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VIGABATRIN- TREATMENT FOR ADULT SEIZURE IN TUBEROUS SCLEROSIS: A CASE REPORT

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ABSTRACT

Tuberous sclerosis complex (TSC) is rare autosomal dominant genetic neurocutaneous syndrome that is caused by mutations in tumour suppressor genes TSC1 or TSC2 which lead to development of benign tumors affecting different body systems affecting the brain, skin, retina, and viscera. The main goal of this article was to state the newer drug for treatment of TSC. Herein, we report a cases of TSC, which presented with seizures at age of 12 year. Case presented with multiple types of seizures from 12 years of age and was diagnosed by multiple calcified subependymal nodules (SENs) detected by magnetic resonance imaging (MRI) and treated with Vigabatrin. In conclusion, the literature review revealed that neurological manifestations were the main feature that led to investigation and diagnosis of TSC and the seizure controlled with Vigabatrin after many anti epileptic drug given.

KEYWORDS

Tuberous Sclerosis, Seizures, Vigabatrin, cortical tubers

INTRODUCTION

Tuberous sclerosis complex (TSC) is a rare autosomal dominant disorder. In 1880, Bourneville coined the term "sclerose tubereuse" based on the pathologic features of the sclerotic tubers found in the postmortem investigation of patients with epilepsy and mental retardation, henceforth known as Bourneville's disease. TSC affects the central nervous system in the majority of patients causing a wide range of structural abnormalities such as cortical tubers, and subependymal nodules (SENs) and functional manifestations such as seizures, intellectual disability. Seizure, and facial angiofibromas present in three quarters of patients and intellectual disorders presents in half of patients; the full triad is only seen in a minority of patients. The classic triad of TS is seizures, mental retardation, and angiofibromas. The main goal of case presentation is to know the advance drug which is used in seizure episode in tuberous sclerosis as vigabatrin used in infantile seizures but in this case we give to adult to control seizure.

Case Report

A 21-year-old female presented with 15 days history of loss of speech with irrelevant behavior and poor communication with attendant recurrent intractable seizures. The patient had had 2-3 attacks per day of multiple types of seizures from 12 years of age. Each attack lasts for seconds to 5 minutes. She was on polytherapy i.e Tab. Levetiracetam 1000 mg BD , Tab. Lacosamide 200mg BD, Tab. Phenytoin 300mg BD, Tab Carbamazepine 200 BD and Tab. Perampanel 6mg HS for several years. But frequency of generalised convulsions not reduced, but she continues to have 6-8 tonic seizures during her sleep every night, four generalised tonic clonic seizure (GTCSz) a month and daily atypical absence seizures. Seizure was on increasing in frequency over time.

Hospital course on admission we started with anti epileptics drug in injectable form as well as oral dose {Inj. Lacosamide 200mg IV BD, Inj. Levetiracetam 500mg IV TDS , Tab. Phenytoin 300mg BD, Tab Carbamazepine 200mg BD and Tab. Perampanel 6mg HS. And we check for frequency of seizure. On next day patient had a episode of seizure in which there is stiffening and jerking of muscle of face and hand. Severity of seizure is still same as it before. Along with injectable and oral formulation of anti epileptic we add with VIGABATRIN POWDER 500 mg BD but before that we get fundus examination of patient and gradually we taper to reduce the dose of other anti epileptics drugs and continue VIGABATRIN POWDER 500 mg at maintenance dose clinically we notice there is decreased in seizure episode with patient responding to verbal command and get interacted with his family member. EEG shows focal epileptiform discharge on right fronto parietal region.

She was averagely built, uncooperative, and not oriented to time, place, and person. Her pulse rate was 126 beats per minute with a regular rhythm, normal volume, and blood pressure of 148/96 mm of Hg, tachypnea with normal temperature and oxygen saturation. Systemic examinations are unremarkable.

On laboratory investigations, hemoglobin level was 14.2 g/dl with hematocrit 15.8%. The rest of the cell counts were normal with a normal erythrocyte sedimentation rate and normal liver function tests. Magnetic resonance imaging (MRI) of the brain showed multiple patchy non-mass abnormal areas in cortical subcortical bilateral frontal, parietal, temporal, occipital lobes and cerebellum representing cortical tubers{figure1}. Few punctiform calcified nodules in bilateral fronto- parieto-occipital lobes representing imaging findings are consistent with the diagnosis of tuberous sclerosis. She was admitted to the ward for management of seizure for which injectable anti epileptic was given.

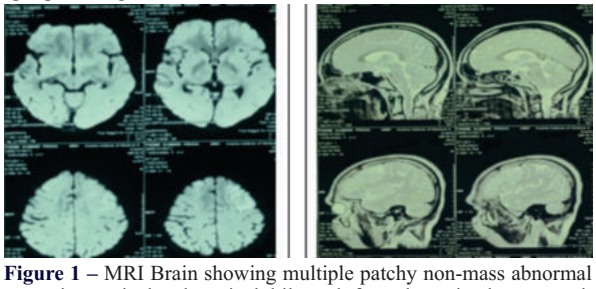

Figure 1 – MRI Brain showing multiple patchy non-mass abnormal areas in cortical subcortical bilateral frontal, parietal, temporal, occipital lobes and cerebellum representing cortical tubers.

As the disease runs a chronic and progressive course, the patient has been under regular follow-up since she was diagnosed with the disease complex. After 1 month follow up the female no longer had seizures presently. She has normal cognitive function and does well household work. She was very active and communicated well for his age.
EEG was done and shows normal findings.

DISCUSSION

TSC is characterized by the development of unusual tumor-like growths (hamartomas) in brain, skin, retina, and viscera. The term "tuberous sclerosis" refers specifically to the presence of multiple sclerotic masses scattered throughout the cerebrum. The abnormal genes have been localized to one of two sites, the long arm of chromosome 9 (9q34), designated as TSC 1 (encoding hamartin), and the short arm of chromosome 16 (16p 13.3) designated as TSC 2 (encoding tuberin). The mutations in these genes result in constitutive mTOR activation leading to the formation of various growths and hamartomas in various organs of the body.

The prognosis of TSC depends on the severity or multiplicity of organ involvement. However, in the case the patient diagnosed 21 years age

with few cutaneous signs, prognosis depends on the associated internal tumors and cerebral calcifications.

CONCLUSION

This case report highlights the drug Vigabatrin is found to most effective single drug therapy for seizure in tuberous sclerosis. This drug generally given to children but in this case we tried in adult and found to be effective for controlling seizure.

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