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A rare case of tuberous sclerosis with rhabdomyoma

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Abstract
Tuberous sclerosis is a neurocutaneous syndrome characterized by abnormalities of both the integument and central nervous system. We present a case of tuberous sclerosis with rhabdomyoma in the heart. A 5 years old female child was presented with seizures. On examination she was found to be having neurocutaneous markers and systolic murmur as positive findings. Other examinations were with in normal limits. On investigations findings were suggestive of rhabdomyoma heart, on echocardiography, dysplastic cortex on MRI and hypsarrthmia on EEG. These findings were consistent with diagnosis of tuberous sclerosis.

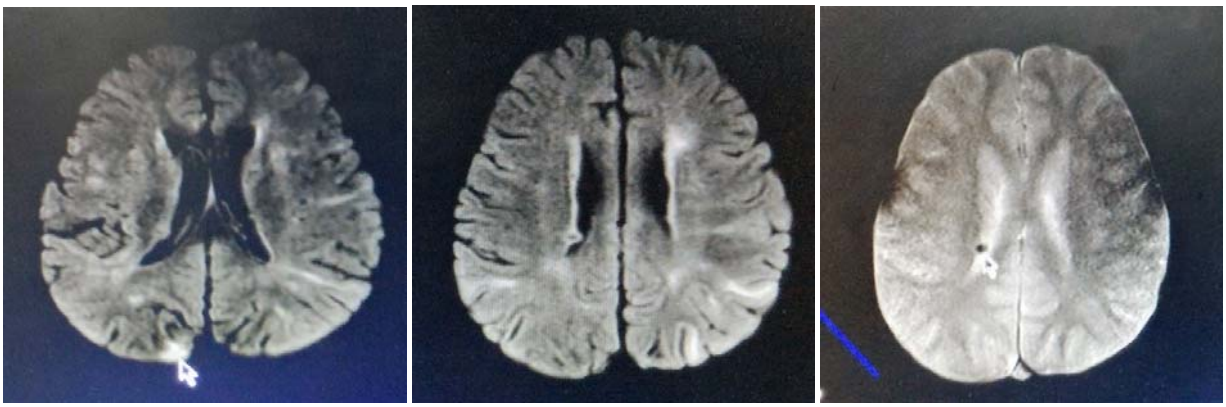
Keywords: Infantile seizures, rhabdomyoma, neurocutaneous

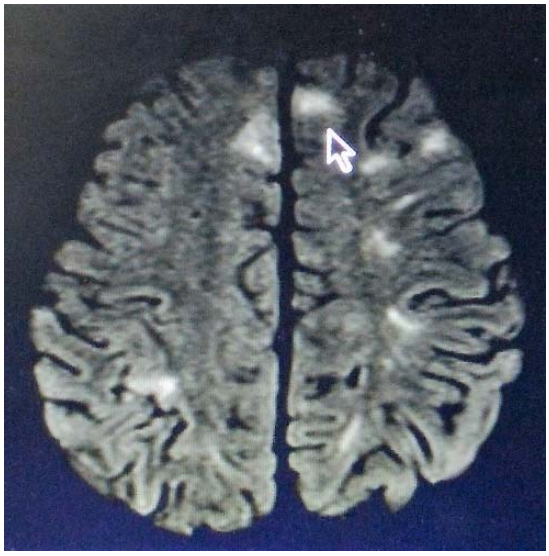
Introduction
Tuberous sclerosis complex is an autosomal dominant neurocutaneous syndrome with a high incidence of sporadic cases and variable clinical expression [1]. It has an estimated frequency of 1/6000 [2]. Major manifestations of tuberous sclerosis include skin lesions in more than 95%, autism and seizures in 85%, kidney disease in 60%, mental retardation in 50%, and cardiac rhabdomyoma in 50% of the cases [3]. Mental retardation and autism are more in tuberous sclerosis patients who presents with generalized seizures in the first 2 years of life. We present a 5 years old female child with features suggestive of tuberous sclerosis associated with rhabdomyoma heart.

Case report
A 5 years old female child born out of a non-consanguineous marriage to a primi mother at 38 weeks of gestation with normal antenatal scans presented with abnormal jerky movements of both upper and lower limb for past 3 months. The development was normal for age, except for social adaptations and speech and language delay. On examination vital parameters were with in normal limits. Patient had one hypo-pigmented patch on face in front of left ear. Clinically central nervous system examination was normal. On examination of cardio vascular system systolic murmur was present. Other systems were normal. During hospital stay child was found to have infantile spasms. On history and clinical examination of parents, mother had adenoma sebaceum on face and Shagreen patch in right lumbosacral region.

Investigations
Haemogram was normal except haemoglobin was 7.7gm% other parameters were with in normal limits. Electroencephalography (EEG) carried out showed hypsarrhythmic pattern. Magnetic resonance imaging (MRI) brain findings showed cortical sub-cortical tubers with radial lines in bilateral frontoparietal lobes and a calcified sub-ependymal nodule s/ o tuberous sclerosis. Ultra sonogram abdomen was normal. Electrocardiogram showed ST segment elevation in leads V1, V2, V3, III and aVR and ST segment depression in leads I and aVL. A two dimensional Echocardiography (2d-ECHO) revealed huge intra-pericardial, myocardial mass seen in left ventricular anterior wall homogenous and smooth in appearance with no intra-tumor haemorrhage measuring 31mm X 36mm with no inflow or outflow obstruction. It was suggestive of rhabdomyoma. Ophthalmological examination was normal. Brain stem Evoked Response Audiometry (BERA) findings were with in normal limits.
Based on the criteria a diagnosis of tuberous sclerosis was made and child was started on oral valparin and clobazam. On follow up after 3 months. There was no further episode of infantile spasms. On follow-up the child was seizure free and without any evidence of cardiac failure.

MRI Findings





MRI showing cortical sub-cortical tubers with radial lines in bilateral frontoparietal lobes and a calcified sub-ependymal nodule

Discussion

The first complete description of tuberous sclerosis was given by Bourneville in 1880 [2]. Tuberous sclerosis is a disorder of cellular differentiation and proliferation that can affect the brain, skin, kidneys, heart, and other organs. Abnormal neuronal migration plays a major additional role in neurological dysfunction. Two genes responsible for tuberous sclerosis are TSC1 at chromosome 9q34 (hamartin) and TSC2 on 16p13.3 (tuberin) [4]. Diagnostic criteria include major and minor features.

Major Features

1. Facial angiofibroma or forehead plaques.
2. Non-traumatic ungual or periungual fibroma.
3. Hypomelanotic macules.
4. Shagreen patch (connective tissue nevis).
5. Multiple retinal nodular hamartomas.
6. Cortical dysplasias (include tubers and cerebral white matter radial migration lines).
7. Subependymal nodule.
8. Subependymal giant cell astrocytoma.
9. Cardiac rhabdomyoma, single or multiple.
10. Renal Angiomyolipoma.
11. Lymphangioleiomyomatosis.

Minor Features

1. More than three randomly distributed pits in dental enamel.
2. Hamartomatous polyps.
3. Intra oral fibromas.
4. Non-renal hamartoma.
5. Retinal achromic patch.
6. “Confetti” skin lesions 1-2 mm hypomelanotic plaques.
7. Multiple renal cysts.

Diagnosis of tuberous sclerosis is established when two major features or one major plus two minor features can be demonstrated.

This child had three of the major features: skin lesions, rhabdomyoma heart and changes on MRI brain dysplastic cortex.

Recent two-dimensional (2D) ECHO studies have reported that 50% of the patients with tuberous sclerosis have cardiac rhabdomyomas. Incidence of tuberous sclerosis is as high as 59-80% in patients with confirmed fetal rhabdomyomas [5]. Cardiac rhabdomyomas are hamartomas; they tend to be multiple and evidence exists that they involute with time.

These lesions sometimes are evident on prenatal ultrasound testing and most of the patients with cardiac dysfunction present soon after birth with heart failure. A few children later develop cardiac arrhythmias or cerebral thromboembolism from the rhabdomyomas. Some patients stabilize after medical treatment with digoxin and diuretics and eventually improve while others require surgery. For existing rhabdomyomas surveillance studies should be done every 6-12 months until stabilization or involution occurs. In tuberous sclerosis vigabatrin is the effective treatment option for infantile spasms. Other drugs which may be useful are topiramate and Adrenocorticotropic hormone. Oral rapamycin has been shown to cause regression of astrocytomas associated with tuberous sclerosis and may eventually be an alternative to operative therapy. This diesease advances slowly. Of the severe cases, approximately 30% die before the 5th year and 50-75% before attaining the adult age. The child with infantile spasm is at great risk of later intellectual deficit [6].

Conclusion

Tuberous sclerosis is a rare disorder manifestations may vary. Severe cases can present at birth. Progression of disease slow long term follow up required. Neurodevelopment testing at school entrance, MRI brain,

and renal ultrasound every 1-3 years during the childhood and adolescence should be carried out.

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