Case Report

Retroperitoneal malignant perivascular epithelioid cell tumor: a rare case report and literature review

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Background: Perivasuclar epithelioid cell tumor (PEComa) is a rare group of mesenchymal tumors with unique histological and immunophenotypes. Retroperitoneal PEComa is extremely rare and almost benign lesion. Herein, we will present a case from China, focusing on histological and immunohistochemical characteristics of the tumor, follow-up and long-term outcome and the related literature review.

Case Description: This 27-year-old Chinese female patient suffered chronical abdominal pain. Transvaginal ultrasound revealed a neoplasm of size 15 cm × 8 cm × 10 cm in the right adnexal region. Following laparoscopy confirmed the mass with multilocular cyst retroperitoneal, adhesion to the right ureter and iliac vessels, and the boundary is unclear, containing about 500 mL turbid fluid. Pathologic immunohistochemical staining indicated HMB-45 (26% positive), SMA (82% positive), Caldesmon (78% positive), Desmin (27% positive), Ki-67 (25% positive), ER (15% positive), PR (12% positive), CDK-4 (5% positive), MiTF (5% positive), S-100 (negative), CD117 (negative), CD34 (negative), EMA (negative), MDM2 (negative), WT-1 (negative) and fluorescence in situ hybridization (FISH) test revealed negative with *MDM2* gene. A retroperitoneal malignant perivascular epithelioid cell tumor was confirmed finally. The patient was well recovered post the operation.

Conclusions: Based on a literature review, there were rare retroperitoneal malignant PEComa reported before, especially the one from China. We wrote this report to arose the attention to this disease, including the diagnosis and antidiastole.

Keywords: Perivasuclar epithelioid cell tumor (PEComa); retroperitoneal; pathology; diagnosis and antidiastole; case report

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Introduction

Perivasuclar epithelioid cell tumor (PEComa) is a rare group of mesenchymal tumors with unique histological and immunophenotypes, which was first proposed by Zamboni in 1996 (1). PEComa can be occured in any ages, mostly in the middle-age. The lesions can involve in various parts of bodies, and uterus is the most common organ (2,3). Retroperitoneal PEComas are extremely rarely seen and almost all benign lesions (2,3).

Herein, we present a case of malignant retroperitoneal PEComa with literature review to draw attention to this disease. Based on a literature review in *PubMed* and *Web of Science* with the terms of ("malignant perivascular epithelioid cell tumor" or "malignant PEComa") and ("Post-peritoneal" or "retroperitoneal"), similar report of retroperitoneal malignant PEComa in this paper was rare reported before, especially the one from China (4,5). Compared with former cases, we focused on histological and immunohistochemical

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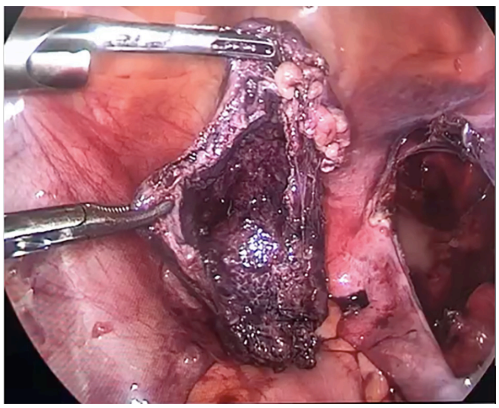

Figure 1 The retroperitoneal mass with 12 cm × 10 cm × 10 cm in size was removed during the surgery.

(IHC) characteristics of the tumor, follow-up and long-term outcome. We present the following case in accordance with the CARE reporting checklist (available at https://gpm.amegroups.com/article/view/10.21037/gpm-21-44/rc).

Case description

A 27-year-old Chinese female patient, gravida 0 para 0, was admitted to our hospital with a chief complain of chronical abdominal pain in Jan. 20, 2019. This symptom had occurred over a period of 3 months. She refused any medical and family history. Physical examination presented that the patient got an adnexal mass. Remaining physical examination did not reveal any additional abnormalities. Trans-vaginal ultrasound revealed a neoplasm of size 15 cm × 8 cm × 10 cm in the right adnexal region. The serum tumor markers were normal. The medical history and family history was normal. With a suspicion of ovarian cyst, a surgery then arranged.

At the explorative laparoscopic, the right ovary is atrophied of 2 cm × 2 cm × 2 cm in size; and the left ovary and uterus were normal in size. The rest exploration of pelvic and abdomen was normal. A retroperitoneal mass was noticed, with 12 cm × 10 cm × 10 cm in size. Following laparoscopy confirmed the mass with multilocular cyst retroperitoneal, adhesion to the right ureter and iliac vessels, and the boundary is unclear, containing about 500 mL turbid fluid (*Figure 1*). Subsequent IHC staining indicated HMB-45 (26% positive) (*Figure 2A*), SMA (82% positive) (*Figure 2B*), Caldesmon (78% positive) (*Figure 2C*), Desmin (27% positive) (*Figure 2D*), Ki-67 (25% positive), ER (15% positive), PR (12% positive), CDK-4 (5% positive), MiTF (5% positive), S-100 (negative) (*Figure 2E*), CD117 (negative) (*Figure 2F*), CD34 (negative) (*Figure 2G*), EMA (negative) (*Figure 2H*), MDM2 (negative), WT-1 (negative) and fluorescence in situ hybridization (FISH) test revealed negative with *MDM2* gene (*Figure 2I*). There was no existence of necrosis in the mass.

After a clinic-pathological debate, the patient was diagnosed with retroperitoneal malignant PEComa. Three months later of the treatment, the patient returned for follow-up and trans-vaginal ultrasound revealed with ascites. The second review was undertaken at six months later of the surgery, and no ascites was detected. And then, she received another 3 follow-ups every 6 months. During the 2 years post-surgery, the patient recovered well without any chemotherapy, radiation therapy, or other drug interventions. The patient claimed she was quite satisfied with the treatment and the prognosis she received.

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

Discussion

Origin of PEComa

In 1991, Pea first reported that renal angiomyolipoma commonly expressed HMB-45, a relatively specific marker of melanocyte-differentiated tumors (6). In 1992, Bonetti proposed the concept of "perivascular epithelial cells" to name these cells (7). In 1994, Bonetti further unified the constitutive cells of angiomyolipoma, lung clear cell "sugar" tumor, and lymphangiomyomatosis as "perivascular epithelioid cells", which are characterized with "unique muscle cells, different expressions of Melanoma-associated antigen" (8). In 1996, Zamboni *et al.* firstly used the term of PEComa to describe a pancreas-derived clear epithelioid cell tumor with HMB-45 (+) (1).

In the following years, some reports on PEComa satisfied these basic histological features, and those tumors extensively distributed in bodies. Nowadays, based on the definition of the World Health Organization tumor classifications, PEComa refers to an interstitial tumor composed of perivascular epithelioid tissue with histological

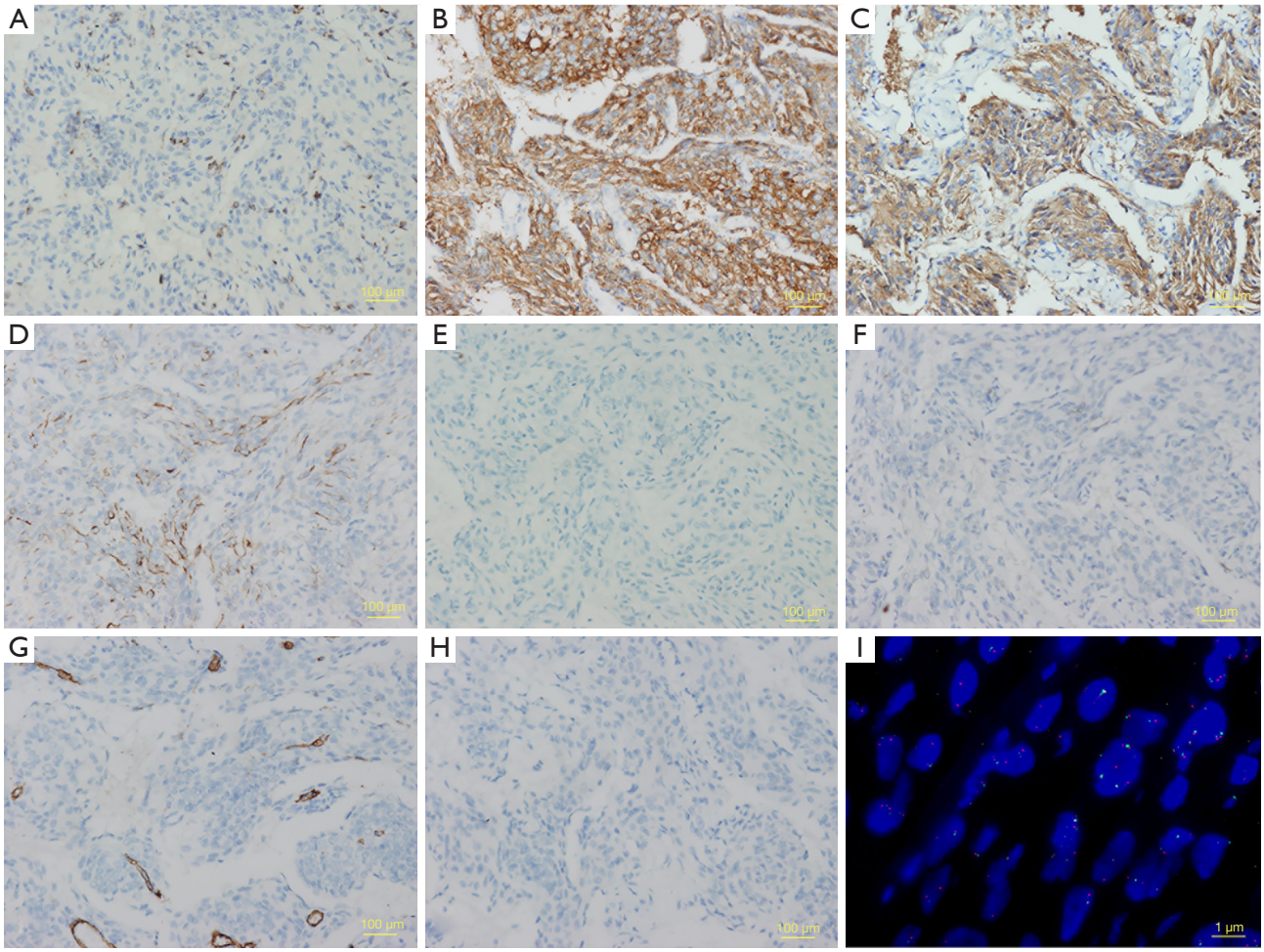

Figure 2 The IHC stains and FISH results of the mass. (A) HMG-45, ×200. (B) SMA, ×200. (C) Caldesmon, ×200. (D) Desmin, ×200. (E) S-100, ×200. (F) CD-117, ×200. (G) CD-34, ×200. (H) EMA, ×200. (I) FISH result, the green signal is CEP12 probe, the red signal is MDM2 probe. IHC, immunohistochemistry; FISH, fluorescence in situ hybridization.

and immunohistochemical features (9). Angiomyolipoma, lymphangioleiomyoma, and clear-cell sugar tumor of the lung are typical members of the PEComa family (7,8,10,11).

Pathological characteristics

Most patients have no obvious symptoms. Some can only accompany with abdominal pain, abdominal distension and diarrhea. PEComa is mostly benign. Due to the reports were limited, the diagnostic criteria for malignant PEComa have not been establishment. Folpe *et al.* classified PEComa tumors as benign, uncertain malignant potential and malignant (12).

- Benign: tumor diameter <5 cm, non-infiltrative, mild nuclear grade and cellularity, mitotic index ≤1/50 high power fields (HPF), no necrosis and vascular invasion.
- Uncertain malignant potential (nuclear pleomorphism/ multinucleated giant cells only or tumor diameter >5 cm only).
- Malignant: tumor diameter >5 cm, infiltrative, high nuclear grade and cellularity, mitotic rate ≥1/HPF, necrosis and vascular invasion).

Grossly, the interior surface is yellowish to grayish red and the boundary is unclear, with or without focal hemorrhage and necrosis.

Under the microscope, PEComa is often located around the blood vessels, and the cells are arranged around the

lumen of the blood vessel. Typical perivascular epithelioid cells are epithelial-like cells around the vicinity of the blood vessels, and the fusiform cells tend to be far from the blood vessels. In the uterus, kidney, perirenal or peritoneum, PEComa cells can exhibit a large number of interstitial transparency (7,8,11). In immunohistochemistry, nearly 100% of PEComas expressed HMB-45 and the positive rate of SMA was 59%, Melan-A was 41%, CD117 was 33%, Desmin was 31%, S-100 was 10%, and negative of cytokeratins (12).

Antidiastole

Malignant melanoma

Due to the strong positive expression of HMB-45, PEComa is easily misdiagnosed as malignant melanoma. In malignant melanoma immunohistochemistry, HMB-45 and S-100 expression are positive, and SMA expression is negative. However, in PEComa, SMA expression is generally positive, and S-100 expression is negative or rarely positive (3).

Epithelioid leiomyoma

The expression of SMA in epithelioid leiomyoma is nearly 100% positive, and the expression of HMB-45 is often negative, while HMB-45 expression in PEComa is diffuse positive strongly (13).

Clear cell carcinoma

Clear cell carcinoma lacks of the perivascular epithelial-like cell; keratin and carcinoembryonic antigen expressions are positive; while HMB-45 and SMA expressions are negative (3).

Endometrial stromal sarcoma

The CD10 expression of endometrial stromal sarcoma is diffusely strong, while the CD10 expression of PEComa is negative. In addition, PEComa was positive for both HMB45 and Melan-A expressions (13,14).

Treatment and follow-up

Surgical resection is still the most reliable and effective treatment currently, including removal of primary and metastatic lesions. The roles of chemotherapy and immunotherapy are still unclear. Although most PEComas are benign, the biological behavior is not clear, and may accompany with potential behaviors, thus a long-term follow-up is recommended.

Strengths and limitations

In this case, we preformed enough immunohistochemistry stains to reach the final diagnosis and presented antidiastole characteristics. Some previous literature revealed ^{18}F-FDG-PET/CT can help characterize these lesions and new therapies with inhibitory m-TOR may open a hopeful therapeutic window (15). But we failed to get the PET/CT scan and immune therapy, due to the patient was lack of insurance and cannot afford it herself.

Conclusions

Retroperitoneal malignant PEComa was rare reported before. Specially attention should be paid to the retroperitoneal malignant PEComa. For take home massage, immunohistochemistry stain is strongly recommended for the diagnosis of PEComa. A surgical removement of the mass is the main treatment method. The roles of chemotherapy and immunotherapy are still unclear and a long-term follow-up is suggested. However, with limited clinical and follow-up data, further cases need to be collected.

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Footnote

Reporting Checklist: The authors have completed the CARE reporting checklist. Available at https://gpm.amegroups.com/article/view/10.21037/gpm-21-44/rc

Conflicts of Interest: Both authors have completed the ICMJE uniform disclosure form (available at https://gpm.amegroups.com/article/view/10.21037/gpm-21-44/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and accompanying

images. A copy of the written consent is available for review by the editorial office of this journal.

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