Original Research Article

Nutshell of imaging spectrum in tuberous sclerosis complex

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Abstract

Background: Tuberous sclerosis is a multisystem autosomal dominant neurocutaneous syndrome. The classical triad (Vogt's triad) of tuberous sclerosis which include epilepsy, mental retardation, and adenoma sebaceum occurs seldomly together in a patient which makes the diagnosis difficult. Here radiologic examinations play an important role in the proper diagnosis and treatment of tuberous sclerosis. The major imaging manifestations include cortical dysplasias (include tubers and cerebral white matter migration lines), subependymal nodule**,** subependymal giant cell astrocytoma, cardiac rhabdomyoma, lymphangioleiomyomatosis (LAM), angiomyolipoma. The purpose of this study is to demonstrate the imaging spectrum of tuberous sclerosis complex using multiple imaging modalities for better diagnosis and treatment. **Materials and methods:** A retrospective study was done at a tertiary care hospital between june 2018 to may 2019 in which 15 patients, both males and females in equal proportion (1 day – 50 years). Ultrasound examinations on Hitachi Aloka Arietta 70 USG machine, MDCT examination was done using SOMATOM definition AS- CT Scanner. MRI of brain and abdomen done using 3T Siemens Verio MRI scanner using head coil and body coil. **Results:** A retrospective study was done at a tertiary care hospital in which 15 patients within the age group of 1 day to 50 years. The presenting complaints included seizure, headache, abdominal lump, breathlessness and cough. Male: Female ratio was 1:1. Combination of multisystem evaluation is needed for the diagnosis of the complete disease spectrum and treatment. So the availability of multimodality imaging techniques makes the process of diagnosis faster and easier. **Conclusion:** By the optimum use of various imaging modalities like ultrasound, MDCT and MRI, early diagnosis and management of Tuberous sclerosis disease spectrum can be made.

Keywords: MDCT, MRI, Tuberous sclerosis.

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INTRODUCTION

Tuberous sclerosis complex is a multisystem autosomal dominant neurocutaneous syndrome with high phenotypic variability. It is characterised by wide spread hamartomas and benign neoplasms distributed in several organs throughout the body especially in the brain, retina, skin, lungs, heart and kidney. Since we could rarely identify the classical triad (Vogt's triad) of epilepsy, mental retardation, and adenoma sebaceum, radiologic examinations can play an important role in the diagnosis of tuberous sclerosis and in its treatment. Cardiac rhabdomyoma, renal angiomyolipoma, and neurologic involvement encompassing cortical or subependymal tubers and white matter abnormalities are the common radiologic findings. Detection of these entities can be strong evidence for suspecting tuberous sclerosis. The presence of pulmonary lymphangioleiomyomatosis, multifocal micronodular pneumocyte hyperplasia, or multiple renal cysts also raises suspicion of tuberous sclerosis. Thus radiological imaging becomes an important aid in presumptive diagnosis and defining of extend of involvement as well as treatment planning.

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AIMS AND OBJECTIVE

The purpose of this article is to demonstrate the imaging spectrum of tuberous sclerosis complex using multiple imaging modalities. The combination of both clinical and imaging features will direct to prompt diagnosis and treatment planning.

MATERIALS AND METHODS

A retrospective study was done at a tertiary care hospital between june 2018 to may 2019 in which 15 patients, both males and females in equal proportion (1 day – 50 years) who presented with multiple CNS (central nervous system) symptoms like seizure, headache, respiratory symptoms and abdominal lump. Ultrasound examinations on Hitachi Aloka Arietta 70 USG machine,MDCT examination was done using SOMATOM definition AS- CT Scanner. MRI of brain and abdomen done using 3T Siemens Verio MRI scanner using head coil and body coil.

RESULTS

A retrospective study was done at a tertiary care hospital which 15 patients within the age group of 1 day to 50 years . The presenting complaints included seizure, headache, abdominal lump, breathlessness and cough. Male: Female ratio was 1:1.

Out of 15 cases studied 10 cases (66.6 %) showed cortical tubers. 95 % of tubers were multiple and 90 % showed frontal lobe involvement. 11 cases (73.3 %) showed subependymal nodules, 3 cases (20 %) showed subependymal giant cell astrocytoma with obstructive hydrocephalous, 8 cases (53%) showed radiation migration lines, 1 case (6.6 %) showed cardiac rhabdomyoma, 4 cases (26.6 %) showed lymphangioleiomyomatosis (LAM) and multifocal micronodular pneumocyte hyperplasia, 3 cases (20 %) showed renal angiomyolipoma and 1 case (6.6%) of hepatic angiomyolipoma.

DISCUSSION

With the evolution of multimodality imaging technologies, it is important to identify the radiologic features of multiorgan involvement in patients with Tuberous sclerosis complex. The extent and severity of involvement is important in planning of the treatment.

Central nervous system involvement (CNS)

A variety of CNS involvement are seen in patients with Tuberous sclerosis. It includes cortical and subcortical nodules, subependymal nodules, subependymal giant cell astrocytoma, radial migration lines.

Cortical tubers, Subependymal nodules and radial migration lines

Cortical tubers are benign hamartomas most commonly found in frontal cortex of cerebrum. Magnetic resonance (MR) imaging, cortical tubers have increased signal intensity on T2-weighted images and decreased signal intensity on T1-weighted images. After administration of contrast material, only 10% of cortical tubers show enhancement [1]. Calcification and central cystic degeneration can sometimes occur. Subependymal nodules are discrete or confluent areas of hypertrophic tissue seen along the ventricular surface. Radial migration lines represent heterotopic glia and neurons along the expected path of cortical migration [3].

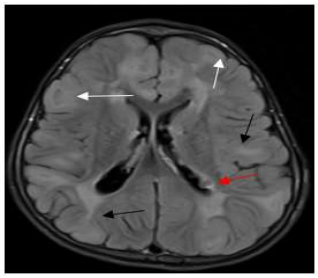

Figure 1: Axial fluid attenuation inversion recovery image demonstrating cortical tubers (white arrow) and subcortical and deep white matter showing circumscribed areas of hyperintense areas. Subependymal nodules along the ventricular surface (red arrow) and radial migration line (black arrow)

Subependymal giant cell astrocytoma (SGCAs)

They are the abnormal proliferation of astrocytes and giant cells. The typical location of SGCAs is in the foramen of Monro, leading to obstructive hydrocephalus. Typically, the initial symptom of SGCAs is increased intracranial pressure [2]. SGCAs can evolve in utero which suggests the importance of antinatal fetal skull ultrasonography. Serial CT scans have shown subependymal nodules growing into SEGAs 9. MR spectroscopy may be helpful in differentiating SEGAs from subependymal nodules, given that SEGAs have been found to have a high choline-to-creatine ratio.

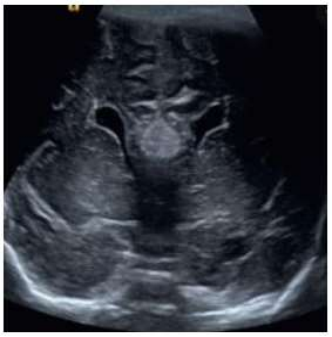

Figure 1: Well circumscribed echogenic mass at the level of foramen of Monro with enlargement of frontal horns of bilateral lateral ventricles

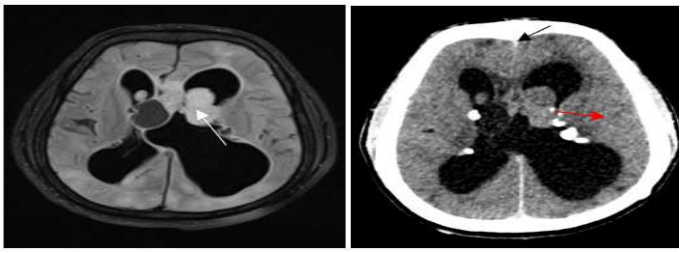

Figure 2: MRI brain FLAIR image and Non contrast CT brain showing well-circumscribed solid cystic mass at the foramen of Monro with extension into bilateral lateral ventricles (white arrow). Calcified subependymal nodule(red arrow)

Pulmonary manifestation

Lymphangioleiomyomatosis and micronodular pneumocyte hyperplasia

It is the diffuse interstitial proliferation of smooth muscle cell bundles and cystic changes in the pulmonary parenchyma. The hallmark feature of LAM is the presence of diffuse, well-circumscribed, thin-walled lung cysts distributed uniformly throughout the lungs. Recurrent pneumothorax can be seen in patients with LAM [6].

Micronodular pneumocyte hyperplasia are multiple noncalcified pulmonary nodules scattered randomly throughout the lung.

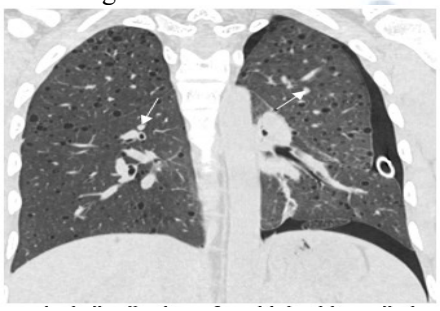

Figure 3: Bilateral symmetrical distribution of multiple thin walled cysts of variable size, scattered in normal lung parenchyma. Randomly distributed multiple lung nodules (white arrow) suggestive of Micronodular pneumocyte hyperplasia

Cardiac manifestations

Rhabdomyomas are benign striated muscle tumour. It is the most common benign primary cardiac tumour in infants and children [5]. The incidence of tuberous sclerosis in patients with cardiac rhabdomyomas is 60-80%. Multiplicity of cardiac rhabdomyomas has stronger association with tuberous sclerosis. In ultrasonography they appear as well-defined echogenic foci in myometrium.

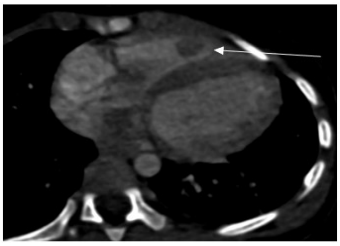

Figure 4: Well circumscribed hypodense mass at the free ventricular wall of right ventricle projecting towards the septum

Renal manifestations

Angiomyolipomas (AML) are characterized by immature smooth muscles, fat and abnormal vessels [7]. Aneurysms may develop from the abnormal vessels raising the risk of haemorrhage. AML can be diagnosed with the variable amount of fat within the tumour. AML can be fat rich or minimal fat. AML due to tuberous sclerosis typically occur in younger patients and are frequently multiple and bilateral 8. Conversely, approximately 20% of patients with angiomyolipomas have tuberous sclerosis. The remainder of angiomyolipomas are sporadic (nontuberous sclerosis). Sporadic angiomyolipomas are usually unilateral and solitary and occur in middle-aged women [10]. Other renal manifestations include bilateral multiple simple cysts and rarely renal cell carcinoma.

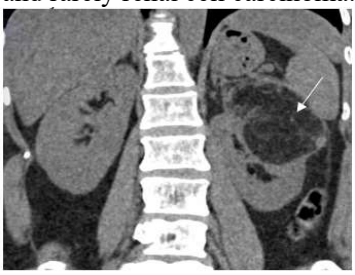

Figure 5: Coronal unenhanced CT image shows large solid left renal mass (white arrow) measuring -67HU owing to fat tissue, strongly suggestive of fat rich angiomyolipoma (AML)

Hepatic manifestations

Hepatic manifestations include hepatomegaly, mesenchymal tumours like angiomyolipoma[4], lipoma and fibromas.

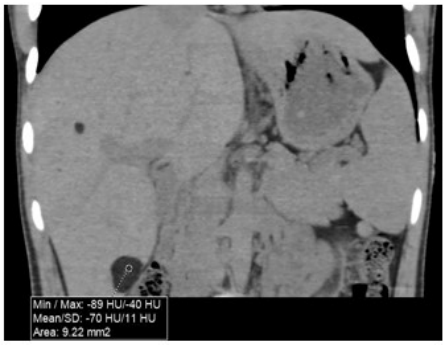

Figure 6: Coronal non enhanced CT showing few well defied variable sized hypodense lesions in the liver HU-89, suggestive of lipomas

CONCLUSION

Tuberous sclerosis is a multisystem complex disorder, with a number of clinical and imaging features. Clinical examination findings and radiological imaging findings acts complementary in diagnosis of tuberous sclerosis. In the cases of any clinical suspicion of tuberous sclerosis, complete radiological investigation should be performed for the prompt diagnosis and treatment planning.

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