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Original Research Paper

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A CASE REPORT OF TUBEROUS SCLEROSIS WITH ATTENTION DEFICIT HYPERACTIVITY DISORDER IN A CHILD

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ABSTRACT Tuberous sclerosis is an autosomal dominant genetic disorder characterized by the formation of hamartomas. These mostly affect the skin, eyes, brain, heart, kidneys and lungs. Cutaneous manifestations like adenoma sebaceum, shagreen patch etc., are apparent on physical examination. Tuberous sclerosis is associated with a wide range of behavioral, psychiatric, neurological, intellectual and psychosocial difficulties. Around 90% of individuals with tuberous sclerosis have neuropsychiatric manifestations. The diagnostic triad of tuberous sclerosis consists of mental retardation, adenoma sebaceum and epilepsy. Attention deficit hyperactivity disorder is also found as a comorbidity in tuberous sclerosis in higher percentage. Here we present a case of a child suffering from tuberous sclerosis and presenting with attention deficit hyperactivity disorder and seizures.

KEYWORDS : Tuberous sclerosis, attention deficit hyperactivity disorder, seizure.

INTRODUCTION

Tuberous sclerosis is a multisystem genetic disorder. It results in growth of hamartomas in multiple organs-skin, eyes, brain, heart, kidneys, lungs [1]. It is inherited in an autosomal dominant fashion. Cutaneous manifestations like adenoma sebaceum, shagreen patch are usually apparent on physical examination. It is usually diagnosed in childhood and infancy and affected individuals may present with varied neuropsychiatric manifestations. These may include behavioral, intellectual, neurological, psychiatric symptoms [2].

It affects approximately 1 in 6000 to 1 in 10,000 live births, with an overall prevalence of 1 in 20,000 [3]. Tuberous sclerosis results from mutations in the genes TSC1 and TSC2 and is known for causing epilepsy and intellectual disability [3]. Attention deficit hyperactivity disorder (ADHD) is a neuropsychiatric condition highly prevalent in the general population (3-7%) [4], but with a significantly higher prevalence in patients with tuberous sclerosis (30-50%) [5]. ADHD is characterized by inattention, hyperactivity and impulsivity. The exact mechanism of this underlying comorbidity is not fully understood,but cortical tubers, frontal epileptiform abnormalities and genetic mutation are thought to be responsible [6].

CASE STUDY

A 5 years and 3 months old, male child hailing from rural lower socio-economic background presented to the OPD of a tertiary care center in North-eastern part of India with complaints of loss of consciousness, stiffening of body, jerky movements of bilateral upper and lower limbs, up rolling of eyes, and incontinence. These episodes lasted for around five minutes followed by sleepy state of the child. The child has been having the episodes since last two months. Initially at an interval of 1-2 weeks between each episode. Since the last week, he had 2-3 episodes per day which prompted the parents to visit the hospital.

On further asking for any comorbidities, he was reported to have restless behavior, running and unable to sit still at one place, even at school. Touching and breaking things, disturbing others, interrupting others conversations, difficulty sustaining attention at class and at play, making mistakes in school work and poor academic performance. The child was also unable to follow simple instructions as per his peers. These symptoms were evident since the last 1 year.

Birth history was uneventful, while milestones including motor and language were delayed. Similar family history was not available and he was the only child of the parents.

Mental status examination showed hyperactivity, inability to follow instructions and poor eye contact. His language also showed poor sentence formation and poor vocabulary.

On physical examination, the child was found to have multiple small erythematous papules over the face especially over the nasolabial folds and the cheek (Figure 1). In the back, the child was seen to have an irregular, thickened, slightly elevated brownish colored patch (Figure 2)

ADHD Rating Scale-IV: Home Version was applied. The child scored 98 percentile each on hyperactivity -impulsivity and inattention domains. The Vineland Social Maturity Scale showed social age of 2.3 years. MRI brain showed sub-ependymal nodules (Figure 3), which is a common feature in tuberous sclerosis. EEG showed generalized epileptiform discharges.

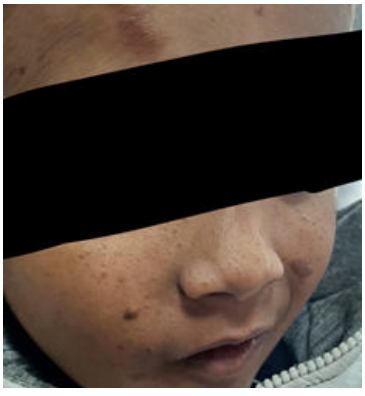

Figure 1: Adenoma sebaceum

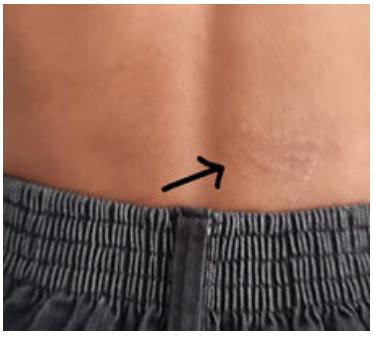

Figure 2: Shagreen patch

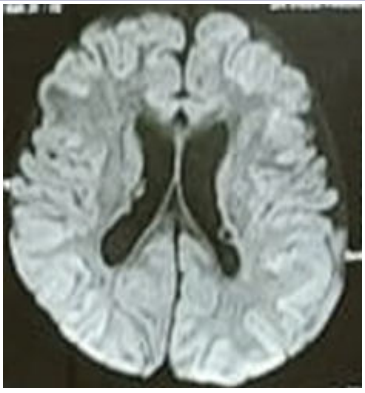

Figure 3: Subependymal Nodules on MRI brain.

CONCLUSIONS

The classic diagnostic triad for tuberous sclerosis complex, consisting of mental retardation, epilepsy and adenoma sebaceum were present in the patient. Presence of sub-ependymal nodules on MRI brain and shagreen patch also supported the diagnosis of tuberous sclerosis. ADHD was diagnosed as per DSM 5 criteria and application of ADHD rating scale.

The patient was started on Tab. Carbamazepine 300mg for seizures, Tab. Clonidine 75 mcg and Syrup

Risperidone 0.5 mg. After 3 weeks of follow-up there was some improvement in his symptom profile.

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