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Case report

Perivascular epithelioid cell tumor of the gastrointestinal tract

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ABSTRACT

Perivascular epithelioid cell tumor (PEComa) is a rare subtype of soft tissue sarcoma that coexpresses melanocytic and smooth muscle markers. We report a case of malignant PEComa arising from the gastrointestinal tract. Definitive diagnosis of malignant PEComa is based on pathological examination and the standard treatment is complete surgical resection. Cytotoxic chemotherapy has limited efficacy in recurrent or metastatic malignant PEComa. Overactivation of the mammalian target of rapamycin (mTOR) pathway is observed in PEComa, and mTOR inhibitors have been recommended as an effective systemic treatment.

1. Introduction

Perivascular epithelioid cell tumor (PEComa), a rare subtype of mesenchymal neoplasm, is composed of perivascular epithelioid cells that coexpress myogenic and melanocytic markers on immunohistochemical staining.[1] Bonetti et al. initially described these distinctive cells in 1992.[2] Currently, the family of PEComas includes angiomyolipoma (AML), pulmonary lymphangioleiomyomatosis (LAM), clear cell sugar tumor of the lung, extrapulmonary clear cell sugar tumor, and clear cell myomelanocytic tumor.[3] Although rare, PEComas have been reported in a variety of anatomical locations and are considered to be ubiquitous.[4] The clinical behavior of PEComas is highly heterogeneous. Most are benign and are cured by surgical resection,[5] but a minority recurs or metastasizes. Thus, various definitions of the malignant potential of these tumors have been proposed.[1,3,6] PEComa patients have been treated with conventional chemotherapy used in other types of sarcoma. However, recognition of aberrantly activated mammalian target of rapamycin (mTOR) pathway in malignant PEComa led to the use of mTOR inhibitors, such as sirolimus, for systemic therapy, and promising anti-tumor effects have been demonstrated.[5] Here, we report a rare case of PEComa arising from the gastrointestinal tract and review the literature regarding this rare subtype of soft tissue sarcoma.

2. Case report

A 60-year-old woman was in her usual state of health until late 2013 when she started to experience acid regurgitation and postprandial fullness. Esophagogastroduodenoscopy showed only mild reflux esophagitis. Her symptoms failed to respond to symptomatic treatments, including metoclopramide, antacids, and proton pump inhibitors, and progressed to easy vomiting over two months and experienced unintentional weight loss of 4 kg in 1 week. There was no tarry stool or change in stool caliber. She was examined at a local hospital, and initial examination with abdominal ultrasonography revealed a large intra-abdominal tumor. Computed tomography (CT) of the abdomen showed a 12 × 12 × 9 cm irregular mass in the left abdomen, with heterogeneous enhancement and mass effect on the adjacent structures. No ascites or enlarged lymph nodes were seen on the CT image (Fig. 1A). The results of all her laboratory tests were within the normal range. A diagnosis of gastrointestinal stromal tumor (GIST), leiomyosarcoma, or adenosarcoma was suspected. Surgical resection was suggested, and she was then referred to our hospital for further management.

The patient was admitted to our gastrointestinal ward in November 2013, and a follow-up abdominal ultrasonography

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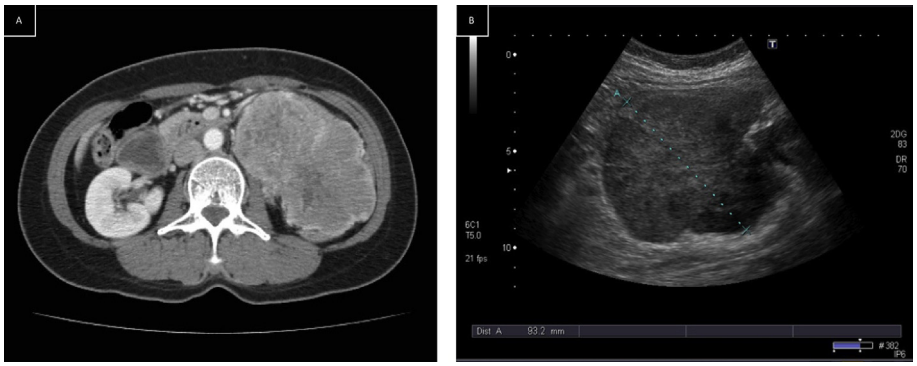

Fig. 1. (A) CT showed a large tumor, measuring up to 12 cm, in the left abdomen (B) Abdominal echo showed a heterogeneous and hypoechoic lesion with hypervascularity arising from the small bowel wall.

showed a large heterogeneous and hypoechoic lesion with hypervascularity arising from the small bowel wall over the left upper quadrant area (Fig. 1B). A tumor marker panel showed levels within normal limits, as follows: carcinoembryonic antigen (CEA), 1.25 ng/mL (normal range <5 ng/mL); CA 19-9, 6.34 U/mL (normal range < 37 U/mL); CA 15-3, 6.8 U/mL (normal range 0–31 U/mL); and CA-125, 7.1 U/mL (normal range 0–35 U/mL). A small bowel GIST or leiomyosarcoma was suspected, and a general surgeon was consulted for further resection.

The patient underwent surgery on December 2, 2013. A 13 × 13 cm elastic lobulated yellowish tumor, arising from the upper jejunum with suspected local invasion into the transverse colon and mesentery, was detected intraoperatively. Thus, small bowel segmental resection and left hemicolectomy were performed as radical treatment. Under gross examination by our pathologist, the tumor was observed to arise from the jejunal muscle layer with a focally jagged and infiltrative tumor border and that it extended to the subserosa, serosa, and nearby mesocolon.

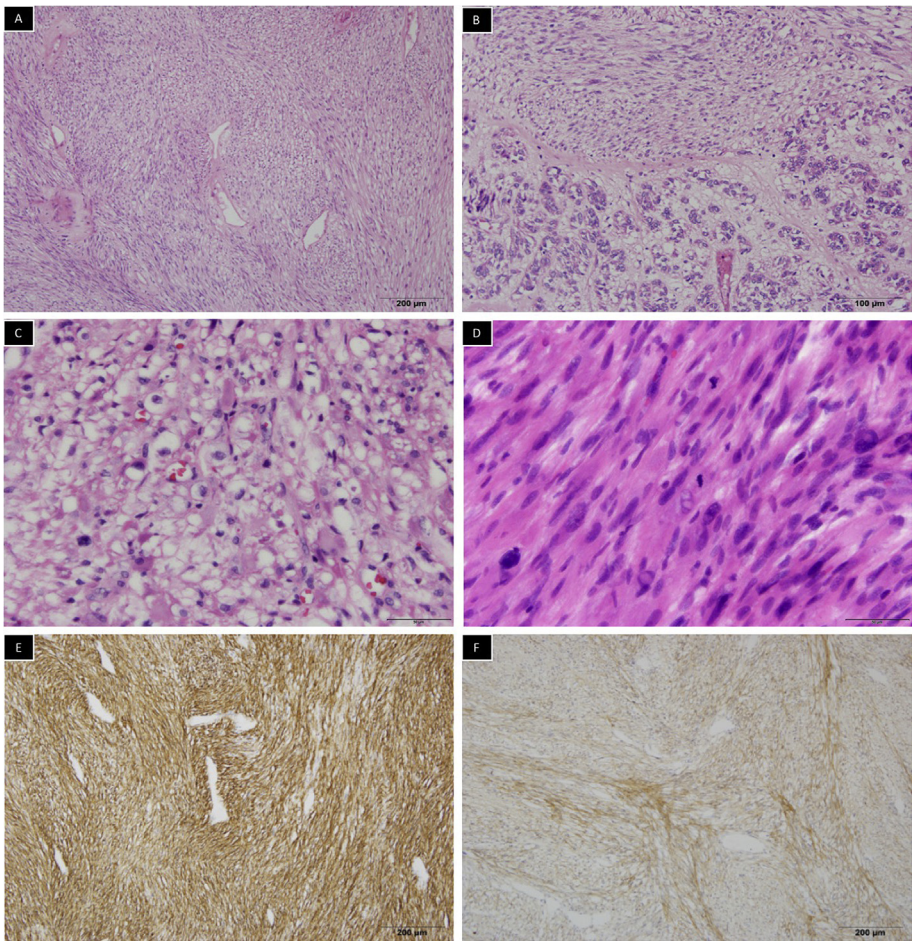

Fig. 2. (A) Epithelioid tumor cell radiating from staghorn-like vessels (100×). (B) Admixture of spindle cells and epithelioid cells (200×). (C) Epithelioid cells with mild nuclear atypia (400×). (D) Spindle cells with mitoses (400×). (E) Tumor cells staining positive for smooth muscle actin (100×). (F) Tumor cells staining positive for Melan-A (100×).