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Case Report

Tuberous Sclerosis Complex with Multiple Psychiatric Co-Morbidities: A Case Report

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ABSTRACT

Tuberous sclerosis complex is an autosomal dominant neurocutaneous disorder characterized by the classical triad of seizure, mental retardation and facial angiofibromas, Psychiatric co-morbidities like Autism Spectrum disorder, learning disabilities, attention deficit hyperactivity disorder, behavioral problems and anxiety disorders have been associated with this. We report a rare case of tuberous sclerosis with multiple psychiatric comorbidities and their successful management.

Keywords: Tuberous sclerosis, physical comorbidities, psychiatric comorbidities, management.

INTRODUCTION

Tuberous sclerosis complex (TSC) or Bourneville disease is an autosomal dominant neurocutaneous disorder characterized by appearance of non-malignant hamartomas in various organs of the body like brain, kidneys, lungs, skin, eyes, and heart. [1] The incidence ranges from 1 in 6000-10000 live births. [2] It is characterized by the classical triad of seizure, mental retardation and facial angiofibromas, although the full triad is manifested is less than one-third patients. [3] Nearly 75-80% of the affected individuals have a mutation in either of TSC1 (chromosome 9) or TSC2 (chromosome 16) genes coding for two cellular proteins - hamartin and tuberin, respectively. [4] Psychiatric co-morbidities like Autism Spectrum disorder(ASD), learning disabilities, attention deficit hyperactivity disorder(ADHD), behavioral problems like acting out, aggression and anxiety disorders have been reported with TSC. [5] We report a rare case of tuberous sclerosis with multiple psychiatric comorbidities and their successful management.

CASE REPORT

A 16 year old boy with no formal education was brought by her mother to the psychiatry outpatient department with re-emergence of generalized tonic clonic seizures since last 2 months and low intelligence from childhood onwards with behavioral oddities since last six years. He was born at term out of a non-consanguineous marriage without any prenatal, perinatal or postnatal complications. At six months of age, he developed generalized tonic clonic seizures occurring 2-3 times in a day for which he was taken to a pediatrician and started on Valproate 100mg/day. His mother noticed significantly delayed motor, language and adaptive milestones but she assumed it to be because of the seizures and did not seek any treatment for the same. The child first stared speaking disyllables at 4 years of age and started walking without support at 5 years of

age. The seizures too were under control and thereafter anti-epileptics were discontinued at 5 years of age. However the child would not make eye-contact, would remain engrossed in himself, not play with his siblings and hardly ever interact with anyone except with his mother. He would only speak in one or two words when he needed something. He would often keep flapping his arms or twisting his limbs in a peculiar manner and would be irritable if stopped. Around 7 years of age his mother noticed that he was very hyperactive, would not sit still and keep running about. He started attending school at 7 years of age but would struggle because of his poor academic performance and his hyperactivity and had to drop out after a year. When he presented to the psychiatry department he had recurrence of seizures after being seizure free since almost 8-9 years. On physical examination he had facial angiofibromas around his nose and cheeks. Mental status examination revealed ill-sustained eye to eye contact, hyperactivity with markedly reduce amount of speech and low cognitive profile. He also had motor tics which went unnoticed by parents. Vineyard Social Maturity Scale revealed a Social Quotient of 40 suggesting Moderate Intellectual Disability. Hematological investigations like hemogram, liver, kidney and thyroid function tests, blood sugar levels etc. were within normal limits. Magnetic resonance imaging of brain revealed multiple calcified and non-calcified sub-ependymal nodules of varying sizes in both cerebral hemispheres suggestive of Tuberous Sclerosis (Fig).

The diagnosis of TSC with Moderate Intellectual Disability along with ASD, ADHD predominantly hyperactive type, motor tic disorder and seizure disorder was kept as per DSM 5. [6] The seizures were controlled with Valproate 1000mg/day in divided doses. Atomoxetine 40mg/day was started in divided doses for control of hyperactivity and haloperidol 0.5mg twice a day was started for tic disorder. The boy had marked reduction in his hyperactivity and motor tics after a month and at 6months he was stable enough to start social skills therapy.

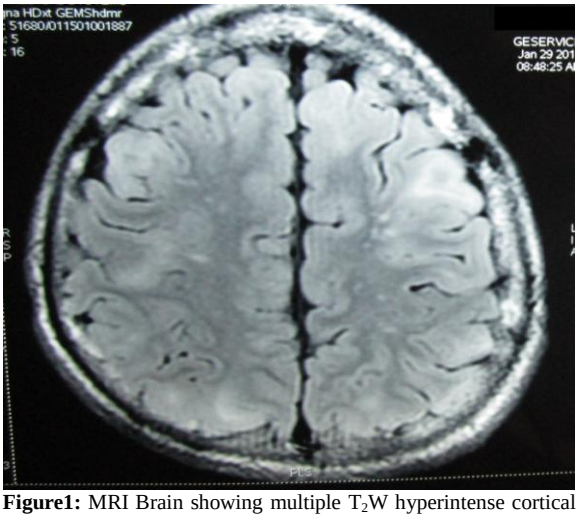

Figure1: MRI Brain showing multiple T_2W hyperintense cortical tubers in bilateral cerebral tubers

DISCUSSION

We highlight a case of TSC presenting with the full triad along with psychiatric co-morbidities like ASD, ADHD and motor tic disorder. Neurodevelopment disorders have a high prevalence in TSC with ADHD seen in 20-30%, [7] ASD in nearly 25%[5] where as tic disorders is a rare occurrence (3%). [8] DSM 5 has included the provision of dual diagnosis of ADHD and ASD following overwhelming research demonstrating the co-occurrence of these disorders (30-50%). [9] Co-occurrence of these conditions leads to lower quality of life, poorer adaptive functioning and greater social impairment. [9] Therefore the family members need to be prognosticated accordingly about the expectations from their ward. This is particularly useful for our case since it helps in management and prognosticating the parents. Valproate was selected as the antiepileptic of choice since the patient had previously shown good response to it. Studies report that Atomoxetine is tolerated well in patients with comorbid ADHD and ASD with minimal effect on tic disorder and therefore was selected for our patient. [10,11]

The present case stresses on detailed evaluation to avoid overlooking of co-morbid psychiatric diagnoses and

formulation of an optimum treatment regime consisting of pharmacological and non-pharmacological interventions.

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