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TUBEROUS SCLEROSIS: A RARE CASE REPORT

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Abstract

Background:
Tuberous sclerosis complex (TSC) is a rare autosomal dominant neurocutaneous disorder caused by mutations in either the TSC1 or TSC2 genes. It is characterized by hamartomas affecting multiple organ systems, most commonly the brain, skin, kidneys, heart, and lungs. Although seizures and neurodevelopmental delays are common, presentations can be variable, and dermatological signs may serve as early clues.

Case Presentation:
A 6-year-old male child presented with abdominal pain at a previous surgical site and notable facial skin lesions. On examination, characteristic dermatological findings included a hypopigmented ash leaf macule, facial angiofibromas (adenoma sebaceum), and a shagreen patch. Radiological investigations revealed bilateral renal angiomyolipomas, subependymal calcified nodules, and cortical tubers. Despite the absence of seizures, diagnostic criteria for TSC were fulfilled based on dermatological and neuroimaging findings.

Conclusion:
This case underscores the importance of thorough dermatological and imaging assessments in the early diagnosis of TSC, even in the absence of classic neurological symptoms. Multidisciplinary management is crucial to improving long-term outcomes.

Keywords: Tuberous Sclerosis Complex, Ash Leaf Macule, Shagreen Patch, Angiomyolipoma, Cortical Tuber, Subependymal Nodule, Pediatric Neurocutaneous Syndrome

Introduction

Tuberous sclerosis complex (TSC) is a multisystem genetic disorder resulting from pathogenic variants in either the TSC1 gene on chromosome 9q34 (encoding hamartin) or the TSC2 gene on chromosome 16p13 (encoding tuberin), both of which regulate the mTOR pathway [1]. Loss of

function in this pathway leads to unregulated cellular growth, resulting in hamartomas in various organs, including the brain, kidneys, lungs, heart, and skin.

The classical Vogt triad of seizures, intellectual disability, and facial angiofibromas is present in only about 29% of cases [2]. Cutaneous manifestations are often the earliest and most accessible signs of TSC, making dermatological examination crucial for early suspicion. The estimated incidence is approximately 1 in 6,000–10,000 live births globally, and thus classified as a rare disease [3].

Case Presentation

Figure 1. Facial angiofibromas in a child with TSC

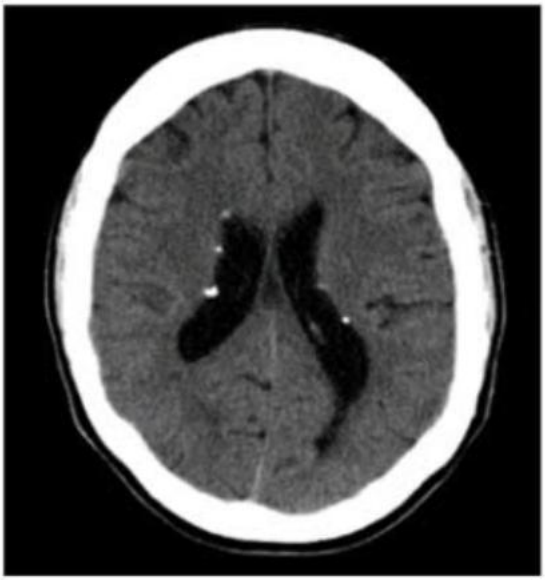

Figure 2. NCCT head showing subependymal calcified nodules

Below are clinical and radiological findings supporting the diagnosis of TSC:
A 6-year-old male child, weighing 19.5 kg, was referred to the Department of Pediatrics with complaints of abdominal pain localized to a prior surgical scar from the excision of a hemangioma on the anterior abdominal wall. Parents also reported multiple facial skin lesions that had gradually increased over time.

Cutaneous Examination:
- One hypopigmented ash leaf macule (>6 mm) observed under Wood's lamp.
- Multiple reddish-brown papular lesions over the nasolabial folds and cheeks, consistent with facial angiofibromas (adenoma sebaceum).
- A leathery, thickened area with orange peel texture (shagreen patch) over the lumbosacral region.

Systemic Examination:
- Normal findings on general, cardiovascular, and respiratory exams.
- No history of seizures, delayed milestones, or cognitive impairment.

Investigations:
- Ultrasound Abdomen: Bilateral kidneys showed multiple hyperechoic lesions consistent with angiomyolipomas [4].
- NCCT Brain: Subependymal calcified nodules along the lateral ventricles and cortical tubers in the left frontoparietotemporal lobe [5].
- Echocardiography, ophthalmic examination, and EEG: Normal.

Diagnosis:
Based on the 2021 International TSC Consensus Diagnostic Criteria [1], the presence of ≥2 major features (facial angiofibromas, hypomelanotic macule, shagreen patch, renal angiomyolipomas, and subependymal nodules) confirmed the diagnosis of definite TSC.

Management:-
A multidisciplinary management plan was initiated involving:
- Pediatrics: Clinical monitoring for developmental progress, seizures, and systemic involvement.
- Dermatology: Education regarding skin care; consideration of topical treatments for cosmetic improvement.
- Radiology: Periodic imaging to monitor renal and intracranial lesions.
- Genetic Counseling: Parents were advised genetic testing and familial screening.
- Neurology Follow-Up: Baseline EEG was advised despite no seizure history.

mTOR inhibitors like everolimus were not initiated due to the small size of renal lesions and absence of SEGA. Regular follow-up was planned every 6 months.

Discussion:-
TSC displays variable expressivity and incomplete penetrance. Although neurological symptoms are hallmark features, 15–20% of patients may present without seizures initially [2]. The dermatologic signs such as hypomelanotic macules and angiofibromas often manifest earlier, making them vital in raising suspicion [3].

Renal angiomyolipomas are present in up to 80% of patients by adulthood and may lead to life-threatening hemorrhage if undetected [4]. Subependymal nodules carry a risk of progression to subependymal giant cell astrocytomas (SEGA), necessitating serial neuroimaging [5].

Early diagnosis allows timely surveillance and, if necessary, initiation of mTOR inhibitor therapy, which has been shown to significantly reduce tumor volume and improve outcomes [6].

Conclusion:-

This case highlights the wide clinical spectrum of TSC and the critical role of dermatological and radiological assessments in diagnosis. Pediatricians and primary care providers should be vigilant for early cutaneous clues. Timely diagnosis allows for appropriate monitoring and improves prognosis through multidisciplinary care.

Conflict of Interest:-

The authors declare no conflict of interest related to this case report...

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