

**Tuberous Sclerosis Complex with Intracranial Aneurysm-A Case Report**Dr. P. Beno Jefferson<sup>1\*</sup>, Dr. M.S. Hemhnath MDRD<sup>2</sup><sup>1</sup>Final year resident, Department of Radiodiagnosis, Sree Balaji medical college and hospital, Chrompet, Chennai-600044 India<sup>2</sup>Assistant professor, Department of Radiodiagnosis, Sree Balaji medical college and hospital, Chrompet, Chennai-600044 India**\*Corresponding author**

Dr. P. Beno Jefferson

**Article History**

Received: 04.09.2018

Accepted: 16.09.2018

Published:30.09.2018

**DOI:**

10.36347/sjmcr.2018.v06i09.014



**Abstract:** Tuberous sclerosis complex is an autosomal dominant disorder of neuro cutaneous syndrome characterized by epilepsy, multiorgan tumours and hamartomas. It is presented with childhood clinical triad of seizures, mental retardation and skin lesions. Here we discuss a 35-year-old female who presented with complains of unable to move right upper and lower limbs, she is also a known case of seizure disorder and mental retardation since her childhood. Patient was referred for CT brain which revealed cortical tubers and subependymal nodules. MRI study of the brain was done which confirmed features suggestive of tuberous sclerosis complex with giant intra cranial aneurysm. This case further aims to substantiate the clinical and radiological features of this syndrome.

**Keywords:** Tuberous sclerosis complex, epilepsy, hamartomas, intracranial aneurysm, Magnetic resonance imaging (MRI).

**INTRODUCTION**

Tuberous sclerosis complex is a neurocutaneous syndrome formerly described by Bourneville in 1880 also known as Bourneville disease [1]. This entity is an inherited autosomal dominant disease caused by mutation or deletion of two genes: TSC1 located on chromosome 9 with encodes for the protein hamartin, and the other one TSC2 located on chromosome 16 with encodes for the protein tuber[2].

Tuberous sclerosis is a rare syndrome with an estimated incidence of 1 in 6000 to 1 in 10,000 live births. The name tuberous sclerosis is derived from the characteristic tuber like growth occurring in the brain which calcifies with age and become sclerotic[3].

**CASE REPORT**

A 35-year-old female was brought to our department with chief complaints of unable to move right upper and lower limbs for past three days. She was also a known case of mental retardation and history of seizure disorder since her childhood.

On clinical examination, there was frontal perturbation and pigmented skin lesions suggestive of café-au-lait spots were seen on the posterior chest wall was clinically diagnosed as tuberous sclerosis.

Patient was referred for computed tomography (CT) of brain, which showed multiple calcified sub

ependymal nodules of varying sizes and shapes, cortical tubers and a well-defined lesion measuring 3.5 x 2.6 cm with peripheral curvilinear calcification noted involving left parasellar region. Skull vault shows right frontal bossing.

MRI study of brain was done, which revealed multiple linear areas of hyperintensity noted on FLAIR in juxta cortical white matter of right fronto - parieto temporal and left parieto - occipital region. A well-defined oval shaped extra axial rim of calcified foci noted in left parasellar region. The lesion appears hyperintense on T2, isointense on T1, shows flow void on FLAIR and blooming on GRE. The lesion causes mass effect over the left temporal lobe and optic nerve.

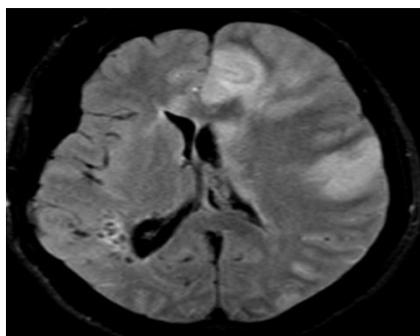
MR angiogram showing aneurysm involving the left internal carotid artery. With these findings tuberous sclerosis with intracranial aneurysm to be considered.



**Fig-1A:** Non contrast CT axial section shows multiple hyperdense, calcified small subependymal nodules



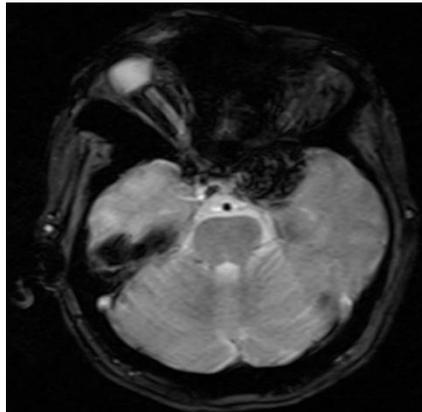
**Fig-1B:** Non contrast CT axial section shows well defined lesion with peripheral curvilinear calcification noted involving the left parasellar region



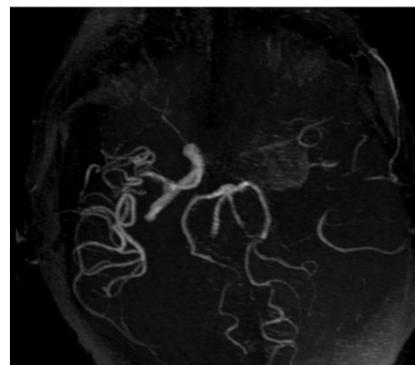
**Fig-2 A:** MRI Brain axial section FLAIR T2 Multiple linear areas of hyperintensity noted in juxtacortical white matter of left fronto-parieto-temporal region



**Fig-2 B:** MRI Brain axial section partially thrombosed giant cavernous sinus aneurysm involving left cavernous portion of internal carotid artery



**Fig-2 C: MRI Brain axial section GRE Well defined oval shaped lesion shows blooming involving left cavernous portion of internal carotid artery**



**Fig-3: MR Angiography Thrombus involving left internal carotid artery**

**DISCUSSION**

Tuberous sclerosis complex is a neurocutaneous disorder or phakomatosis developed from the abnormal growth cell of the embryonic ectoderm producing multiple tumours in the skin, eyes, heart, nervous system and kidneys [4].

Criteria for making the diagnosis of tuberous sclerosis have been categorized into major and minor features, of which two major, or one major and two minor features are diagnostic (Table-I)

**Table-I: Criteria for tuberous sclerosis**

Major Criteria	Minor Criteria
Facial angioma	Multiple pits in dental enamel
Ungual fibroma	Hamartomatous rectal polyps
Shagreen patch	Bone cysts
Hypomelanotic macule	Multiple renal cysts
Internal carotid artery tuber	Gingival fibromas
Subependymal nodule	Aneurysms
Subependymal giant cell astrocytoma	
Retinal hamartoma	
Cardiac rhabdomyoma	

Patients with tuberous sclerosis complex (TSC) can develop a number of renal lesions, the most common being angiomyolipomas and cysts[6].

The intracranial features of TSC are cortical or subcortical tubers, subependymal nodules, subependymal giant cell astrocytomas, and white matter radial migration lines[7]. Tubers are most commonly found in the cerebrum, 90% being present in the frontal lobes. On NECT in early stages tubers appear as low

density subcortical masses. In late stages they appear isodense to brain. 50% calcify by the age of 10 years. On T2weighted and FLAIR MR Images, tubers typically appear as areas of increased signal intensity in the cortical and subcortical regions.

Subependymal nodules (SEN) are found on the walls of the lateral ventricles and are either discrete or roughly confluent areas of rounded hypertrophic tissue. Typically benign, subependymal nodules can

degenerate into sub ependymal giant cell astrocytomas in 5–10% of cases. On NECT the nodules occur anywhere along the ventricular surface but are most commonly found at the caudothalamic groove in the region of the foramen of Monro. 50% calcify. Enhancing SEN may be suspicious for sub ependymal giant cell astrocytoma [3].

Ophthalmic features associated with TSC can be divided into retinal and non-retinal. The retinal lesions are known as astrocytic hamartomas [8]. Non retinal lesions include coloboma, angiofibroma of the eyelid and papilledema (related to hydrocephalus).

TSCofbones involvement shows multiple sclerotic lesions, Hyperostosis of inner table of skull, scoliosis, bone cyst, periosteal new bone formation and bone cysts have also been described [9].

The three main pulmonary lesions found in TSC are LAM, multifocal micronodular pneumocyte hyperplasia (MMPH), and clear cell “sugar” tumor (CCST) of the lung [10].

A cardiac rhabdomyomas have been diagnosed by two-dimensional echocardiography in the fetus[11]. However, the incidence in the newborn may be as high as 90% and in adults as low as 20%.

Vascular lesions (stenosis, ectasia, and aneurysm) are not common in TSC and have been found predominantly in peripheral vessels. Vascular histopathology has demonstrated abnormal architecture, including fragmented or deficient elastic fibers, mucopolysaccharide deposition, dense fibrous tissue, and calcifications [12].

## CONCLUSION

Intracranial aneurysms are a rare finding in tuberous sclerosis, but should be considered in patients with new cranial neuropathies. Neither surgical nor endovascular outcomes have been well described in the literature [13]. Only 17 prior cases of intracranial aneurysms in tuberous sclerosis have been reported in the literature [14]. We report a large aneurysm extending from the left cavernous to distal internal carotid artery causing right upper and lower limb weakness in a 32 year old female with tuberous sclerosis. Once diagnosed, aneurysm occlusion with internal carotid artery surgical clipping and endovascular treatment are two mainstays to treat, either rupture or unruptured[15].

## REFERENCES

1. Hart B, Depper M, Clericuzio C. Síndromes neurocutáneos. En: Orrison WW, editor. *Neurorradiología*. Madrid: Harcourt; 2001. p. 1732-49.
2. Barkovich AJ. *Facomatosis*. En: Barkovich AJ. *Neuroimagenología pediátrica*. Madrid: Marbán; 2001. p. 417-29.
3. Shahara Haque, Tarannum Marshed, Hasina Begum-Tuberous sclerosis complex A case report *Chattagram Maa-O-Shishu Hospital Medical College Journal*. 13(3), 2014. p.89-92
4. Gupta S, Bhowate R, Degwekar SS. Clinical and radiological findings related to tuberous sclerosis complex: a case report. *The journal of contemporary dental practice*. 2008;9(4):85-91.
5. Crino PB, Nathansosn KL, Henske EP. The tuberous sclerosis complex. *N Engl J Med*. 2006; 355: 1345–56.
6. Keith A, Casper MS, Lane FD, BinChen, John JB. Tuberous Sclerosis Complex: Renal Imaging Findings. *Radiology*. 2002; 225.
7. Babak NK and Noriko S. Neuroimaging of Tuberous Sclerosis: spectrum of Pathologic Findings and Frontiers in Imaging. *American Journal of Roentgenology*. 2008; 190(5): 304-309.
8. Rowley SA, O'Callaghan FJ, Osborne JP. Ophthalmic manifestations of tuberous sclerosis: a population based study. *Br j Ophthalmol*. 2001; 85: 420-423.
9. Roach ES, Gomez MR, and Northrup H. Tuberous sclerosis complex consensus conference: Revised clinical diagnostic criteria. *J Child Neurol*. 1998;13:624-8.
10. Rupa R, Sadhna V. Clinically relevant Imaging in tuberous Sclerosis. *Journal of Clinical imaging Science*. 2011; 1: 39.
11. Maryum M, Mohammad D, Mahmood M. Prenatal Diagnosis of Tuberous Sclerosis by Fetal Echocardiography with an Unusual Clinical course After Birth *arch cardiovasc Image*. 2013; 1(2):83-85.
12. Jarrett ME, Libertiny G, Gould SJ, Morris P. Carotid artery aneurysm in a child with tuberous sclerosis. *European journal of vascular and endovascular surgery*. 1998 Jul 1;16(1):80-1.
13. Alam MJ, Das S, Satter AR, Rahman MH, Mahmud E. Tuberous sclerosis with giant internal carotid artery aneurysm resulting left third nerve palsy: A case report. *Bangladesh Medical Journal*.;43(2):103-5.
14. Juneyoung LY, Galgano MA, Tovar-Spinoza Z, Deshaies EM. Coil embolization of an intracranial aneurysm in an infant with tuberous sclerosis complex: A case report and literature review. *Surgical neurology international*. 2012;3.
15. Chen YL, Luo CB, Hsu SW, Rodesch G, Lasjaunias P. Tuberous sclerosis complex with an unruptured intracranial aneurysm: manifestations of contiguous gene syndrome. *Interventional Neuroradiology*. 2001 Dec;7(4):337-41.