Tuberous Sclerosis

MAH MOLLAH[a] , S RAHMAN[b]

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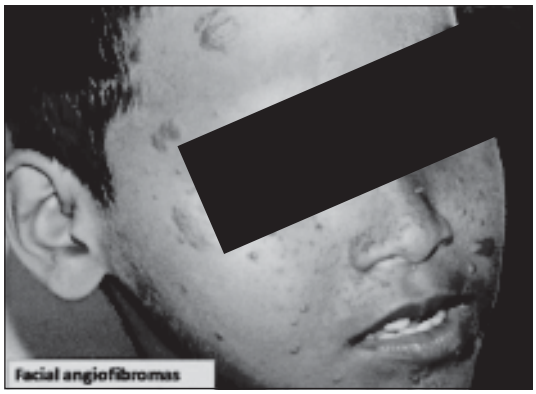
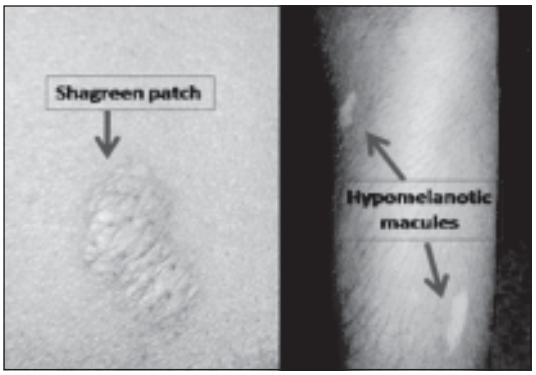
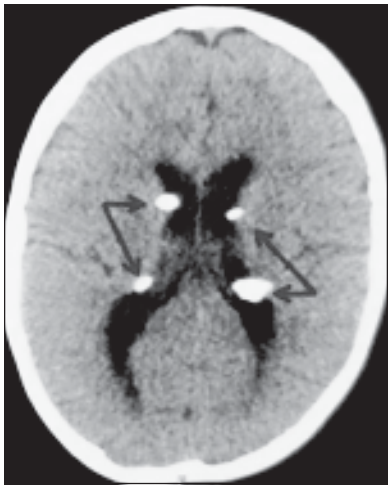
A 12½ years old boy presented with recurrent seizures, which had started since 7 months of age and gradually became more frequent and difficult to control. On examination the boy was found apathetic, non-communicating and severely mentally retarded. He had frequent myoclonic seizures, which involved all 4 limbs lasting for 15-20seconds.On skin survey there were multiple tiny red nodular angio-fibromas over nose and cheeks, 4-5 hypomelanotic macules (ash leaf macule)seen on the back of trunk and thigh. In addition, few*shagreen patches* were also found on back.

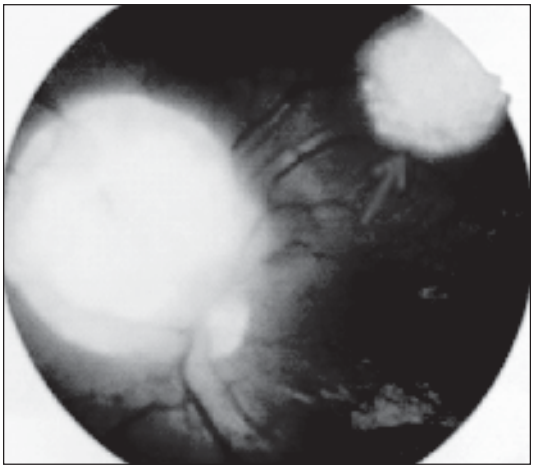




EEG was done which showed hypsarrhythmic pattern. On CT scan of brain there were subependymal noduleswith calcification projecting into the lateral ventricular cavity from the wall with candle-dripping appearance. Fundoscopic examination revealed left sided retinal hamartoma.



Subependymal nodules (calcified)



Left sided retinal hamartoma

a. Prof. Md Abid Hossain Mollah, Prof. of Paediatrics, Dhaka Medical College, Dhaka.

b. Dr. Shegufta Rahman, Neonatologist, National University, Singapore.

Address of Correspondence: Prof. Md Abid Hossain Mollah, Prof. of Paediatrics, Dhaka Medical College, Dhaka, Mob- 01711530323

Based on the clinical and investigation finding the boy was diagnosed as a case of Tuberous sclerosis complex (TSC) and vigabatrin was added to control seizure.

TSC is inherited as an autosomal dominant trait with variable expression. Spontaneous genetic mutations occur in 2/3rd of the cases. It is an extremely heterogeneous disease with a wide clinical spectrum ranging from totally asymptomatic to severely affected patients. The diseaseaffects many organ systems mainly skin and brain,also heart, kidney, eyes, lungs, and bone.TSC is diagnosed when at least 2 major or 1 major plus 2 minor features are present as follows-

Major Features	Minor Features
Cortical tuber	Cerebral white matter migration lineMultiple dental pits
Subependymal nodule	Gingival fibromas
Subependymal giant cell astrocytoma	Bone cysts
Facial angiofibroma or forehead plaque	Retinal achromatic patch
Ungual or periungual fibroma (nontraumatic)	Confetti skin lesions
Hypomelanotic macules (>3)	Nonrenalhamartomas
Shagreen patch	Multiple renal cysts
Multiple retinal hamartomas	Hamartomatous rectal polyps
Cardiac rhabdomyoma	
Renal angiomyolipoma	
Pulmonary lymphangioleiomyomatosis	

The hallmark of TSC is the involvement of the CNS that includes cortical tuber and subependymal nodules, which are seen on Brain MRI or CT scan.[3,5] These lesions do not cause any problem; but in 5-10% of cases subependymal nodules can grow into subependymal giant cell astrocytomas (SEGAs) thatwhen blocks CSF circulation, gives rise to hydrocephalus and requires immediate neurosurgical intervention. The common neurologic manifestations of TSC consist of epilepsy, cognitive impairment, and autism spectrum disorders. Many a times, the seizures are difficult to control and, at a later age, they may turn into myoclonic epilepsy. [1,5]

A careful search for the typical skin and retinal lesions should be done in suspected case of TSC presenting with seizure disorder or autism spectrum disorder. [2,3,5] More than 90% of cases hypomelanotic macules (**ash leaf)** are seen on trunk and extremities. Facial angiofibromas develop between 4 and 6 yr of age, which appear as tiny red nodules over the nose and cheeks and are sometimes confused with acne. Later, they enlarge, coalesce, and assume a fleshy appearance. A *shagreen patch* is also characteristic of TSC and consists of a roughened, raised lesion with an orange-peel consistency mostly in the lumbosacral region. During adolescence or later, small fibromas of skin may form around fingernails or toenails in 15-20% of cases.[1,2,3]

Retinal lesions in TSC are of 2 types: hamartomas (elevated mulberry lesions or plaquelike lesions) and white depigmented patches (similar to the hypopigmented skin lesions).[4]

Approximately 50% of children with TSC have cardiac rhabdomyomas, which can cause congestive heart failure and arrhythmias. 75-80% of patients crossing 10 years of age have benign angiomyolipomas tumors in kidneys. Lymphangioleiomyomatosis (LAM) is the classical pulmonary lesion in TSC that affects only women after the age of 20 year.[1,2,3,6]

There is no cure to TSC. Treatment is symptomatic. To optimize quality of life parents need to be educated and counseled properly about the disease. A routine follow-up has to be planned as per recommendation.[4,6]

References:

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