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Case Report of Tuberous Sclerosis

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1. Introduction

Tuberous sclerosis is an autosomal dominant neurocutaneous syndrome characterized by various abnormalities, including benign hamartomatous tumors in multiple organs. Also known as Bourneville disease, named after Désiré - Magloire Bourneville, the French physician who discovered the potatolike appearance of cortical lesions in the brains of patients with this condition [1], tuberous sclerosis is the second most common phakomatosis behind neurofibromatosis type 1. It has a prevalence estimated to be 1 in 6000 with approximately 1.5 million people affected worldwide [2]. The diagnosis of tuberous sclerosis carries a variable prognosis depending on the severity of symptoms. However, mortality can be as high as 40% by age 35 [3].

We report a case of 46 years old female patient who presented with pain b/l flanks. CECT abdomen was done which showed bilateral renal angiomyolipoma. HRCT chest showed lymphangioleiomyomatosis and NCCT head showed multiple subependymal nodules.

2. Case Report

A 46 years old female patient with history of pain bilateral flanks presented to surgery department. Clinical examination showed multiple skin lesions like periungual fibroma, facial angiofibroma, shagreen patch and ash leaf macules. Patient was sent for radiological investigation. CECT Abdomen was done which showed fat density lesions diffusely involving bilateral kidneys with pseudoaneurysm involving branches of left renal artery. HRCT chest shows multiple well defined innumberable cystic lesions in b/l lungs. NCCT brain shows multiple calcified lesions involving bilateral subependymal regions.

Based upon skin manifestations, CECT abdomen, HRCT Chest and NCCT head findings, a diagnosis of tuberous sclerosis complex is kept.

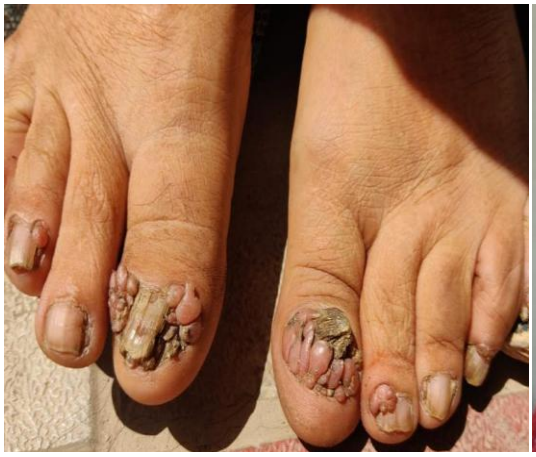

Figure 1 (a)

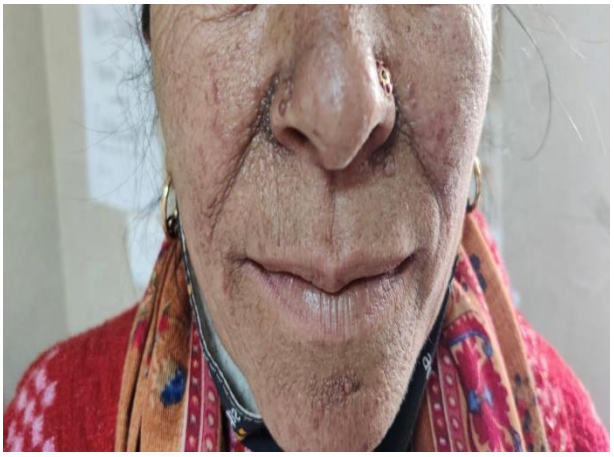

Figure 1 (b)

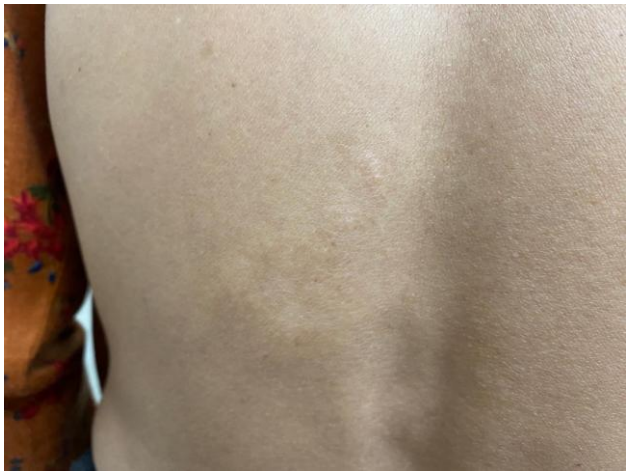

Figure 1 (c)

Figure 1 (a, b and c): shows skin manifestations of tuberous sclerosis including periungual fibroma (a), facial angiofibroma (b) and shagreen patch.

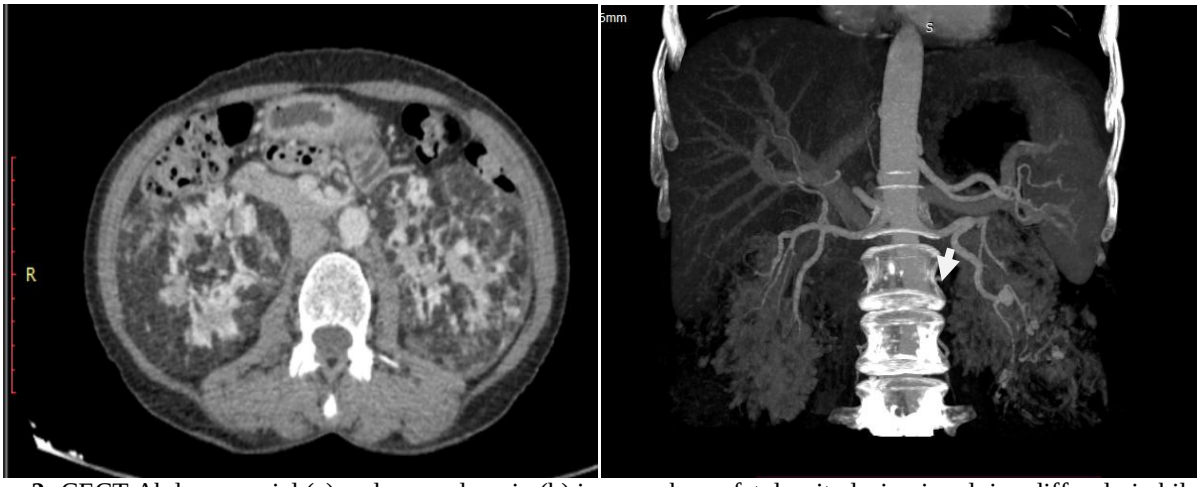

Figure 2: CECT Abdomen axial (a) and coronal angio (b) images shows fat density lesion involving diffusely in bilateral kidneys and a pseudoaneurysm involving branches of left renal artery (white arrow).

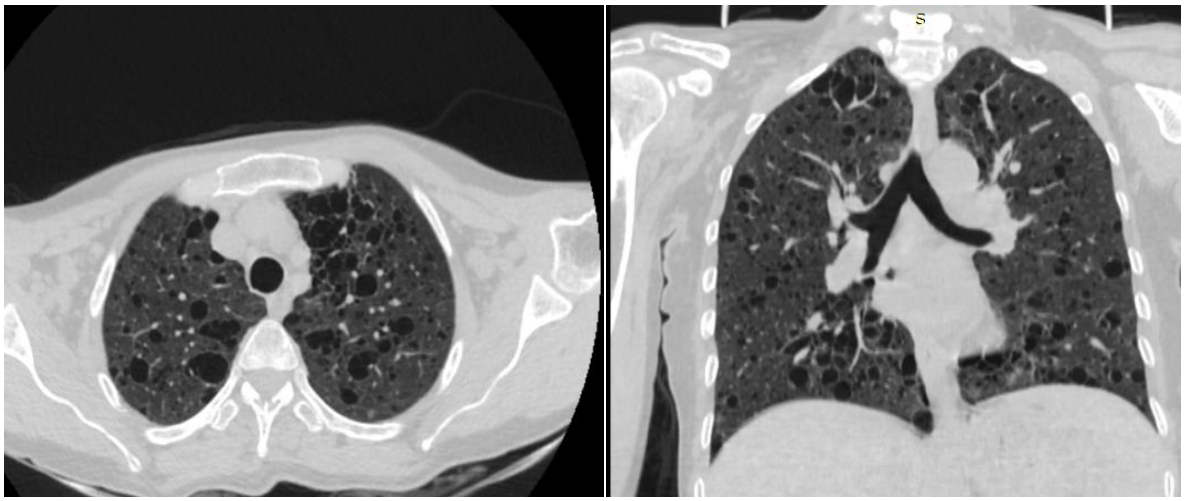

Figure 3: HRCT Chest axial (a) and coronal (b) images shows multiple well defined round cystic lesions involving b/l lungs suggestive of lymphangioleiomyomatosis.

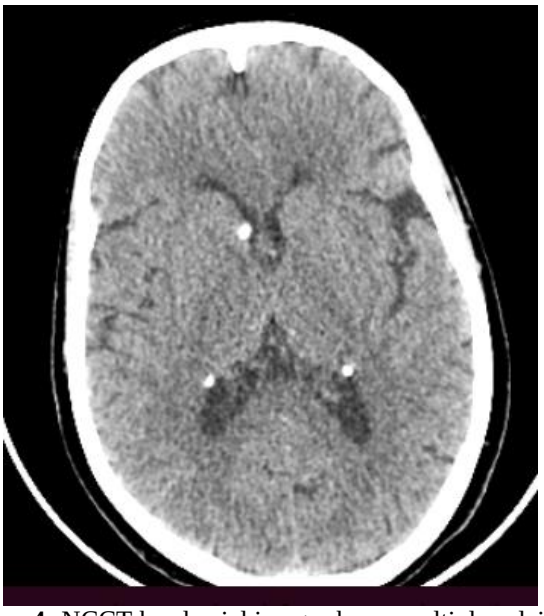

Figure 4: NCCT head axial image shows multiple calcified subependymal nodules involving bilateral cerebral hemispheres.

3. Discussion

Tuberous sclerosis complex (TSC) is an uncommon multiorgan disorder that may present many and different manifestations on imaging. Radiology plays an important role in diagnosis and management, and can substantially improve the clinical outcome of TSC. The literature reports that SENs are present in about 80% of TSC patients. SENs frequently show calcifications, particularly at an early stage [4]. SENs are typically less than 1 cm in size and better detected by CT than MRI.
Renal AML is an uncommon benign tumor, which consists of fat, abnormal blood vessels and smooth muscle elements in varying proportions.

As to the pulmonary lesions, LAM is a rare disease that affects young women of child - bearing age. Cysts are usually 2–5 mm but can be as large as 25–30 mm in size. Cysts are typically round or ovoidal. Pulmonary LAMs are characterized by progressive widespread proliferation of smooth muscle throughout the perilymphatic, peribronchiolar, and perivascular regions of the lungs [5]. In addition, cardiac rhabdomyoma occurs in at least 60% of children with TSC.

4. Conclusion

TSC is an uncommon multiorgan disorder that may present itself with many and different manifestations on imaging scan. Some image manifestations can easily be confused with other diseases. The findings in our case report suggest that the patient clinical features, family history and all multiorgan images should be fully integrated. Radiology plays an important role in diagnosis and management, and can substantially improve the clinical outcome of TSC.

References

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