Kidney International (2012) **82,** 613–614; doi:10.1038/ki.2012.228

The Case | **A woman with bilateral flank pain**

Heng-Pin Chiang[1], Hung-Tien Kuo[1,2], Jer-Ming Chang[1,2,3] and Hung-Chun Chen[1,2]

[1]Division of Nephrology, Department of Internal Medicine, Kaohsiung Medical University Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan; [2]Department of Renal Care, Kaohsiung Medical University, Kaohsiung, Taiwan and [3]Department of Internal Medicine, Kaohsiung Municipal Hsiao-Kang Hospital, Kaohsiung Medical University, Kaohsiung, Taiwan

Correspondence: *Hung-Tien Kuo, Division of Nephrology, Department of Internal Medicine, Kaohsiung Medical University Hospital, Kaohsiung Medical University, No.100, Tzyou, 1st Road, Kaohsiung 807, Taiwan. E-mail: hutiku@kmu.edu.tw*

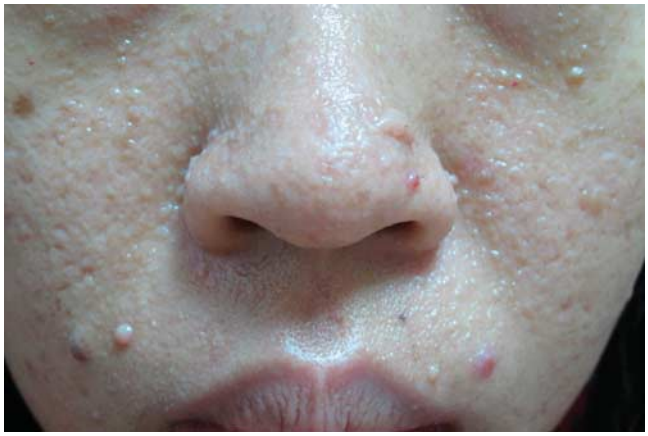

Figure 1 | Multiple skin-colored facial angiofibromas.

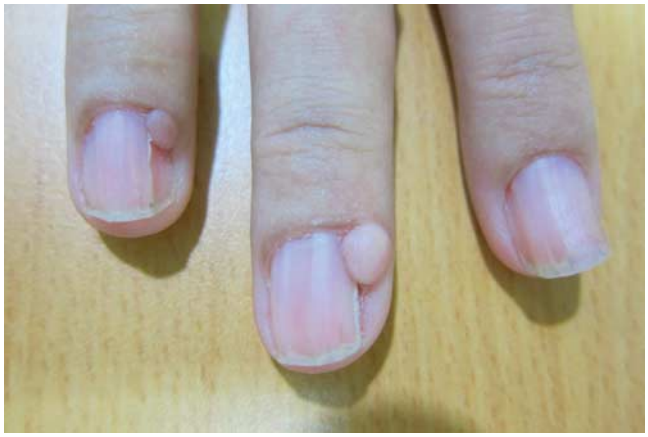

Figure 2 | Skin-colored periungual fibromas.

A 32-year-old woman with seizure history, controlled with phenobarbital since childhood, presented to the emergency department because of acute onset of bilateral flank pain without any recent trauma. There were skin-colored facial angiofibromas (Figure 1) and periungual fibromas (Figure 2). Physical examination was otherwise unremarkable, except for her pale conjunctiva and moderate bilateral flank tenderness, especially on the left side. Her body temperature was 37 °C (98.6 °F), and the laboratory investigation revealed an acute drop of hemoglobin level from 9.8 to 7.7 g/dl, without evidence of gastrointestinal bleeding. Serum creatinine concentration was 0.71 mg/dl, and blood urea nitrogen was 4 mg/dl. Urinalysis showed no hematuria, proteinuria, or pyuria. Abdominal radiograph revealed huge abdominal radiolucent masses.

What is the diagnosis and likely cause of the flank pain?

SEE NEXT PAGE FOR ANSWERS

The Diagnosis | Tuberous sclerosis complex with bleeding renal angiomyolipoma

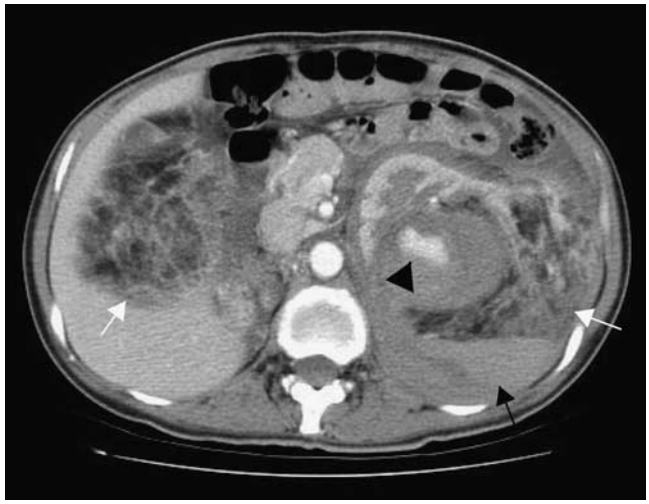

Figure 3 | Post-contrast abdominal computed tomography demonstrated huge fat-containing masses arising from both kidneys (white arrows), suggesting bilateral renal angiomyolipomas. Contrast extravasation (black arrowhead) noted from one of the left renal angiomyolipomas with retroperitoneal hemorrhage (black arrow).

Abdominal computed tomography (CT) scan showed multiple, huge fat-containing tumors in bilateral kidneys, with active bleeding from the left renal tumor and retroperitoneal hematoma (Figure 3).

Angiomyolipoma (AML) is by far the most common fat-containing tumor to arise from the kidney. The combination of seizure history, facial angiofibromas, periungual fibromas, and bilateral renal AMLs points to the diagnosis of tuberous sclerosis complex (TSC).[1]

TSC, an autosomal dominant inherited disorder characterized by widespread hamartomas, affects 1.1 million people worldwide. Its incidence is about 1 per 6000 individuals. This complex is caused by mutations in the *TSC1* or *TSC2* gene, which encode hamartin and tuberin, respectively. These genes control cell growth and division. Therefore, mutations of either of these genes predispose the patients to tumor formation in a variety of organs. The mutations can be sporadic (similar to our case) or inherited. Typical examples include facial angiofibroma, ungual fibroma, cortical tubers, or subependymal nodules in the brain, which may cause seizure, renal angiomyolipoma, pulmonary lymphangioleiomyomatosis, and cardiac rhabdomyoma, as well as many other less common features. As there are no pathognomonic features of TSC, diagnosis is based on a combination of symptoms and signs attributing from the multi-systemic tumor formation, as well as genetic testing. Supportive management is the mainstay treatment for TSC, although new treatments using mTORC1 inhibitors have been shown to be effective.[2] Although the prognosis of TSC depends on the numbers and severity of the organs being affected, with appropriate medical support, most patients can have a normal life expectancy.

Renal angiomyolipomas occur in 70 to 80% of patients with TSC. Both CT and magnetic resonance imaging are useful imaging modalities for diagnosing these tumors, and magnetic resonance imaging has the additional advantage of detecting minimal/microscopic fat-containing angiomyolipomas[2] without the risk of radiation from CT. When the AMLs enlarge with time, these lesions invade adjacent normal renal parenchyma, leading to tortuosity and aneurysmal dilatation of the surrounding vessels. The hemorrhage risk increases with large aneurysms, and aneurysm formation seems to be related to tumor size.[3] Transcatheter arterial embolization is frequently used to stop the acute bleeding. Surgical intervention is indicated if hemorrhage is not responsive to embolization or if there is suspicion of malignancy.

In our case, the renal bleeding was successfully treated by transcatheter arterial embolization, and the patient was discharged home with maintenance antiepileptic medications to control her preexisting seizures.

REFERENCES

1. Roach ES, Gomez MR, Northrup H. Tuberous sclerosis complex consensus conference: revised clinical diagnostic criteria. *J Child Neurol* 1998; **13**: 624–628.
2. Dixon BP, Hulbert JC, Bissler JJ. Tuberous sclerosis complex renal disease. *Nephron Exp Nephrol* 2011; **118**: e15–e20.
3. Yamakado K, Tanaka N, Nakagawa T *et al.* Renal angiomyolipoma: relationships between tumor size, aneurysm formation, and rupture. *Radiology* 2002; **225**: 78–82.