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(CASE REPORT)

Imaging-based diagnosis of late-onset tuberous sclerosis complex presenting with acute renal hemorrhage

Kawtar Khaissidi [1,*], Ismail Chaouche [1], Hajar Ouazzani [2], Amal Akammar [2], Nizar El Bouardi [1], Meriem Haloua [2], Badreddine Alami [1], Moulay Youssef Alaoui Lamrani [1], Meryem Boubbou [2] and Mustapha Maaroufi [1]

[1] *Department of Radiology, Specialty Hospital, Hassan II University Hospital Center, Fez, Morocco.*
[2] Department of Radiology, Mother and Child Hospital, Hassan II University Hospital Center, Fez, Morocco.

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Abstract

Tuberous sclerosis complex (TSC) is a rare multisystem genetic disorder characterized by hamartomatous lesions affecting multiple organs, most commonly the brain, kidneys, skin, heart, and lungs. Although typically diagnosed in childhood, late-onset presentations may occur, particularly when renal manifestations predominate. We report the case of a 64-year-old patient admitted for gross hematuria with acute kidney injury, in whom imaging findings strongly suggested previously undiagnosed TSC.

Keywords: Tuberous sclerosis complex; Renal angiomyolipoma; Spontaneous renal hemorrhage; Embolization

1. Introduction

Tuberous sclerosis complex (TSC) is an autosomal dominant neurocutaneous syndrome caused by pathogenic variants in the TSC1 or TSC2 genes, leading to dysregulation of the mTOR signaling pathway. The disease is characterized by the development of benign tumors and malformations in multiple organs. Renal involvement represents a major cause of morbidity and mortality in adult patients and may occasionally constitute the initial mode of presentation in late-diagnosed cases.

2. Case Presentation

A 64-year-old patient with a history of bilateral inguinal hernia repair and a history of polycystic kidney disease was admitted for management of gross hematuria complicated by clot retention and acute kidney injury.

Laboratory investigations revealed severe anemia, with a hemoglobin level of 6 g/dL compared with a baseline value of 9 g/dL.

Abdominal contrast-enhanced CT demonstrated bilaterally enlarged polycystic kidneys, containing several hemorrhagic cysts and bilateral renal calculi (figure 1).

* Corresponding author: Kawtar Khaissidi

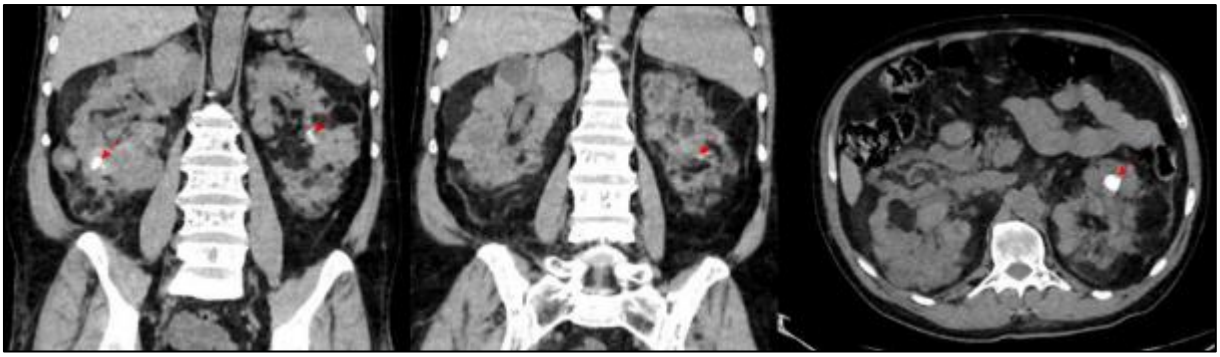

Figure 1 Non contrast axial CT images demonstrating bilaterally polycystic kidneys, with some hemorrhagic cysts and the presence of bilateral calcified renal calculi (red arrow)

In addition, multiple bilateral exophytic renal parenchymal lesions were identified, exhibiting marked heterogeneity with a predominant fatty component associated with a soft-tissue component showing heterogeneous contrast enhancement. These findings were suggestive of renal angiomyolipomas.

One of the right-sided lesions had bled and contained an intralesional hematoma, with marked perirenal and pararenal fat infiltration. A tortuous right inferior polar artery with aneurysmal dilatation was also identified, a finding associated with an increased risk of spontaneous hemorrhage (figure 2).

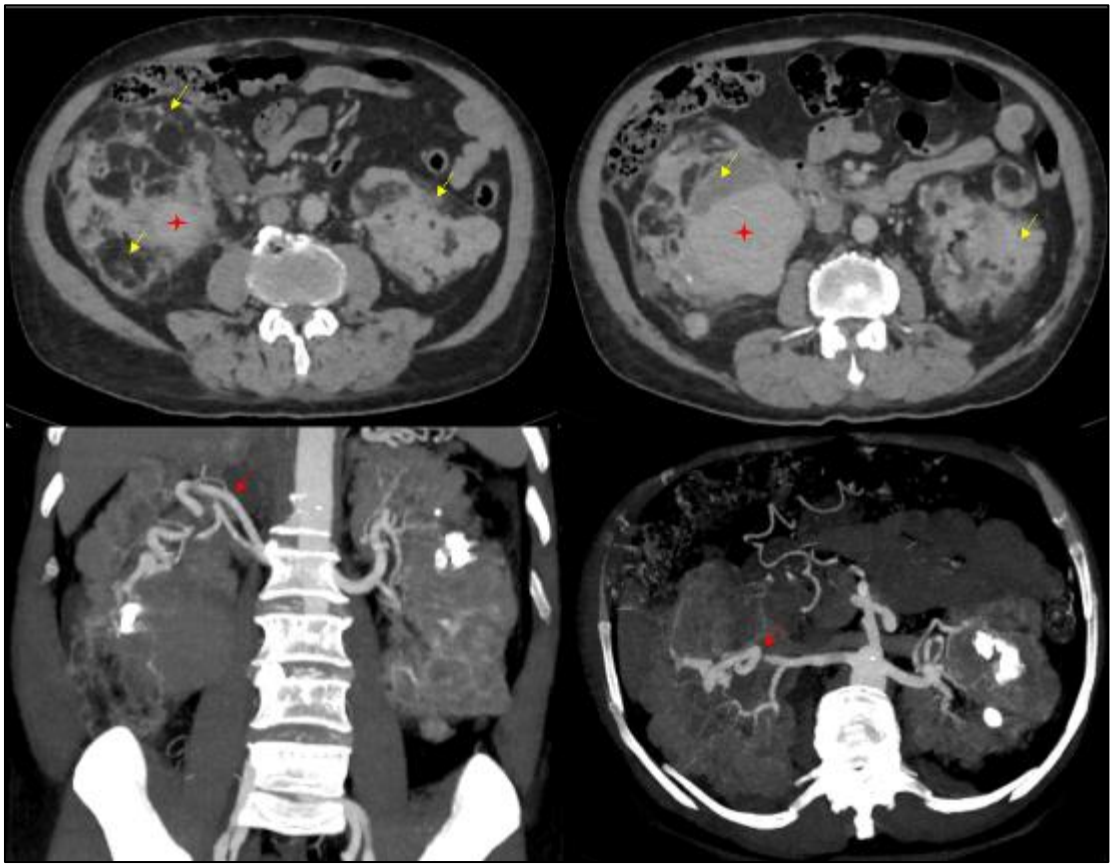

Figure 2 Axial contrast-enhanced CT images demonstrating multiple heterogeneous renal lesions with fatty and solid components and heterogeneous contrast enhancement (yellow arrow), with the right lower pole lesion containing a heterogeneous intralesional hematoma (Red Star). A tortuous right inferior polar renal artery with intravascular pseudoaneurysms was also identified (Red Arrow), a finding associated with an increased risk of spontaneous hemorrhage

The patient underwent renal angiography, which demonstrated a dysplastic right mid-renal artery with multiple aneurysmal formations. Selective embolization of the right mid-renal artery was subsequently performed, with satisfactory technical outcome. (figure 3)

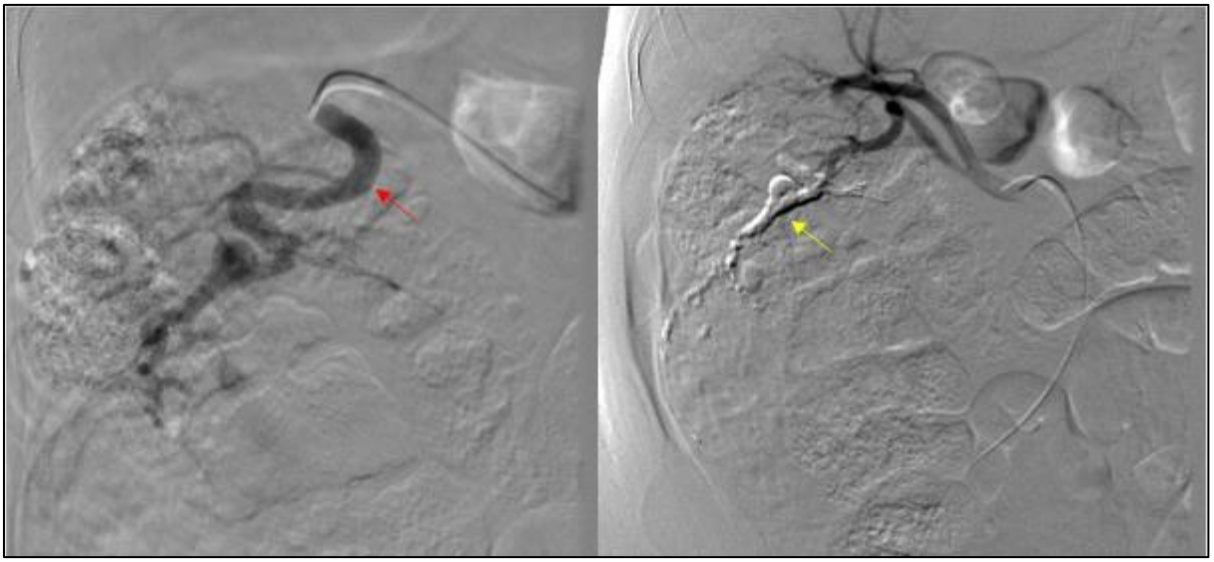

Figure 3 Angiographic images demonstrating a tortuous and dysplastic right mid-renal artery (Red Arrow), which was successfully treated by embolization, with satisfactory post-procedural angiographic and CT scan control (yellow arrow)

Following embolization, the patient showed progressive clinical stabilization with improvement of hematuria and renal function. Long-term multidisciplinary follow-up was recommended.

Additional abnormalities were identified on comprehensive radiological evaluation. Brain CT revealed several calcified nodular lesions along the lateral walls of both lateral ventricles, consistent with subependymal nodules, as well as a right cerebellar calcification. Brain MRI confirmed the persistence of these subependymal and right cerebellar lesions, which appeared calcified and hypointense on all sequences.

Orbital MRI demonstrated elevation of the right optic disc, suggestive of a retinal astrocytic hamartoma (Figure 4).

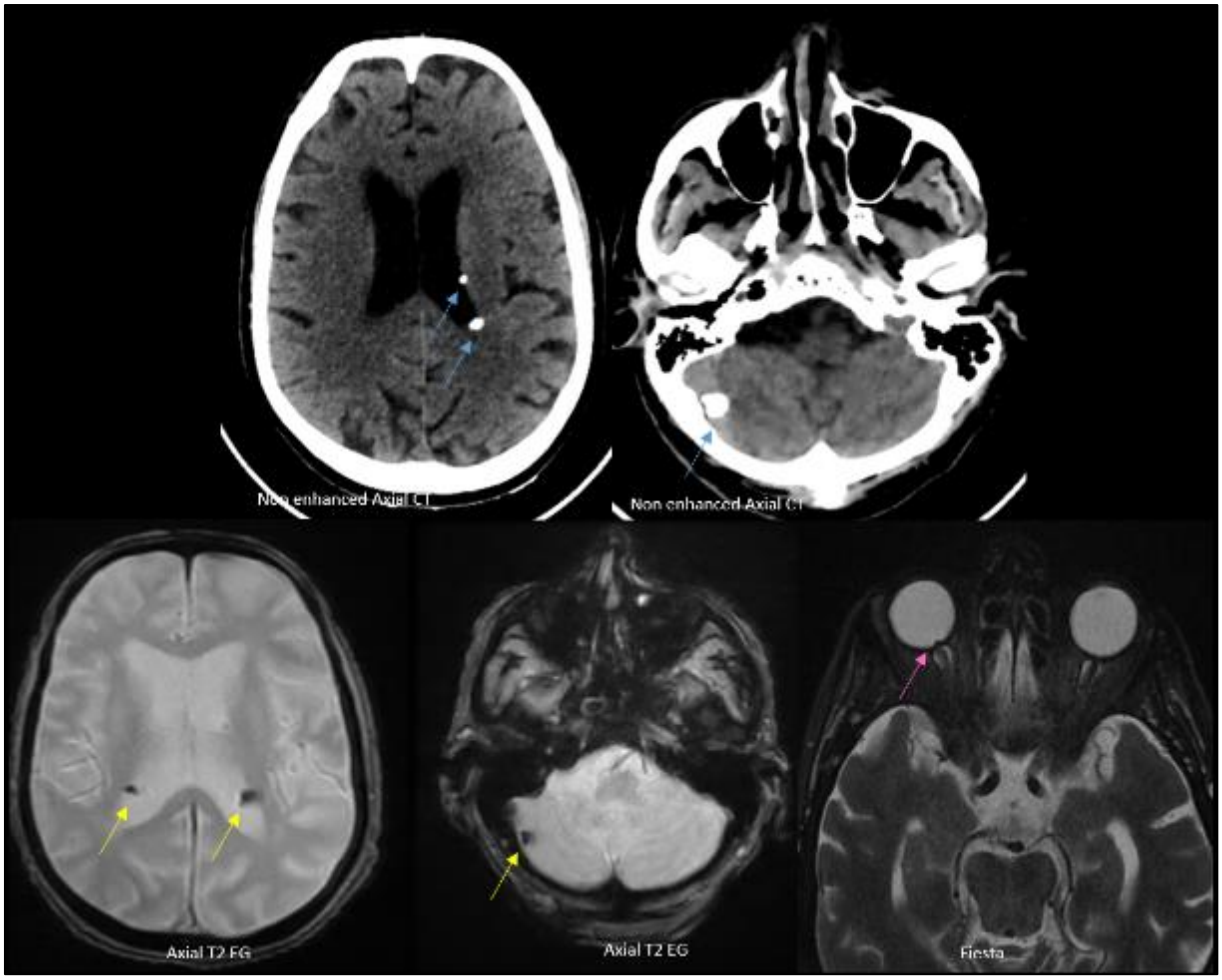

Figure 4 Axial CT images of the supratentorial and infratentorial compartments demonstrate the presence of calcified subependymal nodules (blue arrow). These findings are corroborated by axial T2*weighted MRI sequences, which show the same nodules as signal voids. Orbital MRI demonstrates features consistent with a retinal hamartoma (Pink arrow)

Additional findings included a heterogeneous hepatic lesion involving segment VIII with a fatty component, as well as an intracardiac lesion of fatty density suggestive of a cardiac rhabdomyoma (figure 5).

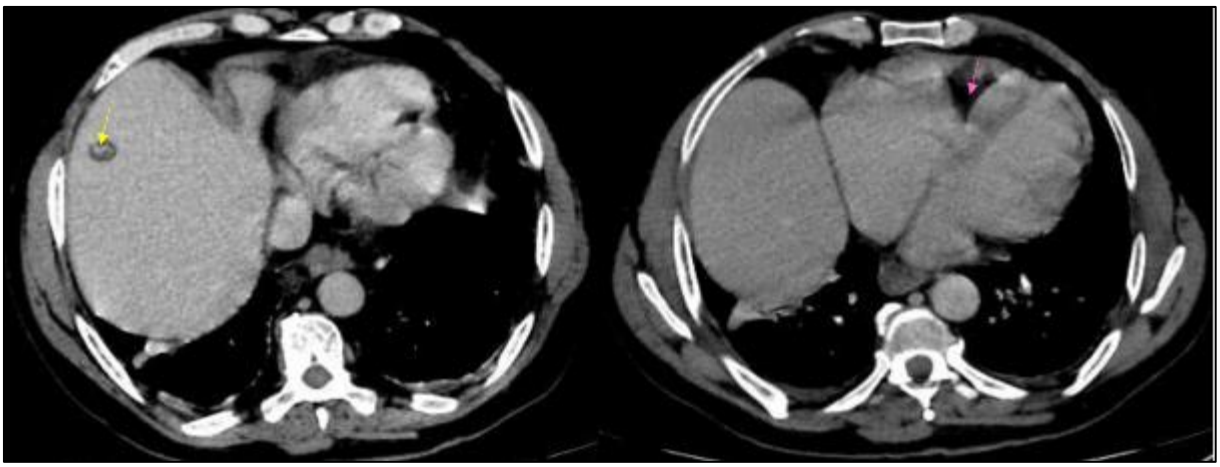

Figure 5 CT images through the liver and cardiac chambers demonstrate a hepatic lesion with fatty and soft-tissue components suggestive of an angiomyolipoma, as well as a second intracardiac lesion consistent with a rhabdomyoma

Taken together, the above-described findings are highly suggestive of tuberous sclerosis complex.

3. Discussion

Tuberous sclerosis complex (TSC) is a multisystem genetic disorder caused by pathogenic mutations in the *TSC1* or *TSC2* genes with highly variable clinical expression.

Although tuberous sclerosis complex is typically diagnosed early in life, recognition may be delayed in oligosymptomatic forms particularly in the absence of prominent neurological or cutaneous manifestations and the disease may instead be revealed in adulthood by visceral complications, notably renal involvement, as illustrated in our patient [1,2].

Renal involvement is a common and clinically significant feature of tuberous sclerosis complex, predominantly characterized by angiomyolipomas and renal cysts. In TSC, angiomyolipomas are typically bilateral, multifocal, and larger than sporadic forms, and may lead to progressive parenchymal damage and acute complications such as hemorrhage, anemia, and acute kidney injury, as observed in our patient [3].

In tuberous sclerosis complex, large angiomyolipomas particularly those exceeding 4 cm or containing intratumoral aneurysms carry a high risk of spontaneous hemorrhage.

Tumor growth increases intralesional blood flow, promoting vascular dilatation and aneurysm formation, whose rupture may precipitate acute bleeding and further tumor enlargement, as illustrated by the giant hemorrhagic angiomyolipoma observed in our patient [4].

Neuroimaging plays a central role in establishing the diagnosis of TSC in adults. Subependymal nodules, especially when calcified, are a hallmark feature and are readily identified on non-contrast CT scans [1,5] Their detection is particularly valuable in patients lacking overt neurological symptoms, as in our case, where intracranial findings were incidental but decisive for diagnosis. The additional cerebellar calcification, although less typical, further reflects the widespread hamartomatous nature of the disease [5].

This case underscores the importance of a pattern-recognition approach in radiology, whereby the association of characteristic brain lesions with complex renal findings should prompt consideration of TSC, even in elderly patients. Early recognition is crucial, as it allows appropriate multidisciplinary management, including nephrological follow-up, screening for other organ involvement, and consideration of targeted therapies such as mTOR inhibitors, which have been shown to reduce angiomyolipoma volume and bleeding risk [2, 3].

4. Conclusion

This case highlights the pivotal role of imaging in the diagnosis of late-onset tuberous sclerosis complex, where characteristic multisystem findings may be incidentally revealed through acute renal complications. Recognition of these imaging patterns is essential to ensure timely diagnosis, appropriate multidisciplinary management, and prevention of potentially life-threatening complications.

Compliance with ethical standards

Disclosure of conflict of interest

The authors declare no conflicts of interest.

Statement of informed consent

Oral informed consent was obtained from the patient for publication of this case report and accompanying images.

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