

# Renal angiomyolipoma presenting as acute abdomen in a previously undiagnosed patient of tuberous sclerosis

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## DESCRIPTION

A 28-year-old man, with no previous comorbidities, presented to our emergency clinic with pain abdomen. The pain was predominantly central, gradually progressive and associated with abdominal distention. The blood pressure was 80/50 mm Hg with pulse rate of 120 beats per minute. General examination revealed presence of periungual fibroma, adenoma sebaceum, multiple skin tags and severe pallor (figure 1A). Abdominal examination revealed bilateral tender flank masses which were ballotable. Laboratory parameters revealed haemoglobin of 62 g/L, haematocrit of 25%, total count of  $13.2 \times 10^9/L$ , platelet count of  $345 \times 10^9/L$  and serum creatinine of 1.9 mg/dL with an estimated glomerular filtration rate (eGFR) of 62 mL/min. Abdominal ultrasonography showed bilateral kidneys replaced by heterogeneous hyper-echoic fatty lesion with no residual renal parenchyma with likely colour flow within left kidney, possibly suggestive of pseudo aneurysm. CT with angiography revealed bilateral giant renal angiomyolipoma (left kidney measuring  $14 \times 13.7 \times 23.5$  cm and right kidney  $9.3 \times 9.6 \times 16.3$  cm) with contained rupture of left intratumoural pseudoaneurysm with left perinephric haematoma (figure 1B). Based on the cutaneous stigmata and bilateral renal angiomyolipoma, diagnosis of tuberous sclerosis was made.<sup>1</sup> Digital subtraction angiography (DSA) revealed multiple pseudoaneurysms in the upper and middle pole of left kidney which were embolised with gelfoam and coils. Post-DSA, there was acute deterioration of kidney function with serum creatinine

rising up to 5.5 mg/dL (eGFR 20 mL/min) and drop in urine output. The urine routine examination showed 1+ albumin in dipstick and 1–2 granular casts/high power field (HPF). He received two sessions of haemodialysis and was transfused with two packs of packed red blood cells to maintain haemodynamic instability. The kidney functions improved over the next 7 days and serum creatinine settled at 1.5 mg/dL (eGFR 78 mL/min) 1-month postdischarge. The patient had no history of seizures/intellectual disability but a screening brain imaging revealed bilateral subependymal nodules and cortical tubers. Funduscopy did not reveal retinal tubers and echocardiogram was normal. The patient was started on mammalian targets of rapamycin (mTOR) inhibitors everolimus 5 mg/day and was targeted to achieve a trough concentration of 5–15 ng/mL.

Renal angiomyolipoma is benign fat containing slow growing tumours. In tuberous sclerosis complex (TSC) syndrome, angiomyolipomas are usually bilateral, asymptomatic, rapidly growing and tend to present with life-threatening haemorrhages. Life-saving interventions like partial/total nephrectomy or embolisation are required in most of these patients.<sup>2</sup> Angioembolisation remains one of the commonly used modalities in emergencies in approximately 50% of the patients. The kidney function decline in Angiomyolipoma (AML) patients is commonly due to focal segmental glomerulosclerosis due to hyperfiltration of the viable nephrons, invasion of kidney tissue by tumours and cysts, surgeries in addition to repeated kidney injuries during embolisation procedures. Because AML has a highly vascular tissue, repeated embolisation procedures may lead to rapid kidney function decline through ischaemic acute tubular necrosis.<sup>3</sup> Acute kidney injury (AKI) post embolisation can be due to contrast related AKI, vascular compromise of nephrons due to coiling of vessels, inflammation caused due to embolisation agents, formation of haematoma, perioperative haemodynamic instability and atheroembolic phenomenon.

Prevention of kidney function decline plays an important role in management of these patients as renal AMLs are one of the important cause of morbidity and mortality in TSC patients. High clinical suspicion should be kept in patients presenting as acute abdomen in known or previously unknown patients of renal angiomyolipoma (RAML). Selective arterial embolisation, as done in our patient, remains the first-line therapy but nephrectomy



**Figure 1** (A) Photograph of patient showing left hand thumb with periungual fibroma (B) CT of the abdomen (coronal section) showing heterogeneously enhancing lesion showing areas of fat involving the entire bilateral enlarged kidneys with left kidney measuring  $14 \times 13.7 \times 23.5$  cm and right kidney measuring  $9.3 \times 9.6 \times 16.3$  cm.



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should be considered in size >4 cm.<sup>4</sup> We report a case of bilateral giant renal angiomyolipoma presenting as acute abdomen in a previously undiagnosed patient of tuberous sclerosis.

### Learning points

- ▶ Ruptured renal AML is a rare but serious complication in tuberous sclerosis patients and selective arterial embolisation should be performed immediately.
- ▶ Treatment with mTOR inhibitors offers patients a noninvasive pharmacotherapeutic option to retard the progression of RAML.
- ▶ Renal-related disease is the most common cause of death tuberous sclerosis complex related deaths in adults.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

### REFERENCES

- 1 Samuelli S, Abraham K, Dressler A, et al DA, et al. Tuberous sclerosis complex: new criteria for diagnostic work-up and management. *Wien Klin Wochenschr* 2015;127:619–30.
- 2 Lee S-Y, Hsu H-H, Chen Y-C, et al. Embolization of renal angiomyolipomas: short-term and long-term outcomes, complications, and tumor shrinkage. *Cardiovasc Intervent Radiol* 2009;32:1171–8.
- 3 Seyam R, Khudair WA, Kattan SA, et al. The impact of renal angiomyolipoma on estimated glomerular filtration rate in patients with tuberous sclerosis complex. *Ann Saudi Med*. In Press 2016;36:356–63.
- 4 Lin L, Li X, Guan H, et al. Renal function, complications, and outcomes of a reduction in tumor size after transarterial embolization for renal angiomyolipomas: a meta-analysis. *J Int Med Res* 2019;47:1417–28.

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