**Tuberous Sclerosis Complex**

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DOI: 10.29322/IJSRP.12.02.2022.p1224  
http://dx.doi.org/10.29322/IJSRP.12.02.2022.p1224

**ABSTRACT**

**Background:** Tuberous sclerosis is an autosomal dominant neuro-cutaneous syndrome characterized by variable phenotype due to development of hamartomas in eyes/skin/kidneys/heart. Epilepsy, mental retardation and dermatological manifestations are seen in majority of patients.

**Introduction:** We describe Tuberous sclerosis complex as an unusual cause of refractory seizure in early adulthood.

**Diagnosis:** Tuberous sclerosis complex diagnosis was made based on correlation between clinical findings and imaging. (Computed Tomography and Magnetic resonance imaging).

**Intervention:** Several anti-epileptic drugs like phenytoin levetiracetam, sodium valproate.

**Outcome:** Patient experienced generalized tonic clonic convulsions even while he was on polytherapy with anti-epileptic drugs (phenytoin + levetiracetam + sodium valproate) without any complications observed over follow up period.

**Conclusion:** It was concluded that combination therapy of anti-epileptic drugs could significantly reduce the frequency of seizures while not being able to eliminate them completely.

**INTRODUCTION:**

Tuberous sclerosis complex is a multisystem autosomal dominant disorder affecting children and adults resulting from mutation in one of two genes TSC 1 (encoding Hamartin) TSC 2 (encoding Tuberin). Tuberous sclerosis often causes disabling neurologic disorders like epilepsy, mental retardation and autism. It is characterized by hamartomatous lesions that can affect any organ system like CNS, eyes, skin, kidneys. Additional features include facial angiofibromas, renal angiomyolipoma, pulmonary lymphangiomyomatosis. Skin lesions are present in 90% patients and may be seen at all ages. Ash-leaf spots (Hypopigmented macules) are generally detected in infancy/early childhood. Facial angiofibromas formerly called adenoma sebaceum may be seen at any age but more common in late childhood or adolescence.

**CASE PRESENTATION:**

A 26-year-old male presented with h/o recurrent seizures. He also complains of multiple growths in upper and lower gums since 5 years. Patient noticed the growth for past 5 years which gradually increased to present size. It was not associated with h/o pain/blood/purulent discharge from growth.

Patient is a known epileptic since the age of 10 years and under anti-convulsant therapy for same. His parent’s give history of recurrent seizures despite being on treatment. Also, they noticed learning difficulties because of which he had been dropped out of school at age of 12 years.

**ON GENERAL PHYSICAL EXAMINATION:**

Multiple well defined reddish brown nodular growths were noted on the fore head/nose/cheeks in a butterfly pattern. Also, Hypopigmented macule was seen over back region.

All routine biochemical investigations including serum electrolytes and serum calcium were done and were normal. Following this the patient was subjected to radiological examination.
NCCT brain was s/o Hypodense areas in subependymal region of both ventricles indicating calcified tuberous lesions. Fundus and USG were found to be normal.

**DISCUSSION:**

Tuberous sclerosis is an autosomal dominant multisystem disorder affecting children and adults which often causes disabling neurologic disorders like epilepsy, mental retardation and autism. It also has skin manifestations viz. facial angiofibromas, renal involvement in the form of angiomyolipoma, and pulmonary lymphangiomyomatosis. Epilepsy/cognitive disability/neurobehavioural abnormalities which are there in almost 80% patients with TSC are found to be the most common presenting complaints. These are related to the cortical tubers (developmental abnormalities of cerebral cortex characterized histologically by loss of normal six layered structure of cortex and dysmorphic neurons). Tubers might calcify and undergo cystic degeneration. For diagnosis of tuberous sclerosis requires correlation between clinical findings and imaging. Major criteria: Facial angiofibromas, ungual fibroma, renal angiomyolipoma, pulmonary lymphangiomyomatosis, cortical tubers, Shagreen patch, Hypomelanotic macule, retinal hamartoma, cardiac rhabdomyoma, subependymal giant cell tumour
Minor criteria: Dental enamel pits, bone cysts, rectal polyps, gingival fibromas, retinal acromic patch. One major plus two minor/ two major criteria when fulfilled are diagnostic.

For management of seizures mono or polytherapy using anti-epileptic drugs are used.

Seizures have found to be refractory even to polytherapy and these are to be considered for surgical evaluation. Cortical resection of area containing tubers is indicated in specific cases.

**CONCLUSION:**

Despite of taking polydrug therapy with Phenytoin, levetiracetam, sodium valproate the patient experienced generalized tonic clonic convulsions with a frequency of once / twice per month which was not followed by any complications during a follow up period. It can be henceforth concluded that the combination of phenytoin/levetiracetam/sodium valproate can reduce the frequency of seizures but cannot eliminate them completely.

**REFERENCES**