

Research Article

FACIAL HYPERPIGMENTATION IN A CASE OF TUBEROUS SCLEROSIS

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ABSTRACT

A 16 year old female a patient of tuberous sclerosis complex presented to our department with complaints of darkish lesions on the face since the age of two years.

Cutaneous manifestations occur almost universally in tuberous sclerosis complex, but hyperpigmented macules and papules along the nasolabial folds in association with the said complex have seldom been documented.

Keywords: *Tuberous Sclerosis Complex, Hamartomas, Angiofibromas*

INTRODUCTION

Tuberous sclerosis complex (TSC), an autosomal dominant disease characterized by hamartomas in different organs- mainly the brain, skin, kidney, liver, lung, and heart- affects one in 10,000 people in the general population. It is caused by mutations in tuberin and hamartin genes which form a complex that inhibit mammalian target of rapamycin (mTOR), a protein kinase whose constitutive activation due to mutations lead to uncontrolled cellular proliferation (Wataya-Kaneda *et al.*, 2011). The cutaneous signs of TSC and the diagnostic criteria have been well delineated, but TSC has rarely been associated with dark hyperpigmented macules on the naso-labial folds. We herein present such a case.

CASES

A 16 year old female, the second product of a non-consanguineous marriage presented to our department with complaints of darkish lesions on the face since the age of two years. She also complaint of an episode of epilepsy at the same time. On examination, multiple symmetric angiofibromas were seen over the face, hyper pigmented macules and papules along the nasolabial folds with multiple hypomelanotic patches over the back, chest and limbs (Figure 1). There was no history of similar complaints in the family. The patient was suspected as having TSC and was further investigated; Electro cardiogram showed T wave inversion in lead three and ST sagging in V5 and V6, ultrasound of the abdomen and pelvis demonstrated bilateral renal cortical subcentimetric ill-defined echogenic lesions, while MRI of the brain revealed multiple small subcentimeterized calcified subependymal nodules along bodies of bilateral lateral ventricles and multiple patchy T2/FLAIR hyperintense areas seen in bilateral cerebral hemisphere involving cortex and subcortical white matter at few places. Rest of the investigations were unremarkable. The patient was then counselled, her angiofibromas removed via electro cautery and referred to a neurologist and cardiologist for further management.



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DISCUSSION

TSC, also known as Bourneville–Pringle disease, is a genetic neurocutaneous disorder characterized by the presence of histologically benign hamartomas in different organs (Borowoska *et al.*, 2011). The clinical symptoms of TSC may appear gradually during life and hence the diagnosis may not become evident until adulthood. The most important clinical features of TSC are facial angiofibromas, renal angiomyolipomas, hypo pigmented macules, cardiac rhabdomyomas, cortical tubers and subependymal glial nodules in the brain (Hengstschläger, 2001). The greatest sources of morbidity are brain tumours, which cause seizures in 80–90% of affected individuals, mental retardation occurring in about half of affected individuals, and behavioural abnormalities (mostly autism) in over half of the affected individuals (Cheadle *et al.*, 2000).

Cutaneous manifestations occur almost universally in TSC, but hyper pigmented macules and papules along the nasolabial folds in association with the said complex have seldom been documented, the patient being more troubled by these hyperpigmentation than her other manifestations.

Rapamycin, also known as sirolimus, and its analogs, inhibitors of mTOR pathway could have been an exciting possibility to treat the angiofibromas and see whether the hyper pigmented macules also respond to the same, underscoring a common pathway for their development, but could not be used due to the high cost associated with the drug.

REFERENCES

Borowoska J, Schwartz RA, Kotulska K and Jozwiak S (2011). Tuberous sclerosis complex: tumors and tumorigenesis. *International Journal of Dermatology* **50**(1) 13-20.

Cheadle JP, Reeve MP, Sampson JR and Kwiatkowski DJ (2000). Molecular genetic advances in tuberous sclerosis. *Human Genetics* **107** 97–114.

Hengstschläger M (2001). Tuberous sclerosis complex genes: from flies to human genetics. *Archives of Dermatological Research* **293** 383–386.

Wataya-Kaneda M, Tanaka M, Nakamura A and Matsumoto S (2011). A topical combination of rapamycin and tacrolimus for the treatment of angiofibroma due to tuberous sclerosis complex (TSC): a pilot study of nine Japanese patients with TSC of different disease severity. *British Journal of Dermatology* **165**(4) 912-916.