

Clinical Image

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Cutaneous manifestations of tuberous sclerosis complex: An uncommon entity

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Case summary

A middle-aged female from Northern India, presented to the Outpatient department with cutaneous lesions typical of Tuberous Sclerosis Complex (TSC) without intellectual disability. She had multiple skin lesions which are enlisted below:

- a. Hypomelanotic macule on medial aspect of left forearm (Figure 1)
- b. Periungual fibromas (Figure 2)
- c. Typical lesions of adenoma sebaceum distributed along and around the centre of face causing disfigurement (Figure 3)
- d. Shagreen patch over the lower back (Figure 4).

Discussion

Tuberous Sclerosis Complex (TSC) is a neurocutaneous syndrome with a prevalence of around nine in 100,000 cases [1]. It is characterized clinically by a triad of seizures, intellectual dis-

ability, and adenoma sebaceum. However, approximately 50% patients have normal intellect, and 15% with no seizures [2]. Cutaneous findings are the most common as well as readily appreciable manifestations of Tuberous Sclerosis Complex (TSC). These cutaneous manifestations compose most of the major and minor criteria (Table 1) for the diagnosis of TSC [3]. Of these, hypomelanotic macules are usually the earliest and most frequently observed cutaneous findings [4]. Other skin lesions do occur at variable frequencies and manifest at different ages.

Timely identification of such lesions not only helps identify the disease per se, but also enables us to prevent the disfigurement of the body parts especially face by timely application of various treatment modalities. Therefore, detailed dermatological examination is recommended upon the time of diagnosis of TSC, followed by at least annual skin examinations. Infact, patients are advised to use sun protective measures to minimise the appearance of some skin lesions. With these clinical image, we aim to make the clinicians in our part of world more familiar to the disease and its cutaneous manifestations.

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Figure 1: A hypomelanotic macule on the medial aspect of left forearm.



Figure 2: Multiple periungual fibroma.



Figure 3: Typical lesions of adenoma sebaceum distributed along and around the centre of face.



Figure 4: Shagreen patch over the lower back (Dorsum).

Table 1: Tuberous sclerosis complex diagnostic criteria.

Major features	Minor features
Hypomelanotic macules (≥ 3 , at least 5 mm diameter)	Confetti' skin lesions
Angiofibroma (≥ 3) or fibrous cephalic plaque	Dental enamel pits (>3)
Ungual fibromas (≥ 2)	Intraoral fibromas (≥ 2)
Shagreen patch	Retinal achromic patch
Multiple retinal hamartomas	Multiple renal cysts
Cortical dysplasias	Non-renal hamartomas
Subependymal nodules	
Subependymal giant cell astrocytoma	
Cardiac rhabdomyoma	
Lymphangiomyomatosis	
Angiomyolipomas	
Definite diagnosis= 2 major features or 1 major feature with ≥ 2 minor features.	
Possible diagnosis= 1 major feature or ≥ 2 minor features.	

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