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Retroperitoneal hamartoma mimicking angiomyolipoma: a case report and review of literature

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Abstract

Background: Hamartoma refers to the excessive focal growth of cells and tissues in organs, which is between malformation and benign tumor. Hamartomas are commonly seen in the lung, but rare in the retroperitoneum.

Main text: Angiomyolipoma was also called hamartoma in the early years. Hamartoma was not separated from the concept of angiomyolipoma until perivascular epithelioid cells were found in angiomyolipoma. We reported a case of retroperitoneal hamartoma, which was suspected to be angiomyolipoma in the initial imaging findings and pathological frozen section, but the paraffin section was finally diagnosed as hamartoma. Due to the fusion of the tumor with the right psoas major muscle, it may need to be cut off for complete resection of tumor, which may damage the sacrococcygeal nerve and affect the postoperative activity function, so the tumor was just partially removed. After 6 and 22 months of follow-up, the patient was in good condition. CT re-examinations showed that the epibiotic right retroperitoneal mass size was $40 \times 42 \times 72$ mm^3 (6 months, length × width × height) and $40 \times 45 \times 81$ mm^3 (22 months, length × width × height). We describe this case and review the literature.

Conclusions: In conclusion, retroperitoneal hamartoma is rare and difficult to differentiate from angiomyolipoma, in which perivascular epithelial cells could be found on pathological paraffin sections.

Keywords: Hamartoma, Retroperitoneal, Angiomyolipoma, Phosphatase and tensin homolog

Background

Hamartoma is a pathological change between malformation and benign tumor, which is defined as the excessive focal formation of cells and tissues in organs (Kumar and Fausto 2004; Sigdel et al. 2012). Hamartomas are common in the lung. Microscopically, hamartomas are tumor-like hyperplasia composed of well-differentiated and disordered tissue components including blood vessels, fibrous tissue, fat, smooth muscle, nerve, and cartilage. Since the tissue composition of retroperitoneal hamartoma overlaps with retroperitoneal angiomyolipoma (AML), it is difficult to differentiate them by radiological examination. In the earlier years, AML was also called hamartoma, but now it is considered that AML belongs to the perivascular epithelioid tumors (PEComas) family (Lienert and Nicol 2012). We reported a case of retroperitoneal hamartoma. The initial imaging and pathological frozen section reports were tended to AML. It was diagnosed as cavernous hemangioma by surgeons during the operation for the tumor tissue was loose, spongy and containing a large amount of blood exudation. But it was finally diagnosed as retroperitoneal hamartoma by paraffin section and immunohistochemistry. Here, we also reviewed the literature of retroperitoneal hamartoma.

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Case presentation

A 20-year-old male patient was admitted to the hospital with acute persistent pain in the right lower quadrant of abdomen in April 2020. Laboratory results showed leukocytes: 21.09×10^{-9}/L and tumor markers were in normal range. In addition to the discovery of appendicitis, the full abdominal CT scan also found a large soft tissue mass with irregular shape located in the retroperitoneum and below the right kidney but did not cross the midline. The mass size was about $51 \times 45 \times 100$ mm^3 (length × width × height), and present as heterogeneous soft tissue lesion. The non-enhanced CT findings presented the spotty suspicious fatty particles in the mass, and no enhancement was observed after injection of contrast agent. The mass was poorly demarcated from the right psoas major muscle and the posterior erector spine muscle, and the right kidney was displaced upward by compression (Fig. 1A–D).

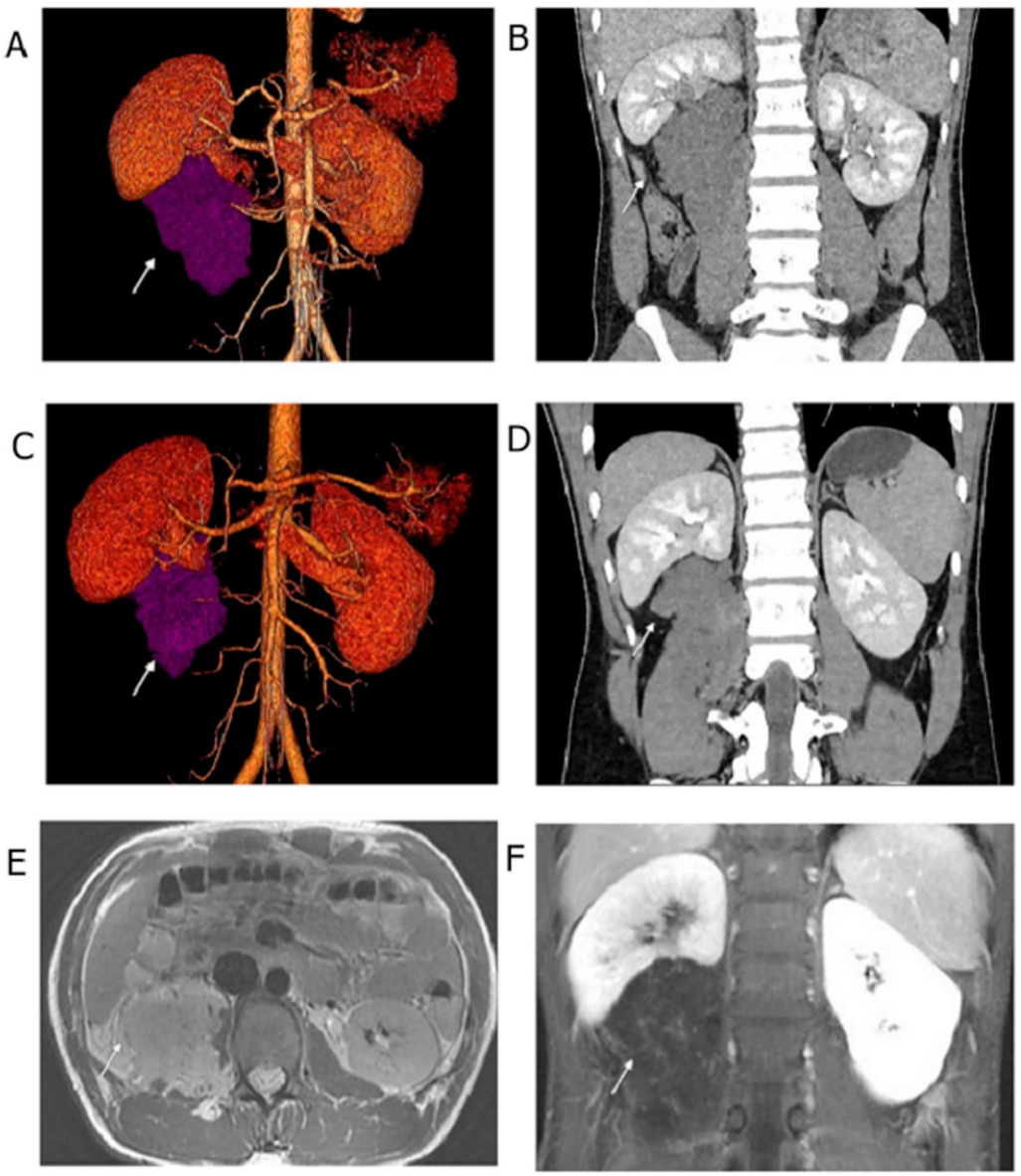

Fig. 1 **A**–**D** A large soft tissue dense mass (white arrow) was seen below the right kidney of the retroperitoneum on CT images, with an ill-defined boundary with the psoas major muscle. No definite enhancement was observed after iodine contrast injection. **E** MRI T1WI showed hyperintense signal mass with ill-defined boundary with psoas major muscle and flowing void effect was revealed in the tumor. **F** After gadolinium injection, cluster and speckled enhanced focal shadows were observed in the mass on coronal MRI

Magnetic resonance imaging (MRI) revealed that there were fat signals and flowing void effect in the tumor, and a large number of progressive enhanced, linear and spotty vascular signals could be seen in the tumor (Fig. 1E, F).

Laparoscopic surgery revealed that the mass was located in the right retroperitoneum and below the right kidney. The main body of the tumor was fused with the right psoas major muscle and its boundary was not clear. No invasion of adjacent blood vessels, right kidney or ureter was found. The cavernous hemangioma was diagnosed intraoperatively for the tumor tissue was loose, spongy and containing a large amount of blood exudation, and the AML was diagnosed by the pathological frozen section. Due to the fusion of the tumor with the right psoas major muscle, it may need to be cut off for complete resection of tumor, which may damage the sacrococcygeal nerve and affect the postoperative activity function, so the tumor was just partially removed.

The paraffin section showed mature adipocytes, thick-walled blood vessels, and a small amount of smooth muscle under a microscope (Fig. 2A–D). The thick-walled blood vessels which were malformed and tortuous without complete muscular layer, fibrosis and hyalinosis were observed. The arrangement of thick-walled vessels was disordered and showed angiomatous changes. Thrombus and hemorrhage could be seen inside, and no peripheral epithelioid cells were observed. Immunohistochemistry showed positive CD31, CD34, ERG, D2-40, FLI1, S-100, 3% positive for Ki67, and focal positive for SMA, yet the HMB-45 was negative. The final pathological diagnosis was hamartoma.

After 6 months and 22 months of follow-up, the patient was in good condition. CT re-examination showed that the epibiotic right retroperitoneal mass was slighter than before operation with the size of $40 \times 42 \times 72$ mm^3 (length × width × height) and $40 \times 45 \times 81$ mm^3.

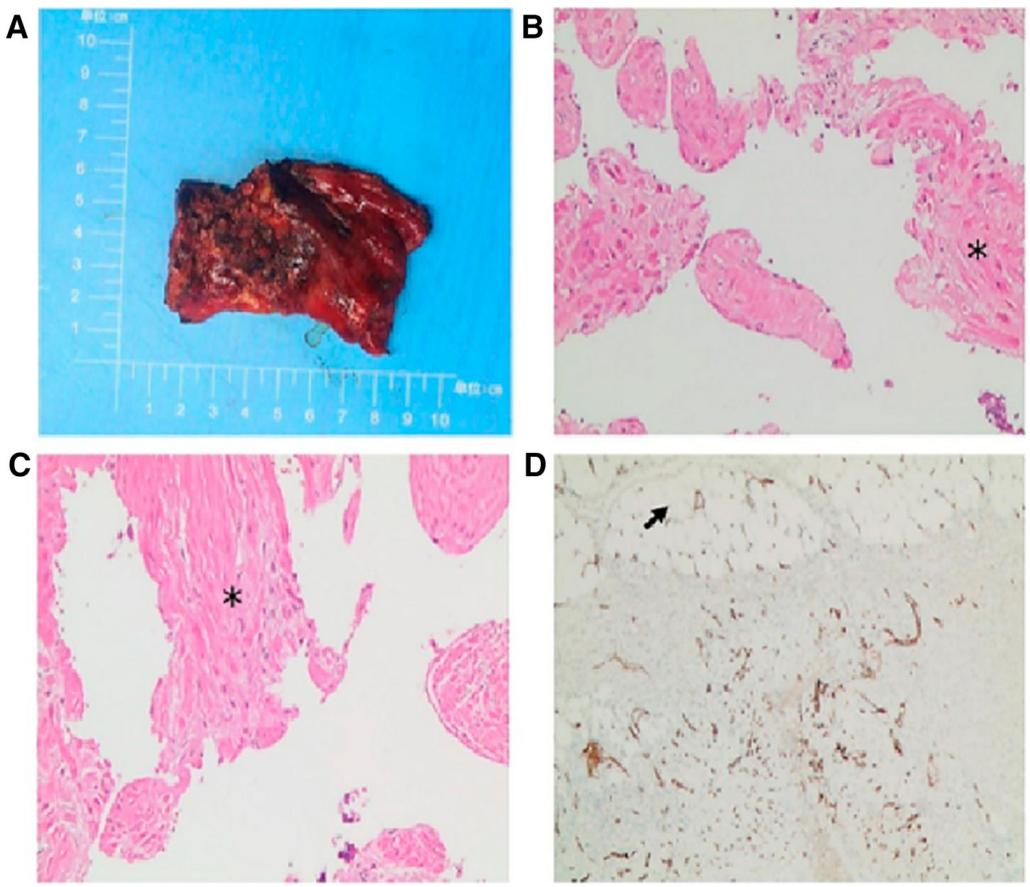

Fig. 2 **A** Gross image of the tumor and right psoas major muscle which is difficult to be isolated after excision showing the size of 90 mm × 50 mm × 12 mm. **B** Section in tumor mass showing hemangioma like changes (asterisk key) (H&E, 20 × 20). **C** Section in tumor showing thick-walled vessels with thrombosis and bleeding (H&E, 20 × 20). **D** Section in tumor showing mature adipocytes (black arrow), and perivascular epithelioid cells were not found (immunohistochemistry for CD34, × 20)

Discussion

Hamartoma is a kind of pathological change between malformation and benign tumor. It is the abnormal differentiation of cells and produces a large number of scattered but mature tissues. Although the mature cells are similar to the surrounding organs and tissues, they are not normal tissues. Generally, intrapulmonary hamartomas are more common than retroperitoneal hamartomas. Only few retroperitoneal hamartomas have been reported in PubMed, Embase, and Scopus databases (Gupta et al. 1980; Hamilton and Mclnerney 1981; Javery and Lee 2010; Lange et al. 1988; Mallory and Spink 1968; Sigdel et al. 2012).

Retroperitoneal hamartoma usually does not cause any symptoms unless it grows to a certain extent and oppresses the surrounding tissue and organs. Hamartoma also contains blood vessels, smooth muscle, fat, and other mature tissue components which is similar to the histological components of AML, causing overlaps in imaging findings and identification difficulty. Although Wang et al. (2002) summarized imaging features of retroperitoneal angiomyolipoma such as linear vessels, aneurysmal dilatation, bridging vessel sign, hematoma, beak sign, and discrete intrarenal fatty tumors, these were not unique to AML. Moreover, in the earlier years, AML had been considered as hamartoma (MMKAB 1999) until perivascular epithelioid cells were found in it, and the differentiation characteristics of AML cells were shown by immunohistochemistry and electron microscopy. Molecular biology showed clonal proliferation, indicating that the lesion was a real tumor rather than a simple hamartoma (Takahashi et al. 2003). It was classified into the PEComa tumor family (Lienert and Nicol 2012).

When the retroperitoneal hamartoma containing abundant blood vessels is enlarged to a certain extent and the boundary with the surrounding tissue is unclear, the CT and MRI imagings just mimic retroperitoneal AML (Lemaitre et al. 1995; Safak et al. 2018) that will increase the difficulty of surgical resection for the high risk of excessive bleeding. As in this case, there is a large amount of blood exudation during the operation. Preoperative interventional embolization was a valuable choice (Javery and Lee 2010; Tseng et al. 2004) in reducing the size of the tumor and the risk of intraoperative bleeding. Only complete resection of the tumor is the radical treatment to avoid possible recurrence in the future.

Preoperative needle biopsy is helpful to judge the nature of retroperitoneal mass, but it should be cautious because there is a risk of bleeding. It is only suitable for low-risk patients, and the patient's physical condition should be comprehensively evaluated before operation. In our case, the blood supply in the mass is abundant, which is not suitable for needle biopsy.

Although the preliminary diagnosis of AML by rapid frozen section is different from that of hamartoma by paraffin section and immunohistochemistry, they are not contradictory. Because the most important difference between them is the finding of perivascular epithelioid cells, while the latter does not contain, the other tissue components could be similar and that is beyond the capability of pathological frozen section. Immunohistochemistry was also inconsistent with AML (Kodzo-Grey 2016). AML has unique immunohistochemistry, which can express human melanin-associated antigen (HMB-45) and smooth muscle actin (SMA) at the same time.

The latest gene detection found that hamartoma is associated with phosphatase and tensin homolog (PTEN) (Javery and Lee 2010), which is called the PTEN hamartoma tumor syndrome and has the potential to develop into various benign and malignant tumors (Tan et al. 2007). PTEN gene on chromosome 10q23.31 encodes a tumor suppressor protein. It regulates the phosphoinositide 3 kinase (PI3K) pathway, which is involved in cell cycle regulation, angiogenesis, cell growth, and proliferation. It was found and isolated in 1997 (Li and Hong 1997; Li et al. 1997). PTEN is now found to be associated with two common diseases: Bannayan–Riley–Ruvalcaba syndrome and Cowden syndrome.

Conclusions

In conclusion, retroperitoneal hamartoma is exceeding rare difficult to differentiate from angiomyolipoma, in which perivascular epithelial cells could be found on pathological paraffin sections.

Abbreviations

AML: Angiomyolipoma; CT: Computed tomography; MRI: Magnetic resonance imaging; PTEN: Phosphatase and tensin homolog; PEComas: Perivascular epithelioid tumors; HMB-45: Human melanin-associated antigen-45; SMA: Smooth muscle actin.

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Author contributions

LH was involved in drafting the manuscript. JJ, WZ, SP, and HP were involved in acquisition of data and preparing the figures. LH and JC designed the manuscript. WZ and JT were involved in analysis and interpretation of data. TL and HL were involved in review and revise the manuscript. All authors have read and approved the final manuscript.

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Availability of data and materials

Some or all data, models, or code generated or used during the study are available from the corresponding author by request (HL, Email: lhy15@mails.jlu.edu.cn).

Declarations

Ethics approval and consent to participate
This study was approved by the ethics committee of Guangdong Hospital of Traditional Chinese Medicine.

Consent for publication
Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Competing interests
The authors declare that they have no competing interests.

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