October 2015. Magnetic resonance imaging (MRI) indicated lesions around the right armpit, subscapularis, deltoid, and infraspinatus muscle, with oozing surrounding soft tissues. The mass was diagnosed by biopsy as sarcoma and treated with 3 cycles of neoadjuvant chemotherapy. Each cycle included daily intravenous infusions of ifosfamide, for 5 days at 5.0 g/day, and epirubicin for 3 days, at 60 mg/day for the first 2 days and 50 mg/day for the third day. Tumor regression was observed after the treatment, and in January 2016, the residual tumor was excised and confirmed by postoperative pathological analysis as PRMS (Fig. 1A–F), with post-chemotherapy features. Immunohis-

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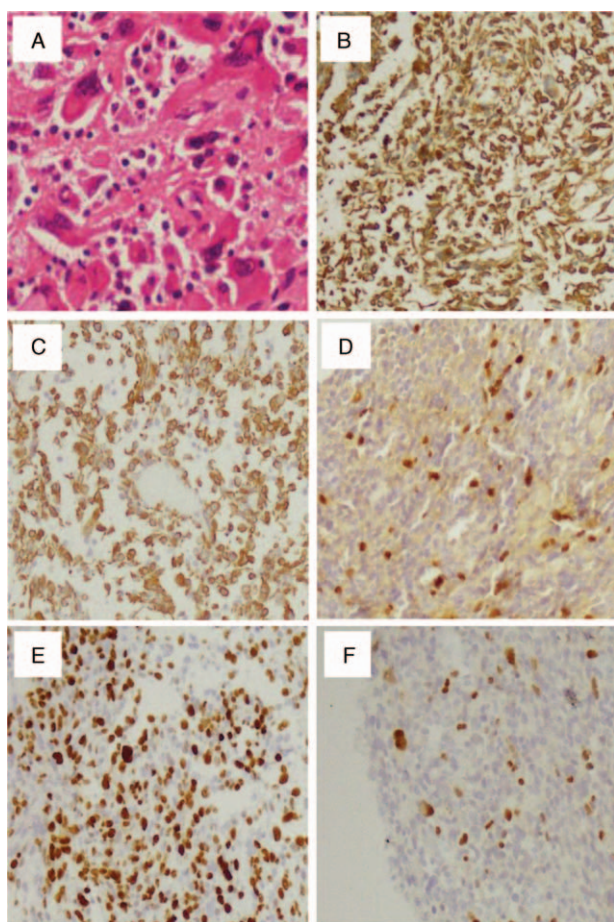
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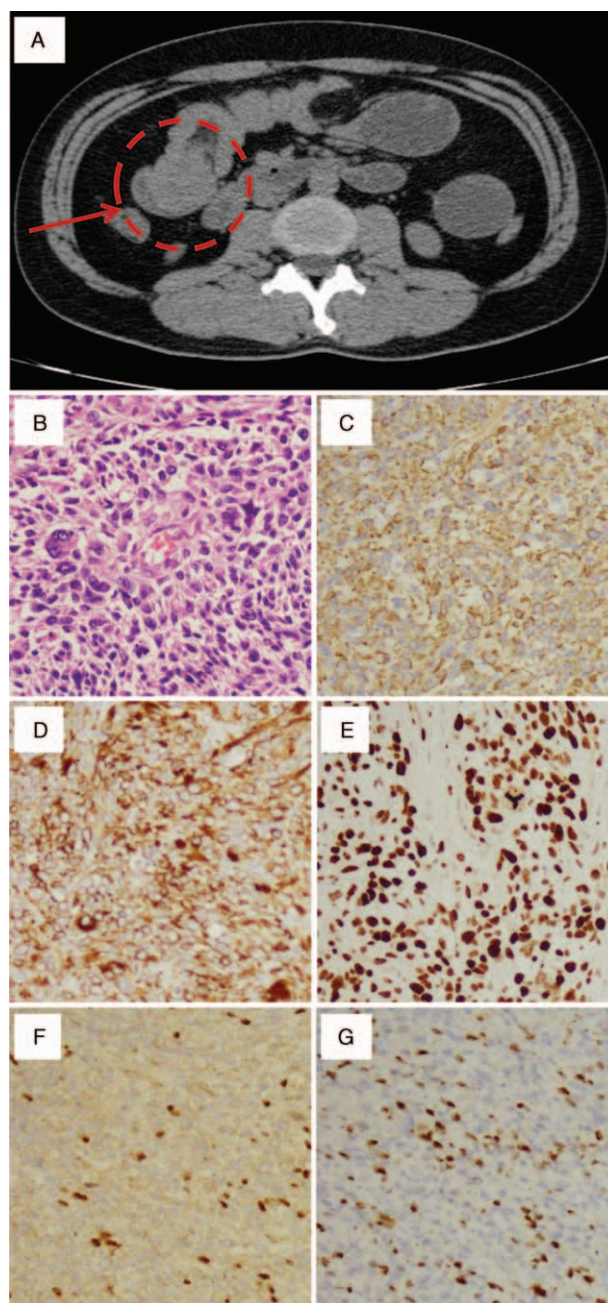
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**Figure 1.** Histopathological and immunohistochemical analysis of the primary pleomorphic rhabdomyosarcoma. Formalin-fixed paraffin-embedded tumor tissue was sectioned and stained with hematoxylin and eosin (A) or antibodies against vimentin (B), desmin (C), myogenin (D), Ki-67 (E), and MyoD1 (F) and imaged (Magnification: 200 ×).

tochemistry analysis of the tumor demonstrated positive staining for vimentin (Fig. 1B), desmin (Fig. 1C), smooth muscle actin (SMA) and CD99, scattered positive staining for myogenin (Fig. 1D), 30% positivity for Ki-67 (Fig. 1E), and minor positivity for MyoD1 (Fig. 1F), but there was no expression of AE1/AE3, CD34, CD68, S100, and H-caldesmon. The patient was further treated with 1 cycle of radiotherapy, at a target volume dose of 60 Gy per day at 2Gy/fraction, and 3 cycles of ifosfamide and epirubicin as aforementioned.

On June 5, 2017, the patient was re-admitted complaining about discomfort in the left abdomen for 2 months with obstructed defecation for 4 days. Physical examination revealed abdominal tenderness over the left lower quadrant but with no rebound tenderness or muscle tension. Full abdominal X-ray computed tomography (CT) showed small intestine intussusception (Fig. 2A), which caused proximal obstruction, while chest CT revealed no neoplastic lesion but scattered bilateral lower lobe inflammation and localized emphysema in the right lower lobe. Laparotomy performed on the same day identified an intussusception of a 20-cm section of intestine, about 100 cm away from the Triesz ligament, into distal intestine. A 4 × 4 × 3 cm mass and a 0.5 cm gray nodule were found in the lumen and removed by small bowel resection and anastomosis. Resection margins were ensured to be free of tumors, and no invasion into the intestinal lymph nodes was found (0/2).



**Figure 2.** Intestine intussusception was detected under abdominal CT (A). Metastatic PRMS was excised surgically, fixed, and paraffin-embedded. Histopathological and immunohistochemical analysis was performed by staining the sections with hematoxylin and eosin (B) or antibodies against vimentin (C), desmin (D), Ki-67 (E), myogenin (F), and MyoD1 (G) and imaged (Magnification: 200 ×). CT=computed tomography, PRMS=pleomorphic rhabdomyosarcoma.

Postoperative pathological analysis of the tumors (Fig. 2B–G) confirmed two malignant lesions with hemorrhage and necrosis. The larger one measured 4 × 4 × 3 cm, involving full thickness of the intestinal wall, while the smaller one was 0.5 cm in the longest diameter. No involvement of vessels and nerves was observed. Immunohistochemical analysis showed pathological changes consistent with PRMS, with positive staining for vimentin (Fig. 2C), desmin (Fig. 2D) and SMA, 90% positivity for Ki-67 (Fig. 2E), partial positivity for myogenin (Fig. 2F) and MyoD1



(Fig. 2G), and focal positivity for CD117, but there was no staining for Dog-1, CD34, and S100.

Half a year after the small bowel resection and anastomosis, the patient remained alive with no further adjuvant radiotherapy or chemotherapy. Re-examination by abdominal and chest CT revealed no tumor recurrence and metastasis.

### 3. Discussion

Pleomorphic rhabdomyosarcoma (PRMS) is a rare but highly malignant soft tissue cancer, accounting for 3% of soft tissue sarcomas which make up less than 1% of all adult malignancies.<sup>[4]</sup> Arising from striated muscle cells, PRMS is mostly located in the large muscles of the extremities, particularly the lower limbs and the trunk, such as the abdomen/retroperitoneum, chest, and abdominal wall,<sup>[3]</sup> although its occurrence around the head and neck area<sup>[5]</sup> and parenchymal organs, including the testis,<sup>[6]</sup> uterus,<sup>[7]</sup> and bladder,<sup>[8]</sup> has also been reported. In this study, we report a case of PRMS originated from the right shoulder but metastasized to the intestine, which is the first case reported to the best of our knowledge. In addition, the metastatic tumor caused intussusception, which is another rare clinical observation in adults.

This case of PRMS was originally diagnosed as sarcoma and treated with ifosfamide and epirubicin. The PRMS histotype was confirmed by histopathology and a series of immunohistochemistry analyses, which demonstrated the skeletal muscle phenotype. Immunohistochemical analysis revealed expression of vimentin, desmin, SMA, myogenin, and MyoD1, but not S100 and CD117, in consistence with previous studies.<sup>[3]</sup> Reliance on multidisciplinary approaches in the diagnosis of this case mirrors the difficulty in the early detection of PRMS. Since firstly reported by Stout in 1946<sup>[9]</sup> limited number of cases of adult PRMS have been reported.<sup>[1,3]</sup> Our knowledge of this rare cancer is based on case reports and retrospective studies with small cohorts of patients.<sup>[1,3]</sup> The lack of comprehensive knowledge of this lethal disease contributes to the controversies in regards to its diagnostic criteria. Future genomic and proteomic profiling of PRMS and characterization of molecular pathways involved in its pathogenesis are expected to identify specific markers for this rare malignant disease.

Metastasis of PRMS is commonly seen in the lungs, lymph nodes, and bone marrow.<sup>[6]</sup> This study reports a unique case of metastasis to the intestine, highlighting the importance of gastrointestinal examination by positron emission tomography (PET)-CT or endoscopy in future practice for patients with PRMS.

Both primary and metastatic PRMS, in this case, were treated by surgical resection in combination with ifosfamide and

epirubicin. Complete surgery represents the standard treatment for PRMS,<sup>[4]</sup> and data on the chemotherapeutic treatment of PRMS is extremely rare. Among the 7 PRMS patients treated with neoadjuvant doxorubicin, ifosfamide, and vincristine, 2 showed a complete response and 2 a partial response.<sup>[10]</sup> This case of PRMS also showed sensitivity to ifosfamide and epirubicin although the long-term outcome remains to be defined.

In conclusion, PRMS is a rare but aggressive cancer in adults. Surgical resection with multidrug chemotherapy represent potential treatment options, however, the chemotherapy regime is yet to be optimized, and the long-term effect of the combinational treatment on patient outcome is yet to be defined.

### Author contributions

**Conceptualization:** Weihua Tong.

**Data curation:** Shiwen Xi.

**Investigation:** Shiwen Xi, Weihua Tong.

**Writing – original draft:** Shiwen Xi.

**Writing – review & editing:** Shiwen Xi, Weihua Tong.

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