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An Unusual Cutaneous Presentation of Tuberous Sclerosis with Normal Neurological Function

Dr. Mohit Singla, Dr. Arashdeep Virk, Dr. Prasun Bhattacharjee

Pg resident pediatrics
Email- Msingla6688@gmail.com
Institute: Ananta institute of medical sciences and research centre. Rajsamand . Rajastan

Pg resident pediatrics
Email- arashdeepvirk22@gmail.com
Institute: Ananta institute of medical sciences and research centre. Rajsamand . Rajastan

Professor and Head of department
Department of pediatrics
Email- prasun9999@gmail.com
Institute- Ananta institute of medical sciences and research centre. Rajsamand . Rajastan

Abstract

Tuberous Sclerosis Complex (TSC) is a rare neuro-cutaneous disorder characterized by the presence of hamartomas in multiple organ systems. The diagnosis is based on established major and minor clinical criteria. Cutaneous manifestations play a crucial role in early recognition. Here, we present a rare case of a 6-year-old male with fibrous hamartoma of the abdominal wall and other cutaneous signs but without any neurological symptoms, an uncommon presentation of TSC. This case highlights the necessity of considering TSC in pediatric patients with unusual subcutaneous masses and characteristic skin lesions, even in the absence of neurological deficits. A multidisciplinary approach, including dermatological, radiological, and genetic evaluations, is essential for accurate diagnosis and long-term management.

Keywords: Tuberous sclerosis, fibrous hamartoma, cortical tubers, neurocutaneous disorder, pediatric neurogenetics.

Introduction

Tuberous Sclerosis Complex (TSC) is an autosomal dominant neurocutaneous syndrome with incidence of approximately 1 in 5000 children (1). It is caused by mutations of the *TSC1* or *TSC2* genes, leading to abnormal cellular growth and the formation of benign lesions (hamartomas) in various organs, including the skin, brain, kidneys, heart, and lungs (2). The diagnosis of TSC is based on a combination of major and minor clinical criteria, as per the 2012 International Tuberous Sclerosis Complex Consensus which include Brain MRI, ECHO, renal sonography and systemic evaluation (3).

Cutaneous markers of Tuberous sclerosis, such as facial angiofibroma's shagreen patch and Ash Leaf spots(hypomelanotic macules), are often the earliest detectable signs and may

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precede neurological manifestations (3,4). However, in some cases, patients may present with non-neurological symptoms, such as subcutaneous masses or renal cysts, necessitating careful clinical assessment (5). Here, we present a case of TSC in a child with a fibrous hamartoma of the abdominal wall, renal cysts, and other characteristic cutaneous findings but without neurological symptoms, emphasizing the need for dermatological vigilance in early diagnosis.

Case Report

Patient Demographics:

we present a case of 6 yrs old male child with tuberous sclerosis in which child presented with fibrous hamartoma of abdominal wall with other cutaneous signs without neurological symptoms which is a very unusual presentation.

Clinical Findings:

- Child was apparently asymptomatic till 1yr of age when he developed a progressively increasing painful swelling at anterior abdominal region extending to anterior superior iliac spine.
- The swelling measured approximately 6x5 cm, had an irregular granular surface, and was covered with normal overlying skin.
- No history of learning disabilities, behavioural issues, or seizures.
- Family history was unremarkable.
- On general physical examination:
    - Multiple well-defined brownish papules arranged in a butterfly pattern over the nose and cheeks.
    - Hypomelanotic macules were noted on the back and legs.
    - Elevated skin lesion at back(shangreen patch)
    - Ocular examination was normal
    - No other systemic abnormalities were identified.

Investigations:

- **CT Scan (Abdomen):** Subcutaneous mass around anterior abdominal wall with suspected arteriovenous malformation and predominant fibrosis
- **Histopathology (Excision Biopsy):** Fibrotic infiltrating hamartoma.
- **USG Kidneys:** Bilateral renal cysts.
- **Echocardiography:** Normal findings.

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- **CT Brain:** Multiple cortical tubers, Subependymal nodules confirming TSC diagnosis based on clinical criteria.
- **Genetic Testing:** Not performed but recommended to confirm mutations in *TSC1* or *TSC2* genes.

Management:
The patient was operated to remove the abdominal mass. The post-operative period was uneventful, and the patient is currently under regular follow-up for monitoring of potential disease progression. Genetic counselling was recommended for family members.

Discussion

TSC is typically diagnosed based on a combination of dermatological, neurological, renal, and cardiac findings (6,7). The 2012 diagnostic criteria include both major and minor features. Diagnosis is confirmed with either two major or one major and two minor features (3).

This patient met the diagnostic criteria for TSC due to the presence of multiple cortical tubers (major criterion), facial angiofibroma's (major criterion), shangreen patch (major criteria), subependymal nodules (major criteria) and bilateral renal cysts (minor criterion). However, the absence of neurological symptoms is an unusual feature in TSC and has been rarely documented in literature (7,8).

Table 1: Diagnostic Criteria and Findings in Our Patient

Diagnostic Criteria (2012)	Our Patient
Major Criteria:	
Cortical tubers	Present
Facial angiofibroma	Present
Fibrous cephalic plaque	Absent
Hypomelanotic macules(Ash Leaf Spots)	Present
Shagreen patch	Present
Multiple retinal hamartomas	Absent
Subependymal nodules	Present
Cardiac rhabdomyomas	Absent
Lymphangioleiomyomatosis	Absent
Renal angiomyolipoma	Absent

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Ungual fibromas	Absent
Minor Criteria:	
Bilateral renal cysts	Present
Dental pits	absent
Multiple intraoral fibromas	Absent
Confetti skin lesions	Absent
Retinal achromic patches	Absent
Non renal hamartoma	Absent

The cutaneous markers of TSC often precede neurological symptoms, which is why dermatological examination plays a pivotal role in early detection (9). In our case, despite the presence of multiple cortical tubers, the child exhibited no neurological deficits. Long-term follow-up is necessary as neurological symptoms, including epilepsy or cognitive impairment, may develop later in life.

The presence of renal cysts also highlights the importance of nephrological monitoring in TSC patients (10). Tuberous sclerosis associated renal disease can range from benign cysts to more severe angiomyolipoma, which may require intervention. Regular ultrasound screenings are essential in such cases.

Conclusion

TSC should be considered in children presenting with cutaneous features and unexplained subcutaneous masses, even in the absence of neurological symptoms. Early recognition is crucial for timely intervention and monitoring of systemic complications. In cases of fibrous hamartoma, a thorough evaluation for underlying TSC is warranteda. A multidisciplinary approach involving dermatologists, neurologists, geneticists, and nephrologists is vital for optimal long-term management.

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