RADIODIAGNOSIS

INDEX COPERNICUS INTERNATIONAL

ICV value for 2024 = 82.51

Case Report | International Journal of Medical Case Reports

Multisystem Involvement in Tuberous Sclerosis Complex: Incidentally Detected Subependymal Giant Cell Astrocytoma with Extensive Abdomino-visceral Manifestations

Authors: Surinder Singh[1], Bhawna Heer[2], Tripti Jain[3], Harinder Singh Chhabra[4]

Affiliations:

[1,2]Department of Radiodiagnosis, Gian Sagar Medical College and Hospital, Rajpura, Patiala, India.

[3]Department of Pathology, Gian Sagar Medical College and Hospital, Rajpura, Patiala, India.

[4]Department of Medicine, Adesh Institute of Medical Sciences and Research, Bathinda, India.

ISSN 2455-0574

ABSTRACT

Background

Tuberous sclerosis complex is a multisystem genetic disorder characterized by hamartomatous lesions involving the brain, kidneys, skin, lungs, heart, and hepatobiliary system. Although typically recognized in childhood, attenuated phenotypes may remain undiagnosed until adulthood, particularly when seizures, cognitive impairment, or cutaneous stigmata are absent.

Case Report

A 47-year-old woman presented with insidious dull aching right flank pain without haematuria, fever, weight loss, seizures, headache, visual symptoms, focal neurological deficit, or altered sensorium. She had no family history or clinical cutaneous features suggestive of tuberous sclerosis complex. Contrast-enhanced computed tomography of the abdomen revealed bilateral enlarged kidneys with multiple well-defined fat-containing lesions consistent with multifocal renal angiomyolipomas, including a dominant 8 cm exophytic right renal angiomyolipoma with increased hemorrhagic risk. Additional findings included multiple hepatic fat-density lesions, a mildly enhancing fat-rich hepatic angiomyolipoma, and multiple tiny pancreatic lipomas. In view of multisystem fat-containing lesions, an underlying phakomatosis was suspected. Non-contrast computed tomography of the brain demonstrated a well-defined partially calcified hyperdense lesion near the right foramen of Monro, consistent with subependymal giant cell astrocytoma, along with multiple calcified subependymal nodules showing a characteristic "candle-guttering" appearance and a right parietal cortical tuber. There was no hydrocephalus. The combined intracranial and abdomino-visceral imaging findings established the diagnosis of tuberous sclerosis complex. The patient was advised multidisciplinary follow-up and imaging surveillance, particularly for the large renal angiomyolipoma and intracranial lesion.

Conclusion

Tuberous sclerosis complex may present for the first time in adulthood with extensive multisystem involvement despite absence of classical clinical manifestations. Comprehensive cross-sectional imaging is crucial for diagnosis, assessment of disease burden, risk stratification, and long-term surveillance.

Keywords: Angiomyolipoma, Astrocytoma, Hamartoma, Subependymal Giant Cell Astrocytoma, Tuberous Sclerosis Complex.

INTRODUCTION

Tuberous sclerosis complex (TSC) is a multisystem genetic disorder characterized by the development of hamartomatous and dysplastic lesions in the brain, kidneys, skin, lungs, heart, and hepatobiliary system as a consequence of pathogenic variants in *TSC1* or *TSC2*, with subsequent dysregulation of the mammalian target of rapamycin signalling pathway.[1] Although classically described as an autosomal dominant phakomatosis, a substantial proportion of cases arise sporadically, and the phenotypic spectrum is remarkably heterogeneous. The traditional clinical triad of seizures, intellectual disability, and facial angiofibromas is now recognized to represent only a minority of patients and does not adequately capture the broad and age-dependent manifestations of the disease. TSC is usually identified in childhood because of neurological symptoms, dermatological stigmata, or prenatal and infantile cardiac findings; however, delayed recognition in adulthood continues to occur, particularly in patients with attenuated or atypical phenotypes. Such late presentations are clinically important because they may conceal substantial visceral disease burden despite the absence of overt neurological or cutaneous symptoms.[2]

International Journal Of Medical Case Reports (ISSN 2455-0574) is an indexed medical journal indexed in Index Copernicus

Access this Journal Online

Quick Response Code

Website: www.ijomcr.net

Email:ijomcr@gmail.com

Dr Surinder Singh

Department of Radiodiagnosis, Gian Sagar Medical College and Hospital, Rajpura, Patiala, India.

email : surinder.aiims@gmail.com

Central nervous system involvement is reported to be one of the defining features of TSC and it is known to contribute substantially to long-term morbidity. The most characteristic intracranial lesions include cortical tubers, subependymal nodules and subependymal giant cell astrocytoma (SEGA).[3] Among these, SEGA is of particular relevance because of its typical location near the foramen of Monro and its potential to cause obstructive hydrocephalus, raised intracranial pressure as well as progressive neurological deterioration if not identified and managed in a timely manner.[4] Although SEGA is generally considered a tumor of childhood and adolescence published data have shown that it may persist, enlarge or even first be recognized in adulthood. These features make age alone an unreliable basis for excluding this diagnosis. Furthermore, some adults with TSC may lack seizures, cognitive impairment or obvious cutaneous markers leading to under-recognition of associated intracranial lesions until imaging is performed for unrelated indications.[5] In such scenarios, cross-sectional neuroimaging assumes a decisive role in establishing the diagnosis and directing surveillance.

We report an atypical case of TSC presenting in the fifth decade with extensive multisystem involvement detected incidentally.

CASE REPORT

A 47-year-old woman presented with dull aching right flank pain of insidious onset. There was no history of haematuria, fever, weight loss, seizures, chronic headache, vomiting, visual disturbance, focal neurological deficit, or altered sensorium. She had no known prior diagnosis of tuberous sclerosis complex (TSC) and no relevant family history. Physical examination revealed no cutaneous stigmata of TSC, including facial angiofibromas, ash-leaf macules, shagreen patches, periungual fibromas, or forehead plaques. Neurological examination was unremarkable.

Contrast-enhanced computed tomography (CECT) of the abdomen and pelvis showed bilateral enlarged kidneys and multiple well-defined fat-containing lesions consistent with multifocal renal angiomyolipomas. The largest lesion was an exophytic angiomyolipoma arising from the right kidney (measuring approximately 8 cm in maximum dimension) indicating increased risk of spontaneous hemorrhage (Figure 1).

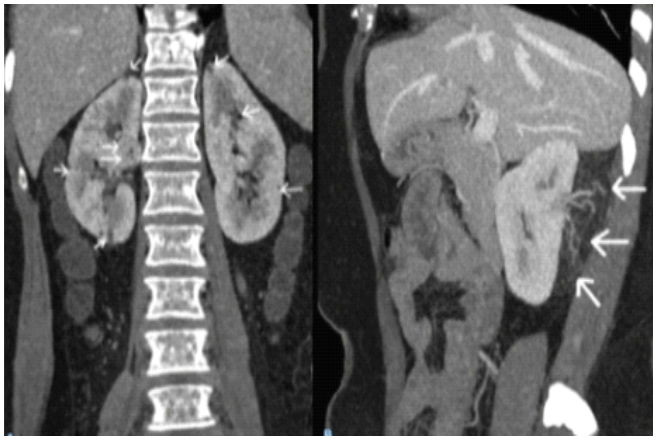

Figure 1: (A) Coronal reformatted contrast-enhanced CT (CECT) of the abdomen showing multiple, well-circumscribed masses in both kidneys containing macroscopic fat-density areas (white arrows) and interspersed enhancing soft tissue components, characteristic of bilateral multifocal renal angiomyolipomas. (B) Sagittal Maximum Intensity Projection (MIP) CECT reformat demonstrating a large, 8 cm exophytic angiomyolipoma in the right kidney.

primarily composed of macroscopic fat-density tissue (white arrows) with conspicuous vascularity.

In addition, multiple discrete fat-density lesions were noted within the liver, suggestive of hepatic lipomas. A separate mildly enhancing fat-containing lesion in the left hepatic lobe was consistent with a fat-rich hepatic angiomyolipoma. Multiple tiny fat-density foci were also identified in the head and body of the pancreas, in keeping with pancreatic lipomas (Figure 2).

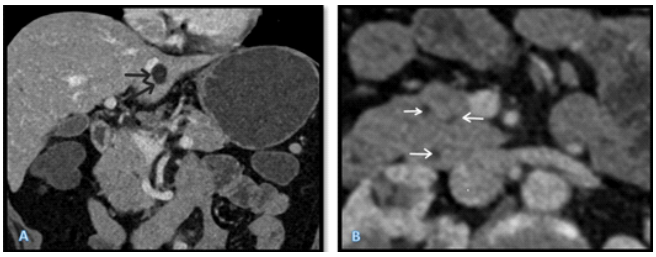

Figure 2: (A) Coronal contrast-enhanced computed tomography (CECT) of the abdomen showing a well-defined, fat-containing lesion in the liver parenchyma with mild soft tissue component consistent with a fat-rich hepatic angiomyolipoma (black arrows). (B) Magnified CECT view of the pancreatic head demonstrating three discrete, small fat-density lesions (white arrows) consistent with pancreatic lipomas, a rare systemic manifestation of Tuberous Sclerosis Complex.

Given the presence of bilateral renal angiomyolipomas and associated abdominovisceral fat-containing lesions, an underlying phakomatosis, particularly TSC, was suspected. Non-contrast computed tomography of the head revealed a well-defined lobulated hyperdense lesion measuring approximately 15 × 15 mm near the right foramen of Monro, showing partial peripheral calcification and imaging features consistent with subependymal giant cell astrocytoma (SEGA). Multiple calcified subependymal nodules lining both lateral ventricles produced the characteristic "candle-guttering" appearance. A focal cortical tuber was also identified in the right parietal lobe. No hydrocephalus was evident (Figure 3).

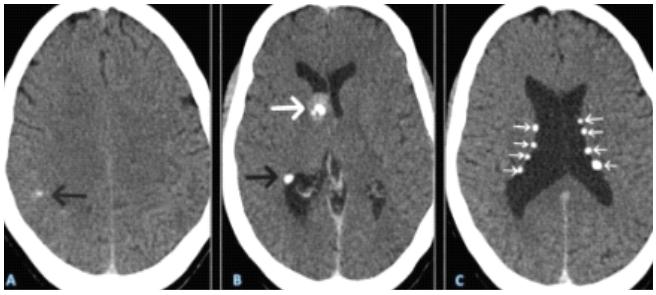

Figure 3: Axial NCCT images demonstrating characteristic neuro-radiological features: (A) A hyperdense cortical tuber in the right parietal lobe (black arrow); (B) A well-defined, partially calcified hyperdense lesion (white arrow) at the right foramen of Monro, consistent with a subependymal giant cell astrocytoma (SEGA). Note the associated calcified subependymal nodule (black arrow) along asymmetrically dilated atrium of right lateral ventricle; and (C) Multiple classic "candle-guttering" calcified subependymal nodules lining the ependymal surface (white arrows).

Based on the combined intracranial and abdominovisceral imaging findings, a diagnosis of TSC was established. This represented an atypical late presentation with incidentally detected SEGA and extensive multisystem involvement in the absence of classical clinical manifestations.

The patient was advised multidisciplinary follow-up and imaging surveillance, particularly in view of the large right renal angiomyolipoma and the need to monitor the intracranial lesion.

DISCUSSION

In this case the diagnosis of tuberous sclerosis complex (TSC) was established primarily through multisystem imaging findings rather than typical clinical features. In the absence of seizures, intellectual disability or cutaneous stigmata the index of suspicion for intracranial pathologies remain low. However eventual neuroimaging showed combination of a foramen of Monro lesion consistent with SEGA, calcified subependymal nodules, cortical tuber and bilateral renal angiomyolipomas strongly suggesting TSC. Adriaensen et al emphasized the characteristic imaging appearance and location of SEGA in TSC.[6] Similarly Northrup et al highlighted that modern diagnosis increasingly depends on identifying major imaging features across organ systems rather than relying on the historical Vogt triad.[7]

The intracranial findings in this patient are particularly important from a neuroradiology perspective. The hyperdense partially calcified lesion seen near the foramen of Monro along with associated subependymal nodules and cortical tuber, is the classic imaging spectrum of CNS involvement in cases of TSC. Jóźwiak et al noted that SEGA may remain clinically silent and be detected only on imaging surveillance, particularly when hydrocephalus is absent.[8] In the present case, CT was sufficient to demonstrate the typical topography and calcific character of the lesion, allowing confident radiologic diagnosis even in an asymptomatic adult.

The abdominal imaging findings in this case further underscores the multisystem nature of TSC. Bilateral multifocal renal angiomyolipomas, especially with a dominant 8 cm exophytic lesion, are highly relevant because lesion size, fat content, and vascularity directly influence hemorrhagic risk and management planning. Rakowski et al showed that renal angiomyolipomas are among the most frequent extracranial manifestations of TSC and a major cause of morbidity in adults.[9] From a radiology standpoint, CT not only establishes the diagnosis by demonstrating macroscopic fat and enhancing soft tissue components, but also helps stratify risk and guide surveillance.

The associated hepatic lipomas, hepatic angiomyolipoma, and pancreatic lipomas make this case unusual and radiologically instructive. Hepatic angiomyolipoma is a recognized but less common manifestation of TSC, whereas pancreatic lipomas are distinctly rare. Black et al described hepatic involvement as part of the broader abdominovisceral spectrum of TSC, and this case expands that imaging phenotype by demonstrating simultaneous renal, hepatic, pancreatic, and intracranial lesions. Recognition of these fat-containing lesions at multiple sites is important because their coexistence on cross-sectional imaging should immediately raise suspicion for an underlying phakomatosis, even when the patient lacks overt clinical markers.[10]

Overall, this case highlights how cross-sectional imaging can be the key to diagnosing atypical or late-presenting TSC. CT of the brain and abdomen not only identified the major diagnostic features but also revealed the full burden of disease in a patient who was otherwise clinically stable. In such cases radiology is not only confirmatory but also often diagnostic as well as surveillance-defining. This report therefore underscores the importance of comprehensive cross-sectional imaging in patients with multisystem fat-containing lesions, as it may uncover occult TSC, detect clinically silent but significant lesions such as SEGA, and guide appropriate long-term multidisciplinary follow-up.

CONCLUSION

Tuberous sclerosis complex, though more commonly presents in pediatric age group, may present for the first time in adulthood with extensive multisystem involvement.

The coexistence of SEGA, subependymal nodules, cortical tuber, and renal angiomyolipomas underscores the radiologic spectrum of disease. A high index of suspicion and comprehensive cross-sectional imaging is therefore important in such atypical cases for establishing diagnosis.

Source of Funding : None

Conflict Of Interest : None

Consent and Ethics

Written informed consent was obtained from the patient for the publication of this case report and any accompanying images. Ethical approval was waived as per institutional policy for single case reports.

REFERENCES

1. Napolioni V, Curatolo P. Genetics and molecular biology of tuberous sclerosis complex. Curr Genomics. 2008 Nov;9(7):475-87. doi: 10.2174/138920208786241243. PMID: 19506736; PMCID: PMC2691673.
2. Nathan N, Burke K, Trickett C, Moss J, Darling TN. The Adult Phenotype of Tuberous Sclerosis Complex. Acta Derm Venereol. 2016 Feb;96(2):278-80. doi: 10.2340/00015555-2203. PMID: 26258325; PMCID: PMC6118340.
3. Menor F, Martí-Bonmatí L, Mulas F, Poyatos C, Cortina H. Neuroimaging in tuberous sclerosis: a clinicoradiological evaluation in pediatric patients. Pediatr Radiol. 1992;22(7):485-9. doi: 10.1007/BF02012989. PMID: 1491903.
4. Moran V, O'Keeffe F. Giant cell astrocytoma in tuberous sclerosis: computed tomographic findings. Clin Radiol. 1986 Nov;37(6):543-5. doi: 10.1016/s0009-9260(86)80009-5. PMID: 3791849.
5. Hanes I, Muir K, Abdeen N, Sell E. Tuberous sclerosis complex associated intracranial lesion found by antenatal ultrasound. Radiol Case Rep. 2020 Apr 22;15(7):816-818. doi: 10.1016/j.radcr.2020.04.003. PMID: 32346461; PMCID: PMC7182691.
6. Adriaensen ME, Schaefer-Prokop CM, Stijnen T, Duyndam DA, Zonnenberg BA, Prokop M. Prevalence of subependymal giant cell tumors in patients with tuberous sclerosis and a review of the literature. Eur J Neurol. 2009 Jun;16(6):691-6. doi: 10.1111/j.1468-1331.2009.02567.x. Epub 2009 Feb 19. PMID: 19236458.
7. Northrup H, Aronow ME, Bebin EM, Bissler J, et al. Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. Pediatr Neurol. 2021 Oct;123:50-66. doi: 10.1016/j.pediatrneurol.2021.07.011. Epub 2021 Jul 24. PMID: 34399110.
8. Jóźwiak S, Mandera M, Młynarski W. Natural History and Current Treatment Options for Subependymal Giant Cell Astrocytoma in Tuberous Sclerosis Complex. Semin Pediatr Neurol. 2015 Dec;22(4):274-81. doi: 10.1016/j.spen.2015.10.003. Epub 2015 Oct 21. PMID: 26706014.

9. Rakowski SK, Winterkorn EB, Paul E, Steele DJ, Halpern EF, Thiele EA. Renal manifestations of tuberous sclerosis complex: Incidence, prognosis, and predictive factors. Kidney Int. 2006 Nov;70(10):1777-82. doi: 10.1038/sj.ki.5001853. Epub 2006 Sep 27. PMID: 17003820.

10. Black ME, Hedgire SS, Camposano S, Paul E, Harisinghani M, Thiele EA. Hepatic manifestations of tuberous sclerosis complex: a genotypic and phenotypic analysis. Clin Genet. 2012 Dec;82(6):552-7. doi: 10.1111/j.1399-0004.2012.01845.x. Epub 2012 Feb 2. PMID: 22251200.

*****

Author Contribution :
SS-Concept, imaging interpretation, manuscript drafting; **BH**-Data collection, manuscript review, final approval; **TJ**-Literature review, manuscript editing; **HSC**- Clinical management, supervision, critical revision of manuscript for important intellectual content

Received : 15-03-2026 Revised: 10-04-2026 Accepted : 30-04-2026