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Case Report

Tuberous sclerosis-A case report

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Abstract

A18 yr old male patient presented with seizures. On thorough clinical evaluation and radiological investigation, he was diagnosed as a case of tuberous sclerosis. This case report emphasizes the importance of complete evaluation of a case presenting with seizures and inclusion of TSC (Tuberous Sclerosis Complex) as a differential diagnosis in children presenting with seizures, developmental delay and mental retardation.
Keywords: tuberous sclerosis, adenoma sebaceum

1. Introduction

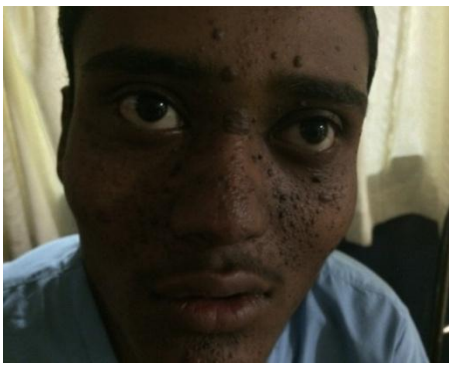
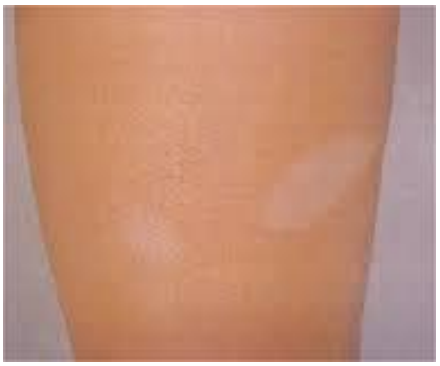
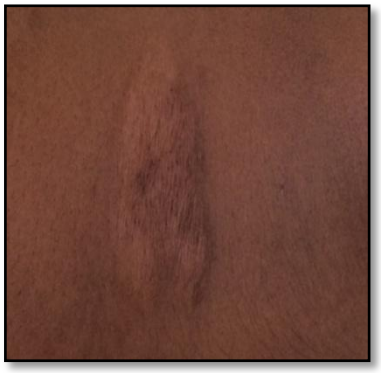
Multiple Von Recklinghausen first described tuberous sclerosis in 1862. Desire-magloire Bournville (a French physician) coined the term sclerosetubercuse, from which the name of disease has evolved. Sherlock coined the term EPILOIA tuberous (Epi-epilepsy, Loi-low intelligence, an adenomasebaceum) encompassing the clinical trial of tuberous sclerosis. As manifestations of this disease are variegated in nature, the term tuberous sclerosis is widely used. It is an autosomal dominant inherited disease, being associated with at least two separate chromosomes (TSC found on chromosome 9q34 and TSC on chromosome16p3). The disease is transmitted either through genetic inheritance or as a spontaneous genetic mutation. Incidence of tuberous sclerosis is 1:6000 births and about 2/3 cases are sporadic, occurring in the absence of family history of the disorder. Clinical diagnosis is easy when patient presents with classical triad of seizures, mental retardation and adenoma sebaceum. However in patient presenting with an incomplete form of tuberous sclerosis, mistakes in the diagnosis are possible. We here report a case of 18 yr old male who presented with seizures and on evaluation was diagnosed as a case of tuberous sclerosis.

2. Case Report

An 18 year old male presented to the emergency department of our hospital with history of convulsions, vomiting, headache and loss of balance since last 15 days. He was known epileptic and had been on anti-epileptic medications for 5 years from the age of three to eight years. After this treatment patient was asymptomatic till 15 days back. Convulsions were on and off being generalized tonic clonic in nature and were associated with up rolling of eyeballs, biting of tongue and frothing from mouth. He had no fecal or urinary incontinence and no history of seeing abnormal objects or hearing abnormal sounds before fits. His past history was significant as he was reported to be mentally retarded. In emergency room diazepam was administered to control convulsions. On physical examination vitals were normal. Examination of skin revealed multiple nodular lesions over face, depigmented spots on the back, yellow thickened area over the back.

1. Multiple small nodular lesions over face-**adenoma sebaceum (figure 1)**
2. A yellowish thickened leathery area over lower back- **shagreen patch (figure 2)**
3. Depigmented patch 2-3 cm over back- **ash leaves (figure 3)**

Fig.1 Adenoma Sebaceum **Fig.2 Shagreen Patch** **Fig. 3 Ash leaves**

After an episode of convulsions he was found to be fully conscious, oriented with muscle tone, reflexes and power being normal. Ataxia was present, tendon gait was not possible and rest of the sensory examination was normal. He had poor past memory and was unable to perform simple arithmetic calculations. Pupils were bilaterally reacting and nystagmus was present bilaterally. Rest of the examination was unremarkable.

His laboratory investigations revealed normal blood counts and routine urinalysis. Renal function test, serum electrolyte and blood sugar levels were normal. CT and MRI of brain showed tuberum sclerosis with pedunculated tuber extending into ventricle. Fundoscopy revealed s/o retinal tuber with papilloedema. ECG and Echocardiography were normal.

All clinical features were suggestive of a classical picture of tuberous sclerosis. Molecular analysis of gene TSC1 and TSC 2 in chromosome 9 and 16 respectively could not be performed due to constraints. Patient was discharged after 2 weeks on carbamazepine and levetiracetam. Condition was stable and regular follow up was advised.

He was thoroughly investigated as follows:

Table 1: Investigation Report

INVESTIGATION	REFERENCE RANGE	LAB VALUE
Haemoglobin	11.9 gm/dl	13.8-17.2 gm/dl
Total WBC count	5600cells/mm3	3500-10500/mm3
Platelate count	3,12,000cells /mm3	1.5-4.5/ mm3
Liver function test	Normal	-
Renal function test	Bl.urea-25 mg/dl Sr.creatinine-1.1mg/dl	10-45 mg/dl 0.6-1.5 mg/dl
HIV	Non reactive	-
HbsAg	Negative	-

Table 2: Investigation Report

ECG	Normal
X-RAY CHEST	Normal
CT BRAIN(PLAIN)	tuberum sclerosis with Astrocytoma
MRI BRAIN(PLAIN)	tuberum sclerosis with pedunculated tuber extending into ventricle.
USG ABDOMEN	Normal
FUNDOSCOPY	Retinal tuber with papilloedema

3. Discussion

Tuberous sclerosis shows a wide variety of clinical expression. Some individuals are severely affected, while others have very few clinical features. Tuberous sclerosis is characterized by the development of unusual tumor like growths (hamartoma) in brain, skin, retina and other viscera. As multiple organs are involved, there is wide variability in presentation. Most important hamartomas are cerebral cortical tubers which are regions of abnormal cortical architectures with distinctive large neuronal cells. These hamartomatous swellings resemble potatoes and hence are referred to as tubers. Cortical tubers cause some of the important clinical manifestations of tuberous sclerosis i.e. epilepsy, mental retardation and abnormal behaviour including autism. Epilepsy occurs in 80 to 90 percent of all patients, with positive correlation with subnormal intelligence. Cutaneous lesions are present in 96 percent of the patients. This include facial angiofibroma (adenoma sebaceum), subungle fibromas, shagreen patches. Two types of renal lesions occur in patients with tuberous sclerosis i.e. angiomyolipomas and renal cysts. They may be found independently altogether and may be unilateral, bilateral, single or multiple. Angiomyolipomas are benign in nature and asymptomatic but spontaneous rupture and subsequent haemorrhage into retroperitoneum may occur. In the heart, the most frequent and characteristic type of tumour is cardiacrhabdomyomas. Incidence of cardiac rhabdomyomasin children with tuberous sclerosis is higher than in adult patients with tuberous sclerosis. It has been suggested that such lesions tend to regress in early infancy and adolescence.

The present patient had tuberous sclerosis characterized by classical features such as seizures, mental retardation and facial angiofibromas. Incomplete forms of tuberous sclerosis may present with acute complications such as hematuria, retroperitoneal haemorrhage or pneumothorax. It is estimated that more than 1 million people are known to suffer from tuberous sclerosis. It is an underestimated figure as many cases remain undiagnosed due to variegated clinical presentation. Intervention programs, including special schooling and occupational therapy may benefit individuals with special needs and developmental issues. Surgery including derma- ablation and laser treatment may be useful for treatment of skin lesions. There is no cure as such for tuberous sclerosis. Drug therapy for some of the manifestations of TSC is currently in the developmental stage. Prognosis of disease depends on severity or multiplicity of organ development. About a quarter of severely affected infants are thought to die before the age of 10 years, and 75 % die before 25 years of age. However prognosis for individuals diagnosed late in life with few cutaneous signs, depends on the associated internal tumours.

This presentation aims at considering the rarity of the disease and listing it in the differential diagnosis of children presenting with seizures, skin manifestations and mental retardation.

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