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Case Report

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IMAGING FEATURES OF TUBEROUS SCLEROSIS WITH A CARDIAC RHABDOMYOMA – A CASE REPORT.

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ABSTRACT

Tuberous sclerosis is a rare autosomal dominant neurocutaneous disorder characterized by benign congenital tumors of multiple organs. The diagnosis is made by a triad of seizures, mental retardation and adenoma sebaceum. Imaging plays an important role in diagnosis of tuberous sclerosis characterized by cortical or subependymal tubers and white matter abnormalities in the brain. Other systemic involvement include cardiac rhabdomyoma and renal angiomyolipoma.

KEYWORDS: Neurocutaneous, sebaceum, rhabdomyoma.

CASE REPORT

A 9 month old male child came with history of seizure disorder. He had recurrent episodes of seizures with first episode at 1 month of age. Patient presented with depigmented area of skin on the left temporal region with irregular margins (Figure 1). Birth history was normal with and no history of NICU admission. History of developmental delay was present. Patient was not able to sit or hold his neck without support. Chest radiograph showed left atrial appendage appeared to be dilated with a lobulated contour which could be a dilated left atrium, anterior mediastinal mass**,** left atrial myxoma**,** partial pericardial defect with herniation of LA appendage (Figure 2A and 2B). Hence 2D Echo was done and it revealed mass in the left ventricle which was suggestive of a myxoma (Figure 3).

MRI and CT scan were performed for the evaluation of seizures and myxoma. MRI Brain revealed thickened cortex in the left frontal region (Figure 4A and 4B). Few subependymal nodules were noted along the bilateral subependymal region, including the left caudo thalamic groove (Figure 5A and 5B). Associated focal indentation of the inter hemispheric fissure was seen. Calcification/hemorrhage beneath the dysplastic cortex also noted (Figure 5C, 6A and 6B).

Radial migration lines were seen radiating from the body of the lateral ventricles bilaterally (left >right) appearing hyperintense on T2W images (Figure 7).

CT brain revealed multiple subcentimeteric focal areas of calcification in the bilateral periventricular region including the caudo-thalamic groove (Figure 8A and 8B). CT thorax showed a diffuse wall thickening of the left lateral ventricle (Figure 9A and 9B).

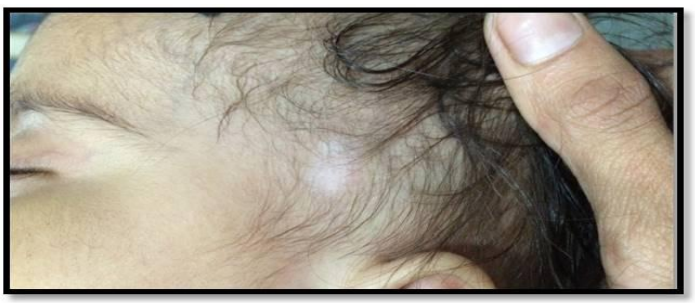

Figure 1:

Patient presented with depigmented area of skin on the left temporal region with irregular margins.The most common skin manifestation of TS are hypodermic macules, also known as ash leaf spots.

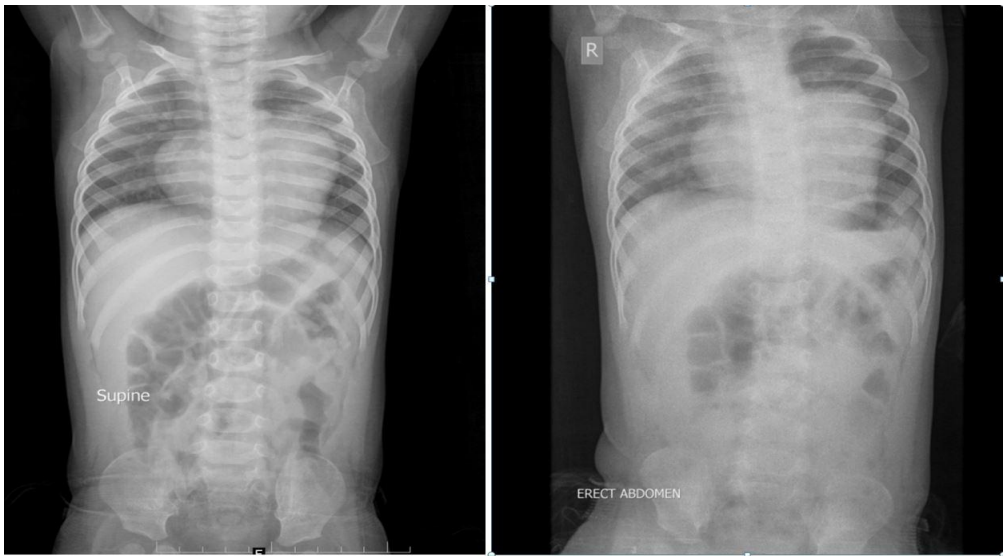

Figure 2: Plain radiograph – Chest supine (A), Erect abdomen (B)

Chest and erect abdomen radiographs showing dilatation of left atrial appendage with a lobulated contour.

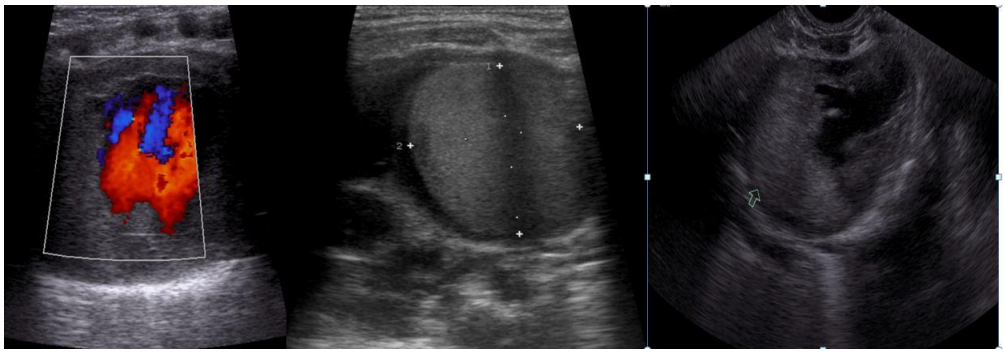

Figure 3: 2D ECHO

2D ECHO showing wall thickening of left ventricle of heart, measuring approx. 32.9 x 30.2 mm.

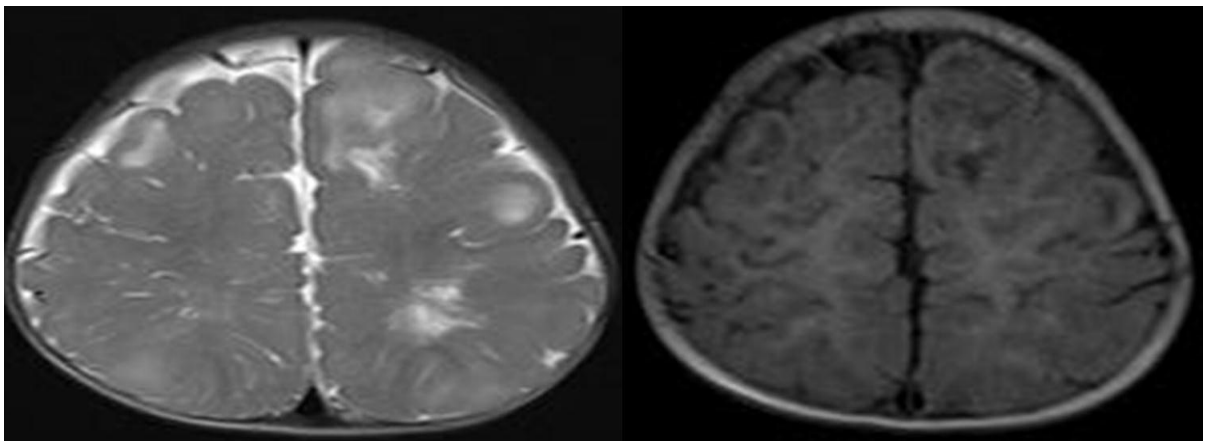

Figure 4: MRI Brain (plain axial) – T2WI (A) and T1WI (B)

MRI brain showed multiple areas of altered signal intensity in the cortical-subcortical regions spread diffusely in bilateral cerebral hemispheres appearing hyperintense on T2WI and hypointense on T1WI. The involved cortex appeared thickened and hyperintense to surrounding cortex on T1WI.

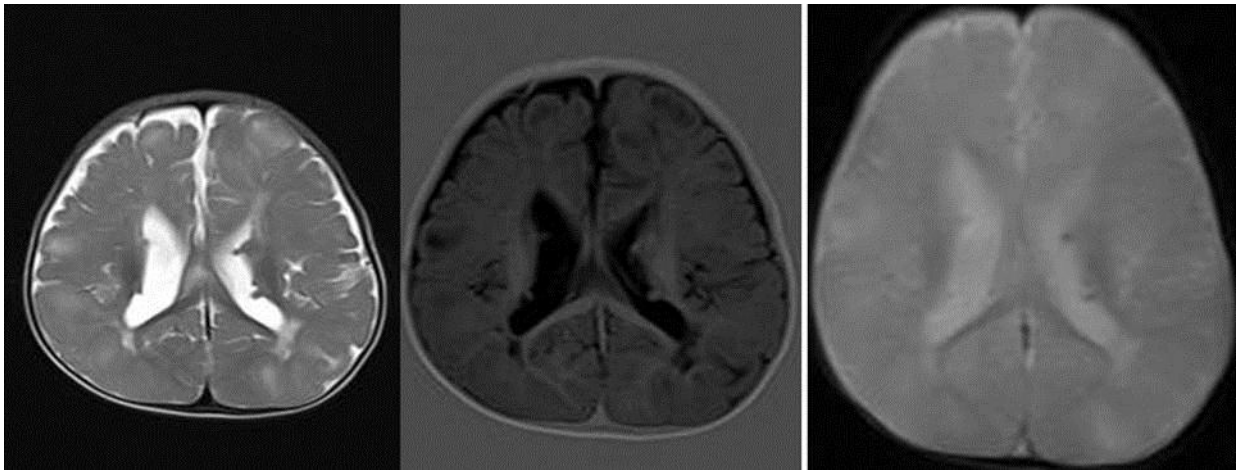

Figure 5: MRI Brain (plain axial) – T2WI (A), T1WI (B) and GRE (C)

Few subependymal nodules were noted along the bilateral subependymal region, including the left caudo thalamic groove. These nodules correspond to focal areas of blooming on GRE suggestive of calcification.

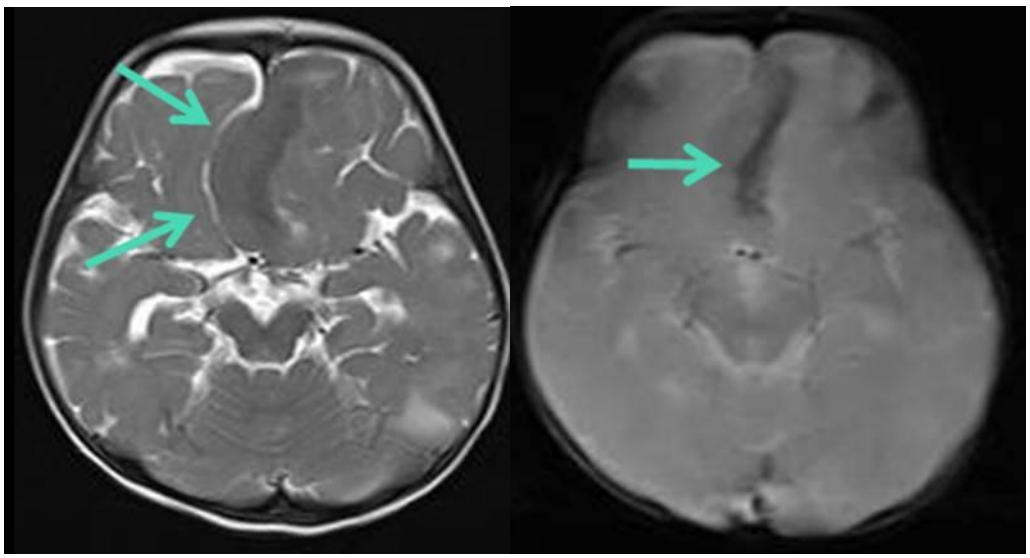

Figure 6: MRI Brain axial A (T2WI) and B (GRE)

Associated focal indentation of the inter hemispheric fissure was also seen – suggestive of cortical dysplasia. Ill defined subcortical areas of blooming on GRE suggestive of calcification were also seen beneath the dysplastic cortex.

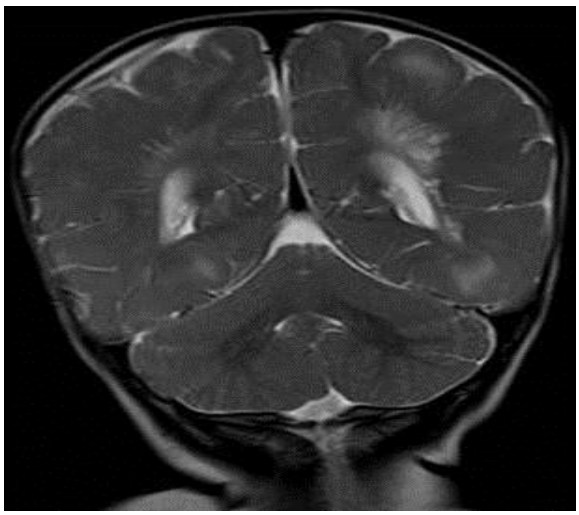

Figure 7: MRI Brain coronal (plain T2WI)

Radial migration lines were seen radiating from the body of the lateral ventricles bilaterally (left >right) appearing hyperintense on T2W images.

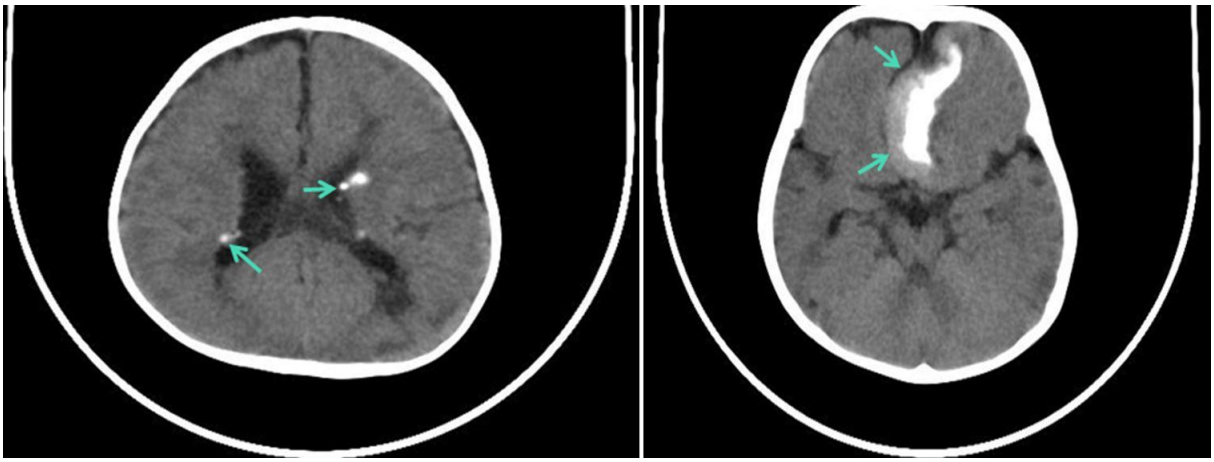

Figure 8: CT Brain (plain axial) – A and B

A large area of subcortical calcification was seen in the left frontal subcortical region measuring approximately 4 (antero-posterior) x 1 (transverse) x 3.5 (cranio-caudal) cm in the left parasagittal region extending to involve the straight gyrus.

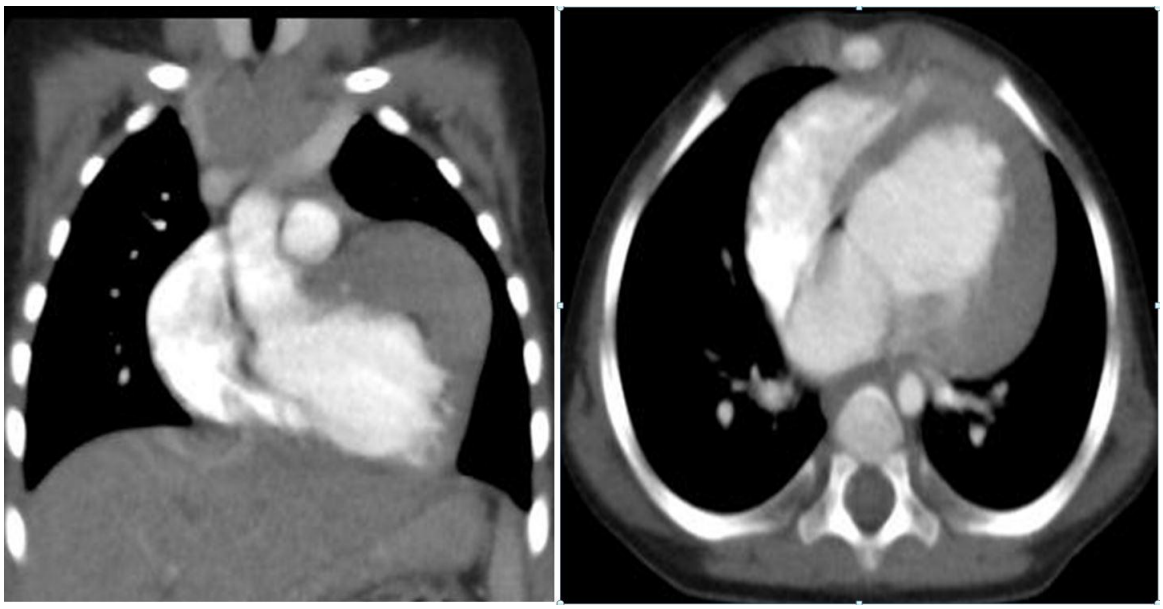

Figure 9: CT thorax – Coronal (A) and Axial (B)

CT thorax showing diffuse wall thickening of the left lateral ventricle along its superolateral wall measuring approximately 2 cm in maximum thickness. This lesion is likely to represent a rhabdomyoma.

INTRODUCTION

Tuberous sclerosis is a neuro-cutaneous disorder with prevalence / clinical penetrance estimated to be from one in 6000 to one in 12,000 live births.[1] We present a case of Tuberous Sclerosis complex diagnosed by MRI and CT. Neuroimaging advances have improved the diagnosis of tuberous sclerosis complex and treatment of patient with this condition. And the purpose of this case is to understand neuroimaging findings in patient with or without symptoms. Although recent advances in treatment have improved morbidity, the prognosis is still quite poor and nearly 40% of patients die by the age of 35 years. The cause of death varies depending on the patient's age.[2]

DISCUSSION

Tuberous sclerosis is an autosomal-dominant disorder. Approximately 50% of cases are inherited and 50% are sporadic, although the incidence of sporadic cases may be as high as 80%. The disease is caused by mutations in either the TSC1 gene, on chromosome 9q34, or the TSC2 gene, on chromosome 16p13.3.[3] Each gene consists of two copies and their mutation prevents the cell from making functional hamartin or tuberin from the

altered copy of the gene. However, enough protein is usually produced from the other normal copy of the gene to regulate cell growth effectively. But in people with tuberous sclerosis complex, a second TSC1 or TSC2 mutation typically occurs in multiple cells over an affected person's lifetime. Hamartin and tuberin form a tumor suppressor heterodimer, that inhibits the mammalian target of rapamycin (mTOR kinase) nutrient signaling input, but how this occur is unclear and their loss in different types of cells leads to the growth of tumors in many different organs and tissues.[4]

The diagnosis of TSC is based upon clinical criteria. In many cases the first clue to recognizing TSC is the presence of seizures or delayed development. In other cases, the first sign may be white patches on the skin (hypomelanotic macules) or the identification of cardiac tumor - rhabdomyoma. Diagnosis of the disorder is based on a careful clinical exam in combination with computed tomography (CT) or magnetic resonance imaging (MRI) of the brain, which may show tubers in the brain, and an ultrasound of the heart, liver, and kidneys, which may show tumors in those organs.[5]

MRI and CT are primary and the most accurate investigative tool to diagnose tuberous sclerosis.

On MRI, brain shows cortical / sub-cortical tubers: 50% are in the frontal lobe, high T2 and low T1 with only 10% of tubers showing enhancement. In subependymal hamartomas, 88% are associated with calcification, although calcification is absent in early childhood that is visible within the first 6 months of age with variable signal, frequently high T1 and iso to high T2 and enhancement is variable. Subependymal giant cell astrocytomas (SGCA) whch are also a feature of tuberous sclerosis, have a peak occurance between 8-18 years and tend to be large and demonstrate growth with intense enhancement.[5]

In white matter abnormalities, MRI shows variable appearance with nodular, ill-defined, cystic and band (radial) like lesions that are thought to be relatively specific for tuberous sclerosis.[6]

Cerebellar atrophy, infarcts (due to vascular occlusive disorders), cerebral aneurysms, dysgenesis of the corpus callosum, Chiari malformations, microcephaly, arachnoid cysts are noted.

Cardiac findings are usually maximal at birth or early in life, may be the presenting sign of tuberous sclerosis complex, particularly in early infancy. 50-60% of individuals with tuberous sclerosis complex have evidence of cardiac disease, mostly rhabdomyomas.

Other findings include Ophthalmic findings in which lesions are actually retinal astrocytomas that tend to become calcified over time. In renal manifestations of tuberous sclerosis complex are the second most common clinical features and includes four types of lesions: autosomal dominant polycystic kidney disease lesions, isolated renal cysts, angiomyolipomas (AMLs) and renal cell carcinomas.[6]

Skin manifestations includes molluscum fibrosum or skin tags. Adenoma sebaticum appearing as red papules on face. They are actually angiofibromas. Patient thinks of it as acne which does not respond to treatment. It usually appears in age < 10 years and grows till puberty.[6]

Lung lesions include lymphangioleiomyomatosis (LAM) and multinodular multifocal pneumocyte hyperplasia (MMPH). LAM is a tumor-like disorder in which cells proliferate in the lungs, and there is lung destruction with cyst formation. There is a range of symptoms with LAM, with many tuberous sclerosis complex individuals having no symptoms, while others suffer with breathlessness, which can progress and be severe.[6]

All individuals with tuberous sclerosis complex are at risk for life-threatening conditions related to the brain tumors, kidney lesions, or lymphangioleiomyomatosis. Continued monitoring by an experienced physician is important. With appropriate medical care, most individuals with the disorder can look forward to normal life expectancy. Recognition of radiologic features of various organ manifestations is essential for making the correct diagnosis and is helpful for detecting additional abnormalities.

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