

Multisystem Imaging Features of Tuberous Sclerosis Complex: The First Case Series from Somalia with a Comprehensive Literature Review

Ahmed Adam Osman

Mogadishu Somali Turkish Training and Research Hospital

Mohamed Osman Dahir Alasow

mcalasow00@gmail.com

Mogadishu Somali Turkish Training and Research Hospital

Ismail Gedi Ibrahim

Mogadishu Somali Turkish Training and Research Hospital

Abdiwahid Ahmed Ibrahim

Mogadishu Somali Turkish Training and Research Hospital

Case Report

Keywords: Neurocutaneous syndrome, Renal angiomyolipoma, The mammalian target of rapamycin (mTOR), Tuberous sclerosis complex, Magnetic resonance imaging

Posted Date: September 8th, 2026

DOI: <https://doi.org/10.21203/rs.3.rs-10515800/v1>

License:   This work is licensed under a Creative Commons Attribution 4.0 International License.

[Read Full License](#)

Additional Declarations: No competing interests reported.

Abstract

Background

Tuberous sclerosis complex (TSC) is a rare autosomal dominant multisystem disorder caused by pathogenic variants in the *TSC1* or *TSC2* genes, resulting in hamartomatous lesions involving the brain, skin, kidneys, heart, lungs, and other organs. Early recognition is essential to reduce disease-related morbidity; however, diagnosis remains challenging in resource-limited settings where molecular genetic testing is unavailable. We report the first case series of TSC from Somalia, highlighting the pivotal role of multimodality imaging in establishing the diagnosis.

Case Presentation:

We report here two Somali patients with different clinical phenotypes who met the updated 2021 International Tuberous Sclerosis Complex Consensus diagnostic criteria. The first patient was a 5-year-old girl with refractory epilepsy, developmental delay, facial angiofibromas, hypomelanotic macules, and bilateral renal angiomyolipomas. Brain magnetic resonance imaging (MRI) showed multiple cortical and subcortical tubers, radial migration lines and subependymal nodules. The second patient was a 22-year-old woman with longstanding drug resistant epilepsy, typical facial angiofibromas and a hypomelanotic macule. Brain MRI was performed with multiple cortical tubers, lines of radial migration and subependymal nodules. Abdominal ultrasonography revealed no renal abnormalities. Molecular genetic testing was not performed; however, both patients met the criteria for a definite diagnosis of TSC based on characteristic clinical and radiological findings.

Conclusion

This case series represents the first reported cases of tuberous sclerosis complex from Somalia and underscores the essential role of MRI and ultrasonography in the diagnosis of TSC. Our findings demonstrate that adherence to the updated 2021 International Tuberous Sclerosis Complex Consensus clinical diagnostic criteria allows confident diagnosis even in the absence of molecular genetic testing. Increased awareness of the multisystem imaging spectrum of TSC may facilitate earlier recognition, appropriate surveillance, and multidisciplinary management, particularly in resource-limited healthcare settings.

Introduction

Tuberous sclerosis (TS), which is often referred to as epiloia, is an extremely uncommon genetic ailment that is inherited in an autosomal-dominant manner. The prevalence of this condition ranges from one in 6,000 to one in 12,000 people. All ethnic groups and both sexes are susceptible to being affected. The term "tuberous sclerosis complex" (TSC) was first described by von Recklinghausen in the year 1862,

which is nearly 150 years ago. There is a wide range of symptoms associated with TSC, and it can manifest itself in practically any organ in the body(1–3). It is a condition that involves multiple systems, including the brain, skin, heart, kidneys, eyes, lungs, and liver, and it manifests only in the later stages of childhood, TSC is a prominent genetic factor contributing to epilepsy, with around 85% of individuals affected by TSC experiencing seizures that frequently resist therapy(3–5). The initial manifestations of TSC include cardiac neoplasms and cortical tubers, which may be detected prenatally. Traditional indicators like cortical tubers or subependymal nodules, white matter irregularities, retinal anomalies, cardiac rhabdomyoma, lymphangioleiomyomatosis (LAM), and renal angiomyolipoma (AML) facilitate a tentative diagnosis, especially in the presence of cutaneous symptoms(6). The presentation of diseases across several organ systems might range significantly among even closely related individuals, and the multifaceted characteristics of the ailment can complicate clinical diagnosis. TSC remained underdiagnosed until the 1980s, when cases with milder symptoms started to get acknowledgment. Prior to the 1980s, the prevalence rates for TSC were reported to be between 1 in 100,000 and 1 in 200,000(7, 8). Mutations in either of the two genes, TSC1 or TSC2, which code for the proteins hamartin and tuberin, respectively, are recognized as the origins of TS. These proteins function as inhibitors of tumor growth, serving as agents that modulate cell proliferation and differentiation. The triad of epilepsy, intellectual impairment, and face angiofibromas was used to diagnose TSC. Skin involvement is essential for considering the diagnosis of TS(9–12).

To our knowledge, this is the first published case series of tuberous sclerosis complex from Somalia. We report two patients with characteristic multisystem manifestations demonstrated by MRI and ultrasonography, fulfilling the updated 2021 International Tuberous Sclerosis Complex diagnostic criteria despite the unavailability of molecular genetic testing. These cases underscored the diagnostic importance of imaging in resource-limited settings and the importance of early diagnosis of TSC for timely surveillance and multidisciplinary management. We also carry out a thorough review of the relevant literature to contextualize our results.

Case Presentation

Case 1: Severe Childhood Phenotype in a 5-Year-Old Female

A 5-year-old girl A 5-year-old female patient was referred to the pediatric neurology department from a regional clinic with a two-year history of recurrent and poorly controlled seizure episodes. According to the mother, the seizures started at about 3 years of age with peculiar episodes of inconsolable crying alternating with sudden, inappropriate laughing spells lasting several minutes, followed by generalized tonic-clonic convulsions with stiffening of the limbs, upward deviation of the eyes, and somnolence in the post-ictal period. The events had become more frequent over the past month, with nearly daily episodes despite treatment with phenobarbital prescribed at the referring facility, which initially occurred nearly daily episodes despite treatment with phenobarbital prescribed at the referring facility, initially occurring two to three times weekly. The mother also reported an exaggerated startle response from early infancy, when sudden auditory stimuli or unexpected touch would cause the child to violently jerk

his whole body and cry.

Developmental milestones were markedly delayed. The patient walked independently at the age of 18 months, much later than her unaffected siblings, and spoke her first meaningful words at the age of 3. She was very expressive at presentation in terms of language but was limited to about 10-15 single words; she was unable to make phrases, name common objects consistently, or communicate basic needs such as hunger, thirst, or pain. As soon as the child had acquired some speech, he started with frequent headaches, poorly localized, in the frontal region, two to three times a week, sometimes accompanied by vomiting, according to Mother.

Physical examination revealed several striking abnormalities. The patient demonstrating multiple symmetrical facial angiofibromas (blue arrowheads) distributed over the cheeks, nasolabial folds, chin, and glabella in a characteristic butterfly pattern. An ash leaf-shaped hypopigmented macule is present on the right cheek and chin (green arrowhead), consistent with cutaneous manifestations of tuberous sclerosis complex (Figure 1).

An ophthalmological examination revealed convergent strabismus of the left eye, which, according to the mother, had occurred about two years ago and was treated with patching in the regional clinic. Visual acuity could not be formally assessed due to patient non-cooperation, but visual tracking behavior was appropriate.

The respiratory history was normal in range and X ray reveals normal.

The patient was the second of three children born to parents who were not related to each other. Her two siblings were 8 and 3 years old and were healthy and developmentally normal. First- and second-degree relatives had no history of seizures, developmental disorders, autism, or similar dermatological conditions. Parents did not know of any TSC diagnosis in extended family members. Pregnancy was full term and uncomplicated with spontaneous vaginal delivery and birth weight of 3.2kg. The neonatal period was uneventful with no hypoxic events, no jaundice requiring phototherapy and no hospital admissions.

Given the constellation of seizures, developmental delay, characteristic cutaneous findings, and facial dysmorphism, clinical suspicion for a neurocutaneous syndrome—specifically tuberous sclerosis complex—was raised. Comprehensive imaging evaluation was undertaken.

Imaging Findings

Brain MRI performed on a 1.5-T scanner demonstrated multiple bilateral cortical and subcortical tubers appearing as hyperintense lesions on T2-weighted (T2W) and fluid-attenuated inversion recovery (FLAIR) images with blurring of the gray-white matter junction. Multiple radial migration lines extending from the periventricular white matter to the cortical surface were also identified, consistent with the characteristic

neuroimaging features of tuberous sclerosis complex (Figure 2).

Renal ultrasonography demonstrates well-defined, markedly hyperechoic cortical lesions within both kidneys (green arrowheads) as indicated (figure 3).

Echocardiography, performed to evaluate the persistent cough and exclude cardiac rhabdomyomas, was entirely unremarkable. The heart was structurally normal with no intracavitary masses, no valvular abnormalities, and normal biventricular function. A chest radiograph showed clear lung fields, no evidence of cystic changes, no pneumothorax, and no cardiomegaly.

She met multiple major diagnostic criteria for a provisional diagnosis of tuberous sclerosis complex (TSC): facial angiofibromas, hypomelanotic macules, cortical tubers, subependymal nodules and renal angiomyolipomas [10]. Levetiracetam was started for seizure control (20 mg/kg/day, BID divided) with a plan for gradual phenobarbital taper. The mother received intensive counseling regarding the chronic nature of TSC, the need for lifelong multidisciplinary surveillance and the genetic implications for future pregnancies. She was referred to pediatric dermatology for management of facial angiofibromas, pediatric nephrology for renal surveillance, and ophthalmology for a complete eye exam. But no local genetic testing was available.

Case 2: Adult-Onset Refractory Epilepsy in a 22-Year-Old Female

A 22-year-old female patient was admitted to the adult neurology clinic with a 14-year history of recurrent seizures. The seizures started at approximately age 8 years with episodes of behavioral arrest with vacant staring and lip smacking and right-hand automatisms lasting 30 to 60 seconds and occurring two to three times per month. In the following years, the semiology of the seizures evolved to include secondary generalization with tonic-clonic activity, and the frequency increased to weekly episodes despite treatment with multiple antiepileptic drugs, including carbamazepine, valproic acid, and lamotrigine, in various combinations and dosages.

Her intellectual functioning was normal and she managed to complete secondary education successfully. However, she reported ongoing problems with concentration and short term memory which she attributed to side effects of her antiepileptic medication. There was no history of developmental delay, autistic features or behavioral disturbance in childhood. There was no history of infantile spasms or myoclonic seizures.

Family history was notable for a maternal cousin with "multiple bumps on the face" and unspecified "learning difficulties," although no formal diagnosis of TSC had been established in this relative. The parents were first cousins, raising the possibility of autosomal recessive inheritance patterns for other conditions, though TSC itself follows autosomal dominant transmission. No other family members had seizures or similar dermatological findings.

Physical examination revealed multiple bilateral, symmetrically distributed, skin-colored to reddish papules over the cheeks, nasal bridge, and nasolabial folds, consistent with facial angiofibromas. A large, well-demarcated hypomelanotic (ash leaf) macule was also present over the posterior abdominal wall. No Shagreen patches, ungual fibromas, or dental enamel pits were identified (Figure 4 A and B).

Given the refractory epilepsy and characteristic cutaneous findings, TSC was suspected and comprehensive imaging was performed.

Imaging Findings

Brain MRI demonstrated multiple bilateral cortical and subcortical tubers with associated radial migration lines extending from the periventricular white matter to the cortex. Multiple calcified subependymal nodules were also identified along the lateral ventricular walls.

Abdominal ultrasonography demonstrates normal finding (Figure 5).

Chest radiograph was unremarkable with no evidence of cystic lung disease or pneumothorax. Echocardiography demonstrated no cardiac rhabdomyomas or structural abnormalities.

TSC was diagnosed based on the presence of multiple major criteria: facial angiofibromas, hypomelanotic macule, cortical tubers, subependymal nodules and renal angiomyolipomas [10]. Given the drug resistant nature of her seizures and the presence of discrete epileptogenic tubers, the patient was referred to a comprehensive epilepsy center for evaluation as a surgical candidate. The patient was counselled regarding the availability of mTOR inhibitor therapy (everolimus) for reduction of renal angiomyolipoma but access to this medication in Somalia is severely limited.

Discussion

The terms Bourneville disease, Pringle disease, and epiloia have all been used in the literature to describe tuberous sclerosis syndrome. All of these relate to the TSC entity, which varies greatly even among families. Unusual tumor-like growths (hamartomas) in the brain, skin, retina, and viscera are a hallmark of TSC. The existence of numerous sclerotic masses dispersed throughout the brain is specifically referred to as "tuberous sclerosis." The detection of hamartomas in several organ systems is the basis for the diagnosis of TSC(13,14). Nonetheless, the quantity of organs impacted and the intensity of tumors within a specific organ exhibit considerable variability. As a result, the disease's symptoms are extremely varied and unpredictable(10,15). TSC is an autosomal dominant disorder with variable expressivity and almost full penetrance. The mutated genes have been identified at two specific loci, namely the long arm of chromosome 9 (9q34), labeled TSC 1 (responsible for encoding hamartin), and the short arm of chromosome 16 (16p 13.3), labeled TSC 2 (responsible for encoding tuberin). The gene products create a tumor suppressor complex that converts Rheb (Ras homolog enriched in brain), part of the Ras superfamily, into the inactive state bound to guanosine diphosphate. In its guanosine triphosphate-bound active form, Rheb promotes the mammalian target of rapamycin (mTOR), a protein

kinase that is conserved through evolution and serves as a key regulator of cellular development. The genetic alterations lead to continuous mTOR activation, which contributes to the emergence of numerous neoplasms and hamartomas across different organs(5,16,17). The clinical diagnostic criteria for tuberous sclerosis complex (TSC) were initially revised and divided into major and minor features at the Second International Tuberous Sclerosis Complex Consensus Conference in Washington, DC, in 2012. These criteria were subsequently validated by the 2021 International Tuberous Sclerosis Complex Consensus Conference, which also provided updated recommendations for surveillance, multidisciplinary management and the role of molecular genetic testing for diagnosis. The present diagnostic criteria consist of 11 major and 7 minor clinical features as summarized in Table 1. The 2021 consensus describes a definite diagnosis of TSC as the presence of two major features or one major feature and two or more minor features or the identification of a pathogenic variant in TSC1 or TSC2(1,18,19). A possible diagnosis is defined by the presence of one major feature or two or more minor features. Although molecular genetic testing can confirm the diagnosis, it is not mandatory when patients fulfill the established clinical diagnostic criteria based on characteristic clinical and radiological findings (Table 1).

The clinical manifestations of TSC include those of neurological origin such as epilepsy, cortical dysplasia (ie, in the form of cortical tubers), subependymal nodules (SENs), subependymal giant cell astrocytomas (SEGAs), SEGAs associated with hydrocephalus, and autism spectrum disorder(3,20). In comparison to the other cutaneous manifestations of TSC, they are the most obvious and unattractive of the bunch. Especially in patients of school-going and adolescent age, when peer relationships become more important and self-concept matures, this facial dermatological condition has negative psychosocial impacts, such as anxiety, depression, low self-esteem, and self-isolation. These symptoms are especially prevalent in patients who are experiencing these conditions. Angiofibromas, ash leaf spots, confetti skin lesions, ungual fibromas, and shagreen patches are some of the other cutaneous manifestations that can be seen(21,22). Respiratory complications feature lymphangioleiomyomatosis; cardiovascular complications present as cardiac rhabdomyoma; gastrointestinal complications are characterized by dental enamel pits and intraoral fibromas, and renal complications comprise angiomyolipoma, numerous kidney cysts, and renal failure(23). These characteristics have been associated with a significant amount of educational, occupational, and psychosocial impairment, which includes frequent absences from school, poor academic achievement, decreased work opportunities, social dysfunction, and a significantly reduced quality of life in terms of health(18). Medical imaging modalities like ultrasonography, magnetic resonance imaging (MRI), and multidetector computed tomography (MDCT) are very useful tools for early screening, monitoring disease progression, and confirming the diagnosis of TSC. The consensus conference in 2012 recommended the following surveillance guidelines: (I) brain and abdominal MRI every 1-3 years until age 25, and more frequently in patients with SEGA or AMLs with progressive growth; (II) echocardiography every 1-3 years for pediatric TSC patients to monitor cardiac rhabdomyomas; (III) chest MDCT every 5-10 years for women at risk of LAM; and (IV) yearly comprehensive dermatologic, ophthalmologic, and pulmonary function testing(1,24,25). There is a multidisciplinary approach to the management of TSC. In general, it entails

preventing or reversing the development of the numerous hamartomatous lesions and the impact that these lesions have on practically every organ system in the body(26).

This case series adds to the scant literature of tuberous sclerosis complex (TSC) from sub-Saharan Africa and is, to the best of our knowledge, the first published report from Somalia. Both our patients had very different clinical presentations despite meeting the revised 2021 International Tuberous Sclerosis Complex Consensus diagnostic criteria, which underscores the wide phenotypic variability of this disorder. The pediatric patient had severe early-onset epilepsy, developmental delay and widespread multisystem involvement while the adult patient had preserved cognition despite long-standing drug-resistant epilepsy and characteristic neurocutaneous findings. These contrasting phenotypes emphasize that TSC should be considered in the differential diagnosis of patients presenting with epilepsy accompanied by characteristic cutaneous lesions or suggestive neuroimaging findings, irrespective of age or disease severity. Importantly, our cases demonstrate that comprehensive clinical evaluation combined with brain magnetic resonance imaging (MRI) and renal ultrasonography is sufficient to establish a definite diagnosis according to current international consensus criteria, even in healthcare settings where molecular genetic testing is unavailable.

The diagnosis in both patients was established by means of imaging. Classic cortical tubers, radial migration lines, and subependymal nodules were demonstrated on brain MRI, and angiomyolipomas on renal ultrasonography, providing several major criteria of diagnosis for TSC. These findings support the central role of radiological evaluation in confirming the diagnosis, in assessing multisystem involvement and in directing long-term surveillance. Knowledge of the characteristic imaging spectrum of TSC is important for radiologists and clinicians in many low-resource countries where advanced molecular diagnostics are not yet available, for early diagnosis, appropriate multidisciplinary management and improved patient outcomes.

Study Limitations

The principal limitation of this study is the absence of molecular confirmation of **TSC1** or **TSC2** pathogenic variants because genetic testing was not available locally. Histopathological confirmation was also not obtained because neither patient required surgical intervention or tissue sampling for diagnosis. However, both patients fulfilled several major clinical criteria according to the revised 2021 International Tuberous Sclerosis Complex Consensus guidelines which allowed a definitive diagnosis based on characteristic clinical and imaging findings. However, the current cases provide valuable clinical and radiological insight into TSC in an underrepresented population.

Conclusions

In conclusion, this case series highlights the heterogeneous clinical and radiological presentation of tuberous sclerosis complex, and underscores the fundamental role of MRI and ultrasonography in confirming the diagnosis based on the revised 2021 International Tuberous Sclerosis Complex Consensus criteria. The results of our study suggest that the characteristic clinical and imaging features

are still sufficient for diagnosis in the absence of molecular genetic testing. We present the first case series from Somalia. This study adds to the world literature and highlights the need for increased awareness of TSC among clinicians and radiologists for early diagnosis, appropriate surveillance, and multidisciplinary management, especially in resource-limited health-care settings.

Abbreviations

AML, Angiomyolipoma

FLAIR, Fluid-Attenuated Inversion Recovery

LAM, Lymphangioleiomyomatosis

MDCT, Multidetector Computed Tomography

MRI, Magnetic Resonance Imaging

mTOR, Mammalian Target of Rapamycin

SEN, Subependymal Nodule

SEGA, Subependymal Giant Cell Astrocytoma

T1W, T1-Weighted Imaging

T2W, T2-Weighted Imaging

TSC, Tuberous Sclerosis Complex

TSC1, Tuberous Sclerosis Complex 1

TSC2, Tuberous Sclerosis Complex 2

US, Ultrasonography

Declarations

Ethics Approval and Consent to Participate

Ethical approval for this case series was waived by the Institutional Ethics Committee of **Mogadishu Somali Turkish Recep Tayyip Erdoğan Training and Research Hospital**, as the study involved retrospective reporting of anonymized clinical data and imaging findings. Written informed consent for participation and publication was obtained from the parent/legal guardian of **Patient 1** and from **Patient 2**, who provided consent personally as an adult.

Consent for Publication

Written informed consent for publication of the clinical details and accompanying images was obtained from the parent/legal guardian of **Patient 1**, who was a minor, and from **Patient 2**, who was an adult at the time of publication. Copies of the signed consent forms are available for review by the Editor-in-Chief of this journal upon reasonable request.

Data Availability

All data generated or analyzed during this study are included in this published article. Additional information is available from the corresponding author upon reasonable request.

Competing Interests

The authors declare that they have no competing interests.

Funding

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

Authors' Contributions

All authors contributed to the conception and design of the study. Data collection, radiological interpretation, manuscript drafting, and critical revision were performed by the authors. All authors read and approved the final manuscript.

Acknowledgements

The authors sincerely thank the patients and their families for providing written informed consent for publication of this case series and the accompanying clinical and radiological images. The authors take full responsibility for the accuracy, interpretation, and scientific content of this manuscript.

References

1. Northrup H, Krueger DA, Roberds S, Smith K, Sampson J, Korf B, et al. Tuberous sclerosis complex diagnostic criteria update: Recommendations of the 2012 international tuberous sclerosis complex consensus conference. *Pediatr Neurol*. 2013;49(4):243–54.
2. Dzefi-tetty K, Edzie EK, Gorleku P, Piersson AD, Cudjoe O. Tuberous Sclerosis: A Case Report and Review of the Literature. 2021;13(1):1–8.
3. Dzefi-Tetty K, Edzie EK, Gorleku P, Piersson AD, Cudjoe O. Tuberous Sclerosis: A Case Report and Review of the Literature. *Cureus*. 2021;13(1):1–8.
4. Curatolo P, Jóźwiak S, Nabbout R. Management of epilepsy associated with tuberous sclerosis complex (TSC): Clinical recommendations. *Eur J Paediatr Neurol*. 2012;16(6):582–6.
5. Webb DW, Osborne JP. Tuberous sclerosis. *Arch Dis Child*. 1995;72(6):471–4.

6. von Ranke FM, Zanetti G, e Silva JLP, Neto CAA, Godoy MCB, Souza CA, et al. Tuberous Sclerosis Complex: State-of-the-Art Review with a Focus on Pulmonary Involvement. *Lung*. 2015;193(5):619–27.
7. Krueger DA, Northrup H, Krueger DA, Roberds S, Smith K, Sampson J, et al. Tuberous sclerosis complex surveillance and management: Recommendations of the 2012 international tuberous sclerosis complex consensus conference. *Pediatr Neurol*. 2013;49(4):255–65.
8. Frost M, Hulbert J. Clinical management of tuberous sclerosis complex over the lifetime of a patient. *Pediatr Heal Med Ther*. 2015;139.
9. Wataya-Kaneda M, Uemura M, Fujita K, Hirata H, Osuga K, Kagitani-Shimono K, et al. Tuberous sclerosis complex: Recent advances in manifestations and therapy. *Int J Urol*. 2017;24(9):681–91.
10. Dabora SL, Kwiatkowski DJ, Franz DN, Roberts PS, Nieto A, Chung J, et al. Mutational analysis in a cohort of 224 tuberous sclerosis patients indicates increased severity of TSC2, compared with TSC1, disease in multiple organs. *Am J Hum Genet*. 2001;68(1):64–80.
11. Jóźwiak S, Kotulska K, Domańska-Pakieła D, Łojczyk B, Syczewska M, Chmielewski D, et al. Antiepileptic treatment before the onset of seizures reduces epilepsy severity and risk of mental retardation in infants with tuberous sclerosis complex. *Eur J Paediatr Neurol*. 2011;15(5):424–31.
12. Schepis C. The tuberous sclerosis complex. *Dermatological Cryosurgery and Cryotherapy*. 2016;615–7.
13. Hake S. Cutaneous Manifestations of Tuberous Sclerosis. 2010;200–4.
14. Sarkar S, Khaitan T, Sinha R, Kabiraj A. Tuberous sclerosis complex: A case report. 2016;
15. Mongrain V, Doesburg NH Van, Rypens F, Fallet-bianco C, Maassen J, Dufort-gervais J, et al. A case report of severe tuberous sclerosis complex detected in utero and linked to a novel duplication in the TSC2 gene. 2020;1–9.
16. Cheng TS. Review Article Tuberous sclerosis complex: an update. 2012;61–7.
17. Manuscript A. The TSC1. *Genetics*. 2009;290(5497):1717–21.
18. Zöllner JP, Conradi N, Sauter M, Knuf M, Knake S, Kurlemann G, et al. Quality of life and its predictors in adults with tuberous sclerosis complex (TSC): a multicentre cohort study from Germany. 2021;3.
19. Northrup H, Aronow ME, Bebin EM, Bissler J, Darling TN, Vries PJ De, et al. Pediatric Neurology Updated International Tuberous Sclerosis Complex Diagnostic Criteria and Surveillance and Management Recommendations. 2021;123.
20. Lienda M, Mwila M, Sichula C, Kabengele C, Akombwa M, Zulu C, et al. Diagnosis and Management of Tuberous Sclerosis Complex in a Resource-Limited Setting — A Case Report of a 14-Year-Old Female Zambian Adolescent. 2025;0–5.
21. Monaghan M, Takhar P, Langlands L, Knuf M, Amin S. Impact of facial angiofibromas in tuberous sclerosis complex and reported e cacy of available treatments.
22. Vasani RJ. Facial Angiofibromas of Tuberous Sclerosis Treated with Topical Sirolimus in an Indian Patient. 2015;60(2):165–9.

23. Mahadevappa M, Patil V, Poornima KS. Two Cases of Tuberous Sclerosis With Multiple Rhabdomyoma Succumbing to Sudden Cardiac Death. 2021;
24. Alshoabi SA, Hamid AM, Alhazmi FH, Qurashi AA, Abdulaal OM, Aloufi KM, et al. Diagnostic features of tuberous sclerosis complex: case report and literature review. 12(1):846–61.
25. Kowal DJ. Comprehensive Imaging Manifestations of Tuberous Sclerosis. 2015;(May):933–43.
26. Franz DN. Diagnosis and management of tuberous sclerosis complex. Semin Pediatr Neurol [Internet]. 1998;5(4):253–68. Available from: <https://www.sciencedirect.com/science/article/pii/S1071909198800041>

Table

Table 1 is available in the supplementary files section

Figures

Figure 1-

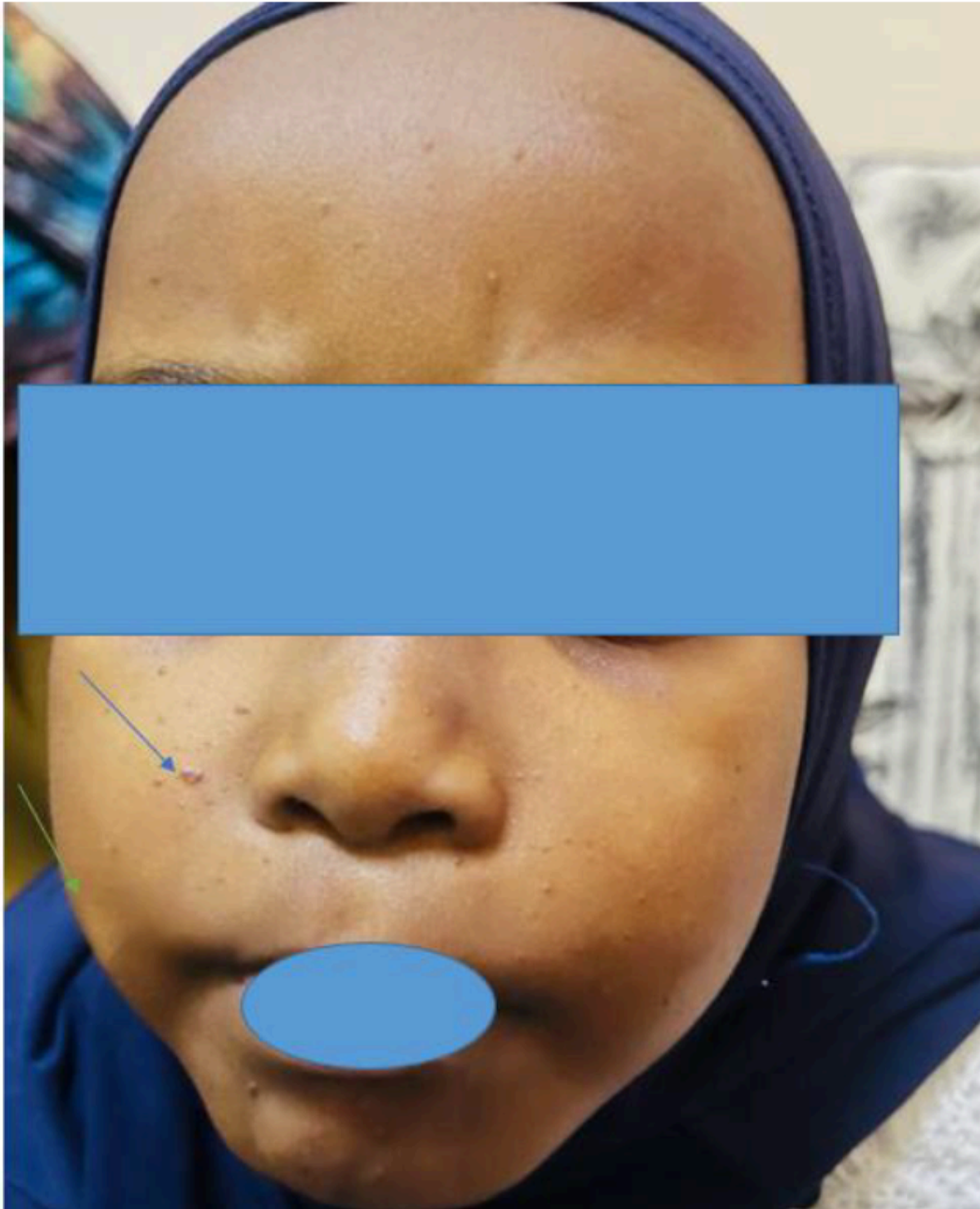


Figure 1

Clinical photograph showing the characteristic facial manifestations of tuberous sclerosis complex, including multiple facial angiofibromas (blue arrowheads) involving the cheeks, nasolabial folds, chin, and glabella, and an ash leaf-shaped hypopigmented macule (green arrowhead) on the right cheek and chin.

Figure 2-

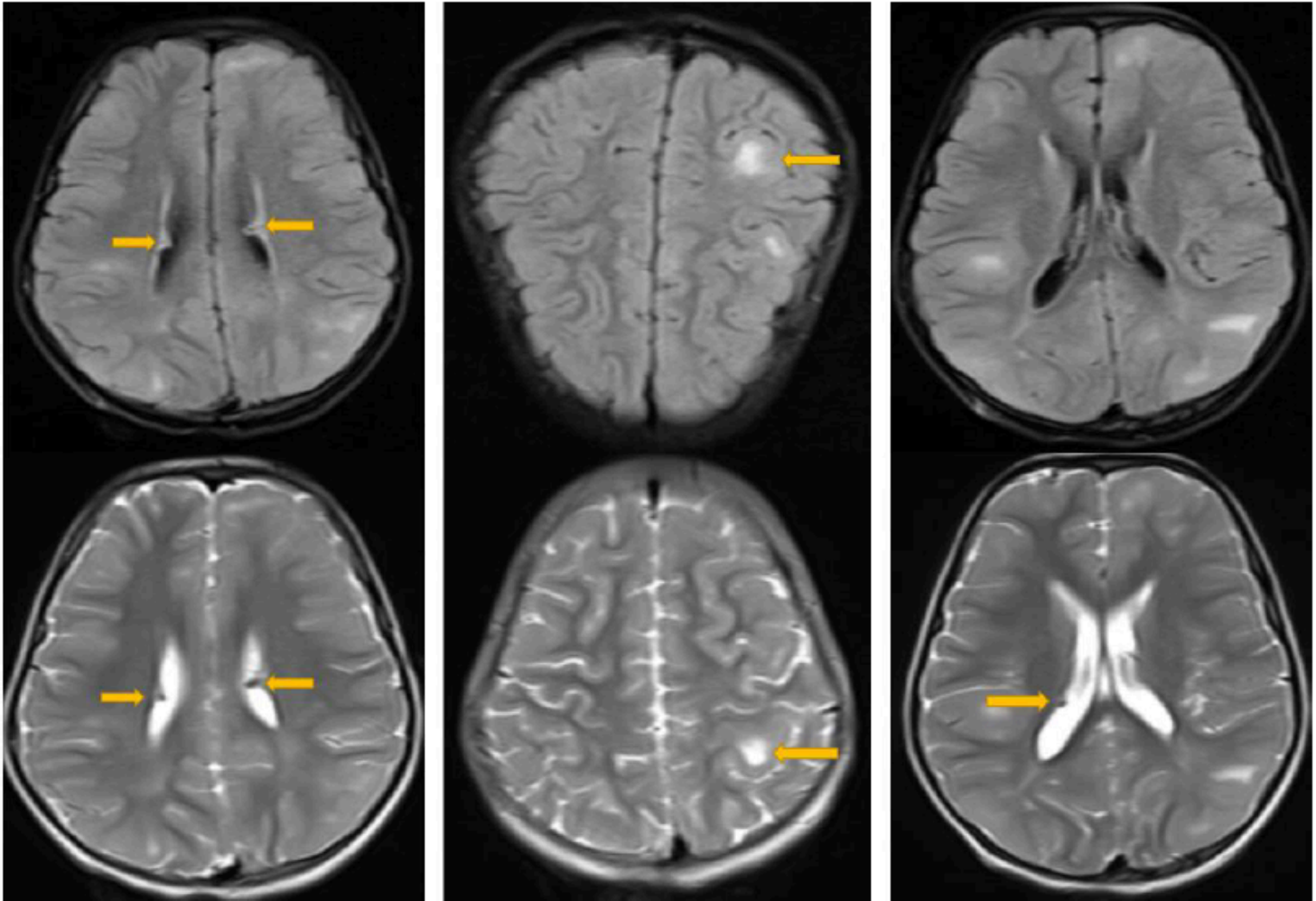


Figure 2

Axial **T2-weighted (T2W)** and **FLAIR** MR images demonstrate multiple bilateral cortical and subcortical tubers (orange arrowheads) appearing as hyperintense lesions with blurring of the gray-white matter interface. Hyperintense radial migration lines (radial band sign) extend from the periventricular white matter to the cortical surface, representing disrupted neuronal migration.

Figure 3-

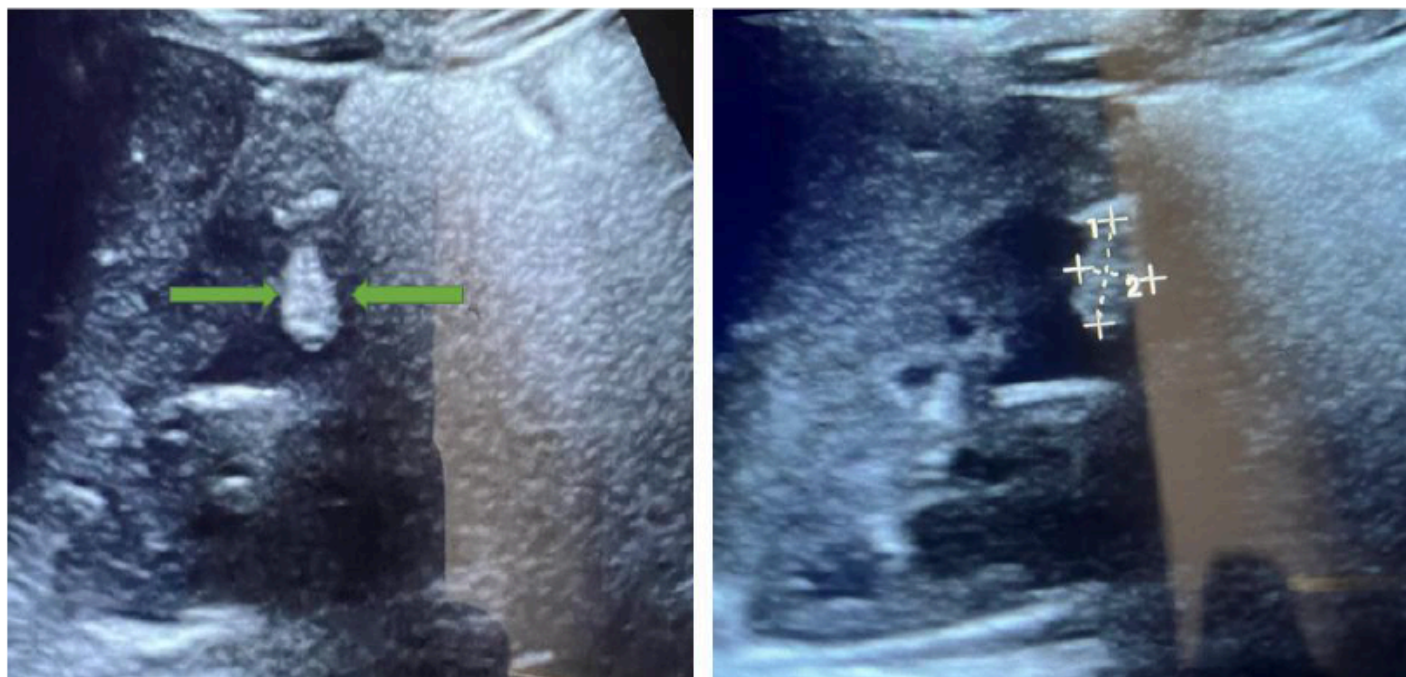


Figure 3

Renal ultrasonography demonstrating a well-defined hyperechoic cortical renal lesion (green arrowheads), consistent with a renal angiomyolipoma. Similar lesions were identified bilaterally.

Figure 4 A&B

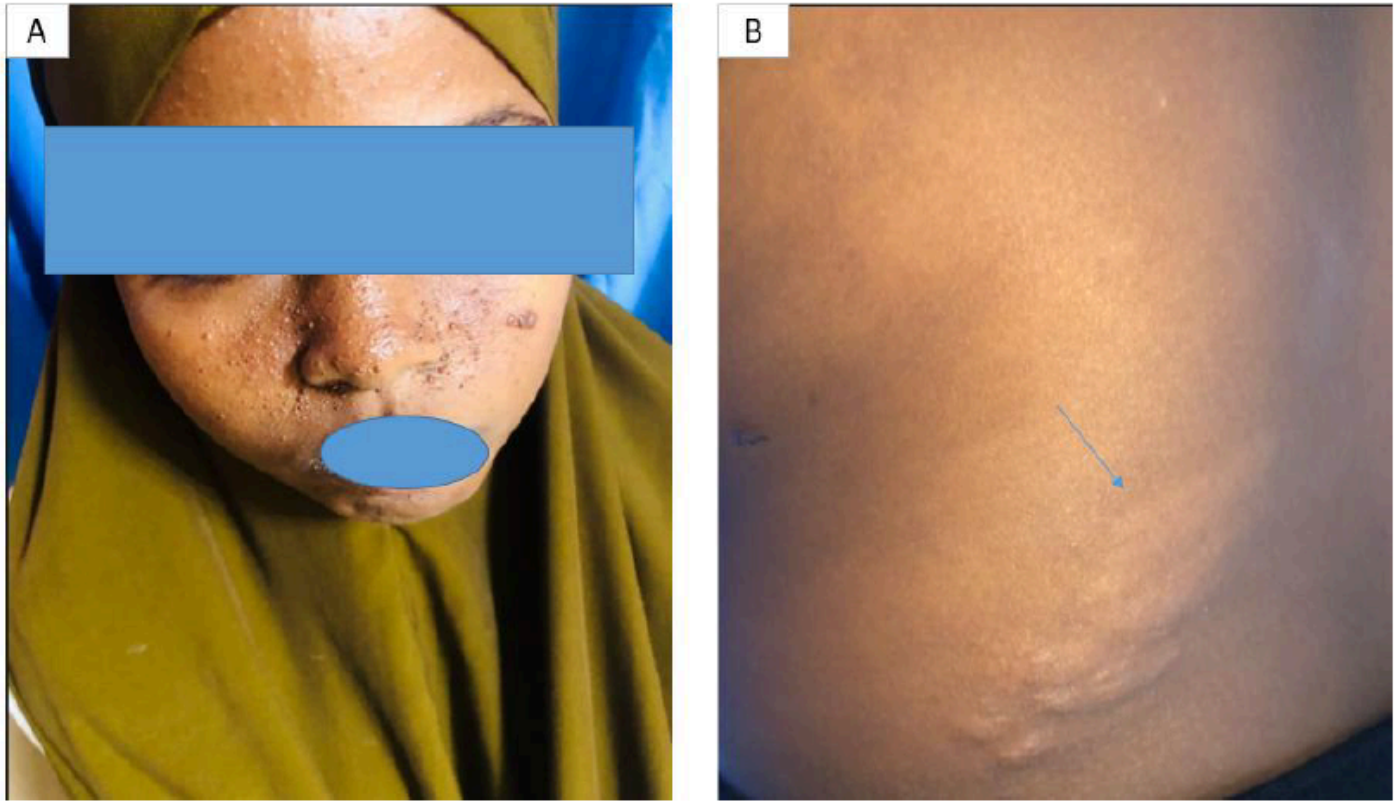


Figure 4

(A) Clinical photograph demonstrating multiple bilateral facial angiofibromas involving the cheeks, nasal bridge, and nasolabial folds, characteristic of **tuberous sclerosis complex**. **(B)** Clinical photograph of the posterior abdominal wall showing a well-defined hypomelanotic (ash leaf) macule (blue arrow), another characteristic cutaneous manifestation of tuberous sclerosis complex.

Figure5-

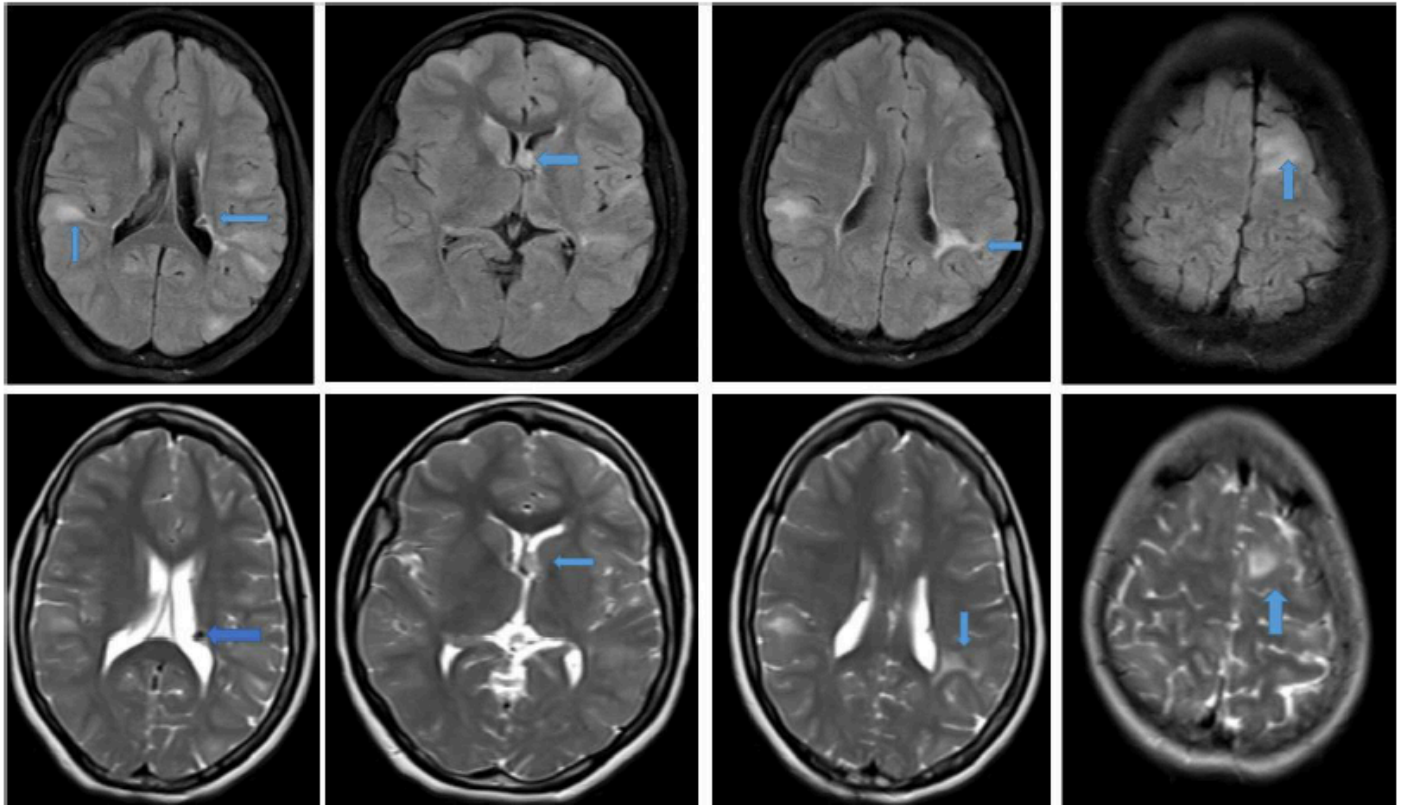


Figure 5

Axial **FLAIR** (upper row) and **T2-weighted** (lower row) MR images demonstrate multiple bilateral cortical and subcortical tubers (blue arrows) with associated radial migration lines extending from the periventricular white matter to the cortical surface. Multiple calcified subependymal nodules are present along the lateral ventricular walls.

Supplementary Files

This is a list of supplementary files associated with this preprint. Click to download.

- [TableTUBEROUSSCLEROSIS.docx](#)