

## Case Report

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# Tuberous Sclerosis: A Case Report

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### Abstract

*Tuberous sclerosis is a disorder affecting multiple organs leading to morbidity and mortality. It is important to make an early diagnosis of tuberous sclerosis so that lifelong monitoring, early recognition of complications and proactive treatment can lower the morbidity and mortality rates. Here we report a case of a 50-year-old male who presented with seizure. This case report emphasizes the importance of complete evaluation of a case presenting with seizures. Tuberous sclerosis must be included in the differentials if patient presents with seizures, skin manifestations and mental retardation.*

**Key words:** Tuberous sclerosis, Cutaneous Angiofibromas.

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### Introduction

Von Recklinghausen first described tuberous sclerosis in 1862. Desire-Magloire Bourneville (a French physician) coined the term sclerosetubereuse, from which the name of the disease has evolved. Sherlock coined the term EPILOIA encompassing the clinical triad of tuberous sclerosis (Epi: epilepsy, Loi: low intelligence, A: adenoma sebaceum). As the manifestations of the disease are variegated in nature, the term tuberous sclerosis complex (TSC) is now

widely used<sup>1</sup>. Tuberous sclerosis (TS) is an autosomal dominant disorder characterized by the formation of hamartomatous lesions in multiple organs, with a birth incidence of around one in 10,000<sup>2</sup>. However, with more sensitive screening the prevalence may be as high as one in 6,000<sup>3,4</sup>. The disease results from mutations in one of two genes, *TSC1* (encoding hamartin) or *TSC2* (encoding tuberin), which have an important role in the regulation of cell proliferation and differentiation<sup>5</sup>. It should be considered as a differential diagnosis if patients present with seizure, mental retardation and adenoma sebaceum. However, in a patient presenting with an incomplete form of tuberous sclerosis, establishing diagnosis is difficult. We report a case of 50-year-old male who presented with seizures and subsequently found to be a case of tuberous sclerosis.

### Case report

A 50-year-old male from Dhaka, Bangladesh presented with the complaints of repeated seizures for 6 hours. He had generalized tonic-clonic seizure associated with uprolling of eyes, tongue biting, urinary incontinence and postictal confusion. There was no history of prodromal aura or auditory and visual hallucination. He was diagnosed as a case of epilepsy since 21 years of age. He had delayed development of milestones and had to quit school due to low IQ and poor memory. His brother is also diagnosed as a case of epilepsy. On examination, he had normal vital signs. Examination of skin revealed multiple nodular lesions on face (Fig: 1)

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**Figure-1:** *Adenoma Sebaceum*

and multiple macular dark brown lesions (Fig: 2) all over the body. On neck region, hypomelanotic white patches suggestive of ash leaf spots were seen. There were small fleshy like growths seen around and under the right main fingernail suggestive of periungual fibromas. On neurological examination, he had poor past memory and was unable to perform simple arithmetic sums. He had whitish patches on retina of left eye suggestive of retinal phakoma. Rest of examination was unremarkable.

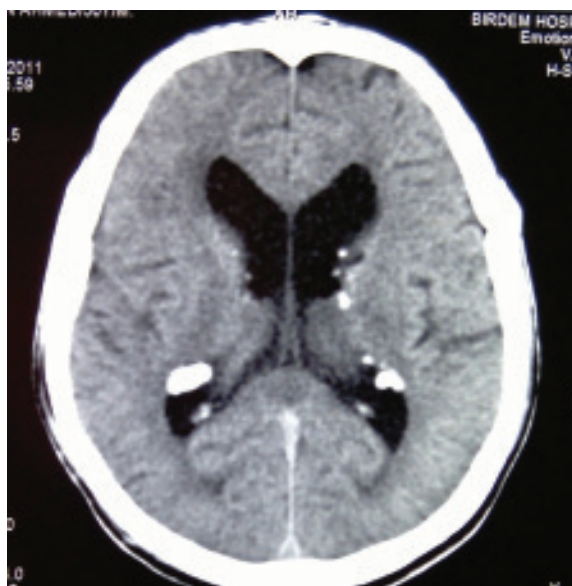


**Figure-2:** *Macular dark brown lesions*

His laboratory investigations including blood counts, routine urinalysis, renal function tests, serum electrolytes and blood sugar level were normal. Radiology of skull showed multiple areas of calcification (Fig: 3). CT scan of brain showed bilateral sub-ependymal nodules (Fig: 4). Abdominal ultrasonography showed multiple cysts and heterogenous high intensity masses consistent with diagnosis of renal angiomyolipoma. ECG and



**Figure-3:** *Calcification on X ray Skull*



**Figure-4:** *Bilateral sub-ependymal nodules on CT scan of brain*

echocardiography were normal. All clinical features suggested the classical picture of tuberous sclerosis. Molecular analysis of the gene *TSC1* and *TSC2* in chromosome 9 and 16 respectively could not be performed due to the availability constraints. Patient was discharged in stable condition on levetiracetam with regular follow-up advice. Genetic counseling was done.

### Discussion

Tuberous sclerosis shows a wide variety of clinical expressions. Some individuals are severely affected, while others have very few features. Tuberous sclerosis is characterized by the development of unusual tumor-like growths (hamartomas) in brain, skin, retina and viscera. As multiple organs are involved, there is wide variability in presentation. Arguably the most important hamartomas are cerebral cortical tubers, which are regions of abnormal cortical architecture with distinctive large neuronal cells. These hamartomatous swellings resemble potatoes and hence, referred to as tubers. Cortical tubers cause some of the most important clinical manifestations of tuberous sclerosis: epilepsy, mental retardation, and abnormal behavior including autism (mosaicism)<sup>6-9</sup>. Epilepsy occurs in 80-90% of all patients; with a positive correlation with subnormal intelligence<sup>10</sup>. Cutaneous lesions are present in 96% of the patients. These include facial angiofibroma (adenoma sebaceum), subungual fibromas, and shagreen patches. Two types of renal lesions occur in patients with tuber sclerosis: angiomyolipomas and renal cysts. They may be found independently or together: they may be unilateral, bilateral, single or multiple. Angiomyolipomas are benign in nature and asymptomatic but spontaneous rupture and subsequent hemorrhage in retroperitoneum may occur<sup>11</sup>. In the heart, the most frequent and characteristic type of tumor is cardiac rhabdomyomas. Incidence of cardiac rhabdomyomas in children with tuberous sclerosis is higher than in adult patients with tuberous sclerosis. It has been suggested that such lesions tend to regress in early infancy and adolescence<sup>12</sup>. Our patient had tuberous sclerosis characterized by classical features: seizures, mental retardation, and facial angiofibromas. He had ash leaf macules, periungual fibroma, retinal phakomas and low IQ which were characteristic features of tuberous sclerosis.

Incomplete forms of tuberous sclerosis may present with acute complications such as hematuria, retroperitoneal hemorrhage or pneumothorax<sup>13</sup>.

Tuberous sclerosis cases remain undiagnosed due to variegated clinical presentation. Thorough history taking and clinical examination is mandatory for all patients. Intervention programs, including special schooling and occupational therapy, may benefit individuals with special needs and developmental issues. Surgery, including dermabrasion and laser treatment, may be useful for treatment of skin lesions. There is no cure for tuberous sclerosis. Drug therapy for some of the manifestations of TSC is currently in the developmental stage. Prognosis of the disease depends on the severity or multiplicity of organ involvement. This presentation aims at considering the importance of neurocutaneous diseases presenting with seizures, skin manifestations and mental retardation as differential diagnoses.

**Conflict of interest:** None

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