

中文題目：結節性硬化症伴腎血管平滑肌脂肪瘤破裂之個案報告

英文題目：A case of tuberous sclerosis complex presented with rupture of renal angiomyolipoma - case report

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## **Introduction**

Tuberous sclerosis complex (TSC) is a rare autosomal dominant genetic disorder characterized by the development of numerous benign tumors that occur in multiple organ systems throughout the lifetime of the affected individuals. Renal angiomyolipomas occur in up to 80% of TSC patients, and chronic kidney disease from increasing tumor burden is the primary cause of TSC-related mortality. A serious and life threatening complication of TSC is intratumoral or retroperitoneal hemorrhage due to renal AML rupture.

## **Case report**

A 48-year-old woman is a case of CKD, bilateral renal angiomyolipomas and HTN. She was followed up at local medical department. Once suffered from sudden onset of abdominal pain for minutes, accompanied with near syncope. Thus she was sent to Cheng Ching H Chong Gang branch where abdominal CT angiography showed bid size renal AML with bleeding causing hemoperitoneum. Due to progression of anemia, she received trans-arterial embolization for hemostasis, and then was referred to Taichung Veterans General Hospital (VGHTC) for further care. There are several typical dermatologic and dental manifestations of tuberous sclerosis complex (TSC) found on physical examination, including facial angiofibromas, fibrous cephalic plaque on forehead and scalp, hypomelanotic macules, Shagreen patches as well as ungal fibromas over extremities. The CT scan showed lesions of lymphangiomyoleiomatosis (LAM) visible in both lungs. Brain MRI disclosed cortical/subcortical tubers and subependymal hamartomas. These clinical features fulfilled the clinical diagnostic criteria, tuberous sclerosis complex is diagnosed. Also, a pathogenic variant in the TSC2 gene was detected by genetic examination.

The patient was discharged under stable condition and is regularly followed up at nephrology OPD in VGHTC. She started mTOR-inhibitors, further followed-up image study for evaluating treatment response would be arranged after a period of mTOR-inhibitor therapy.