Case Report

Tuberous Sclerosis Complex: A Case Report

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Tuberous sclerosis complex is an uncommon neurocutaneous syndrome characterized by development of benign tumors affecting multiple body systems including skin, brain, retina and viscera. The management of these patients is multidisciplinary, involving specialists from different fields. Since it can present with a wide range of manifestations, the quality of life and prognosis depends on the particular abnormalities seen in a patient. Here, we report a case of an 11 year old boy with typical clinical and radiological features of tuberous sclerosis.

Key Words: adenoma sebaceum, subependymal giant cell astrocytoma, nonrenal hamartoma.

Tuberous sclerosis also known as Bourneville disease is a rare phacomatosis characterized by development of benign tumours[1] affecting multiple organ systems including skin, brain, retina, kidney, heart and lungs. It may present in sporadic (60%) or autosomal dominant (40%) manner with a prevalence of 1 in 6000[1] live births affecting both sexes and all ethnicities. We report a case of an 11-year-old boy with distinctive clinical and radiological features of tuberous sclerosis.

CASE REPORT

An 11-year-old boy presented to our OPD with complaints of blurring of vision in both eyes for past few months and headache and vomiting for 15 days. Headache and vomiting were more noticeable in early morning after waking from sleep. He also had abdominal pain for 15 days.

On general physical examination, multiple well-defined brownish papules were seen on nose and cheeks in a typical butterfly pattern. A fibrous patch of around 2cm was present on forehead and right cheek. A skin tag was present in left preauricular area. A nodular growth involving nail bed of left fourth digit was seen. On ocular examination his best-corrected visual acuity was 6/18 OD with -1.00 DS/-0.50 DC x30 and 6/9 OS with -1.00 DS. Anterior segment of both eyes was normal. On dilated fundus examination, established papilledema was seen. He was advised to get an MRI Brain done. MRI Brain revealed non-communicating hydrocephalus secondary to intraventricular mass most likely a sub-ependymal giant cell astrocytoma. It also showed multiple sub-ependymal nodules. For abdominal pain, he was referred to pediatrician who advised him abdominal ultrasound followed by a CT scan abdomen, which revealed the presence of a large extra-renal hamartoma. Parents also gave history of seizures since childhood for which the child had taken sodium valproate for some time. Currently, EEG revealed non-focal seizure pattern. Heart and lung imaging was normal. The child was result of a non-consanguineous marriage and no family member had similar manifestations. On the basis of above findings diagnosis of tuberous sclerosis was made. For the hydrocephalus, ventriculo-peritoneal shunt surgery was planned by neurosurgery department. For the extra-renal hamartoma, observation with yearly CT Abdomen was advised by the pediatric oncologist. For early detection of retinal hamartomas, yearly fundoscopy was advised.

Table 1: Diagnostic criteria for tuberous sclerosis[1].

Major Features	Minor Features
Hypo-melanotic macules (>3 at least 5mm diameter)	"Confetti" skin lesions
Angiofibromas (>3) or fibrous cephalic plaque	Dental enamel pits (>3)
Ungual fibromas > 2	Intraoral fibromas (>2)
Shagreen patch	Retinal achromic patch
Multiple retinal hamartomas	Multiple renal cysts
Cortical dysplasias	Non-renal hamartomas
Sub-ependymal nodules	
Sub-ependymal giant cell astrocytoma	
Cardiac rhabdomyoma	
Lymphangioleiomyomatosis (LAM)	
Angiomyolipomas>2	

DISCUSSION

TSC is characterized by hamartomas of multiple organs from all primary germ layers[2]. It is an autosomal dominant disorder with nearly complete penetrance with variable expressivity. The mutation[3,10] in genes TSC1 encoding hamartin and TSC2 encoding tuberin result in formation of hamartomas in various organs. The classic triad[4,8] of epilepsy, mental retardation and adenoma sebaceum is seen in only a minority of patients.

TSC has dermatological manifestations[1,6] like hypomelanotic macules (Ash leaf spots; 90%), facial angifibromas (adenoma sebaceum; 75%), ungual hamartomas (20%), skin tags, Shagreen patch (50%)

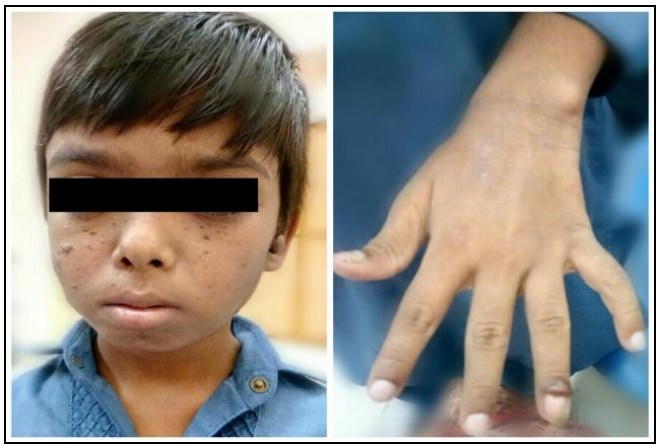

Fig. 1: Clinical photograph of the patient showing adenoma sebaceum, right cheek fibrous plaque, left preauricular skin tag and ungual fibroma.

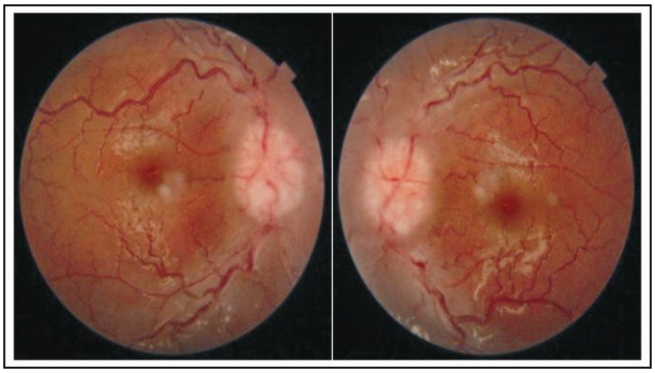

Fig. 2: Fundus photographs showing papilledema.

and café-au-lait macules. Hypo-melanotic macules are at least 5mm in size and typically appear at birth or in infancy on limbs, trunk or scalp. Adenoma sebaceum appear between two and five years as fibro-angiomatous red papules in a butter fly distribution around nose and cheeks. Shagreen patch is an area of diffuse thickening over lumbar region having an orange peel appearance that usually appears in first decade of life. Fibrous cephalic plaques can be seen on forehead as well as face in about 25%. Subungual and

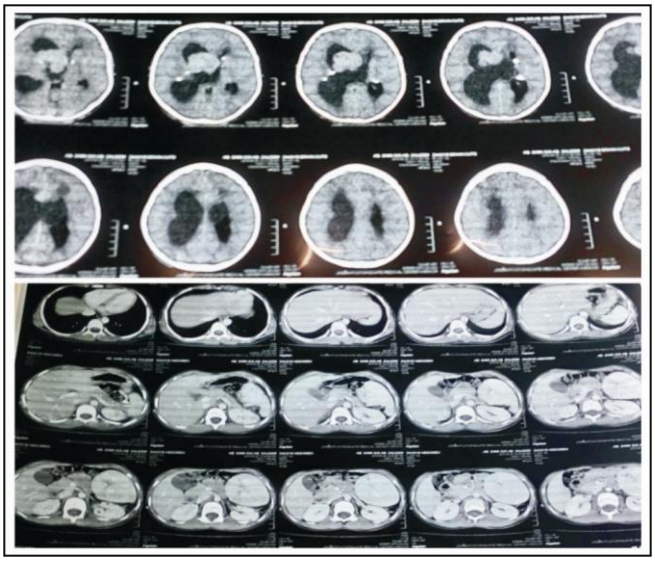

Fig. 3: CT Brain showing subependymal nodules and giant cell astrocytoma. CT Abdomen showing nonrenal abdominal hamartoma.

periungual fibromas[1] appear in second decade or later. Confetti skin lesions are hypo-pigmented macules of 1 to 3 mm in size[5]. In oral cavity, dental enamel pits and fibromas may be seen. Among ocular findings[1], retinal

astrocytoma, retinal achromic patch, patchy iris hypopigmentation and atypical iris coloboma may be seen. Sub-ependymal nodules[1] (SEN; 80%) are benign growths that may be detected prenatally or at birth. Sub-ependymal giant cell astrocytomas[1] (SEGA; 5-15%) arise from SEN mostly during childhood or adolescence. They are benign and slow growing but can cause obstructive hydrocephalus. Seizures, learning difficulties, mental retardation and psychiatric disturbances can be present[8,9]. Cardiac rhabdomyomas[1] are frequently seen during prenatal life but regress later on and may cause arrhythmias. Pulmonary lymphangioleio-myomatosis[1] may be seen. Angiomyolipomas[1] usually affect kidneys but can affect other organs too. Multiple renal cortical cysts can be present too.

Updated diagnostic criteria[1] according to the recommendations of 2012 International TSC Consensus Conference is given in table 1. Two major or one major with two minor features make a definite diagnosis.

Our patient had three major features including facial angiofibromas/fibrous cephalic plaque, subependymal nodules, subependymal giant cell astrocytoma and one minor feature i.e., non-renal abdominal hamartoma which led us to the clinical diagnosis of definite TSC. Ungual fibroma and renal cortical cyst were also present but they did not meet the criterion.

It is quite evident from our case report that an array of findings can be seen in tuberous sclerosis. The approach to management is therefore multidisciplinary and symptomatic to a large extent. Likewise, the quality of life depends on the particular manifestations in a patient.

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