SAS Journal of Medicine
Abbreviated Key Title: SAS J Med
ISSN 2454-5112
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Multisystemic Tuberous Sclerosis Complex: A Comprehensive Case Report

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DOI: https://doi.org/10.36347/sasjm.2026.v12i09.014 | **Received:** 13.07.2026 | **Accepted:** 01.09.2026 | **Published:** 17.09.2026

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Abstract | **Case Report**

Tuberous Sclerosis Complex (TSC) is a rare, multisystemic, autosomal dominant genetic disorder resulting from mutations in the *TSC1* or *TSC2* genes. These mutations lead to the constitutive activation of the mechanistic target of rapamycin (mTOR) signaling pathway, resulting in the formation of benign hamartomas across multiple organ systems. We present the case of a 14-year-old male patient who presented with new-onset generalized tonic-clonic seizures. Clinical evaluation revealed hallmark cutaneous stigmata, specifically multiple café-au-lait macules. Diagnostic neuroimaging (computed tomography) demonstrated pathognomonic central nervous system involvement, including calcified subependymal nodules and cortical tubers. Abdominal ultrasound identified bilateral renal angiomyolipomas. This report provides a detailed analysis of the clinical presentation, radiological findings, and the necessary multidisciplinary surveillance protocols for adolescents with TSC.

Keywords: Tuberous Sclerosis Complex, mTOR pathway, generalized tonic-clonic seizures, subependymal nodules, cortical tubers, renal angiomyolipomas.

INTRODUCTION

Tuberous Sclerosis Complex, historically referred to as Bourneville disease, represents a classic phakomatosis. The pathophysiology is rooted in the loss of function of the hamartin/tuberin complex, which serves as a vital regulator of cellular growth and differentiation. The resulting mTOR pathway dysregulation manifests in widespread tissue disorganization. While the disorder is often identified in infancy, many adolescents present with neurological symptoms—most commonly epilepsy—as the initial sentinel event. Because TSC can affect almost any organ system, clinicians must maintain a high index of suspicion, utilizing multimodal imaging to confirm the diagnosis and assess systemic disease burden.

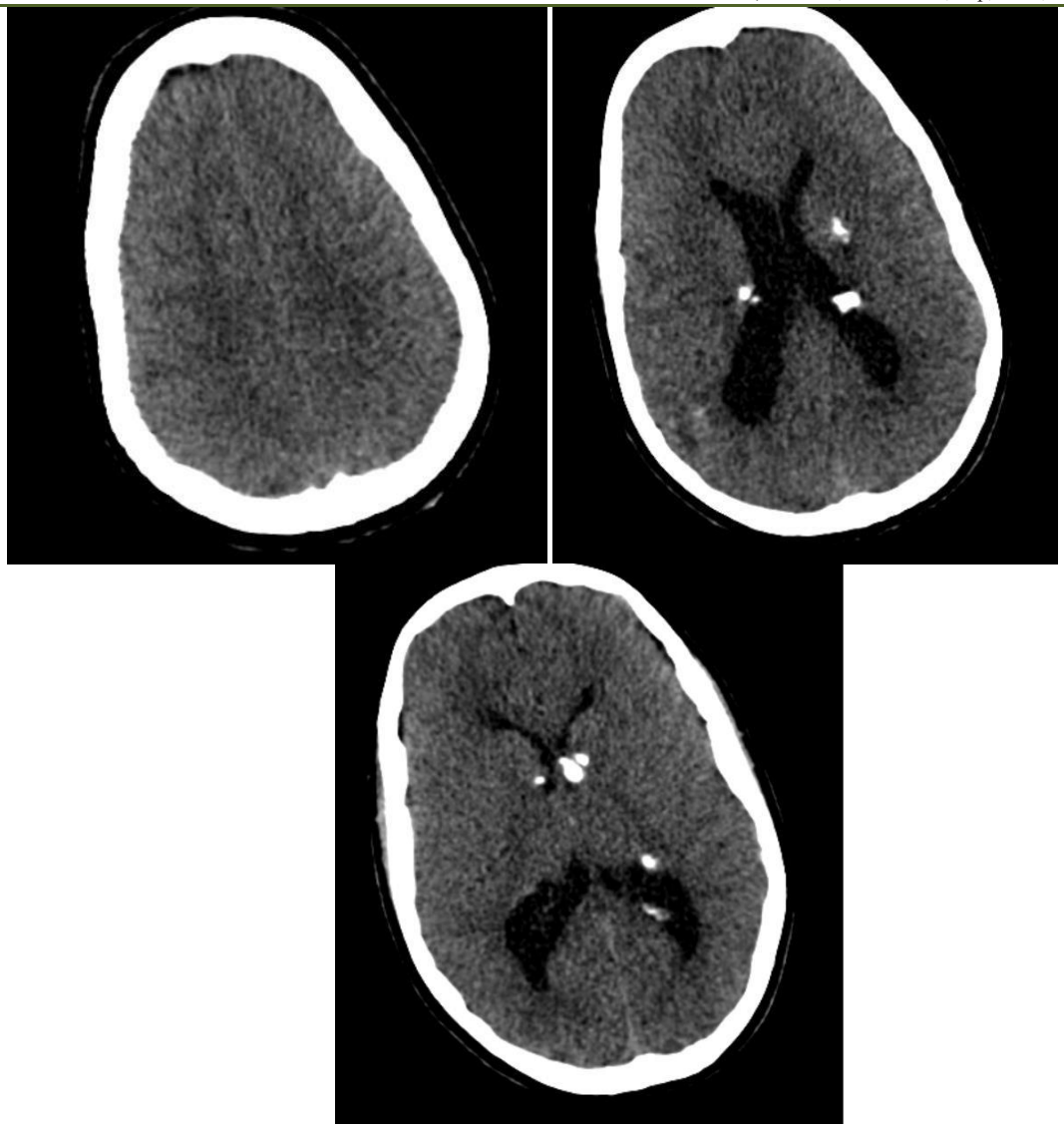
CASE PRESENTATION

Clinical History:

A 14-year-old male was referred to our department following a first-time generalized tonic-clonic seizure. The patient had no prior history of developmental delays. Dermatological inspection revealed several café-au-lait macules.

Radiological Assessment (Brain CT Scan):

Finding	Description
Subependymal Nodules (SENs)	Multiple calcified nodules noted along the ventricular walls, with a primary lesion (4.5 x 6.7 mm) protruding into the lateral ventricle.
Cortical Tubers	Hypodense cortical/subcortical lesions in bilateral fronto-parietal regions, consistent with focal cortical dysplasia.
Calcifications	Extensive parenchymal calcifications observed in the right frontal lobe and basal ganglia, indicating stable, chronic pathology.



Visceral Screening:

Abdominal ultrasonography confirmed bilateral renal angiomyolipomas (AMLs), defined by their characteristic hyperechoic appearance on imaging.

DISCUSSION

Pathophysiological Insights and Diagnostic Criteria

The diagnosis of Tuberous Sclerosis Complex (TSC) in this adolescent patient relies on a combination of major clinical and radiological features. The pathognomonic triad epilepsy, intellectual disability, and cutaneous stigmata serves as the traditional framework, yet the modern diagnosis is increasingly driven by specific imaging biomarkers. The presence of cortical tubers and subependymal nodules (SENs) represent the hallmark central nervous system (CNS) phenotype. These findings are the direct result of the constitutive activation of the mechanistic target of rapamycin (mTOR) pathway, which impairs normal neuronal migration and differentiation during cortical development.

Neuro-radiological Risks and Surveillance

While the identified subependymal nodules are currently calcified and appear stable, they carry a latent risk of transformation into Subependymal Giant Cell Astrocytomas (SEGAs). SEGAs are benign but can be life-threatening if they grow near the foramen of Monro, causing obstructive hydrocephalus. Clinical vigilance is mandatory; we emphasize that annual brain MRI (using FLAIR and T2-weighted sequences) is superior to CT for characterizing these tubers and monitoring the volumetric evolution of SENs. A rapid increase in the size of a nodule should trigger an immediate transition to pharmacological intervention with mTOR inhibitors.

Visceral Management: The Renal Dimension

The finding of bilateral renal angiomyolipomas (AMLs) introduces a significant morbidity risk. AMLs are benign neoplasms composed of dysmorphic blood vessels, smooth muscle, and adipose tissue. In this adolescent patient, the risk is not just one of mass effect, but also of spontaneous retroperitoneal hemorrhage (Wunderlich syndrome), which correlates with the size

and vascular density of the AMLs. International consensus suggests that prophylactic embolization or medical therapy is indicated when lesions exceed 3–4 cm. Consequently, the systematic follow-up of these masses via ultrasound is non-negotiable to prevent acute complications.

CONCLUSION

This case underscores that adolescents presenting with sudden-onset seizures require a comprehensive systemic workup. The use of multimodal imaging facilitates an accurate diagnosis of Tuberous Sclerosis Complex. Early detection of these lesions allows for proactive monitoring, reducing the risks associated with the long-term progression of this multisystemic disease.

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