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Images in Medicine

Tuberous sclerosis: Inside and outside

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Introduction

Tuberous sclerosis is an autosomal dominant neurocutaneous syndrome characterized by hamartomatous lesions in multiple organs. We report an unusual case of tuberous sclerosis in pictorial format wherein most of the diagnostic findings were present. This case would be of immense value in identifying this disease entity at all echelons of medical care.

Case report

A 28-year-old male patient was detected to have hypertension on routine medical examination and was advised ultrasonography of abdomen for further evaluation. The USG abdomen revealed small, well-defined, round hyperechoic lesions in bilateral renal cortices suggestive of angiomyolipoma (AML). Due to presence of facial lesions and bilateral renal AML, a suspicion of tuberous sclerosis was raised and further

radiological and dermatological evaluation was done to confirm the diagnosis. The unusual feature of this case was that a total of 11 out of the 20 diagnostic criteria were demonstrable in this patient who was asymptomatic and did not fulfill the Vogts triad.

Discussion

Tuberous sclerosis was first described by Von Recklinghausen in 1862 when he reported a case of a newborn with tumors in the heart and nodules in the brain. In 1880, Bourneville first used the term tuberous sclerosis in a case where nodules in the brain resembled tubers or roots of a plant.¹

Tuberous Sclerosis Complex (TSC) is a neurocutaneous syndrome where the basic pathology is the abnormal differentiation of the primitive ectoderm which forms the skin and neural tissue.

Two genes have been implicated for this disease, viz. TSC 1 located on chromosome 9 and coding for the protein Hamartin and TSC 2 located on chromosome 16 coding for Tuberin. Even though the genetic basis of this disease is understood, the diagnosis is still based on clinical and imaging findings which are further complicated by the broad spectrum and numerous manifestations of the disease entity.^{2,3}

The Consensus Conference on TSC by the National Tuberous Sclerosis Association in 1998 categorized major and minor diagnostic features for the diagnosis of the tuberous sclerosis complex. Criteria for definite, probable, and possible diagnosis of tuberous sclerosis were proposed. The 2012 International Tuberous Sclerosis Complex Consensus Group further reviewed and updated the diagnostic criteria to include genetic testing and reduced the diagnostic classes to two, that is,

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Fig. 1 – Dermatological features: (a) angiofibroma, (b) dental pits, (c) gingival fibroma, (d) ash leaf macule, (e) confetti lesions, (f) Café-au-lait macule, (g) Shagreen patches, and (h) periungual fibroma in toe.

Fig. 2 – Radiological findings: (a) NCCT head, axial image – calcified subependymal nodules, (b) MRI brain, FLAIR image – subcortical nodules, (c) NCCT chest, lung window – single thin walled cystic lesion in right lung, (d) NCCT chest, lung window – tiny nodules with random distribution representing multifocal micronodular pneumocyte hyperplasia (MMPH), (e) CECT abdomen, axial image – angiomyolipoma in posterior aspect of left kidney showing fat density within, and (f) NCCT PELVIS, bone window – sclerotic foci within bilateral iliac bones.

possible and definite. Definite diagnosis: Two major features or one major feature with ≥ 2 minor features; Possible diagnosis: Either one major feature or ≥ 2 minor feature (Figs. 1–2).⁴

Clinical diagnostic criteria

Major features

1. Angiofibromas (≥ 3) or fibrous cephalic plaque
2. Ungual fibroma (≥ 2)
3. Hypomelanotic macules (≥ 3 , at least 5 mm in diameter)
4. Shagreen patch
5. Multiple retinal hamartomas
6. Cortical dysplasia
7. Subependymal nodule
8. Subependymal giant cell astrocytoma
9. Cardiac rhabdomyoma
10. Lymphangiomyomatosis (LAM)
11. Angiomyolipoma (≥ 2)

Minor features

1. Dental enamel pits (≥ 3)
2. Intraoral fibromas (≥ 2)
3. Nonrenal hamartomas
4. Retinal achromic patch
5. “Confetti” skin lesions
6. Multiple renal cysts

Our patient did not have retinal, cardiac, or rectal lesions. The cystic lesion noted in the lung parenchyma may be considered as ‘forme fruste’ of Lymphangiomyomatosis. The patient is now on follow-up for new lesions.

Conclusion

Tuberous sclerosis has a wide variety of presentations. It is essential to recognize the common manifestations and then evaluate for other lesions even in asymptomatic patients. The mainstay of diagnosis is a detailed dermatological assessment coupled with targeted imaging of various organs. An early diagnosis and subsequent follow-up are essential for administering appropriate treatment and to improve quality of life.

Conflicts of interest

The authors have none to declare.

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