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Case Report

Epithelioid angiomyolipoma with vascular invasion: An aggressive presentation of an unusual AML variant ☆

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ABSTRACT

Renal angiomyolipoma (AML) is a typically benign renal tumor that is divided into 2 classes, the classical variant and the more aggressive epithelioid variant. It is extremely rare for an AML to exhibit aggressive features such as vascular invasion. We present the case of a 36-year-old female who presented with right lower quadrant pain for 9 months and was found to have an AML with tumor extension into the renal vein and the IVC. Diagnosis was confirmed with histopathology and the patient was treated with a total nephrectomy. The epithelioid subtype of AML is a rare variant that should be considered in the differential of a renal mass with vascular invasion.

Introduction

Angiomyolipoma (AML), though rare, is the most common mesenchymal tumor of the kidney and belongs to a larger group of neoplasms defined as perivascular epithelioid cell tumors (PEComas). PEComas are a rare group of neoplasms which include renal angiomyolipoma, lymphangioleiomyomatosis (LAM), and clear cell (sugar) tumor of the lung. These tumors are characterized by distinctive cells that are often closely related to vascular structures and usually express melanocytic and smooth muscle markers [2].

AML is typically a benign disease and usually encompasses 3 components: mature adipose tissues, dysmorphic blood vessels, and smooth muscle fibers [3,4]. AML is characterized into 2 main subtypes: the classical subtype, as previously described, and the more aggressive epithelioid variant. Epithelioid angiomyolipoma (EAML) is predominantly composed of sheets of epithelioid cells that can have a high degree of cytoplasmic pleomorphism and atypia [5–7]. This histological

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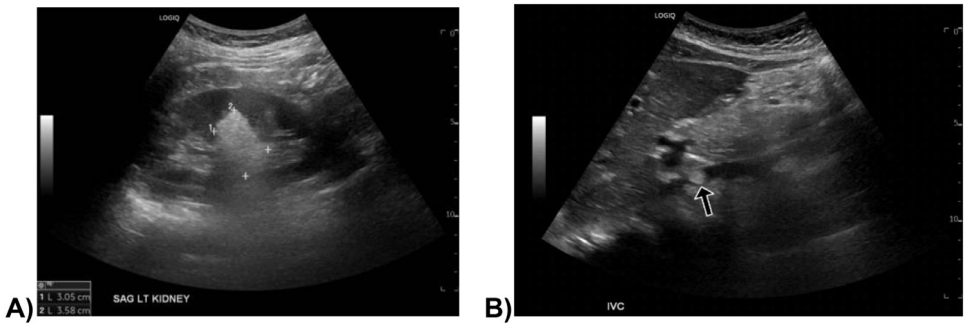

Fig. 1 – Greyscale images of the IVC (A) and left kidney (B). Fig. 1A demonstrates hyperechoic lesion within the IVC (arrow) at the branch point of the left renal vein and IVC. Fig. 1B demonstrates a hyperechoic mass within the left kidney.

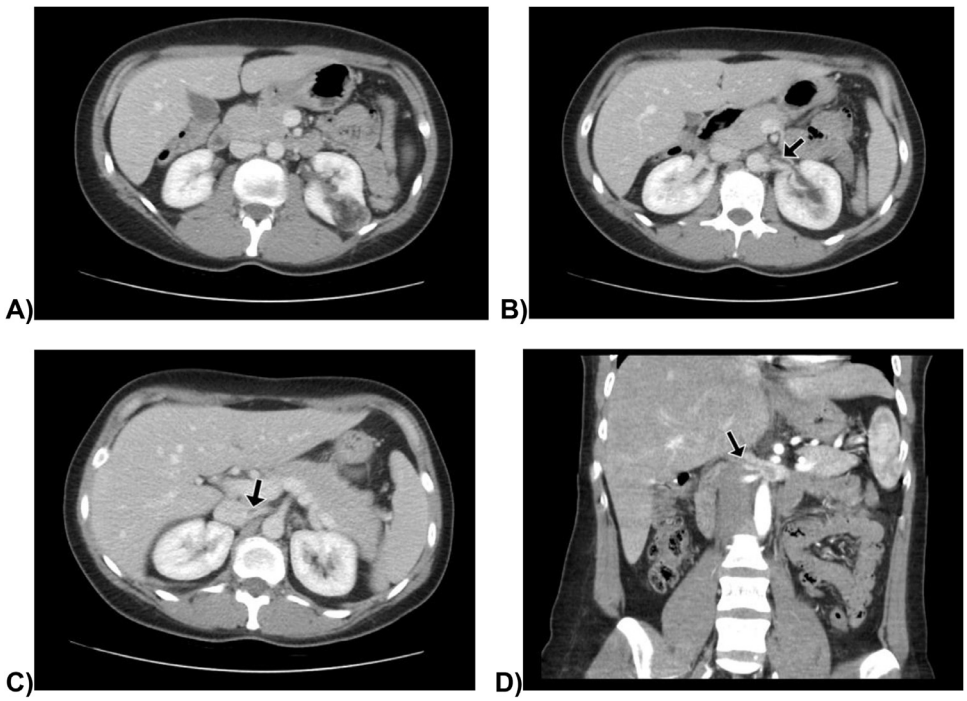

Fig. 2 – Axial and coronal abdominal CT scan images with IV contrast. Axial (A, B, C) images show a heterogenous fat containing mass extending from the renal cortex (A) into the left renal vein (B) and up to the IVC (C, D) (arrow). There is extension of tumor within the left renal vein on coronal reformat (D).

presentation of EAML differentiates it from classical AML, which is solely composed of dysmorphic blood vessels, smooth muscle, and adipose tissue. EAMLs often have similar features to malignant renal cell tumors such as renal cell carcinoma [5,8].

It is uncommon for an AML to invade into the renal vasculature given its typically indolent nature [1]. In this case study we present a 38-year-old female with an epithelioid angiomyolipoma invading into the renal vein and extending to the inferior vena cava.

Case Report

A 36-year-old female with no significant past medical history presented with complaints of intermittent right lower quadrant pain for approximately 9 months. A complete abdominal ultrasound was performed, which revealed a mass in the left kidney (Fig. 1A). The mass appeared largely hyperechoic and measured approximately 3.6×3.1×3.9 cm. Additionally, within the inferior vena cava (IVC), there was a mobile echogenic filling defect initially suspected to be a thrombus (Fig. 1B).

A follow-up computed tomography (CT) scan of the abdomen and pelvis, with and without contrast was performed. The CT revealed a mass originating from the mid left kidney (Fig. 2A). The mass measured 4.6×2.8×3.1 cm and demonstrated predominant internal fat density. This fat density was observed to extend directly from this lesion into the left renal vein (Fig. 2B) and across the midline to the inferior vena cava (Fig. 2C and D).

A subsequent magnetic resonance imaging (MRI) scan of the abdomen, with and without contrast, was conducted (Fig. 3). The MRI demonstrates the presence of the renal mass

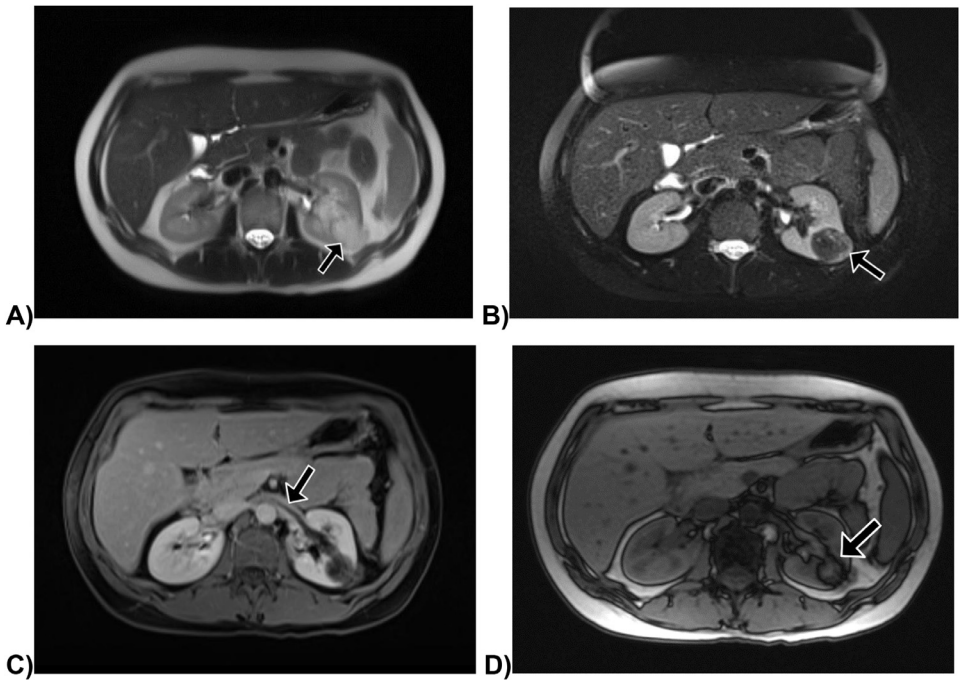

Fig. 3 – MRI axial images of the abdomen. Hyperintense T2 (A) left renal mass (arrow) which appears intermediate on T2 fat saturated image (arrow) (B). Axial LAVA/fat saturated T1 (C) shows a renal mass with invasion of the renal vein (arrow). Out of phase T1 weighted image showing India ink artifact surrounding the fat-containing mass invading into the left renal vein (arrow) (D).

with invasion of the renal vein. The mass demonstrated high signal intensity on both T1 and T2 weighted sequences with loss of signal on fat saturated images, indicative of internal fat composition, a typical imaging appearance of renal AML. As seen on the prior CT and ultrasound, there was fat signal extending into the left renal vein, reaching the margin of the inferior vena cava.

The patient then underwent a left total nephrectomy with tumor thrombectomy. During surgery, a tumor extending from the left renal vein orifice to the IVC was identified.

Histologic examination of the left kidney mass demonstrated an admixture of thick-walled blood vessels, smooth muscle and adipose tissue (Fig. 4). The smooth muscle cells demonstrated a round to oval architecture with prominent nuclei consistent with epithelioid morphology. Immunohistochemical staining revealed positive cathepsin-K, smooth muscle actin (SMA) and Mel C, supporting the diagnosis of epithelioid variant of AML (Fig. 5)

Discussion

EAML is a rare subtype of AML that exhibits more aggressive features than the classical subtype [9]. Renal AMLs can develop sporadically or be associated with other conditions, most commonly tuberous sclerosis [10]. AMLs are often found incidentally, and typical imaging appearances have been thoroughly described in the literature. These appearances often depend on the fat content, which may vary with the epithelioid variant. This patient had a significant fat component within the EAML, which aided the diagnosis prior to the pathology report, though this is not always the case.

The classic ultrasound appearance of AMLs is hyperechoic with posterior acoustic shadowing. On CT, AMLs are often well circumscribed and typically measure -10 HU or less. Intralesional necrosis on CT is rare but is rather typical for epithelioid variants. Necrosis was not evident on imaging in this case. AMLs have a variety of appearances on MRI depending on the sequence used. A typical fat rich AML would appear T1-hyperintense without fat suppression and T1-hypointense with fat saturation. The India ink artifact seen in out-of-phase imaging at the junction of the fat in the AML cells and the water in the renal parenchyma results in sharp delineation of these lesions on this sequence. This can be helpful when AMLs have low fat content or with very small lesions. If there is no fat in the lesion, this artifact will not be present [11]. Due to the EAML often having low fat content, many of the classic radiographic features of AML are sometimes absent and histology is relied on for an accurate diagnosis.

AML usually encompasses 3 components: mature adipose tissues, dysmorphic blood vessels, and smooth muscle fibers. Epithelioid angiomyolipoma (EAML) often consists predominantly of sheets of epithelioid cells that can have a high degree of cytoplasmic pleomorphism and atypia. Often stains for Cathepsin-K, smooth muscle markers and melanocytic markers are positive, as in our case [12].

Given the nature of vascular involvement and the risk of thromboembolism, radical nephrectomy is typically performed [13]. Alternative treatment methods such as emboliza-

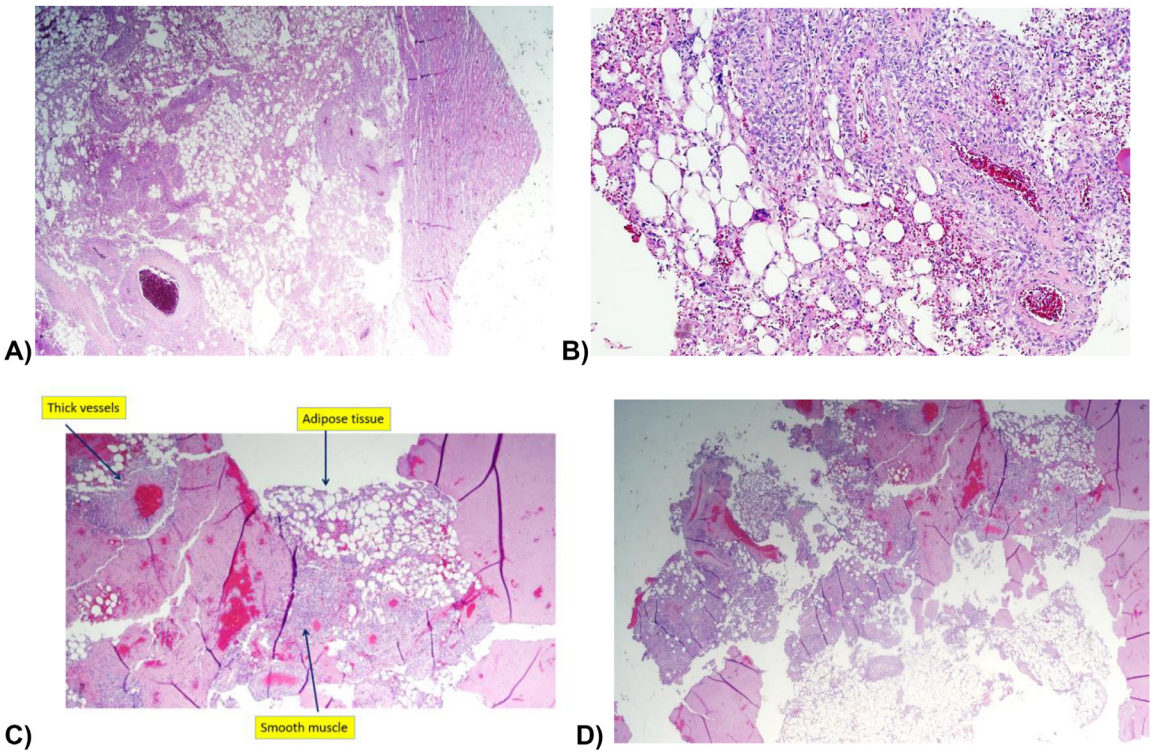

Fig. 4 – Histology slides taken from excised renal tumor. (A) Low power (20×) showing normal renal parenchyma at 1 edge (right), underneath is tumor composed of an admixture of thick-walled blood vessels, smooth muscle and adipose tissue. The smooth muscle cells exhibit epithelioid structure as they are oval and contain prominent nuclei. (B) High power (40×) showing adipose tissue, thickened vessels and epithelioid stromal cells. (C) Labeled thick wall blood vessel, adipose tissue and smooth muscle. (D) Low power (2×) section from tumor thrombus consisting of thickened blood vessels, smooth muscle and adipose tissue.

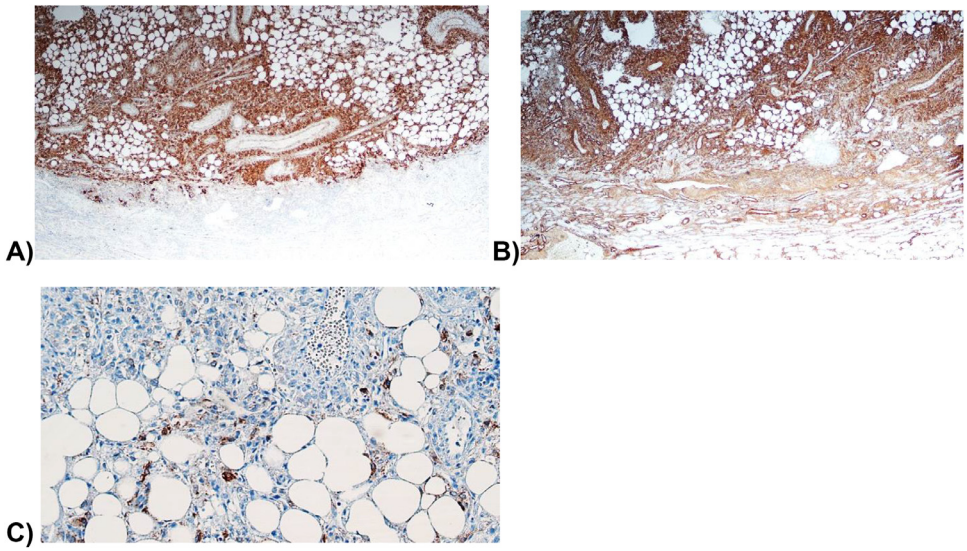

Fig. 5 – Histology slides taken from the excised renal tumor. (A) Cathepsin-K immunohistochemical stain positive in tumor cells. (B) SMA immunohistochemical stain highlighting smooth muscles. (C) Mel-C immunohistochemical stain focally positive in tumor cells.

tion have been recorded in the existing literature [14]. There is currently no long-term data on the surgical prognosis and outcomes. Our patient at the time of this paper is in good health and recovered without complications.

Conclusion

This case report conveys the rarity of vascular invasion with AMLs and highlights the subtle differences between classical and the more aggressive epithelioid AML variant. Three modalities of imaging as well as histology are provided and discussed. This report provides further information to existing literature of vascular invasive epithelioid angiomyolipoma to assist practitioners with future research and clinical endeavors.

Patient consent

The patient who is the subject of this case report was contacted via telephone and informed of the case report. The patient agreed to the case report being written using nonidentifying information such as patient history, imaging and pathology. A verbal consent with witness was taken over the phone and uploaded to the patient's electronic medical record.

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