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Facial Angiofibromas as a Diagnostic Clue to Tuberous Sclerosis in a Child: Images in Medicine

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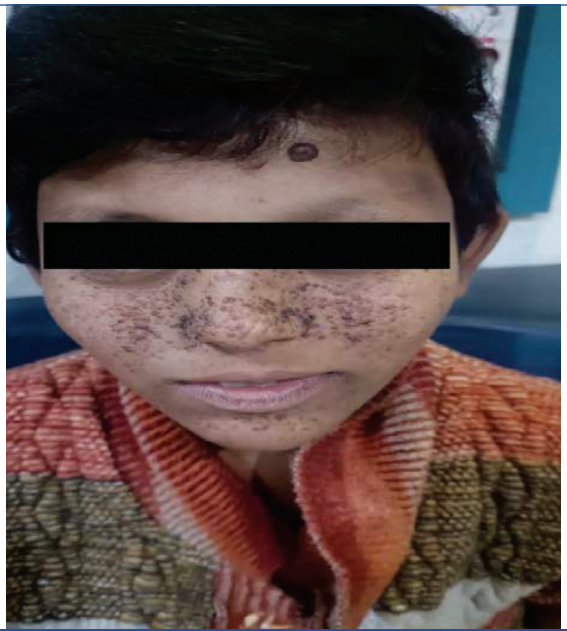
Tuberous Sclerosis Complex (TSC) is an autosomal dominant multisystem neurocutaneous disorder caused by mutations in the TSC1 or TSC2 genes. According to the updated International TSC Consensus diagnostic criteria, the presence of major clinical features such as facial angiofibromas, cortical dysplasia, hypomelanotic macules, and seizures strongly supports the diagnosis [1-3]. Cutaneous manifestations, especially facial angiofibromas, are some of the earliest and most characteristic disease manifestations and often provide an important clue to the diagnosis.

A 10-year-old female child who had several reddish-brown elevated lesions over her central face for the previous six months was brought to the outpatient clinic. The lesions began as small papules over the nose and gradually increased in number and size, spreading symmetrically over both cheeks and the nasal bridge. For the previous year, the child had experienced recurring generalised seizures that happened roughly twice or three times a month. Each episode, which lasted roughly one to two minutes, was marked by widespread tonic-clonic movements and loss of consciousness. Although they were self-limiting, the seizures occasionally needed medical attention. There was a recent decline in academic performance. A contributing family history was absent.

On examination, numerous firm, dome-shaped, reddish-brown papules were seen symmetrically distributed over the nose and bilateral malar areas, giving the classical appearance of facial angiofibromas (previously termed “adenoma sebaceum”). A well-defined hyperpigmented plaque of approximately 1×1 cm was seen on the central forehead [Table/Fig-1]. Furthermore, many hyperpigmented papular lesions were symmetrically dispersed across the nose, bilateral malar regions, and perinasal area. No comedones, pustules, or inflammatory acneiform lesions were present. Neurological involvement was evident in the form of recurrent generalised seizures and recent decline in academic performance. No obvious cardiac or renal manifestations were clinically evident at presentation, although detailed systemic evaluation could not be performed due to financial constraints.



[Table/Fig-1]: A child with TSC with several reddish-brown papular facial angiofibromas symmetrically distributed over the nose and malar regions.

In view of the characteristic facial lesions along with a history of seizures a neurocutaneous syndrome was strongly considered. Due to financial constraints neuroimaging was not possible. Nonetheless, the diagnosis was established clinically according to the updated International TSC diagnostic criteria proposed by Northrup and Krueger, in which facial angiofibromas and neurological manifestations such as seizures constitute major clinical features supportive of TSC diagnosis [2,3]. Differential diagnoses included multiple trichoepitheliomas, rosacea, sebaceous hyperplasia, angiokeratomas, nevus sebaceous, and acne vulgaris. However, because there were no family history and no usual distribution around the nasolabial folds, numerous trichoepitheliomas were ruled out. Since there were no signs of flushing, telangiectasia, or erythema, rosacea was ruled out. The lack of yellowish umbilicated papules made sebaceous hyperplasia unlikely. Since the lesions lacked a keratotic surface and were not violaceous, angiokeratomas were ruled out. Because there was no linear or plaque-like lesion evident from birth, Nevus sebaceous was ruled out. Due to the early age of onset and lack of comedones, pustules, or inflammatory lesions, acne vulgaris was deemed implausible. The presence of multiple symmetrical facial angiofibromas along with seizure history strongly confirmed the diagnosis of TSC.

At the time of presentation, the child was not having active seizure episodes. She was given oral sodium valproate in divided doses of 10-15 mg/kg/day to control her seizures. The cutaneous lesions were treated with topical sirolimus 0.1% ointment once daily. The patient is receiving frequent check-ups, and treatment has been ongoing for the past three months. She is currently under regular neurological and nephrological follow-up for evaluation of systemic involvement.

The TSC is an autosomal dominant neurocutaneous disorder caused by mutations in the TSC1 or TSC2 genes, which cause dysregulation of the mammalian Target of Rapamycin (mTOR) pathway and the development of hamartomas in several organ systems, including the brain, skin, and kidneys [1,2]. Cutaneous symptoms are frequently the first and most distinguishable features, with facial angiofibromas affecting approximately 75% of patients, who are typically aged three to 10 years [3].

Neurological involvement, particularly epilepsy, occurs in up to 80-90% of patients and has a substantial impact on long-term neurodevelopmental prognosis [4]. However, early detection

is sometimes delayed because facial angiofibromas are often misinterpreted as acne or other benign dermatological conditions [5]. This case emphasises the value of a thorough dermatological examination as a straightforward, non-invasive screening method for children who are having seizures. Early diagnosis can be facilitated by quick neuroimaging and systemic assessment when distinctive cutaneous characteristics are identified [2,6].

Topical sirolimus, an mTOR inhibitor, has demonstrated significant improvement in the size, erythema, and cosmetic appearance of facial angiofibromas in patients with TSC, with good long-term safety and tolerability [7,8]. Early identification and multidisciplinary management are essential to prevent complications and improve long-term outcomes [6].

This case highlights how facial angiofibromas can function as an early clinical indication of TSC, and how early diagnosis and effective treatment can be greatly aided by their timely detection.

Ethical Consideration: The case report was prepared in accordance with institutional ethical standards and the principles of the Declaration of Helsinki.

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Diagnosis: Tuberous Sclerosis Complex (TSC)

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