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Case Report

Multisystem Imaging Clues Leading to the Diagnosis of Tuberous Sclerosis Complex in Adulthood: A Case Report

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Abstract

Tuberous sclerosis complex (TSC) is a rare autosomal dominant disorder affecting multiple organ systems. While often characterized by neurocutaneous findings, renal angiomyolipomas (AMLs) are a significant source of morbidity. We present the case of a 20-year-old woman with epilepsy who presented with chest pain and was diagnosed with pulmonary thromboembolism. Imaging revealed bilateral renal AMLs, cortical-subcortical tubers, and widespread sclerotic bone foci, suggesting TSC. These were identified through contrast-enhanced thoracoabdominal computed tomography (CT) and brain magnetic resonance imaging. Eight months later, she re-presented with abdominal pain, and follow-up abdominal CT showed retroperitoneal hemorrhage and a hematoma in the left kidney, consistent with AML rupture. Pulmonary embolism as an initial manifestation is rarely reported in TSC. This case underscores the importance of recognizing TSC based on radiological findings in adulthood and highlights the need for close monitoring of AMLs. Radiology plays a key role in diagnosis and management.

Keywords: Tuberous Sclerosis, Angiomyolipoma, Pulmonary Embolism, Radiology, Computed Tomography.

MAIN POINTS

Pulmonary embolism may rarely be the initial manifestation of tuberous sclerosis complex, as demonstrated in this adult case. Multisystem radiologic findings such as renal angiomyolipomas, cortical-subcortical tubers, and sclerotic bone lesions are key clues for diagnosing previously unrecognized tuberous sclerosis complex.

Renal angiomyolipomas associated with tuberous sclerosis complex carry a high risk of rupture, particularly when lesions exceed 4 cm in size. Timely diagnosis of tuberous sclerosis complex through imaging enables early monitoring and intervention to prevent potentially life-threatening complications.

Radiology plays a central role not only in diagnosing tuberous sclerosis complex but also in assessing disease progression and guiding clinical management.

INTRODUCTION

Tuberous sclerosis complex (TSC) is a rare, autosomal dominant genetic disorder that affects multiple organ systems. It is caused by mutations in the TSC1 or TSC2 genes, resulting in the loss of function of hamartin and tuberin proteins, which regulate cellular growth and proliferation. These mutations lead to the development of hamartomatous lesions, particularly in the brain, skin, kidneys, heart, and lungs. The clinical spectrum is broad, most commonly presenting with neurocutaneous manifestations such as epilepsy, intellectual disability, and cutaneous lesions (1). However, renal involvement is also a significant source of morbidity and is most frequently associated with angiomyolipomas (AMLs) (2). While approximately three-quarters of cases are diagnosed in childhood, diagnosis may be delayed until adulthood in some patients (3). Renal

CITATION

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AMLs in patients with TSC typically present bilaterally, are multiple in number, and often develop at a young age. Although they are usually asymptomatic, the risk of rupture and retroperitoneal hemorrhage increases as the tumor size enlarges. This can result in Wunderlich syndrome, which is characterized by spontaneous perirenal hematoma and hypovolemic shock (4). In this case report, we present a young female patient with a known history of epilepsy and intellectual disability who initially presented with pulmonary embolism. Abdominal computed tomography (CT) revealed renal AMLs and additional systemic findings suggestive of a diagnosis of TSC. Eight months later, she was readmitted with a ruptured angiomyolipoma. This case is notable for its unusual initial presentation with pulmonary embolism and delayed diagnosis in adulthood, which are both rarely reported in the literature. The case is evaluated in light of the literature, with an emphasis on diagnostic and radiologic findings.

CASE REPORT

A 20-year-old female patient with a medical history of epilepsy and intellectual disability presented to the emergency department with complaints of decreased oral intake for approximately one week and pleuritic chest pain localized to the left lower costal region for the past 3–4 days. Her overall clinical status was stable; she was alert, oriented, and cooperative. Physical examination findings were unremarkable.

Initial laboratory evaluation did not reveal any gross abnormalities. No significant growth was observed in blood or urine cultures. Transthoracic echocardiography showed no pathological findings. Thoracic and abdominal imaging was systematically performed to evaluate both respiratory symptoms and possible systemic involvement. Thoracic CT angiography revealed a pleural effusion measuring approximately 2 cm in the left hemithorax. Filling defects consistent with pulmonary embolism were observed in the left main pulmonary artery and segmental branches of the lower lobe. Additionally, widespread sclerotic lesions were identified in the bony structures, including the pelvis and vertebrae. A fat-density lesion consistent with a lipoma was noted in segment 6 of the liver. Multiple cystic structures and solid lesions compatible with angiomyolipomas (AMLs) were detected in both kidneys (Figure 1). Recent brain MRI revealed characteristic cortical and subcortical tubers accompanied by subependymal nodules, consistent with the neuroimaging features of tuberous sclerosis complex (Figure 2). These multifocal radiological findings raised a preliminary diagnosis of tuberous sclerosis. The patient was hospitalized with a diagnosis of pulmonary embolism and anticoagulant therapy was initiated. She was discharged in stable condition.

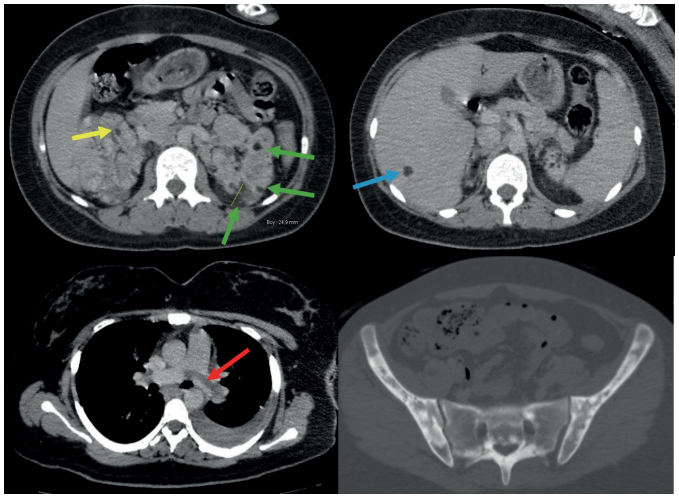

Figure 1. Contrast-enhanced thoracic and abdominal computed tomography (CT) images obtained at the initial presentation of the patient. Multiple fat-density lesions consistent with angiomyolipomas (green arrows), the largest measuring up to 2.5 cm in diameter, are observed in the left kidney. A cortical renal cyst is noted in the right kidney (yellow arrow). In the second image, a hepatic lipoma is seen in the right lobe of the liver (blue arrow). The third image shows filling defects in the main pulmonary artery and left pulmonary artery, consistent with pulmonary thromboembolism (red arrow). In the lower right corner, bone window images reveal widespread sclerotic foci within the pelvic bones.

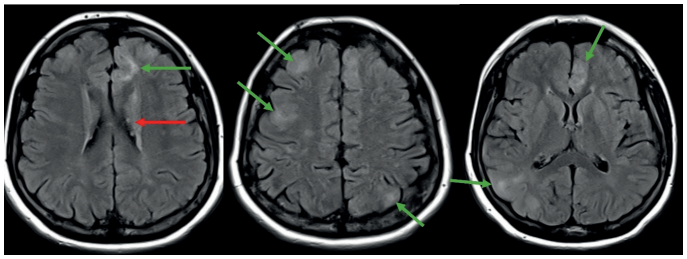

Figure 2. Axial FLAIR images from a recently performed brain magnetic resonance imaging prior to the initial presentation. Signal hyperintensities representing multiple cortical/subcortical tubers (green arrow), predominantly in the frontal lobes, and subependymal nodules in the periventricular region (red arrow) are observed.

Approximately eight months later, she presented again to the emergency department with abdominal pain. Physical examination revealed tenderness in the left lower quadrant. Her hemoglobin level was 8.5 g/dL (reference: 11–15 g/dL). Due to ongoing warfarin therapy, prolonged international normalized ratio (INR) (2.99, reference: 0.8–1.2) and prothrombin time (33.2 seconds, reference: 9.4–12.5 seconds) were detected. No other significant laboratory abnormalities were observed. Contrast-enhanced abdominal CT demonstrated a previously unseen 10 cm lesion in the lower pole of the left kidney, consistent with a hematoma showing internal hemorrhagic layering, along with hemorrhagic fluid in the retroperitoneal and pelvic regions (Figure 3). The clinical and radiological findings supported a diagnosis of AML rupture in the setting of underlying tuberous sclerosis.

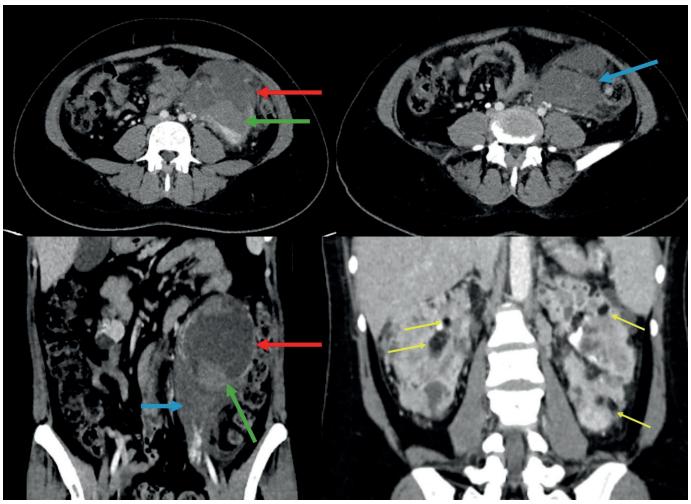

Figure 3. Axial and coronal contrast-enhanced abdominal computed tomography images obtained during the second admission, 8 months after the initial presentation. The first three images demonstrate a collection in the retroperitoneal compartment adjacent to the left kidney, consistent with hematoma (red arrow), containing hyperdense areas suggestive of layering blood products (green arrow). Hemorrhagic free fluid with increased density is observed in the surrounding retroperitoneal space (blue arrow). The bottom right image shows angiomyolipomas in both kidneys (yellow arrows). The patient was under anticoagulant therapy at the time of presentation.

DISCUSSION

Tuberous sclerosis complex (TSC) is a rare genetic disorder characterized by hamartomatous lesions affecting multiple organ systems. According to the 2021 update of the International Tuberous Sclerosis Complex Consensus Conference, the diagnosis can be established either by identifying a pathogenic mutation in the TSC1 or TSC2 genes through genetic testing or by fulfilling clinical criteria specifically, the presence of at least two major features or one major plus two or more minor features (5). Among these diagnostic features, several radiologically demonstrable findings such as renal angiomyolipomas, subependymal nodules, cortical dysplasia, cardiac rhabdomyomas, and sclerotic bone lesions—highlight the pivotal role of radiology in establishing the diagnosis.

Renal AML is the most common abdominal manifestation in individuals with TSC, observed in more than 80% of patients (2). AMLs associated with TSC are typically bilateral, multiple, and display varying fat content. Based on fat composition, they are classified into three subtypes: fat-rich, fat-poor, and fat-invisible. Both CT and MRI play a critical role in the diagnosis of these lesions (6). Notably, the risk of rupture increases significantly in AMLs larger than 4 cm (7). This complication may result in retroperitoneal hemorrhage and a clinical presentation of acute abdominal pain with anemia. In the present case, the hematoma adjacent to the left kidney and the associated retroperitoneal hemorrhagic fluid observed during the second admission were consistent with AML rupture.

One of the distinguishing features of this case is that the combination of the patient's systemic and neurological history, along with CT imaging findings, presented a striking picture suggestive of tuberous sclerosis in an undiagnosed individual. In particular, the presence of multiple renal AMLs in conjunction with subcortical lesions on brain MRI supports the diagnostic criteria. Additionally, the fact that the patient initially presented with pulmonary embolism an infrequently reported finding in the literature underscores the need to consider the vascular manifestations of TSC as well (8). Radiologic evaluation plays a crucial role in both the diagnosis of TSC and the monitoring of its complications. Identifying AMLs with a high risk of rupture is vital for timely follow-up and appropriate intervention. Management strategies may include conservative monitoring, embolization, or surgical approaches (9). This case contributes to the literature by demonstrating the recognition of TSC through radiologic findings in adulthood and emphasizing that renal AML rupture is a complication that requires careful surveillance.

CONCLUSION

This case underscores the critical role of radiological imaging in diagnosing tuberous sclerosis complex in adulthood. Recognition of characteristic multisystem imaging findings such as renal angiomyolipomas, cortical-subcortical tubers, and sclerotic bone lesions can facilitate timely diagnosis and appropriate management. Given the potential for life-threatening complications such as angiomyolipoma rupture, regular imaging follow-up is essential for risk assessment and intervention planning.

Financial disclosures

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Conflict of interest

The authors have no conflicts of interest to declare.

Informed Consent Form

Informed consent was obtained from the patient for this study.

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