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Focal gigantism in tuberous sclerosis



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ABSTRACT

A 38 year lady had recurrent generalized and myoclonic seizures since 6 years of age. On examination she had facial adenoma sebaceum and periungual fibromas, cutaneous angiofibromas on the back. Contrast enhanced magnetic Resonance Imaging of brain was suggestive of multiple cortical tubers with subependymal nodules consistent with the diagnosis of Tuberous sclerosis. Focal hypertrophy of the skin of the dorsal aspect of the left hand with focal gigantism was noted.

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A 38 year lady had recurrent generalized and myoclonic seizures since 6 years of age. On examination she had facial adenoma sebaceum (Fig. 1a) and periungual fibromas, cutaneous angiofibromas on the back. Contrast enhanced magnetic Resonance Imaging of brain was suggestive of multiple cortical tubers with subependymal nodules consistent with the diagnosis of Tuberous sclerosis. Focal hypertrophy of the skin of the dorsal aspect of the left hand with **focal gigantism** was noted (Fig. 1b).

Tuberous sclerosis complex in an autosomal dominant neurocutaneous disorder frequently recognized by presence of facial angiofibromas, periungual fibromas, shagreen patches; and presence of cortical tubers and subependymal nodules in MRI Brain.¹ Focal gigantism in the form of digital enlargement is an uncommon musculoskeletal association in tuberous sclerosis. The mechanism for its occurrence is unknown and it could be implicated to be due to localized tissue hyperplasia.²



Fig. 1. Showing patient of tuberous sclerosis with adenoma sebaceum (Fig. 1a) and focal gigantism and localized digital enlargement (Fig. 1b).

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Conflicts of interest

None.

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None.

Author contribution

Sanat R Bhatkar Drafting the article or revising it intellectual content, analysis or interpretation of the data, manuscript preparation.

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