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Tuberous sclerosis: A case report

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Abstract
Tuberous sclerosis complex (TSC) is a genetic disorder that causes tumors to form in many different organs, primarily in the brain, eyes, heart, kidney, skin and lungs. The aspects of TSC that most strongly impact quality of life are generally associated with the brain: seizures, developmental delay, intellectual disability and autism. However, many people with TSC are living independent, healthy lives and enjoying challenging professions such as doctors, lawyers, educators and researchers. The case report tells us that Mr. y 22yrs old man with tuberous sclerosis presented with a complaints of 30 convulsions from childhood with the duration of 1 minute. Finally from the age of 20 years convulsions got reduced to 5- 7 convulsions in a month. On examination small lumps and masses on his face, Scalp, Nostrils, Ear canal, shoulders and in between the scapula. Patient was under the treatment of medical management. Many TSC patients show evidence of the disorder in the first year of life. However, clinical features can be subtle initially, and many signs and symptoms take years to develop. As a result, TSC can be unrecognized or misdiagnosed for years.

Keywords: Tuberous sclerosis

Introduction
Tuberous sclerosis is a genetic, variably expressed, multisystem disorder that can cause circumscribed, benign, non-invasive lesions in any organ.1, 2 The term tuberous sclerosis of the cerebral convolutions was used more than a century ago to describe the distinctive findings at autopsy in some patients with seizures and mental subnormality [1]. The term tuberous describes the potato-like consistency of gyri with hypertrophic sclerosis. The wide range of organs affected by the disease 4 implies an important role for *TSC1* and *TSC2* genes, encoding hamartin and tuberin, in the regulation of cell proliferation and differentiation [2].
The name tuberous sclerosis comes from the characteristic *tuber* or potato-like nodules in the brain, which calcify with age and become hard or *sclerotic*. The disorder--once known as *epiloia* or *Bourneville's disease*--was first identified by a French physician more than 100 years ago [3].

Incidence
At least two children born each day will have tuberous sclerosis complex. Current estimates place tuberous sclerosis complex-affected births at one in 6,000. Nearly 1 million people worldwide are estimated to have TSC, with approximately 50,000 in the United States [4]. Many cases may remain undiagnosed for years or decades due to the relative obscurity of the disease and the mild form symptoms may take in some people [5].

Signs and symptoms
Tuberous Sclerosis include

- **Neurologic findings:** Abnormal neurologic findings result from the location, size, and growth of tubers and the presence of subependymal nodules (SENs) and subependymal giant cell astrocytomas (SEGAs)
- **Cutaneous findings:** The best-known cutaneous manifestation of TSC is adenoma sebaceum, which often does not appear until late childhood or early adolescence
- **Cardiac findings:** Cardiac involvement is usually maximal at birth or early in life; it may be the presenting sign of TSC, particularly in early infancy; 50-60% of individuals with TSC have evidence of cardiac disease, mostly rhabdomyomas.
- **Ophthalmic findings:** At least 50% of patients have ocular abnormalities; these lesions are actually retinal astrocytomas that tend to become calcified over time
- **Pulmonary findings:** Prospective and retrospective studies have found cystic pulmonary abnormalities in as many as 40% of women with TSC [6]. Etc…….

Diagnosis
Major and minor criteria exist to diagnose tuberous sclerosis (panel). The diagnosis is made when two major features, or one major and two minor ones, can be shown. Sometimes, an antenatal diagnosis can be made based on fetal ultrasound and MRI, which show cardiac and brain lesions. Most patients are diagnosed in infancy or early childhood, making

early therapeutic interventions and treatments possible [7].

Management

Since there is no cure early diagnosis and intervention can help overcome developmental delays. Data show that early seizure control in children can improve learning as compared to children without good seizure control. Advancements in research continue to bring new and improved therapeutic options. Some anti-seizure drugs can be effective in individuals with TSC. When drug treatment fails to adequately control seizures, technology can help identify the exact portions of the brain stimulating seizures and creating new therapies to help control seizures [8].

For tumors in the brain, surgery is sometimes used to permanently remove tumors that are relatively few in number and easily accessible by the surgeon. In other cases, drug treatment may be used to shrink brain tumors. In the fall of 2010, the FDA approved the first drug with an indication specifically for TSC to treat a type of brain tumor known as subependymal giant cell astrocytomas (SEGAs) [9]. In 2012, the same drug was approved to treat growing angiomyolipomas, a type of kidney tumor in TSC [10].

Major advancements in treatments such as these require clinical studies to test the effectiveness of experimental drugs, surgery, or other interventions in people with TSC [11]. Because the TSC community is in vital need of new treatments, individuals with TSC frequently volunteer to participate in cutting-edge clinical studies [12]. Some ongoing clinical studies in TSC include testing the effects of drug treatment on neurocognitive function, testing a new combination drug treatment for LAM, finding biomarkers to identify infants at high risk of developing autism or infantile spasms, and testing a topical drug treatment of facial angiofibromas [13]. Thanks to volunteers in these and other studies, every new day brings us one step closer to finding improved treatments for TSC [14].

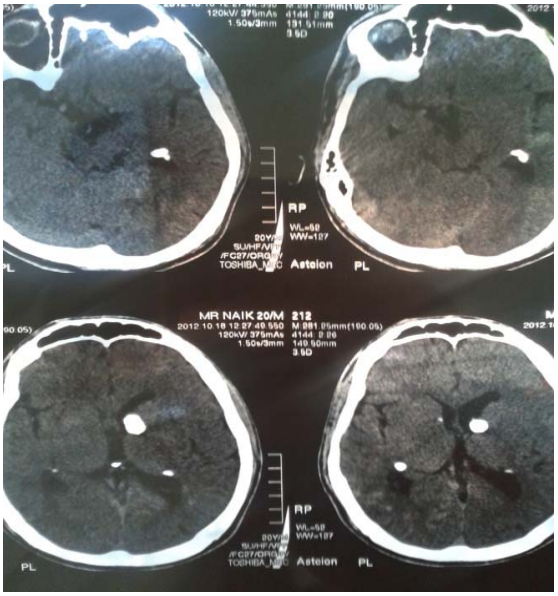
Case report

Mr. y 22yrs old man with tuberous sclerosis presented with a complaints of 30 convulsions from childhood with the duration of 1 minute. Finally from the age of 20 years convulsions got reduced to 5- 7 convulsions in a month. And no other known co- morbidities. Personal history of the client is

- **Bath:** Once a day (with warm water only)
- **Sleep and rest:** 8 hours of sleep at night and 2 hours in the afternoon.

On physical examination he was worried and depressed and he has small lumps and masses on his face, Scalp, Nostrils, Ear canal, shoulders and in between the scapula. He has developmental delay in numerical ability.

MRI scan shows here benign turmors present in the brain.



Patient was under the treatment of medical management

▪ Tab : Zenoxa	300 mg	1-0-1
▪ Tab : Cloba	10 mg	½-0-1
▪ Tab : Levipil	500 mg	1 ½ -0- 1 ½.

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