RESEARCH PAPER | Medical Science | Volume : 4 | Issue : 12 | Dec 2014 | ISSN - 2249-555X

A Case of Tuberous Sclerosis

KEYWORDS: Tuberous sclerosis, seizures

Dr. Pothamsetty Ravikaladhar Reddy
M.D. Assistant Professor of Medicine, Government General Hospital, Kurnool Medical College, Kurnool .

Dr. Kekathi Vidyasagar
M.D. Associate Professor of Medicine,, Government General Hospital, Kurnool Medical College ,Kurnool .

Dr. S.ChandraSekhar
M.D. Professor of Medicine, Government General Hospital, Kurnool Medical College ,Kurnool . A.P.

Dr.P.Prasanthi
Resdent in General Medicine, Government General Hospital, Kurnool Medical College ,Kurnool .A.P.

ABSTRACT *Tuberous sclerosis or tuberous sclerosis complex is an autosomal dominant genetically inherited disorder characterised by hamartoma formation in multiple organs, particularly the skin, brain, eye, kidney and heart, epilepsy, low IQ. We report a case of 14 year old male patient with tuberous sclerosis presented as refractory seizures with hamartoma in multiple organs and cutaneous manifestations.*

CASE REPORT:

- A 14year old male patient came with complaints of Seizures since 5 months about 2 to 3 episodes daily characterised by involuntary movements followed by loss of consciousness for 10mins.No post ictal drowsiness, no tongue bite or involuntary micturition or defecation during the episodes.

No h/o headache or vomiting, fever, trauma, syncope or palpitations

On examination:

- patient was conscious and coherent, moderately built and nourished
- No anemia, cyanosis, jaundice, clubbing, pedal edema or lymphadenopathy
- Facial angio fibromas in the malar regions of the face

Hypo pigmented macules in elliptical shape over the back and left thigh regions

- Thickened areas of skin and sub cutaneous tissue over the lower back (Shagreen patches)
- Linear grooves over finger nails
- Vitals:
- PR-86bpm ,regular in rhythm, normal volume, all peripheral pulses felt, no RR/RF delay

BP-110/70mmhg

- Other system examination was unremarkable. Nervous system-conscious, coherent, no focal deficit diagnosed provisionally as tuberous sclerosis on the basis of seizures with typical cutaneous manifestations.
- Patient was started on sodium Valproate for control of seizures and planned to taper Carbamazepine which he was already using for seizures.
- Advised for regular follow up

INVESTIGATIONS:

Hb-10.gm/dl,Total Count-7000/cmm, Platelet count-2,37,000/cmm,RBS-94mg/dl, Blood Urea-25mg/dl S.creatinine-0.8mg/dl,S.bilirubin-0.8mg/dl,S.electrolytes-Na 130meq/dl,K 4.2meq/dl Cl 105meq/dl ,

U/S abdomen: Multiple hypo echogenic lesions largest 1.2cms in left kidney-renal angiomyolipomas, Rt kidney was normal, Spleen, liver,and rest of the abdomen was normal.

- CECT brain- sub ependymal calcified nodule in right side of foramen of Monro s/o tuberous sclerosis
- Fundus examination-normal,2D ECHO-normal,X-ray chest PA view-normal

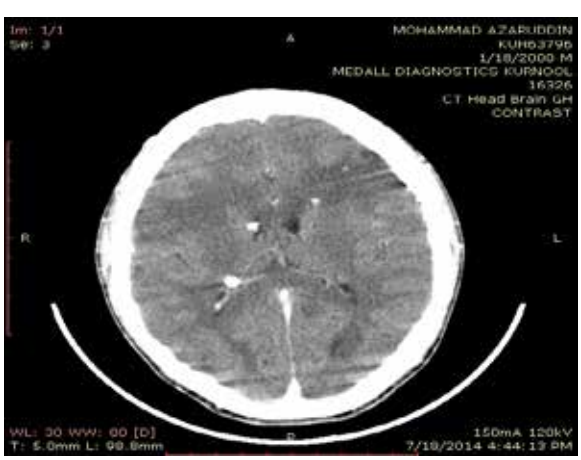

Figure1: CECT brain showing sub ependymal calcified nodule in right side of Foramen of Monro. S/o Tuberous sclerosis

Volume : 4 | Issue : 12 | Dec 2014 | ISSN - 2249-555X

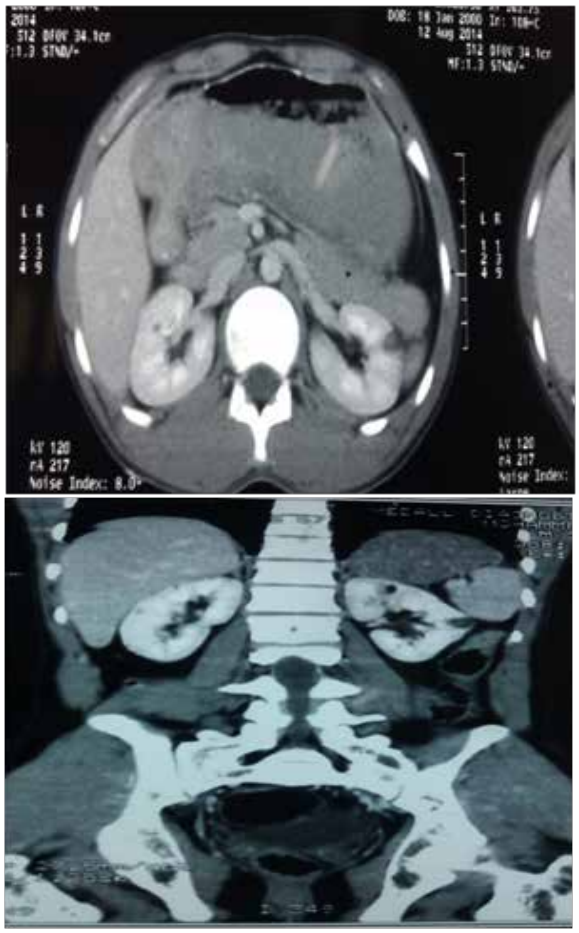

Figure 2: CECT abdomen:left kidney small enhancing hypodense cortical lesion with hypodensity in upper and mid pole regions s/o tuberous hamartomas

DISCUSSION:

Tuberous sclerosis or tuberous sclerosis complex is an autosomal dominant genetically inherited disorder with a birth incidence of around one in 10,000 [1]. However, with more sensitive screening the prevalence may be as high as one in 6,000 [2,3] characterised by hamartoma formation in multiple organs, particularly the skin, brain, eye, kidney and heart, epilepsy, low IQ. It is caused by mutation of two genes TSC1, and TSC2, which encode Hamartin and Tuberin, respectively. Diagnosis can be established by presence of typical cutaneous features in epileptic patient with hamartomas in multiple organs.

CRITERIA TO DIAGNOSIS TUBEROUS SCLEROSIS ARE:

- DEFINITE- either 2 major or 1major+2minor features
- PROBABLE-1major+1minor features
- POSSIBLE-either 1major or 2 or more minor features

Major features-

- Facial angio fibromas
- Hypo melanotic macules(>3)
- Non traumatic ungual or peri ungual fibromas
- Shagreen patches
- Multiple retinal nodular hamartomas
- Sub ependymal nodules
- Cortical tuber
- Sub ependymal gaint cell astrocytomas
- Cardiac rhabdo myomas
- Lymphangiomyomatosis
- Renal angiomyolipomas

Minor features:

- Multiple randomly distributed pits in dental enamel
- Hamartomatous rectal polyps
- Bone cysts
- Cerebral white matter radial migration lines
- Gingival fibromas
- Non renal hamartoma
- Retinal achromic patch
- Confetti skin lesions
- Multiple renal cysts

In our case patient presented with seizures having Facial angio fibromas in the malar regions of the face (fig-3) Hypo pigmented macules in elliptical shape over the back and left thigh regions, thickened areas of skin and sub cutaneous tissue over the lower back (shagreen patches) (Fig-4), Linear grooves over finger nails with small enhancing hypodense cortical lesion with hypodensity in upper and mid pole regions left kidney tuberous hamartoma , sub ependymal calcified nodule in right side of foramen of monro of brain suggestive of tuberous sclerosis fulfilling the criteria of definite case of Tuberous sclerosis.

The commest cause of death in TS is renal complication (3&5). Renal angiomyolipomas are present in 93% of patients with tuberous-sclerosis-associated pulmonary lymphangiomyomatosis [3]. It is important to recognize LAM before renal surgery for angiomyolipoma because of the risk of spontaneous pneumothorax or other perioperative pulmonary complication [5,8]. Pneumothoraces ultimately occur in approximately 60 to 70% of patients with LAM, and the rate of recurrence is > 70%, the highest among all chronic lung diseases [11]. In our case renal hamartomas present not associated with pulmonary lymphangiomyomatosis.

Volume : 4 | Issue : 12 | Dec 2014 | ISSN - 2249-555X

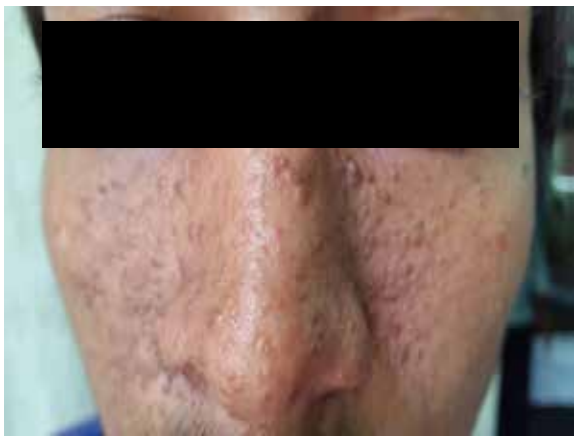

Figure-3 facial angiomas in malar region

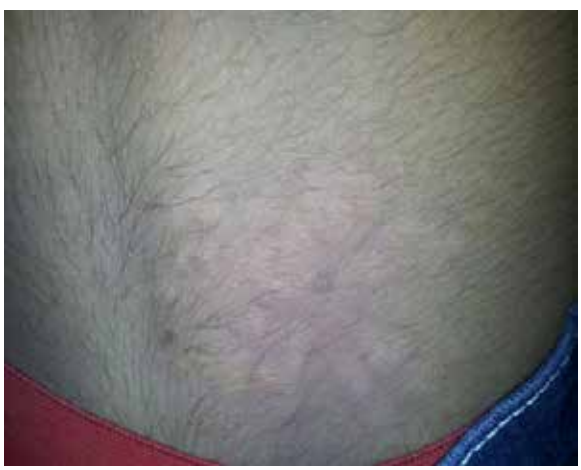

Figure-4 Shagreen patches

Conclusion:

Tuberous sclerosis is a genetically inherited rare disorder with multi system involvement with typical cutaneous manifestations that clinch the diagnosis. Early diagnosis helps in diagnosing the complications and treating in a better way that improves the overall survival. Because of the rarity of the condition we report this case.

REFERENCE

1. Hancock E, Tomkins S, Sampson J, Osborne J: Lymphangioleiomyomatosis and tuberous sclerosis. | Respir Med 2002, 96(1):7-13 | 2. Choyke PL, Glenn GM, Walther MM, Zbar B, Linehan WM: Hereditary renal cancers. | Radiology 2003, 226(1):33-46. | 3. Curatolo P, Bombardieri R, Jozwiak S: Tuberous sclerosis.Lancet 2008, 372(9639):657-68 | 5. Tuzel E, Kirkali Z, Mungan U, Culer C, Sade M: Giant angiomyolipoma associated with marked pulmonary lesions suggesting lymphangioleiomyomatosis in a patient with tuberous sclerosis. Int Urol Nephrol 2000, 32(2):219-22 | 8. Yoshida S, Hayashi T, Ishii N, Yoshinaga A, Ohno R, Terao T, Watanabe T, Yamada T, Osada H:Bilateral renal angiomyolipoma coexistent with pulmonary lymphangioleiomyomatosis and tuberous sclerosis. Int Urol Nephrol 2006, 38(3-4):413-5. | 11. McCormack FX: Lymphangioleiomyomatosis: a clinical update. Chest 2008, 133(2):507-16. |