

Tuberous Sclerosis - A Rare Case Presentation

Dr. Ginni Kalra¹, Dr. Akilesh Patel², Dr. Himanshu Sonim³

¹3rd Year Resident, Department of Radio- Diagnosis, Gujarat Cancer and Research Institute Asarwa, Ahmedabad – 380016, Gujarat, India
Email id: [ginnikalra2018\[at\]gmail.com](mailto:ginnikalra2018[at]gmail.com)

²3rd Year Resident, Department of Radio- Diagnosis, Gujarat Cancer and Research Institute, Asarwa, Ahmedabad – 380016, Gujarat, India
Email id: [akileshkumar.patel\[at\]gcriindia.org](mailto:akileshkumar.patel[at]gcriindia.org)

³Professor, Department of Radio – diagnosis, Gujarat Cancer & Research Institute, Asarwa, Ahmedabad - 380016
Corresponding Author E - Mail address: [himanshucsoni\[at\]yahoo.com](mailto:himanshucsoni[at]yahoo.com)

Abstract: Tuberous sclerosis is a rare case presentation. A 24 year old female patient presented to our hospital with suspected renal angiomyolipoma for further evaluation and management. CECT thorax, abdomen and pelvis with renal dynamic study was done. On examination, presence cutaneous lesions on face, periungual fibroma was noted. Patient also revealed history of similar lesions in her siblings and mother. Patient was further investigated, HRCT thorax and NCCT brain, MRI brain was done to look for other characteristic lesions in lung, brain to rule out tuberous sclerosis complex. Presence of renal angiomyolipoma, multiple subcortical and subependymal calcified lesion, dermal lesions confirmed the diagnosis of tuberous sclerosis. **Introduction:** Tuberous sclerosis also known as Bourneville disease is a neurocutaneous syndrome characterised by formation of variety of hamartomatous lesions in several organs. Patient generally present with seizure, intellectual disability and adenoma sebaceum. This classical triad is uncommonly seen therefore radiological examination plays important role in diagnosis and treatment of tuberous sclerosis. **Case Report:** A 24 year old female patient presented to our hospital with suspected renal angiomyolipoma for further evaluation and management. CECT thorax, abdomen and pelvis with renal dynamic study was done. On examination, presence cutaneous lesions on face, periungual fibroma was noted. Patient also revealed history of similar lesions in her siblings and mother. Patient was further investigated, HRCT thorax and NCCT brain, MRI brain was done to look for other characteristic lesions in lung, brain to rule out tuberous sclerosis complex. Presence of renal angiomyolipoma, multiple subcortical and subependymal calcified lesion, dermal lesions confirmed the diagnosis of tuberous sclerosis. **Conclusion:** On the basis of various clinical and radiological features diagnosis is made as definite, possible and probable tuberous sclerosis.

Keywords: Tuberous sclerosis, cortical tubers, subependymal nodules, periungual fibroma

1. Introduction

Tuberous sclerosis also known as Bourneville disease is a neurocutaneous syndrome characterised by formation of variety of hamartomatous lesions in brain, heart, skin, kidney, lung and other organs. About 50% of cases are inherited and follow an autosomal dominant pattern and other half represent de novo mutations and germline mosaicism with mutations or deletions in TSC 1 (encodes for hamartin protein) and TSC 2 (encodes for tuberin protein) genes. Clinically, patient presents with seizures, intellectual disability and adenoma sebaceum (Vogt triad).

2. Radiological Findings

A. Neurological Involvement

The four major features of TS in brain:

1) Cortical Tubers

These are glioneuronal hamartomas found in over 90% of patients.

CT findings - hypodense cortical/subcortical masses within broadened and expanded gyri. Lucency decreases with age, so that tubers become isodense with cortex in older children and adults.

MRI findings - In infants - tubers appear as hyperintense cortex compared to underlying unmyelinated WM on T1W,

become moderately hypointense on T2W. Streaky" linear or wedge - shaped T2/Flair hyperintense bands may extend from the tuber all the way through the WM to the ventricular ependyma.

In older children and adults, Signal intensity changes after myelin maturation. Tubers become more isointense relative to cortex on T1WI. Tubers demonstrate mixed signal intensity on T2/FLAIR. The periphery of the expanded gyrus is isointense with cortex while the deeper component is strikingly hyperintense.

2) Subependymal Nodules

These are hamartomatous changes in subependymal tissue and occur as multiple nodules. They are located immediately beneath the ependymal lining of the lateral ventricles.

CT findings - NCCT - SEN are mostly associated with calcification. On NCCT, multiple small foci with dense calcification are noted along the b/l lateral ventricles

CECT - Typically, they do not enhance on CT scan. Enhancing or enlarging SEN on CECT (if located near foramen of monro) is suspicious of malignancy.

MRI findings - in unmyelinated brain - appear hyperintense on T1WI and hypointense on T2WI. In myelinated brain - become isointense to WM on T1WI and iso - to hyperintense on T2 - weighted images. They may show moderate to intense enhancement on T1W C+ (does not indicate malignancy)

Volume 10 Issue 11, November 2021

www.ijsr.net

Licensed Under Creative Commons Attribution CC BY

SEN are stable lesions. For nodules located near foramen of Monro, close interval follow up is required as they are likely to become malignant.

3) White Matter Lesions

They are seen in nearly all patients of TS. They represent altered development along the migratory pathways of neurons and glial cells. They occur as fine lines radiating from ependymal ventricle surface to cortical tubers.

CT findings - They are small therefore difficult to detect on CT.

MRI findings - Streaky linear or wedge - shaped lesions extend radially from the ventricles to the undersurfaces of cortical tubers.

In unmyelinated brain - they appear mildly hyperintense to WM on T1WI.

In older children and adults - they are hyperintense on T2/FLAIR sequences.

4) Subependymal Giant Cell Astrocytoma -

SEGAs are well - circumscribed solid intraventricular masses located near the foramen of Monro. They are WHO grade II tumors that can cause obstructive hydrocephalus without invading adjacent brain.

CT findings - SEGAs show mixed density on NCCT scans and demonstrate focal calcification. They show moderate enhancement on CECT. SEGAs are larger tumours (>1cm) as compared to SEN.

MR Findings - SEGAs show mixed signal intensity on both T1WI and T2WI. THEY all show moderate enhancement on T1 C+. Changes of hydrocephalus may be seen.

B. Renal Involvement

Renal involvement of TS includes renal angiomyolipoma (AML), renal cysts and renal cell carcinoma. Renal AML is one of the common manifestations.

RENAL AML - Benign tumours of kidneys, characterised by abnormal vessels, immature smooth muscles and fat cells.

Typical CT findings include - mixed density non calcified cortical tumours containing fat (CTHU = - <20), blood vessels and smooth muscles. Absence of calcification favours the diagnosis of renal AML.

C. Pulmonary Involvement

Pulmonary involvement of TS includes lymphangiomyomatosis (LAM) and multifocal micronodular pneumocyte hyperplasia (MMPH).

Pulmonary LAM - Characterised by diffuse interstitial proliferation of bundles of smooth muscle cells and cystic changes in lung parenchyma. **CT findings include** - round, thin walled round cysts showing symmetrical distribution throughout the lungs.

MMPH - Characterised by multicentric, well demarcated nodular proliferation of type II pneumocytes along alveolar

septa. **CT findings include** - multiple tiny nodules (1 - 8 mm in diameter) diffusely scattered throughout lung in random manner.

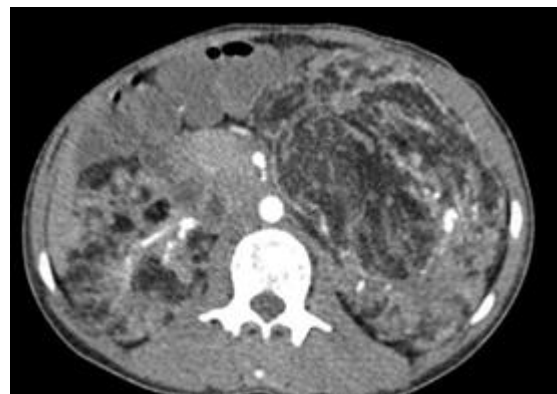
D. Dermatological Involvement & Cardiac Involvement

The most common skin lesions are hypomelanotic macules - ovoid depigmented areas with irregular margins. Other cutaneous findings include forehead plaques, shagreen patches, adenoma sebaceum, periungual fibroma. Cardiac rhabdomyoma is a benign striated muscle tumour. Seen in 50 - 65 % cases of tuberous sclerosis, commonly located on ventricular septum. Most cardiac rhabdomyoma regress before birth or by the age of 4 years.

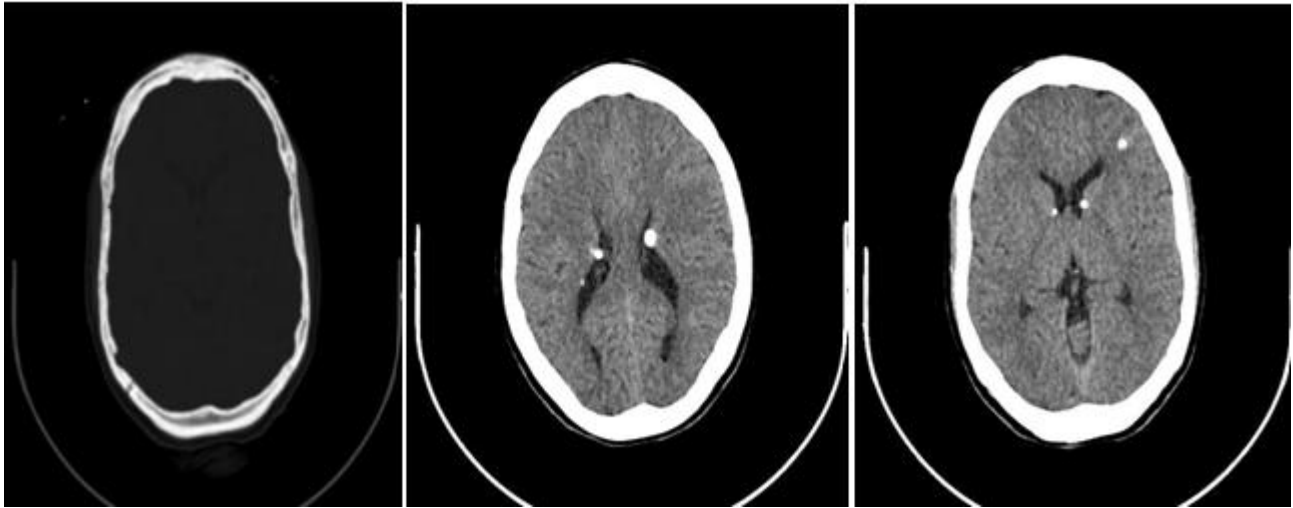
3. Case Report

A 24 year old female presented to our hospital with abdominal pain and palpable mass in left lumbar region.

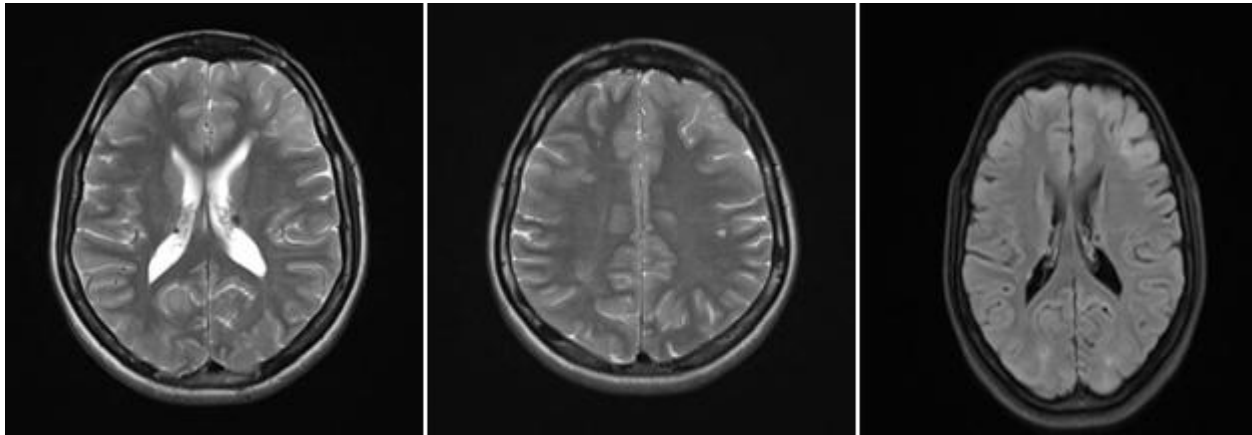
- **USG - Abdomen & pelvis** was done outside which suggested large heterogeneous mass in abdomen and retroperitoneum, both kidneys were not seen separately from the lesion. Patient was referred to our cancer hospital for further management. CECT thorax, abdomen pelvis was done with renal dynamic study.
- **On CECT Abdomen & pelvis** - Presence of mixed density lesion was noted involving left kidney. Lesion showed internal fat density areas (CTHU = - 88). Lesion showed internal enhancing soft tissue density lesions. There was no e/o internal calcification. Presence of similar characteristic lesion was noted involving right kidney. Above findings s/o P/o b/l renal angiomyolipoma.



- **Clinically** - Patient also showed cutaneous manifestations in the form of adenoma sebaceum and periungual fibromas since childhood. Patient also had family history of similar cutaneous lesions in her mother, siblings and son. The above finding raised suspicion of TS. Patient was advised NCCT brain, MRI Brain and HRCT thorax to look for other manifestations of TS and confirm the diagnosis.
- **On NCCT brain** - Presence of multiple subcortical and subependymal calcified lesions were noted in b/l frontal region and b/l lateral ventricle s/o subependymal and subcortical tubers. Presence of multiple sclerotic lesions were noted involving skull vault and base of skull. There was associated hyperostosis of inner table of skull vault noted.



- **On MRI Brain** - Few cortical tubers were noted in b/l parietal regions and few subependymal nodules were noted in b/l frontal region and b/l lateral ventricle.



- **On HRCT thorax** - presence of few tiny well - defined cystic lesions were noted in b/l lung fields. i/v/o P/o TS, P/o pulmonary lymphangiomyomatosis was suspected.



TSC: Diagnostic Clinical Features

Diagnosis

- Definite TSC - 2 major features or 1 major + 2 minor
- Probable TSC - 1 major + 1 minor feature
- Possible TSC - 1 major or ≥ 2 minor features

Major Features

- Identified clinically - ≥ 3 hypomelanotic ("ash leaf") macules, Facial angiofibromas or forehead plaque, Shagreen patch, Ungual/periungual fibroma, Multiple retinal hamartomas
- Identified on imaging - Subependymal nodules, Cortical tubers, Cardiac rhabdomyoma, Renal angiomyolipoma, Subependymal giant cell astrocytoma, Lymphangiomyomatosis

Minor Features

- Identified clinically - Gingival fibromas, Affected first - degree relative, Pitting of dental enamel, Retinal achromic patch
- Identified on imaging - WM hamartomas, radial migration lines, Hamartomatous polyps, Nonrenal hamartomas, Bone cysts, Renal cysts

4. Conclusion

The various clinical features of TSC are designated as major or minor features. Based on these features, the diagnosis is divided into definite, probable, and possible:

This 24 year old patient showed presence of following -

Major Features: Periungual fibroma, Renal AML, Cortical tubers, SEN, Pulmonary LAM.

Minor Features: Affected first - degree relative.

Hence presence of 2 major features confirms the diagnosis of Tuberous sclerosis in this case.

References

- [1] Osborns brain 2nd edition.
- [2] Radiopedia <https://radiopaedia.org/articles/tuberous-sclerosis>.
- [3] Imaging of tuberous sclerosis NCBI <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5347503/>
- [4] The American Journal of Neuroradiology <http://www.ajnr.org/content/20/5/907>
- [5] CT and MRI of the whole body - John R Haaga 6th edition