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Tuberous Sclerosis with Multisystem Manifestations A Case Series

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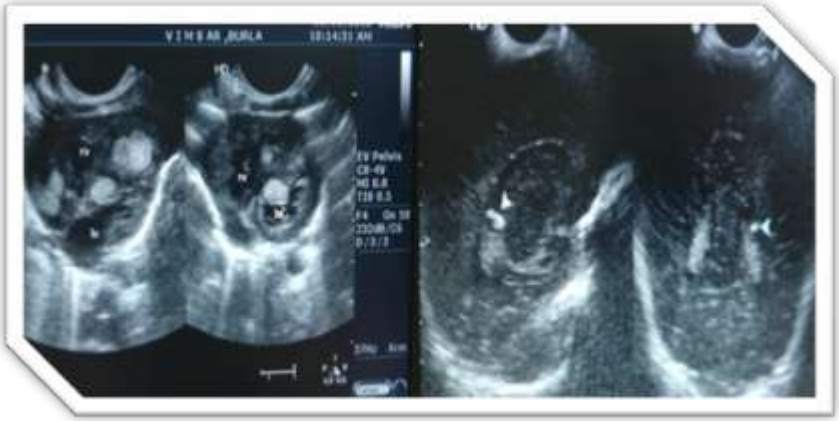
Abstract : *Tuberous Sclerosis is a hereditary neuro-cutaneous disorder with multi-system manifestations, esp. the brain, heart and kidneys. Here a case series of tuberous sclerosis with multi-system involvement is reported.*
In Case 1: *A neonate on 10th day of postnatal life was referred to our department for screening of the abdomen due to complaints of intermittent vomiting. Incidentally multiple echogenic mass lesions were found related to her myocardium. Ultrasound and Computerised tomography of her brain were also performed and revealed positive findings.*
In Case 2: *A 15 year old male with red papules over his cheeks and a two year history of recurrent seizures underwent MRI of the brain and subcortical tubers were observed.*
In Case 3: *Computerised Tomography of the Brain, Thorax and the Abdomen were carried out in a 30 year old female presenting with seizure. Characteristic findings were observed. To conclude it is suggested that a thorough multimodal screening of brain, chest and abdomen is necessary for accurately diagnosing this rare condition.*
Keywords: *angiomyolipoma, subependymal nodules, rhabdomyoma*

I. Introduction

Tuberous sclerosis is an autosomal dominant hereditary neurocutaneous disorder of abnormal histogenesis **w**ith documented neurological, cardiac, renal, pulmonary and cutaneous manifestations [1,2]. Classic clinical triad of epilepsy, mental retardation and skin manifestations such as adenoma sebaceum are seen in only half of the patients [2,3]. We report a case series that illustrates the spectrum of radiological features of tuberous sclerosis in patients of diverse age groups - a neonate, an adolescent and an adult.

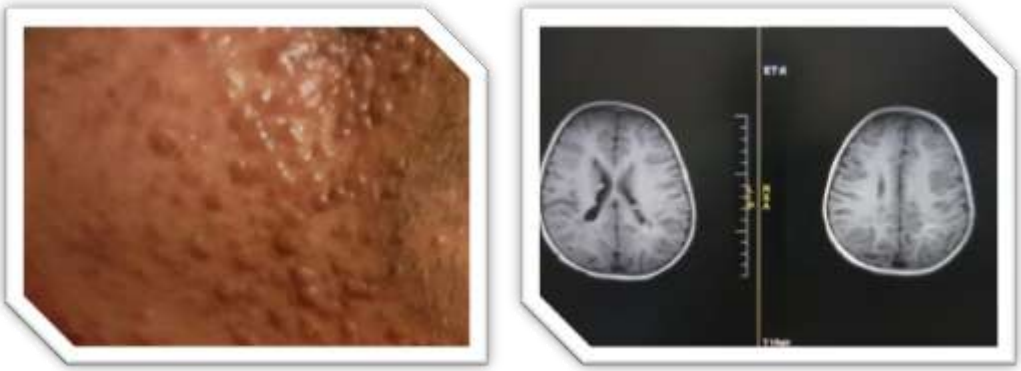
II. Case Series

Case 1: A female neonate, born out of a non-consanguineous marriage to a 26 year old primigravida, was referred to our department, on 10th day of postnatal life, for screening of the abdomen due to episodes of vomiting. No abnormality was detected in the abdomen by ultrasound. Out of curiosity, the chest was scanned and incidentally parasternal long as well as short axis views of the heart revealed multiple hyperechoic mass lesions (rhabdomyomata) related to mid and apical interventricular and interatrial septa (Fig 1a). Imaging of the brain was performed to rule out any abnormalities. Transcranial ultrasonography revealed bilateral subependymal echogenic foci with calcification in one of them (Fig 1b). Computerised tomography (CT) of the brain revealed densities corresponding to the lesions seen on ultrasound consistent with subependymal nodules. Her parents were counselled and advised for regular follow-up.



(Fig 1a). USG Parasternal Views - rhabdomyomas **(Fig 1b).** Transcranial USG - subependymal nodules

Case 2: A 15 year old male presented with a two year history of recurrent seizures. Physical examination revealed numerous red papules over his cheeks (adenoma sebaceum) which had previously been mistaken for acne (Fig 2a). Magnetic resonance imaging of the brain revealed multiple nodular subependymal lesions which were hyperintense to gray matter on T1 weighted images and hypointense on T2 weighted image. Multiple small subcortical areas of altered signal intensity were identified in bilateral fronto-temporo-parietal lobes suggestive of subcortical tubers (Fig 2b). CT scan of the abdomen ruled out abnormality of any organ.



(Fig 2a) Adenoma sebaceum **(Fig 2b)** MRI brain (T1W) - hyperintense subependymal nodules and subcortical hamartomas

Case 3: Computerised tomography of the brain was carried out in a 30 year old female presenting with repeated attacks of seizure. Dense calcification was seen in subependymal, subcortical and cortical locations (Fig 3a). Further CT scan of the Abdomen revealed heterogenous mass lesions with well defined fat density over bilateral kidneys suggestive of angiomyolipomas (Fig 3b). Computerised tomography of the thorax depicted multiple small rounded cystic lesions throughout bilateral lung parenchyma without sparing of costophrenic angles suggestive of lymphangioleiomyomatosis (LAM) (Fig 3c). She also had reddish papules suggestive of adenoma sebaceum over her face.

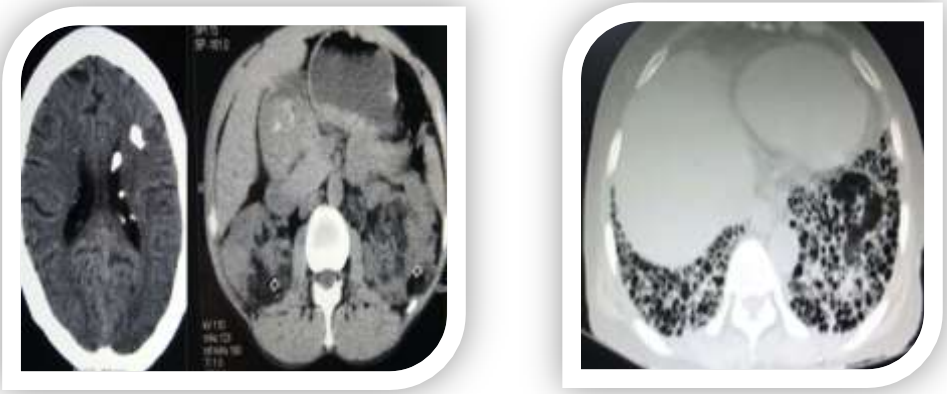


Fig 3(a,b) CT Scan Brain showing calcified subependymal nodules and subcortical tubers, CT Scan Abdomen showing bilateral renal angiomyolipoma **(Fig 3c)** CT Thorax Lung Window showing LAM

III. Discussion

The multisystem manifestations of tuberous sclerosis may involve the brain, heart, kidneys, lungs, and the skin. Central Nervous System involvement is evidenced by presence of cortical or subcortical tubers which develop before birth and progressively increase in size and shape. These lesions often undergo calcification, in 50% of patients by the age of ten [4]. Subependymal nodules and giant cell astrocytomas are also characteristic.

Cardiac rhabdomyomas, benign tumors of striated muscle origin, are seen in 30% of cases with tuberous sclerosis. Reports suggest that cardiac rhabdomyomas may be the earliest finding of this condition in utero and may precede the detection of the brain or kidney lesions They are usually diagnosed before 1 year of age and undergo spontaneous regression by early childhood [5].

An identifiable renal lesion, most commonly angiomyolipomas (hamartomatous masses composed of blood vessels, smooth muscle and adipose tissue) followed by renal cysts, with progressive increase in size, are found in 55% of children with tuberous sclerosis. In adolescents and adults they are prevalent in around 80 % of

cases [6]. Tuberous sclerosis accounts for 20 % of all angiomyolipomas which tend to be multiple and bilateral [7]. An association with renal cell carcinomas and oncocytomas has been reported [5].

Pulmonary involvement is rare (<5% of women with TSC) and is seen in the form of multiple lung cysts with normal intervening parenchyma- a condition almost exclusively seen in females called Lymphangioleiomyomatosis(LAM) [8]. Involvement of the costophrenic angles is typical and in sharp contrast to langerhans cell histiocytosis. Retinal hamartomas and vascular lesions such as aneuryms of the aorta have been reported rarely.

The diagnosis of tuberous sclerosis is definite on fulfillment of either 2 major or 1 major with 1 minor criteria (mentioned below) according to 2012 International Tuberous Sclerosis Complex Consensus Conference.

Major Criteria includes: hypomelanotic macules (≥3, at least 5-mm diameter), angiofibromas (≥3), ungual fibromas (≥2), shagreen patch, multiple retinal hamartomas, cortical dysplasia (including tubers), subependymal nodules, subependymal giant cell astrocytoma, cardiac rhabdomyoma, lymphangioleiomyomatosis (LAM), angiomyolipomas(AML). Exclusive coexistence of LAM and AML isn't sufficient for the diagnosis.

Minor Criteria includes: confetti skin lesions, dental pits, intraoral fibromas, retinal achromic patch, multiple renal cysts, nonrenal hamartomas. [9].

Our case series demonstrates various neurological (tubers, nodules), renal (angiomyolipomas), cardiac (rhabdomyoma) and pulmonary (LAM) presentations of tuberous sclerosis. Each of our cases remarkably manifested two major criteria for certain diagnosis of this condition.

IV. Conclusion

Knowledge of various systemic manifestations of tuberous sclerosis can contribute to diagnosing this condition with a high degree of accuracy. The importance of antenatal and neonatal ultrasound examination of the heart is of great significance and periodic surveillance is essential.

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