Renal angiomyolipoma with inferior vena cava and right atrium extension in patient with tuberous sclerosis complex: a rare case report and literature review.

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Abstract

Angiomyolipoma(AML) is one of the most common benign renal tumors. Classical AML is benign, but it can be locally invasive, extending into perirenal fat, or in rare cases, invading the renal collecting system, renal vein or inferior vena cava and right atrium. About 10% of patients clinically diagnosed with renal AML have tuberous sclerosis complex. Tuberous sclerosis complex can be diagnosed by genetic diagnosis or clinical manifestation. We report a rare case of a 35-year-old woman who was diagnosed with tuberous sclerosis complex caused by TSC2 gene mutation, which was characterized by multiple angiomyolipoma in the right kidney and extended growth to the inferior vena cava and right atrium. Intracardiac extension is often observed in the malignant tumor and only seldom seen in benign tumors. Our case reminds the rare possibility of intracardiac extension in renal AML, which may potentially result in fatal complications if not appropriately managed.

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Abstract

Angiomyolipoma(AML) is one of the most common benign renal tumors. Classical AML is benign, but it can be locally invasive, extending into perirenal fat, or in rare cases, invading the renal collecting system, renal vein or inferior vena cava and right atrium. About 10% of patients clinically diagnosed with renal AML have tuberous sclerosis complex. Tuberous sclerosis complex can be diagnosed by genetic diagnosis or clinical manifestation. We report a rare case of a 35-year-old woman who was diagnosed with tuberous sclerosis complex caused by TSC2 gene mutation, which was characterized by multiple angiomyolipoma in the right kidney and extended growth to the inferior vena cava and right atrium. Intracardiac extension is often observed in the malignant tumor and only seldom seen in benign tumors. Our case reminds the rare possibility of intracardiac extension in renal AML, which may potentially result in fatal complications if not appropriately managed.

KEYWORDS

renal angiomyolipoma, inferior vena cava, right atrium, tuberous sclerosis complex, echocardiography

CASE

A 35-year-old woman presented to our hospital with chest tightness and shortness of breath for a week after exercise. Her medical history and physical examination were unremarkable. The blood test and electrocardiography(ECG) were negative. Transthoracic echocardiography(TTE) revealed a long strip mass in the inferior vena cava(IVC), which extended to right atrium(Video1,Figure1). The mass was 12.3×2.4cm in size with clear boundary, large range of activity and there was no clear pedicle structure. Abdominal ultrasonography revealed several hyperechoic masses in the right kidney and the larger one was about 4cm in size, with regular shape and clear boundary, and some of them were close to the renal vein(Figure2). There was no obvious thrombus in the deep veins and gynecological ultrasound showed no obvious abnormality in uterus and double adnexal area. Abdominal computed tomography(CT) confirmed a larger tumor involving the right kidney with invasion of the right renal vein and extension into the IVC and right atrium(Figure3). The patient finally underwent radical nephrectomy and extraction of tumors in the inferior vena cava and right atrium. Postoperative pathology confirmed that the tumors in the right kidney, inferior vena cava and right atrium were angiomyolipoma. The gene detection is somatic TSC2 gene mutation. Finally, the patient was diagnosed with tuberous sclerosis complex caused by TSC2 gene mutation, which was characterized by multiple angiomyolipoma in the right kidney and extended growth to the inferior vena cava and right atrium.

DISCUSSION

Angiomyolipoma is one of the most common benign renal tumors and accounts for 0.3-3% of all renal masses.[1] It contains fat, blood vessels, and smooth muscles in different proportions.[2] It mainly includes two histology types: classical and epithelioid [3]. Classical AML is benign, but it can be locally invasive, extending into perirenal fat, or in rare cases, invading the renal collecting system, renal vein or inferior vena cava and right atrium. Only a few cases of giant AML involving the IVC have been reported in the available literature.[4-5]This might be attributed to multifocal genesis of tumor, instead of direct vascular involvement.[6]

About 10% of patients clinically diagnosed with renal AML have tuberous sclerosis complex[7]. Tuberous sclerosis complex(TSC) is an autosomal dominant disease with an estimated prevalence of 1 in 12000 with a birth rate as high as 1 in 6000.[8]The molecular genetics of TSC have been well characterized with mutations

to TSC1 found at chromosomal location 9q34 and TSC2 found at 16p13.3 identified. And the TSC2-deficient smooth-muscle cells have higher migration potential than normal cells in vitro.[9] The manifestations are diverse and individual differences are large, mainly involving the brain, skin, heart, lungs, kidneys, as well as seizures, tuberous sclerosis-related neuropsychiatric disorders and so on. It can be diagnosed by clinical manifestation or genetic detection. This patient belongs to this rare case, and tuberous sclerosis was diagnosed by genetic diagnosis.

Most AML patients are asymptomatic, and the tumor is incidentally detected on ultrasonographic or radiological imaging.[1] The diagnosis depends on showing the fat composition in the tumor. On ultrasound (US), AMLs are almost always hyperechoic compared to renal parenchyma due to the presence of macroscopic fat. Areas within a lesion of -15HU or less are generally considered diagnostic of macroscopic fat on computed tomography(CT).[10]Magnetic resonance imaging(MRI) is essentially equivalent in accuracy to CT in the diagnosis of AML and can be particularly helpful in diagnosing fat-poor AMLs.[11]Masses that appear T1 hyperintense without fat-suppression and T1 hypointense after frequency selective fat suppression are consistent with AML [12].

Tumor, thrombosis and intravenous leiomyomatosis(IVL) should be considered in this case. The tumor included primary cardiac tumor and cardiac metastatic tumor. Among them, cardiac metastatic tumor can grow through blood metastasis, direct mediastinal invasion, or the tumor grows into the vena cava and extends into the right atrium[13]. We did not find thrombosis by deep venous ultrasound, and the patients had no risk factors of intracardiac thrombosis such as underlying heart disease, and laboratory tests did not support thrombosis. The patient had no abnormal gynecological ultrasound and no history of hysteromyomectomy, so IVL was basically excluded. The incidence of cardiac metastases is about 20 times higher than that of primary cardiac tumors [14]. Ultrasound found that the boundary of the tumor in the inferior vena cava and the right atrium is very clear, and there is no clear adhesion with the atrium wall. The range of activity of the tumor is very large and no clear pedicle structure is observed. Therefore, we consider that the tumor grows from the inferior vena cava to the right atrium. So it is likely to consider cardiac metastatic tumor. At the same time, we found multiple angiomyolipoma in the right kidney, about 3-4 cm in size, some of which were close to the renal vein. It was highly suspected that the mass in the inferior vena cava and right atrium originated from the angiomyolipoma in the right kidney. CT also supported the ultrasound conjecture.

The enlightenment from this patient is: (1) The tumor of the heart does not necessarily come from the heart, and the possibility of systemic origin should be considered when tumor extends into the right atrium and inferior vena cava at the same time. (2) When there are multiple angiomyolipoma in the kidney and the size is large, extension to the inferior vena cava and cardiac may occur, and at the same time, we should pay attention to whether it is complicated with tuberous sclerosis complex. (3)The correct diagnosis of TSC and identification of AML intracardiac extension are of great significance for its treatment and prognosis.

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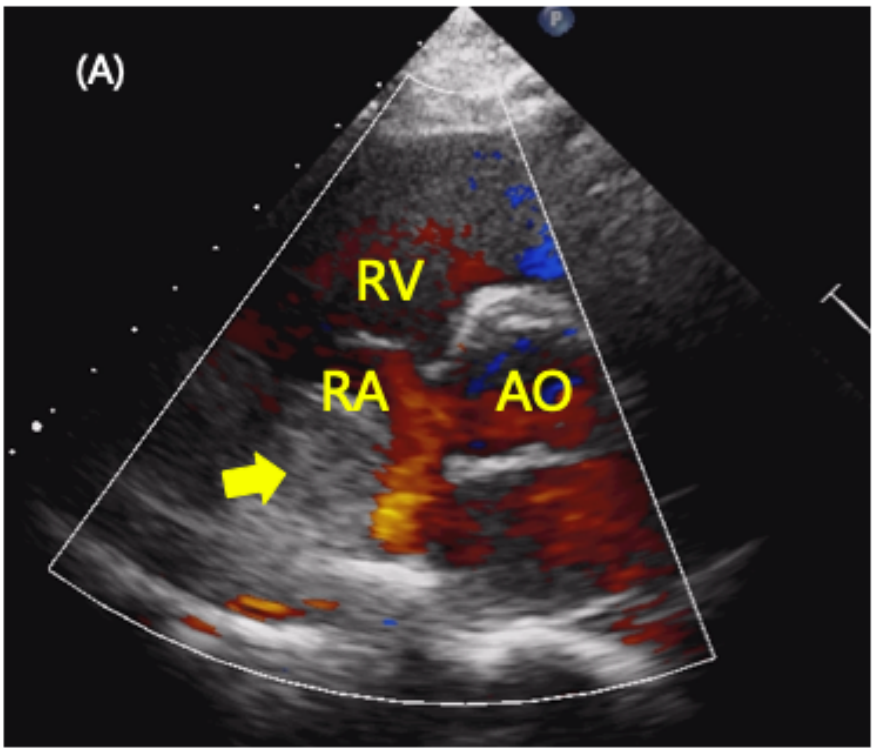
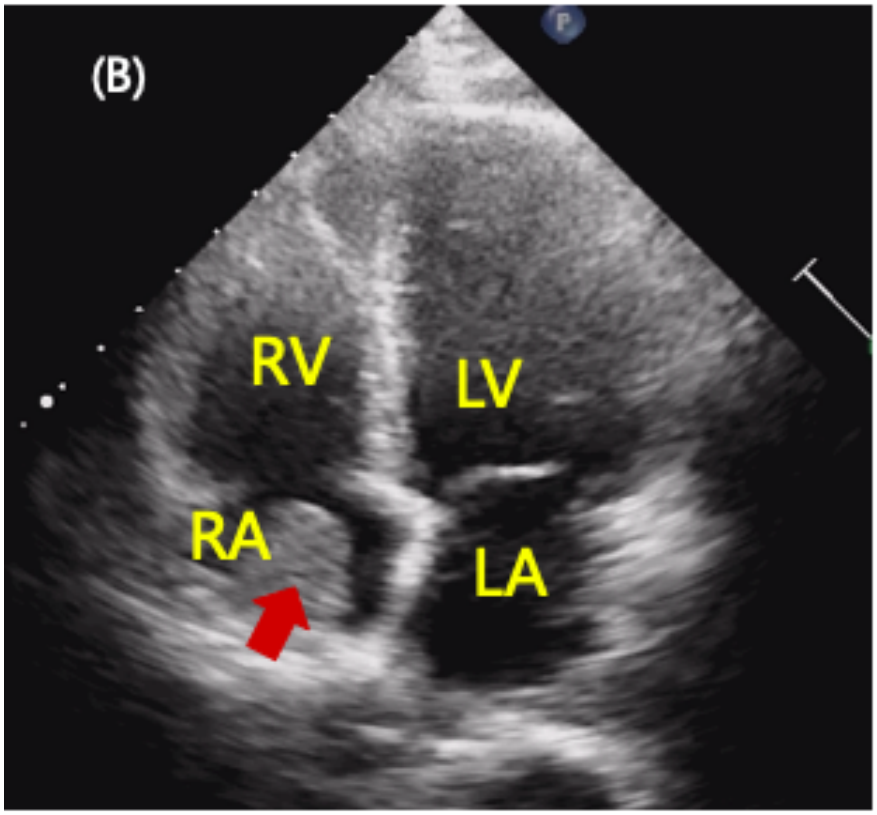
Figure and Video 1 Transthoracic echocardiography(TTE) revealed a long strip mass in the inferior vena cava(IVC), which extended to right atrium. The parasternal short axis view of the aortic valve(A) The apical four-chamber view(B). The subcostal view(Video).RA, right atrium. RV, right ventricle. LA, left atrium. LV, left ventricle. AO, aorta. Red arrow shows mass.

Figure 2 Abdominal ultrasonography revealed several hyperechoic masses in the right kidney and the larger one was about 4cm in size, and some of them were close to the renal vein. RRV, right renal vein. Red arrows show masses.

Figure 3 Abdominal computed tomography(CT) confirmed a larger tumor involving the right kidney with invasion of the right renal vein and extension into the IVC and right atrium. RK, right kidney. IVC, inferior vena cava. Red arrows and stars show masses.





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