TUBEROUS SCLEROSIS DIAGNOSED BY INCIDENTAL COMPUTED TOMOGRAPHY SCAN

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ABSTRACT

Tuberous sclerosis or tuberous sclerosis complex or Bourneville disease was first described in 18th century by Bourneville, a French physician. The neurocutaneous syndrome is named for the firm whitish tuber like nodules arising from the cerebral convolutions. The most frequently involved organs are brain, kidneys, lungs, heart, skin and skeleton. It is a neurocutaneous disorder or phakomatosis which includes multiple benign tumours of the embryonic ectoderm. The clinical trial (Vogt Triad) of popular facial nevus, seizure and mental retardation is found in less than half of the patient. In this case report of Tuberous Sclerosis, it is detected at 2 years of age in a male patient presented with seizure and mental retardation by magnetic resonance imaging brain study.

KEYWORDS

Tuberous Sclerosis, Computer Tomography.


INTRODUCTION

Tuberous Sclerosis or Tuberous Sclerosis Complex is a genetic disorder characterized by the growth of numerous benign tumours in many parts of the body; including the brain, heart, lungs, eyes, kidneys, skin and other organs, leading to significant health problems like seizures, intellectual disability, autism or developmental delay. It is an autosomal dominant disorder. Its incidence is 1:10,000-50,000. It is thought to be associated with disordered migration of dysgenetic neurons along abnormal radial glial fibres. In the majority of cases nearly 80% of the mutation has been narrowed down to two tumour suppressor genes that are TSC1 encoding HAMARTIN on chromosome 9q32-34 and TSC2 encoding TUBERIN on chromosome 1q13.3 (Account for most cases). Spontaneous mutation occurs in 50-80% of cases with remainder inherited as autosomal dominant condition. Epidemiologically it has an incidence of 1:6000-12000, with most of them are sporadic cases.

A 2 year old male baby presented with seizure disorder since birth and delayed developmental milestones with no history of familial genetic disorders was referred to radiology department. MRI brain study reveals multiple well defined T1 hypointense, T2 hyperintense lesions involving subcortical white matter of bilateral cerebral hemispheres. Most of these lesions showing central area of suppression on FLAIR sequence which suggests cystic degeneration with surrounding FLAIR hyperintensities around these lesions, all this features suggests white matter degenerative changes with most of them showing cystic changes. Multiple T1 intermediate T2 hypointense and FLAIR hyperintense lesions arising from the lateral wall of bilateral lateral ventricles which suggests subependymal nodules.

On Ultrasound there is presence of Cardiac Rhabdomyoma arising from proximal part of interventricular septum, which is confirmed later by echocardiography.

DISCUSSION

The classical clinical triad of TSC symptoms are mental retardation, seizures, and cutaneous angiofibroma (Formerly called adenoma sebaceum), reported by Vogt in[3]. Mental retardation and seizures are both neurologic manifestations of TSC.[5] The overall incidence of mental retardation is 38 percent to 80 percent in TSC, while epilepsy is one of the most prevalent manifestations of TSC, occurring in more than 80 percent to 90 percent of patients with TSC.[3,4] These neurological manifestations are highly related to cortical tubers, which are detected in 80 percent of patients.[3] In this case, mental retardation and seizures were detected, also cortical and subcortical tubers were found on brain MRI scans. The skin lesions of TSC, such as facial angiofibroma and hypomelanotic macules, are detected in more than 90 percent of patients with TSC.[3] but only hypomelanotic macules observed in this case. It has been reported that 6 percent of patients with TSC have none of these three findings.[3] The MR imaging appearance of intracranial manifestations of tuberous sclerosis varies with age[6]. The nodular subependymal and linear parenchymal tuberous sclerosis lesions in children and adult differ in the form of hyperintense on T1 weighted images and hypointense on T2 weighted images as opposed to reverse signal intensity in adults or older patients. The scarce myelination helps to identify white matter anomalies, which become less evident as myelination progresses. Tuberous sclerosis or tuberous sclerosis complex is a lifelong condition, therefore individuals should be regularly monitored[7]. It must be included in the differentials of children presenting with seizures, developmental delay, and mental retardation[8].
T2 weighted plain MRI axial, sagittal and coronal image of brain showing white matter degeneration in the form of hyperintense signal in the cortical and subcortical white matter.

T1 weighted and FLAIR axial image of brain showing subependymal nodules arising from the lateral wall of the ventricles associated with suppression of white matter degeneration suggestive of cystic degeneration.

Differential Diagnosis
1. Complex partial seizure.
2. Hydrocephalus.
3. Infantile spasm (West syndrome).
4. Intellectual disability.
5. Lennox Gastaut syndrome.

CONCLUSION
Tuberous sclerosis is an important genetic disorder which affects the patient and the family in various ways. Now, due to an understanding of its pathogenesis, multiple drug therapies are available for certain manifestations of the disease. But the patient, along with symptomatic control of seizures, should also be offered special schooling, and occupational therapy. Surgery like laser treatment, may be useful for treatment of skin lesions. Multiple cases have been reported highlighting involvement of different organs. In majority, there was probable diagnosis with one major plus and one minor positive feature. In our case, however, the interesting feature was the presence of four major and 2 minor criteria, making it a very conspicuous presentation of tuberous sclerosis complex.

REFERENCES