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TUBEROUS SCLEROSIS - MYRAID PRESENTATIONS

Radiodiagnosis

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KEYWORDS

INTRODUCTION

Tuberous sclerosis is a multisystem hamartomatous neurocutaneous disease with one-third of the cases showing familial pattern of inheritance due to mutations in two tumor suppressor genes, TSC1 and TSC2. It is also known as tuberous sclerosis complex or Bournville disease and is characterized by development of multiple tumors of embryonic ectoderm which are mostly benign in nature or rarely malignant and involving several organs throughout the body, especially in the brain, skin, retina, kidney, heart and lung.Accounting for a prevalence of about 1 in 6000 with approximately 1.5 million people affected worldwide.The classical triad of manifestation ,which is also known as the Vogt's triad consist of facial adenoma sebaceum, epilepsy, and mental retardation.However, this triad is found in less than half of the patients.TSC1 has a locus on chromosome 9q34 and encodes the protein hamartin whereas TSC2 is found on chromosome 16q13 and encodes for the protein tuberin.These two proteins react to form a heterodiamer , which inhibits the mammalian target of rapamycin(mTOR)-signaling cascade, which is responsible for regulating cell growth and differentiation.[1] The absence of either hamartin and tuberin leads to loss of inhibition which results in hamartomatous tumors of tuberous sclerosis.

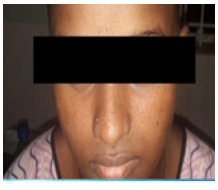
We report 4 cases from our department who had presented with various clinical manifestations of this disease spectrum.

Case report

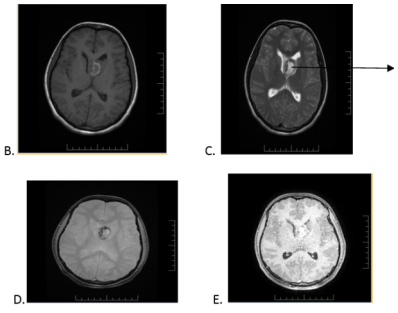
Case 1

A 25 year old female patient had presented complaint of seizures. Clinical examination had showed adenoma sebaceum on face.

MRI brain of the patient demonstrated a relatively well defined lesion at the level of foramen of monroe which is T1 and T2 heterogenously hyperintense and showed foci of blooming on On GRE images within the lesion suggestive of haemorrage.On contrast administration the lesion showed heterogenous moderate enhancement. These findings were consistent with subependymal giant cell astrocytoma with haemorrhage within it.



A. Clinical photograph demonstrates adenoma sebaceum.

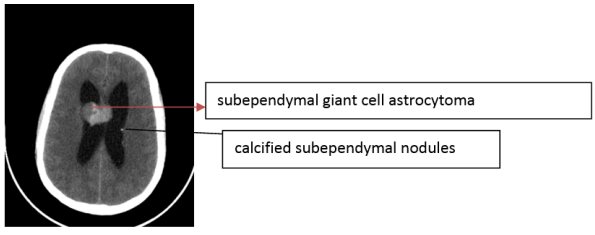


MRI BRAIN axial section demonstrates a relatively well defined lesion(→) at the level of foramen of monroe which is T1 (B.) and T2 (C.) heterogenously hyperintense . On GRE images (D.) blooming is noted in the lesion suggestive of haemorrage.On contrast administration (E.)the lesion is showing heterogenous moderate enhancement.

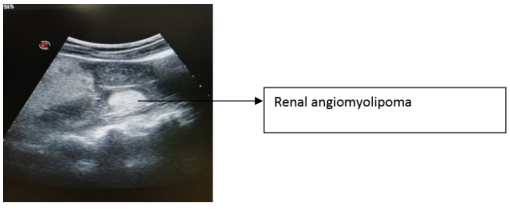
CASE 2:

A 25 year old female patient came with complaint of seizures but mental retardation was not present.Clinical examination shows adenoma sebaceum on face.Noncontrast CT brain showed a relatively well defined hyperdense lesion at foramen of monroe on right side consistent with subependymal giant cell astrocytoma and few small foci with dense calcification along the left lateral ventricle suggestive of subependymal nodule.

Ultrasound abdomen showed multiple well defined hyperechoic lesions in bilateral kidneys with posterior acoustic enhancement. Pathologically they were confirmed to be angiomyolipoma



A. NCCT BRAIN axial section demonstrates a relatively well defined hyperdense lesion at foramen of monroe on right side suggestive of subependymal giant cell astrocytoma.Few small foci with dense calcification is noted along the left lateral ventricle suggestive of subependymal nodule.



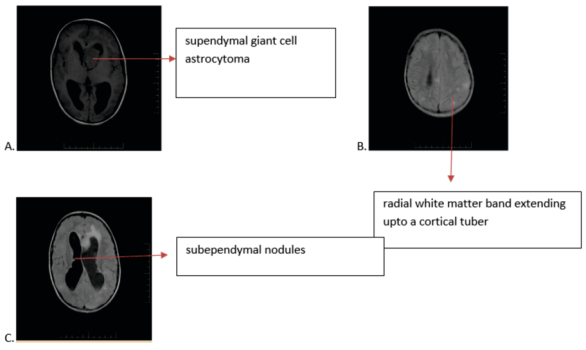
B. Ultrasound abdomen shows a well defined hyperechoic lesion noted in the lower pole of right kidney

CASE 3:

A 25 year old male patient presented with seizures and mental retardation. MRI brain of the patient showed a relatively well defined lesion at foramen of monroe which was isointense with grey matter on all the sequences , this was suggestive of subependymal giant cell astrocytoma.

Thin straight bands of hyperintensity was seen on T2 FLAIR images which extended from ventricular surface to deep surface of cortical tubers or normal appearing cortex.

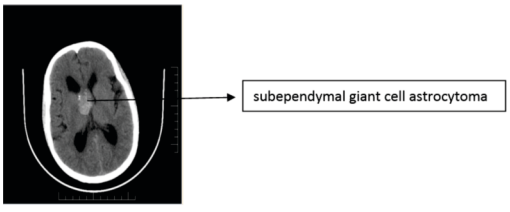
Subependymal nodules were noted which were hyperintense on T1 weighted images and iso to hyperintense on T2 weighted images.



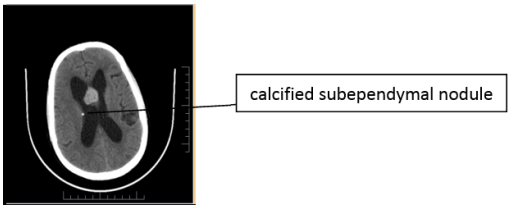
MRI brain A. T1WI axial section demonstrates a relatively well defined lesion at foramen of monroe which was isointense with grey matter, this was suggestive of subependymal giant cell astrocytoma.B.T2 FLAIR demonstrates linear radial white matter bands extending from the lateral ventricles to cortex , cortical tubers are also appreciated.C. T2 FLAIR axial section shows supendymal nodules which are isointense to grey matter.Hydrocephalus is also appreciated

CASE 4:

A 15 year old patient came with complaint of headache with no complaint of seizures or mental retardation.Noncontrast CT brain showed a relatively well defined hyperdense lesion at foramen of monroe few calcific foci within consistent with subependymal giant cell astrocytoma and few small foci with dense calcification along the left lateral ventricle suggestive of subependymal nodule.



NCCT BRAIN axial section demonstrates a relatively well defined hyperdense lesion at foramen of monroe few calcific foci within with suggestive of subependymal giant cell astrocytoma.



BRAIN axial section shows Few small foci with dense calcification along the right lateral ventricle suggestive of calcified subependymal nodule.

DISCUSSION:

The most frequently involved organs are brain, kidney, lungs ,cardiac, skin and skeleton.

CENTRAL NERVOUS SYSTEM:

Classical CNS involvement includes four entities 1. cortical tubers, 2. subependymal nodules, 3. subependymal giant cell astrocytoma and 4.benign white matter lesions in the form of cerebral white matter radial migration lines.

Subependymal nodules: noted in about 90% of patients and represent hamartomatous lesions lining the ventricle.These are collections of abnormal, swollen glial cells and giant cells which cannot be differentiated as normal neural tissue. They have a tendency to calcify and can progress into subependymal giant cell astrocytomas, which are histologically indistinguishable from SENs but distinguishable based on their larger size, higher growth rate, and potential for mass effect compared to the relatively static course of SENs [2]. CT is the modality of choice in patients with undiagnosed tuberous sclerosis who have subtle neurologic symptoms .On MRI ,T1WI the lesion has is hyperintense ,iso to hyperintense on T2WI and FLAIR if not calcified.

Cortical and Subcortical Tubers :These occur in 90% of patients, and are most commonly visualized in the frontal lobes. The expansive lesions of the central nervous system alone are not diagnostic of TSC and may be seen in other pathological conditions [3].They typically have a triangular configuration with the apex pointing toward the ventricle. These tubers are hypointense on T1-weighted images and hyperintense on T2- weighted and FLAIR images .On contrast administration only about 10% of tubers exhibit enhancement Cerebral White Matter Radial Migration Lines : Incidence is 80% of patients with tuberous sclerosis. They most commonly occur in the cerebral white matter of the frontal lobes bilaterally and are characterized as thin straight lines of T2 hyperintensity and T1 isointensity to hypointensity coursing from the periventricular white matter to the cerebral cortex. These lesions rarely enhance.

Subependymal Giant Cell Astrocytomas:occurs in 10-15 % of patients with tuberous sclerosis and are thought to arise from subependymal nodules.They are slow-growing, enhancing lesions usually located at the foramen of Monro .MR spectroscopy helps to differentiate SEGAs from subependymal nodules, as SEGAs shows high choline-to-creatine ratio and low N-acetylaspartate–to–creatine ratio . SEGAs cause obstructive hydrocephalus because of their size and location .

Renal involvement:

The renal manifestation of tuberous sclerosis consist of simple renal cyst, angiomyolipomas. and renal cell carcinoma. Angiomyolipomas account for 80% of cases of tuberous sclerosis and are benign in nature. They may arise in renal cortex or medulla. Compared with sporadic lesions, AMLs seen in patients with TS tend to manifest at a younger age and to be multiple, larger, and bilateral .[4]

Small angiomyolipomas are usually homogeneously hyperechoic at ultrasound, but larger angiomyolipomas may have a more heterogeneous appearance with posterior acoustic shadowing. At CT, most angiomyolipomas consist of macroscopic fat and measure less than –20 HU, which is pathognomonic of an angiomyolipoma . MRI especially T1-weighted fat-suppressed MR images is useful for the detection of the microscopic fat present in these lesions.

Renal Cysts : are usually multiple in tuberous sclerosis tend to occur in younger children in this condition. On ultrasound imaging they are multiple and anechoic. These cysts may increase in size and number over years ,making the kidneys look enlarged and boss elated..

Renal Cell Carcinoma: Tuberous sclerosis–associated RCCs are predominantly clear cell RCCs , but papillary and chromophobe may also be seen .Clear cell carcinomas are hypervascular and typically show heterogeneous early enhancement with early washout. Papillary carcinomas tend to be hypovascular, enhancing in a gradual manner. The papillary subtype also tends to contain calcifications. The chromophobe subtype frequently exhibits early weak enhancement and early washout Pulmonary Manifestations is lymphangio leio myomatosis which is observed as multiple thin-walled cysts scattered diffusely and surrounded by normal lung parenchyma. Women with tuberous sclerosis are more severely affected by the disease than are men.

Cardiac Manifestations are Rhabdomyomas which can be solitary or multiple and occurs as a well-defined mass or masses on the ventricular septum which are hyperechoic at sonographic evaluation .They are isointense to myocardium on T1-weighted images and hyperintense on T2-weighted images . Childhood tumor regression is the rule, with most of them regressing before birth and 70% by the age of 4 year.[5]

Hepatic Manifestations consist of Angiomyolipomas which are echogenic on ultrasound images, have fat attenuation on CT images, and demonstrate decreased signal intensity on T1-weighted fat suppressed MR images .

Splenic Manifestations consist of hamartomas rarely . These are hypervascular, homogeneously hyperechoic masses at ultrasound examination. At CT, splenic hamartomas have same or lower attenuation in relation to the spleen. At MRI, splenic hamartomas are hypointense on T1-weighted and hyperintense on T2-weighted images.

Osseous involvement: are variable and consist of focal sclerotic lesions found in vertebrae and ribs, bone cysts mostly found in

phalanges of hands and feet , and periosteal new bone formation.

CONCLUSION:

The knowledge of various manifestations of tuberous sclerosis helps in establishing the diagnosis retrogradely , which is highlighted in this case report.

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