ORIGINAL RESEARCH PAPER

General Medicine

TUBEROUS SCLEROSIS : ITS VARIED PRESENTATION

KEY WORDS: Fibromas, Hamartomas, Subependymal Nodules

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ABSTRACT

BACKGROUND: Tuberous sclerosis is clinical triad of papular facial nevus, seizures and mental retardation is found in less then half of patient.

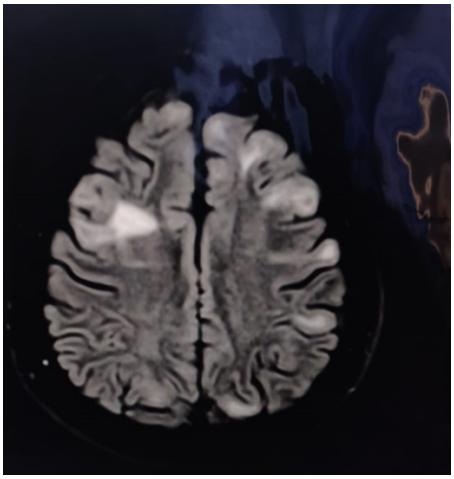
Tuberous sclerosis complex is an unusual autosomal dominant neurocutaneous syndrome characterized by the development of benign tumors affecting different body systems affecting the brain, skin, retina, and viscera. It is characterized by cutaneous changes, neurologic conditions, and the formation of hamartomas in multiple organs leading to morbidity and mortality. The most common oral manifestations are fibromas, gingival hyperplasia, and enamel hypoplasia. The management of these patients is often multidisciplinary involving specialists from various fields. Here, we present a case report of a 26-old-year male patient with characteristic clinical, radiological, and histological features of tuberous sclerosis complex.

INTRODUCTION

- Tuberous sclerosis is a neurocutanous syndrome with an autosomal dominant inheritance.
- It is caused by mutation in one of the two genes TCS1 encoding hamartin or TCS2 encoding tuberin exhibiting multiple organ system such as brain, kidney, heart, lungs, eyes and skin.
- Clinical triad of papular facial nevus, seizures and mental retardation is found in less then half of patient.
- Radiological hallmarks of this neurocutaneous syndrome are universally accepted as sufficient for diagnosis.
- Approximate incidence of less than one in five thousand to ten thousand. It has no cure, but treatment as medicine, educational and occupational therapy is available.
- We report four patients who presented with typical skin lesions, seizures and abdominal lump with pain.

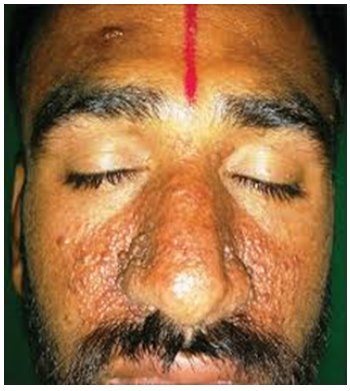
Case 1

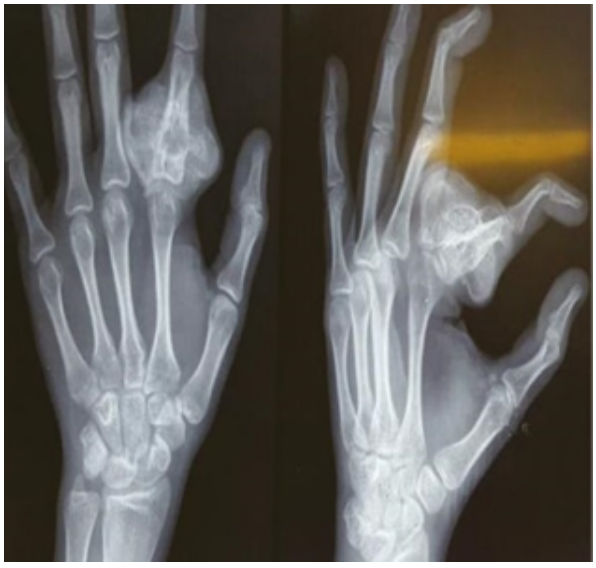
- 29 years male presented with episode of seizures.
- **Clinical examination:** Adenoma sebaceum on the face .
- Hypomalemic patch found on back.
- Patient mentally retarted .
- **Mri brain:** multiple cortical hamartoma & bilateral cerebral hemisphere and sub ependymal nodules



Case 2

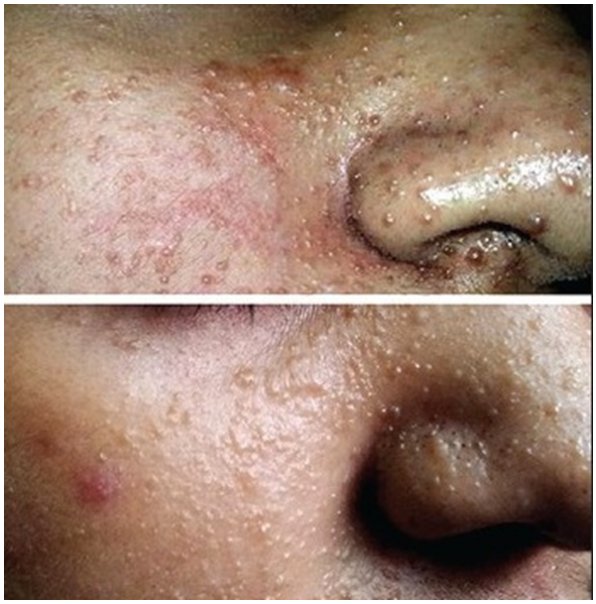
- 25 year/Male presented with abdominal pain.
- Past history of seizures.
- **Clinical examanination:** Adenoma sebaceum on the face.
- Mentally retarded .
- **Non Contrast CT brain:** Subependymal calcified nodules .
- **X-Ray:** Bone cysts in terminal phalanges and flame shaped radio opaque lesions in both iliac wings
- **Usg abdomen:** Mixed echogenic well defined mass arising from upper pole of kidney .
- **Contrast CT scan:** Heterodense lesion with inhomogenous enhancement arising from upper pole of left kidney.
- **Pathologically:** Turned out to be a malignant variety of Angiomyolipoma.





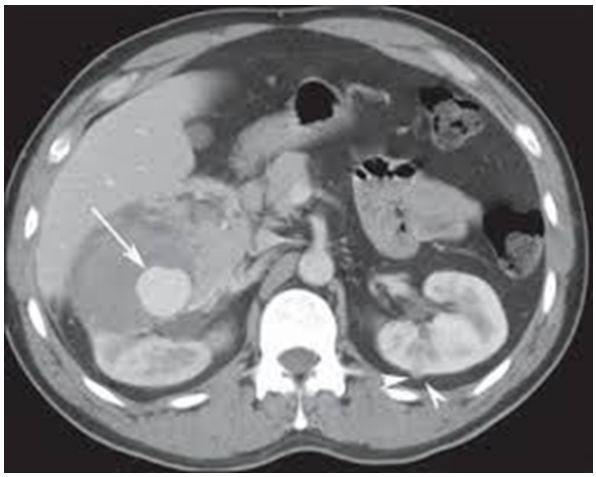
Case 3

- 14 years male presented with lump in abdomen.
- **Clinical examination:** Adenoma sebaceum on face.
- Past history of seizures.
- **Contrast CT abdomen:** Mixed echogenic mass with cystic, solid components and areas of fat density, arising from upper pole kidney.



Case 4

- 20 year female presented with abdominal pain.
- Patient was mentally retarded .
- Clinical examination of the patient revealed presence of facial angiofibromas .
- **Ultrasonography:** Enlarged kidneys, the parenchyma of which was completely replaced by multiple echogenic lesions which was suggestive of angiomyolipomas. Liver showed presence of haemangiomas .
- **Non contrast CT brain:** Calcified subependymal noduless



Clinical Criteria

Major criteria	Minor criteria
Hypomelanotic macules	Confettil skin lesions
Angiofibromas	Dental enamel pits
Ungual fibromas	Intra oral fibromas
Shagreen patch	Muliple retinal lesions
Multiple retinal hamartomas	Multple renal cysts
Cortical dysplasia	
Subependymal nodules	
Subependymal giant cell astrocytoma	
Cardiac rhabdomyoma	
lymphangioleiomyomatos	
angiomyolipoma	

DISCUSSION

- Tuberous sclerosis is an important genetic disorder that affects the patient and the family in various ways.
- The neuro cutaneous syndrome is named for the firm whitish tuber like nodules arising from the cerebral convolutions .
- Most frequently involved organs are brain, kidneys, lungs, heart, skin and skeleton.
- Multiple research projects are being done around the world regarding further work up of the genes involved and treatment strategies.
- Since understanding its pathogenesis, multiple drug therapies are available for certain manifestations of the disease.
- Patient along with symptomatic control of seizures, should be offered special schooling, and occupational therapy.
- Surgery including demabrasion and laser treatment, may be useful for treatment of skin lesions.
- Multiple cases have been reported highlighting involvement of different organs in tsc.
- In majority, there was probable diagnosis with one major plus one minor positive feature .

CONCLUSION

- Tuberous sclerosis is a lifelong condition.
- Individuals should be regularly monitored by an experienced clinician.
- Tuberous sclerosis must be included in the differentials of patient presenting with seizures, development delay and mental retardation.

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