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A Case Series

Tuberous Sclerosis Complex Presenting with Abnormal Body Movements: A Case Series

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Abstract:

Tuberous sclerosis complex (TSC) is a rare autosomal dominant neurocutaneous disorder characterized by multisystem hamartomatous involvement, most commonly affecting the brain and skin. Neurological manifestations such as seizures and abnormal body movements are frequently the earliest presenting features, while characteristic cutaneous lesions provide crucial diagnostic clues.

We report a case series of three female patients with tuberous sclerosis complex presenting with abnormal body movements and classical dermatological and neuroimaging findings. The first case was a 3-year-old female child with recurrent abnormal body movements and hypomelanotic macules, with MRI brain demonstrating cortical tubers. The second case involved a 16-year-old female presenting with intellectual disability, behavioral abnormalities, abnormal involuntary movements, facial angiofibromas, a shagreen patch, and MRI brain findings of cortical tubers with subependymal nodules. The third case involved an 8-year-old female presenting with recurrent abnormal body movements and seizures, associated with hypomelanotic macules and characteristic MRI brain findings.

These cases highlight the importance of combined neurological, dermatological, and radiological evaluation for early diagnosis and management of tuberous sclerosis complex.

Keywords: Tuberous sclerosis complex; Ash-leaf macules; Facial angiofibromas; Shagreen patch; Cortical tubers; Case series

Introduction

Tuberous sclerosis complex (TSC) is a genetic neurocutaneous disorder resulting from pathogenic variants in the TSC1 or TSC2 genes, which cause dysregulation of the mammalian target of rapamycin (mTOR) pathway [1,2]. The disease is marked by the growth of hamartomas in many organ systems, with the most common and clinically important being the nervous system and the skin [3].

Neurological symptoms include seizures, unusual body movements, developmental delays, intellectual disabilities, and behavioural problems [4–6]. Hypomelanotic macules, facial angiofibromas, shagreen patches, and periungual fibromas are important signs that can show up early in life [7–9].

An MRI of the brain is necessary to find typical intracranial abnormalities, such as cortical tubers and subependymal nodules,

that strongly support the diagnosis [10–12]. We present two cases of tuberous sclerosis complex that exemplify the classical neurological, dermatological, and radiological manifestations of the condition.

Case Presentation

Case 1

A 3-year-old female child, Zikra, presented with recurrent abnormal body movements noted since early childhood. The episodes were sudden, involuntary, and recurrent, suggestive of seizure activity. There was no significant perinatal history.

Dermatological examination revealed multiple well-defined hypopigmented macules over the limb and trunk. The lesions were non-scaly, oval to irregular in shape, consistent with ash-leaf macules (Figures 1A and 1B).

Neurological examination demonstrated abnormal motor activity.

MRI brain revealed multiple cortical and subcortical hyperintense lesions on T2-weighted and FLAIR sequences, consistent with cortical tubers (Figure 2A).

Based on the presence of seizures, hypomelanotic macules, and characteristic MRI findings, a diagnosis of tuberous sclerosis complex was established.

Case 2

A 16-year-old female, Shabana, presented with intellectual disability, behavioral abnormalities, and abnormal involuntary body movements since childhood. Developmental delay and behavioral disturbances, including irritability and impaired social interaction, were reported.

Dermatological examination revealed multiple reddish-brown papules symmetrically distributed over the malar region, nasolabial folds, and chin, consistent with facial angiofibromas (Figures 3A and 3B). A thickened, hyperpigmented plaque with a rough, peau-d'orange appearance was noted over the trunk, consistent with a shagreen patch (Figures 4A and 4B).

Neurological evaluation showed abnormal involuntary movements suggestive of seizure activity.

MRI brain demonstrated multiple cortical tubers along with subependymal nodules lining the lateral ventricles (Figure 5A).

Based on the combination of neurological manifestations, characteristic cutaneous lesions, and classical MRI findings, a diagnosis of tuberous sclerosis complex was made.

Case 3

An 8-year-old female child, Sana, presented with recurrent episodes of abnormal body movements and seizures for the past two years. The episodes were sudden in onset, involuntary, and associated with brief loss of awareness. There was no significant antenatal or perinatal history, and no similar illness in the family.

Dermatological examination revealed multiple hypopigmented macules over the trunk and lower limbs, consistent with ash-leaf macules. Few small erythematous papules were noted over the nasal bridge and cheeks, suggestive of early facial angiofibromas.

Neurological examination showed mild developmental delay with poor attention span. MRI brain revealed multiple cortical tubers involving the frontal and parietal lobes along with small subependymal nodules along the lateral ventricles.

Based on the presence of recurrent seizures, characteristic cutaneous lesions, and supportive neuroimaging findings, a diagnosis of tuberous sclerosis complex was made.

Discussion

Tuberous sclerosis complex is a leading cause of epilepsy and neurodevelopmental disorders in children and adolescents [13–15]. Seizures and abnormal body movements often represent the earliest

clinical manifestations and may precede recognition of other systemic features [16].

Cutaneous manifestations are present in the majority of patients and serve as vital diagnostic markers [17–19]. Ash-leaf macules frequently represent the initial lesions, whereas facial angiofibromas and shagreen patches generally manifest subsequently during childhood or adolescence. All three patients in this case series had classic skin problems that made the diagnosis very likely.

MRI of the brain is very important for confirming the diagnosis because it shows cortical tubers and subependymal nodules, which are very typical of TSC [20–22]. Early diagnosis is crucial, as delayed recognition may result in poor seizure control and adverse neurodevelopmental outcomes [23–25].

Figures

Figure 1A–1B: Hypomelanotic (ash-leaf) macules in Case 1.

Figure 2A: MRI brain of Case 1 showing cortical tubers.

Figure 3A–3B: Facial angiofibromas in Case 2.

Figure 4A–4B: Shagreen patch over the trunk in Case 2.

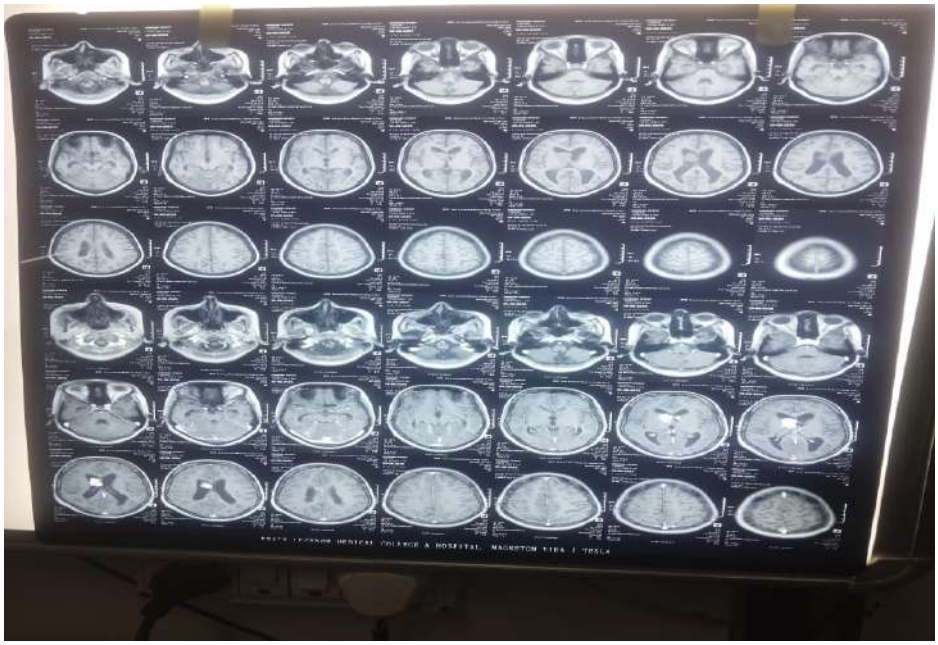

Figure 5A: MRI brain of Case 2 showing cortical tubers and subependymal nodules.

Conclusion

This case series highlights the classical neurological, dermatological, and radiological manifestations of tuberous sclerosis complex. Recognition of characteristic skin lesions in association with abnormal body movements and supportive MRI findings allows early diagnosis and timely intervention, thereby improving long-term outcomes in affected patients.

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