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CASE REPORT: A CASE OF TUBEROUS SCLEROSIS PRESENTED WITH STATUS EPILEPTICUS

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ABSTRACT Tuberous sclerosis complex inherited in an autosomal dominant pattern, it is characterized by the development of benign tumors affecting different systems of the body involving the brain, skin, retina, and viscera organs. TSC characterized by cutaneous changes, neurologic conditions, and the formation of multiple hamartomas in the organs leading to morbidity and mortality. The treatment of these patients should be in a multidisciplinary approach involving specialists from the various medical field. Here, we present a case of a 10-old-year female child with characteristic clinical features of tuberous sclerosis complex.

KEYWORDS : tuberous sclerosis complex, Fibromas, hamartomas, subependymal nodules, adenoma sebaceum, shagreen patch, phakomatosis, and EPILOIA

INTRODUCTION:

In 1862, von Recklinghausenfirst described Tuberous sclerosis complex (TSC) is an autosomal dominant disorder. In 1880, the term "sclerose tubereuse" coined by Bourneville, henceforth known as Bourneville's disease. TSC is classified as a phakomatosis. It is characterized by the triad of epilepsy (EPI), intellectual disability (LOI), and adenoma sebaceum (A); therefore, by combining these features, Sherlock coined the term EPILOIA.

Tuberous sclerosis complex (TSC) is a multisystem disease characterized by an autosomal dominant mode of inheritance, variable expression, and a prevalence of 1 in 6,000 to 10,000 newborns. Spontaneous genetic mutations occur in 65% of the cases. Molecular genetic studies have identified two foci for TSC: the *TSC1* gene located on chromosome 9q34, and the *TSC2* gene is on chromosome 16p13. The *TSC1* gene codes a protein called hamartin, while the *TSC2* gene encodes a protein called tuberin. Within a cell, these two molecules form a complex along with a third protein, TBC1D7. Consequently, a mutation in either the *TSC1* gene or the *TSC2* gene results in a similar disease in patients, though individuals with *TSC2* mutations tend to be more severely affected.

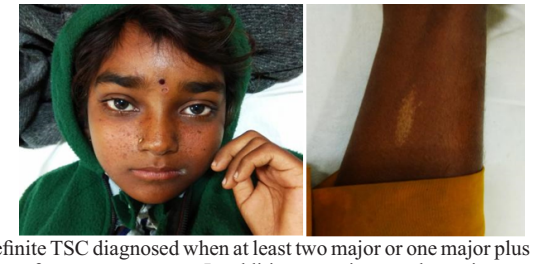
Case Report:

A 10-year-old female child brought to the emergency service room with complaining of seizure activity for nearly one hour with postictal drowsiness since 2hours. On examination of the child her PR 110 per minute, RR 22 per min and BP 110/70. On general physical examination, the child had, multiple well-defined, brownish sessile nodular growths were noted on the forehead, cheeks, and nose in a characteristic "butterfly pattern" suggestive of angiofibroma. Similar firm nodular growths were noted in the upper extremities of varying size. A well-defined rough hypermelanotic patch was noted in the right shoulder area, and the left lumbosacral region was showing an orange peel appearance indicative of shagreen patch. Ashleaf macule is present over the left foot and right leg region.

The patient underwent various radiological, hematological, and histopathological investigations. CT of the brain showed hypodense areas in the subependymal regions of both ventricles, indicating multiple calcified tuberous lesions (subependymal nodules). Ultrasound of the abdomen showed no abnormality. The echocardiogram is normal. Based on all these clinical findings and investigations, the diagnosis of tuberous sclerosis was made.

DISCUSSION

TSC is characterized by the development of unusual tumor-like growths in brain, skin, retina, and viscera. The term "tuberous sclerosis" refers specifically to the presence of multiple sclerotic masses scattered throughout the brain. The diagnosis of TSC is based on the identification of hamartomas in more than one organ system.



Definite TSC diagnosed when at least two major or one major plus two minor features are present. In addition, carrying a pathogenic mutation in *TSC1* or *TSC2* is sufficient for the diagnosis of TSC.

Major Features of Tuberous Sclerosis Complex:

a) Cortical dysplasias (including tubers and cerebral white matter migration lines)
b) Subependymal nodules
c) Subependymal giant cell astrocytoma
d) Facial angiofibromas (≥3) or forehead plaque
e) Ungual fibromas (≥2)
f) Hypomelanotic macules (≥3, ≥ 5 mm in diameter)
g) Shagreen patch
h) Multiple retinal nodular hamartomas
i) Cardiac rhabdomyoma
j) Renal angiomyolipoma
k) Pulmonary lymphangioleiomyomatosis

Minor Features of Tuberous Sclerosis Complex:

1. Dental enamel pits (>3)
2. Intraoral fibromas (≥2)
3. Retinal achromic patch
4. Confetti skin lesions
5. Nonrenal hamartomas
6. Multiple renal cysts

Our case had three major criteria and diagnosed as tuberous sclerosis clinically and radiologically. Seizure activity controlled with antiepileptic drugs and child discharged with further followup advise.

CONCLUSION:

The management of these patients is often multidisciplinary involving the neurosurgeon, neurologist, nephrologist, pulmonologist, cardiologist, ophthalmologist, and the genetic counselor. The knowledge of various associations of tuberous sclerosis can be helpful in retrogradely establishing the diagnosis. The long-term prognosis in

this patient remains uncertain in spite of the timely diagnosis.

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