

Case Report

Scalp shagreen patch: an unusual cutaneous manifestation of tuberous sclerosis complex: a case report

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ABSTRACT

Tuberous sclerosis complex (TSC) is an inherited neurocutaneous disorder with multisystem involvement and highly variable expression of the disease. Common cutaneous manifestations include angiofibromas, periungual fibroma, ash-leaf-shaped macules and shagreen patch which is slightly elevated soft skin-coloured plaque usually found in lumbosacral region. We report a case of TSC in a 21-year-old Saudi man with a 13 years history of asymptomatic skin lesions and generalized tonic-clonic seizures. Dermatological examination revealed facial angiofibromas, ash leaf macules, pedunculated fibromas on trunk, and shagreen patch on the back. Scalp swelling was found and confirmed to be shagreen patch by the histopathology findings. In conclusion, scalp shagreen patch is an unusual presentation of TSC. It should be considered in clinical examination of suspected cases of TSC with relevant histopathology to confirm the lesion.

Keywords: Collagenoma, Shagreen patch, Tuberous sclerosis complex

INTRODUCTION

Tuberous sclerosis complex (TSC) is an inherited neurocutaneous disorder characterized by pleomorphic features that involve different organ systems.^{1,2} It is an autosomal dominant genetic disorder with an incidence of approximately 1 in 5000 to 10,000 live births.^{3,4} It results from mutation of one of the two different genes (TSC-1 and TSC-2) which are located on chromosomes 9 and 16, respectively.⁵ As these genes act as tumor suppressors, their mutation leads to hamartomatous lesions in many organs including the skin.⁶

CASE REPORT

A 21-year-old Saudi man presented with a history of asymptomatic skin lesions which were confined to his face and first appeared at the age of 8 years old, then few months later, a new lesion developed on the scalp

associated with whitish and brownish discoloration on trunk and upper extremities. His parents gave no birth history of skin lesion or discoloration, but there was a history of recurrent attacks of generalized seizures since the age of 4 months for which, he is on control treatment and follow up by a neurologist. The condition was not associated with mental retardation, developmental delay, learning difficulties, behavioral abnormalities or systemic complaints. The patient's parents were first degree relatives and no reported family history of similar condition. Dermatological examination revealed firm non tender skin-colored swelling on scalp, 3X5 cm in size, suggestive of shagreen patch (figure 1-A) with another similar lesion found on the center of back midline (Figure 1-B), multiple facial angiofibromas in a butterfly distribution and brown pedunculated papules on forehead (Figure 2), few macular and patchy brown lesions (café au lait macules) on upper chest (Figure 3-A),

hypomelanotic white patches seen on upper left arm, suggestive of 'ash leaf spots' (Figure 3-B).



Figure 1: Shagreen patch on scalp measuring 3x5cm A) and on the midback B).



Figure 2: Facial angiofibromas in the butterfly area and gingival hyperplasia.

Intraoral examination showed gingival hyperplasia, (Figure 2). Systemic examination including CNS showed no significant abnormality. Brain MRI revealed multiple subependymal hamartomas with focal dots of calcification of the left lateral ventricle, cortical and subcortical hamartomas. The patient was clinically diagnosed as a case of tuberous sclerosis complex syndrome. Histopathology of a punch biopsy taken from the scalp lesion established the diagnosis of collagenoma (Figure 4). The patient was referred for ophthalmological examination, renal ultrasonography, skeletal radiographs, electrocardiograph, and pulmonary function tests. Regular sessions of ND-YAG laser were done for disfiguring facial lesions with acceptable results.

DISCUSSION

Tuberous sclerosis complex (TSC) is an inherited neurocutaneous disorder with multisystem involvement and highly variable expression of the disease. In our reported case, the patient was known to have generalized tonic clonic seizures since four months of age. This is consistent with the literature on TSC which documents that epilepsy is one of the most frequent and significant

causes of morbidity in TSC, affecting 79 to 90 percent of patients in population-based studies.⁷ Seizures begin in the first year of life in over 60 percent of cases; however, a risk for new-onset seizures into adult life was reported.^{8,9} Other common neurological manifestations include learning disabilities, autism, behavioral problems, and psychosocial difficulties; these are collectively termed TSC-associated neuropsychiatric disorders (TAND).^{10,11}

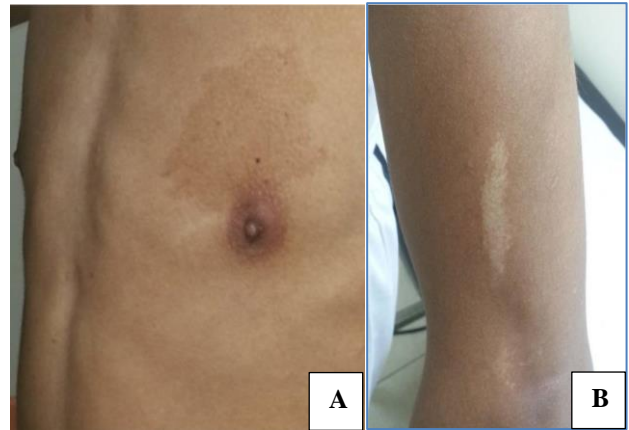


Figure 3: Café au lait on the chest A) and ash leaf spot on left arm B).

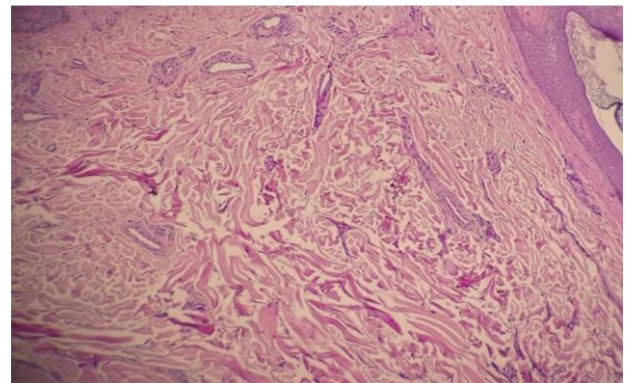


Figure 4: Histopathology of the scalp lesion showing haphazardly arranged collagen bundles in the dermis with mild distortion of the skin adnexa.

TSC has a wide range of mucocutaneous manifestations which include adenoma sebaceum (angiofibroma), connective tissue nevi, hypomelanotic macules, periungual fibromatous lesions, fibromatous plaque on forehead, and oral mucosal fibromas.¹² Shagreen patches are connective tissue nevi exhibited as irregular, slightly elevated areas of skin, 1-10cm in diameter found most commonly in the lower back region. Shagreen patches are not uncommon in patients with TSC; it was observed in up to 80% of patients in an Indian study, and to less frequency in Western literature (40%).¹³ In the present case report, scalp plaque was observed and confirmed by histopathology to be a shagreen patch in an unusual site

for its presentation. Diagnosis of TSC is mainly clinical supported by imaging and other relevant tests.⁵

Patient work up in our case was done in concordance with the published protocols which support the multidisciplinary approach of management including the neurosurgeon, neurologist, nephrologist, pulmonologist, cardiologist, ophthalmologist, and the genetic counsellor. Laser surgery treatment was performed for its usefulness in the treatment of associated skin lesions.¹⁴

CONCLUSION

TSC is a rare neurocutaneous disorder with neurological features commonly presented as epileptiform convulsions as well as variable skin lesions. Shagreen patch as one of the diagnostic skin manifestations which is commonly seen on the lower back and to our knowledge, not described before in the scalp region. Therefore it should be considered in clinical examination of suspected cases of TSC with relevant histopathology to confirm the lesion.

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