

Case Series

Unraveling the Underlying Neuropsychiatric Aspects of Epilepsy from Tuberous Sclerosis Complex to Fahr's Syndrome: A Case Series

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ABSTRACT

Epilepsy is a condition characterized by recurrent unprovoked seizures. It can affect individuals of any age and can manifest with various neuropsychiatric disorders. It can have a varying etiology. Among many reasons, conditions such as tuberous sclerosis complex (TSC) and Fahr's syndrome can present with epilepsy as one of the clinical features. TSC is a genetic disorder which is characterized by multisystem non-cancerous tumors, seizure episodes, and intellectual disability. On the other hand, Fahr's syndrome is a rare autosomal-dominant neurological disorder that is characterized by calcification of the basal ganglia. Here, we present the case series of a 30-year-old male with TSC and a 9-year-old male with Fahr's syndrome, both presenting with epilepsy.

Keywords: Epilepsy, Fahr's syndrome, Tuberous sclerosis complex

INTRODUCTION

Epilepsy is condition that is characterized by recurrent unprovoked seizures. A seizure manifests as uncontrolled abnormal electrical activity in the brain which may lead to changes in behavior, memory, or level of consciousness.^[1] The prevalence of epilepsy in India is 1%, which means that India contributes to one-sixth of the global burden of epilepsy.^[2] There are many etiological factors for the occurrence of epilepsy of which tuberous sclerosis complex (TSC) and Fahr's syndrome are the known conditions. Around 72–85% individuals with TSC tend to have epilepsy.^[3] On the other hand, the prevalence of epilepsy is not well established in cases of Fahr's syndrome with epilepsy as a rare occurrence in Fahr's syndrome cases.^[4]

TSC is an autosomal-dominant genetic disorder which is characterized by the triad of epilepsy, intellectual disability, and Pringle type of sebaceous adenoma (also known as angiofibroma).^[5] TSC is a multisystem disorder that consists of hamartomas or benign tumors in multiple organs such as skin, brain, eyes, lungs, heart, and kidneys.^[5,6] The overall prevalence of TSC is 1 in 20,000 and it affects around 1 in 6,000–1 in 10,000 live births.^[6] TSC is caused by the mutations in *TSC1* or *TSC2* genes. These gene mutations cause disruption of TSC1 - TSC2 intracellular protein complex which leads to overactivation of the mammalian target of rapamycin protein complex.^[3] The genetic testing for the detection of mutations tends to be positive in 75–95% of

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individuals suffering from TSC.^[6] These genes are involved in protein production which regulates growth and cell division in the body. Genetic mutations in these genes disrupt their functions which lead to the development of hamartomas in various organs of the body.^[6] Around 1 in 3 individuals inherit defective copies of TSC1 or TSC2 genes. If a parent has TSC, children will tend to have a 50% risk of inheriting the disorder. Due to autosomal dominant (AD) inheritance, both males and females are equally affected.^[6] TSC is associated with a wide range of neuropsychiatric manifestations called tuberous sclerosis-associated neuropsychiatric disorders (TAND) and almost all of them have some form of neuropsychiatric presentation. The prevalence of psychosis, autism spectrum disorder, and attention-deficit hyperactivity disorder is 1%, 40% to 50%, and 30–50%, respectively.^[3] Nearly 50% individuals with TSC tend to have intellectual disability, and 30% school-going children experience academic difficulties in mathematics, reading, spelling, and writing.^[3]

Fahr's syndrome is an autosomal-dominant neuropsychiatric disorder characterized by bilateral basal ganglia calcification which can be secondary to a pre-existing condition like hypoparathyroidism.^[7] When basal ganglia calcification is idiopathic and etiology is unknown, it is known as Fahr's disease and its prevalence is very low with <1 case per 10 lakh population.^[7] Metabolic disorders related to calcium and phosphorus can lead to bilateral calcification in the basal ganglia, mainly in hypoparathyroidism.^[8] Common clinical features of Fahr's syndrome include cognitive impairment, dysarthria, cerebellar signs, psychiatric illness, pyramidal signs, sensory impairment, and gait disorder.^[7] Epilepsy is a rare occurrence in the case of Fahr's syndrome.^[8]

In this case series, both individuals presented as cases of epilepsy, and on detailed neuropsychiatric and radiological evaluation, turned out to be the individual cases of TSC and Fahr's syndrome.

CASE - 1

A 30-year-old unmarried male was brought to the psychiatry outpatient by his father and brother with chief complaints of clenching of teeth, frothing from mouth, deviation of mouth, uprolling of the eyes, abnormal jerky movements of all the limbs, urinary incontinence, and loss of consciousness. He was having such episodes of generalized tonic clonic seizures for the past 11 years with a frequency of around 1–2 episodes per month. His last seizure episode was 10 days back at the time of first assessment but was not on regular anti-epileptic treatment. He was having multiple facial skin lesions for the past 11 years [Figure 1]. He was also having behavioral symptoms since last 3 years in the form of repetitive thoughts of hands being contaminated by germs, repetitive hand washing, becoming restless and irritable on not letting him wash hands repetitively, spending more time and water while taking bath,



Figure 1: Multiple facial skin lesions – Angiofibromas (Adenoma sebaceum).

repetitively checking his face in the mirror, finger counting behavior, suspiciousness that people talk ill about him, staring look, using abusive language against others and not doing any gainful work. During a mental status examination at the time of hospitalization, he was restless and repeating the same sentence like “what has happened to him and his face.” He had a staring gaze, his behavior was irritable, his speech was spontaneous with increased tone and volume, his affect was restricted, and he had absent insight. He was under treatment with a private psychiatrist but there was no improvement due to poor compliance with treatment and follow-up. He was admitted to the psychiatry ward and started on treatment in the form of Tab Fluvoxamine 100 mg once a day for obsessive compulsive (OC) symptoms, Tab Haloperidol 5 mg twice a day along with Tab Trihexyphenidyl 2 mg twice a day for psychotic symptoms, Tab Divalproex sodium 500 mg twice a day as mood stabilizer for irritability as well as for epilepsy, and Injection Lorazepam was given on emergency basis. He had a family history of epilepsy and TSC in his father. Based on the clinical assessment (individual history, facial skin lesions, epilepsy, and family history), a provisional diagnosis of TSC was made and opinions of other specialists such as radiologists, psychologists, neurologists, dermatologists, and ophthalmologists were sought. The dermatologist's opinion was taken in view of facial skin lesions and the facial skin lesions were confirmed as angiofibromas (adenoma sebaceum) [Figure 1]. For confirmation of the diagnosis, neuroimaging was done. Magnetic resonance imaging (MRI) of the brain showed ill-defined hyperintense lesion in the subcortical region of the left temporal lobe representing “tuber” in axial T2 [Figure 2a] and in fluid-attenuated inversion recovery [Figure 2b] images. Axial non-contrast computed tomography (CT) of the brain showed well-defined “subependymal calcified nodule” in the left lateral ventricle [Figure 3]. Axial CT of the chest showed a small thin-walled cyst in the posterior basal segment of the left lower lobe [Figure 4]. Coronal bone window CT of the abdomen showed few small sclerotic lesions in the bilateral ilium (thin arrow)

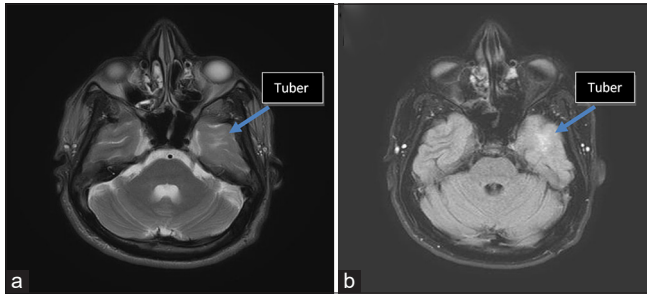


Figure 2: MRI brain - Axial T2 (a) and fluid-attenuated inversion recovery (b) images show ill-defined hyperintense lesions in the subcortical region of the left temporal lobe representing “tubers.”

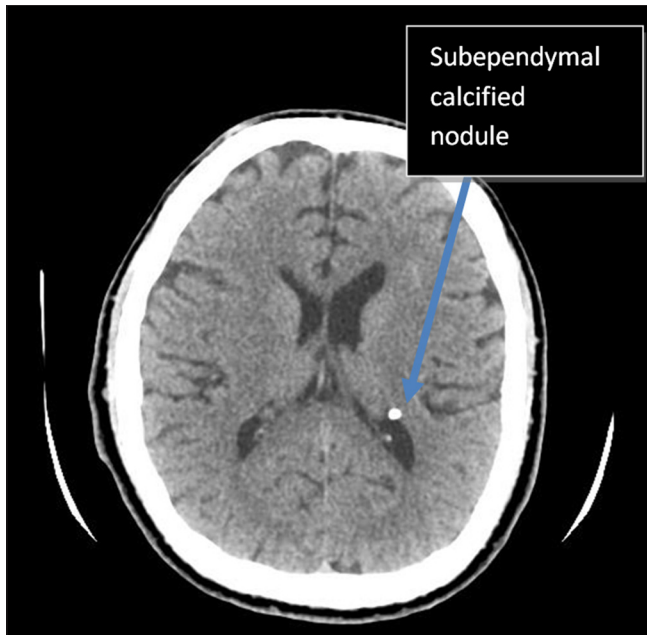


Figure 3: Axial non-contrast computed tomography image of the brain showing well-defined subependymal calcified nodule in the left lateral ventricle.

[Figure 5a] and in vertebral bodies (thick arrow) [Figure 5b]. All these neuroimaging findings confirmed the diagnosis of TSC. His ophthalmological examination was within normal. His intelligence was within the normal range as confirmed by intelligence tests by the psychologist. His intelligence quotient (IQ) and social quotient (SQ) were tested by psychologist using the Binet Kamat Test (BKT) and Vineland Social Maturity Scale (VSMS), respectively. His IQ was 88 and SQ was 86. Index individual with TSC showed significant improvement in behavior over a period of 2 weeks. He was discharged in behaviorally stable condition and regular treatment as well as follow-up was advised.

CASE - 2

A 9-year-old boy was brought by his parents with chief complaints of uprolling of eyes, tongue biting, frothing from

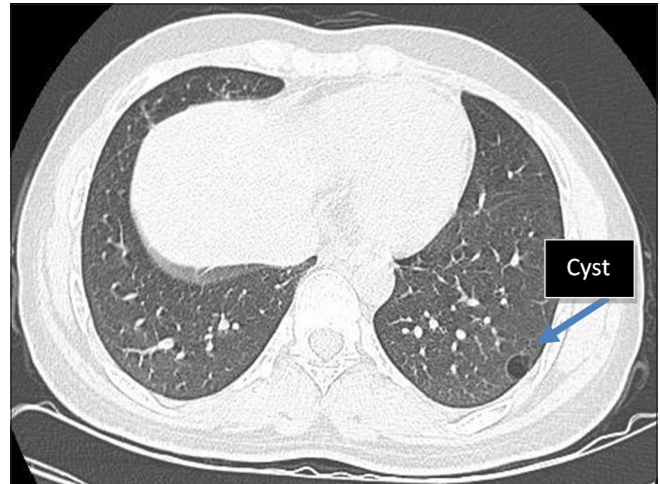


Figure 4: Axial computed tomography image of the chest shows small thin-walled cyst in posterior basal segment of the left lower lobe.

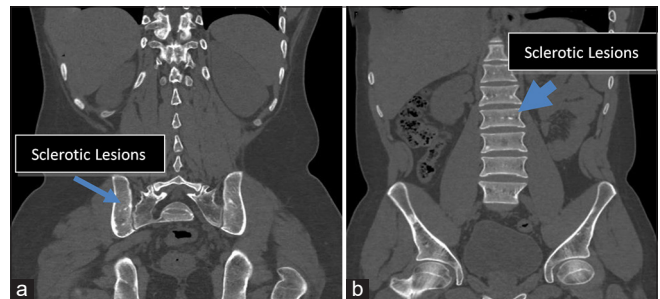


Figure 5: Coronal bone window computed tomography image of the abdomen shows few small sclerotic lesions in bilateral ilium (blue thin arrow) (a) and vertebral bodies (blue thick arrow) (b).

the mouth, abnormal jerky movements of all limbs, urinary and fecal incontinence, and loss of consciousness. He was having such GTCS episodes since last 6-years period with a frequency of around 3-4 episodes per month. He had his last seizure episode about 8 days back. He also had behavioral symptoms such as irritability, restlessness, poor academic performance, passing urine in cloths during night sleep once to twice a week, requiring assistance in consuming food properly, requiring assistance in changing clothes and taking bath, and not able to talk fluently. His IQ and SQ were tested by the psychologist using BKT and VSMS, respectively. His IQ was 67 and SQ was 65. Based on psychological testing, he was diagnosed as a case of mild intellectual disability. His CT scan of the brain was done in view of epilepsy which showed symmetrical foci of calcifications in bilateral basal ganglia [Figure 6]. His blood investigation showed reduced serum parathyroid hormone (PTH) level to 12.60 pg/mL (normal range: 15–65 pg/mL). Based on CT brain finding and serum PTH level, his diagnosis was confirmed as Fahr’s syndrome. The child was started on Tab Risperidone 0.5 mg twice a day for behavioral disturbances and showed moderate

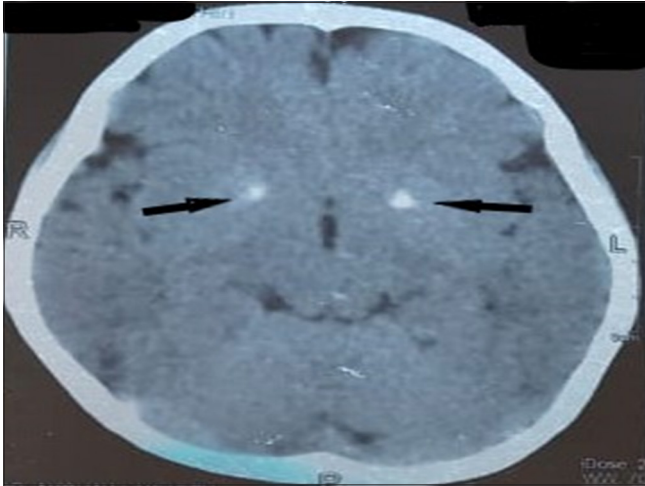


Figure 6: Computed tomography brain showing symmetrical foci of calcifications in bilateral basal ganglia indicated by black arrows.

improvement within a period of the next 1 month. He was given antiepileptic treatment from a neurologist in the form of syrup Phenobarbitone (20 mg/5 mL) 7.5 mL twice a day and syrup Sodium Valproate (200 mg/5 mL) 3 mL in the morning and 5 mL in the evening. He was referred to an endocrinologist for the management of hypoparathyroidism.

DISCUSSION

Epilepsy can have multiple etiologies, and it can manifest with multiple neuropsychiatric disorders. In this case series, epilepsy was associated with underlying neuropsychiatric disorders such as TSC and Fahr's syndrome.

TSC is a disorder that can present with various forms of TAND.^[3] In the index individual (case – 1), TAND was present in the form of psychosis and OC symptoms. The prevalence of psychosis in TSC is 1%^[3] and the prevalence of OC symptoms in TSC is not well established. One study found that out of 241 individuals with TSC, only one had obsessive compulsive disorder (OCD).^[9] To our best knowledge, this index case (case – 1) is the only second case to report a case of TSC that presented with both psychosis and OC symptoms.^[10] Dermatological lesions of TSC may include facial angiofibromas, shagreen patches, unguis fibromas, focal hypopigmentation, and fibrous cephalic plaques in 75%, more than 50%, 20–80%, 90%, and 25% cases, respectively.^[6] MRI brain finding of cortical tubers is the hallmark of TSC.^[11,12] There exists an association between the number of tubers and seizures and cognitive impairment in TSC.^[6] Subependymal nodules are observed in 80–90% individuals with TSC and around 10–20% can develop subependymal giant cell astrocytomas (SEGAs).^[6] Pulmonary cysts can occur in 10–38% of adult male cases of TSC and in around 35% adult female cases of TSC.^[6,13,14]

Although not diagnostic, sclerotic bone lesions can occur in around 89% cases of TSC.^[15] Other body organs that may get affected in TSC include retina, teeth and oral cavity, kidneys, heart, and liver.^[6] According to the recommendations from 2012 International TSC consensus conference, index individual (case – 1) met 2 major (facial angiofibromas and subependymal nodule) and 1 minor (non-renal hamartomas mainly in the form of brain parenchymal tubers) features of the diagnostic criteria for TSC.^[6] In index individual with TSC, intelligence was within the normal range and he did not meet the full triad of TSC. Literature has shown that only 29% cases of TSC fulfill the triad and around 6% cases do not tend to have all the 3 components of the triad.^[16,17] In index individual with TSC (case – 1), genetic testing was not done due to financial issues because the patient belonged to the lower economic background and his family members did not want the testing done. Literature has also shown that there exist hypothetical relations between the financial difficulties and mental illnesses.^[18] His diagnosis was done mainly based on clinical history and radiological evaluation. The management of behavioral disturbances like psychosis as well as OC symptoms and epilepsy involves treatment with antipsychotics, selective serotonin reuptake inhibitors, and antiepileptic medicines, respectively.

Fahr's syndrome is a rare neuropsychiatric disorder that can rarely manifest as epilepsy. The extent of basal ganglia calcification in Fahr's syndrome mainly correlates with psychiatric/behavioral manifestations rather than the neurologic manifestations, which can explain the rare occurrence of epilepsy in cases of Fahr's syndrome.^[7,19] İnci *et al.* reported a rare case of Fahr's syndrome that presented with seizures. They concluded that the clinical diagnosis of Fahr's syndrome is difficult due to its vast neuropsychiatric presentation and occurrence of epileptic seizure is uncommon in Fahr's syndrome, which can make it more difficult to diagnose.^[20]

CONCLUSION

In this case series, both individuals presented with epilepsy and on detailed clinical and radiological evaluations, they were diagnosed as individual cases of TSC and Fahr's syndrome. Epilepsy is just a tip of the iceberg of various underlying neuropsychiatric disorders. In-depth evaluation of the cases of epilepsy by clinical examination as well as by radiological means can help in understanding the phenomenology of various neuropsychiatric aspects of it. In turn, it can help in assessing the prognosis of the disorder and may help in improving the outcome.

Ethical approval: Institutional Review Board approval is not required.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient and parental consent forms. In the

forms, the patients and parents have given their consent for the images and other clinical information to be reported in the journal. The patients and parents understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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