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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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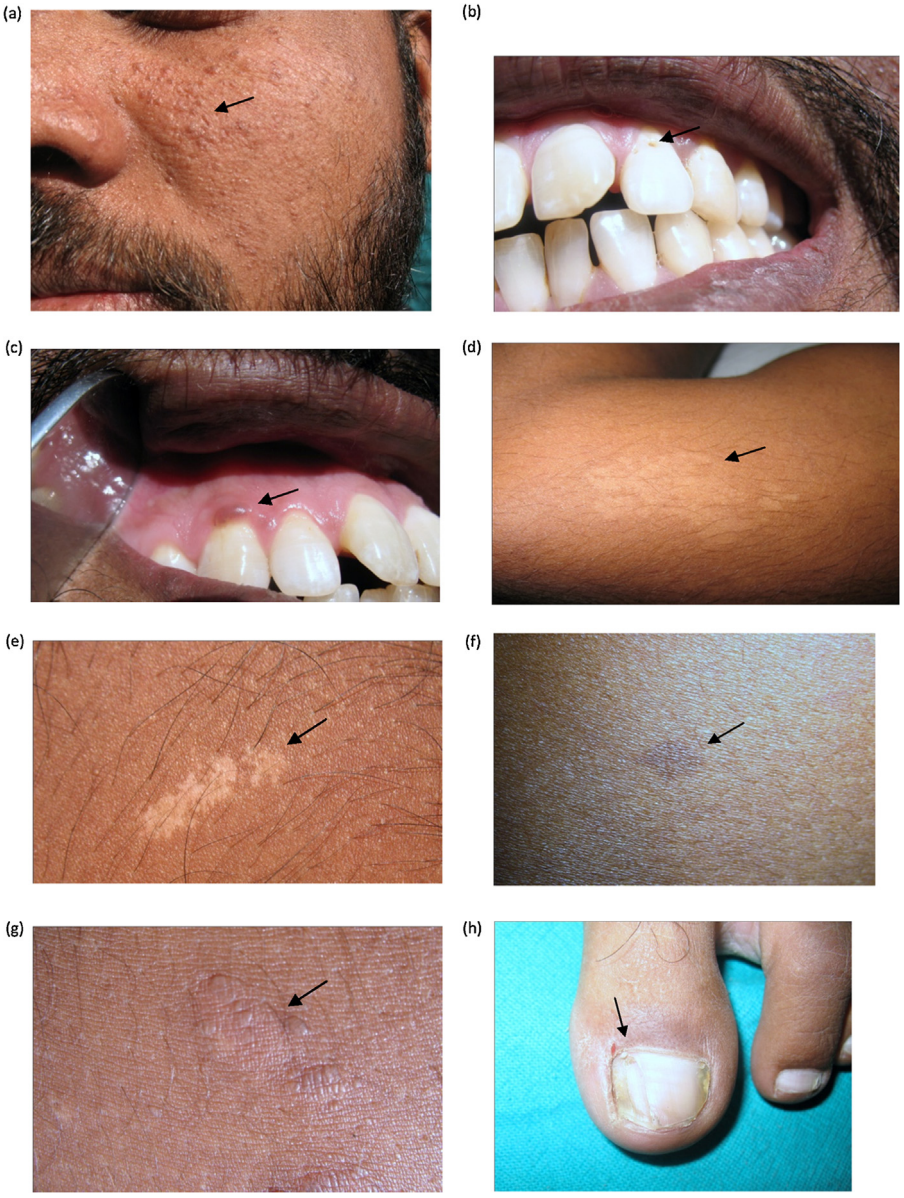

Fig. 1 – Dermatological features: (a) angiofibroma, (b) dental pits, (c) gingival fibroma, (d) ash leaf macule, (e) confetti lesions, (f) Café-au-lait macule, (g) Shagreen patches, and (h) periungual fibroma in toe.

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