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TUBEROUS SCLEROSIS WITH INTERESTING FEATURES

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A case of tuberous sclerosis with intracranial calcification, retinal phakoma and premature canities is reported for its interesting features.

Key Words : Intracranial calcification, Epiloia

Introduction

Tuberous sclerosis was first recognised as a specific disease in the 19th century. In 1880 Bourneville, a french neurologist, reported the case of a mentally retarded girl who also suffered from hemiplegia and epilepsy.[1] This is an autosomal dominant disease resulting from abnormal growth of ectodermal and mesodermal cells which in turn results in benign tumours of various organs. Tuberous sclerosis is also known as epiloia which is a telescopic term representing the triad epilepsy, low intelligence and angiofibroma. The diagnostic criteria for tuberous sclerosis have been considered as primary, secondary and tertiary features. Definite diagnosis of tuberous sclerosis can be made with one primary and two secondary or one secondary and two tertiary features.[2]

The skin lesions include angiofibroma over the face, ash leaf macules, fibromas about the nails and on the scalp and shagreen plaques. Pigmentary changes can be thumb-print, ash leaf, confetti or distributed in dermatomal pattern. Macular hypopigmentation of the retina and iris has also been described.[3]

Phakoma of the eye are very common and were present in about 50% of the patients with tuberous sclerosis.[4] According to Messinger and Clark visual disturbance and ophthalmoscopic findings may give the first clue to the presence of tuberous sclerosis.[5]

With reference to brain involvement, patients with a positive diagnosis of tuberous sclerosis without roentgen evidence of intracranial calcification are of great interest. Patients with lesser tendency to form intracranical calcification have less brain damage and mental retardation along with milder form of epilepsy and a greater chance of survival.[4]

Case Report

A 15-year-old girl presented with asymptomatic skin lesions over the face since childhood. She was born of non-consanguineous parents. At the age of 5 years, she developed asymptomatic hyperpigmented and skin coloured papules over the face which progressively increased in size and number. She subsequently developed asymptomatic plaques over the scalp and lumbar region. There were no systemic complaints. Family history was not relevant.

Dermatologic examination revealed multiple angiofibromas over the face with nearly symmetrical centrofacial distribution (Fig.1). Fibrous plaques were seen over the forehead and scalp. Shagreen patch was

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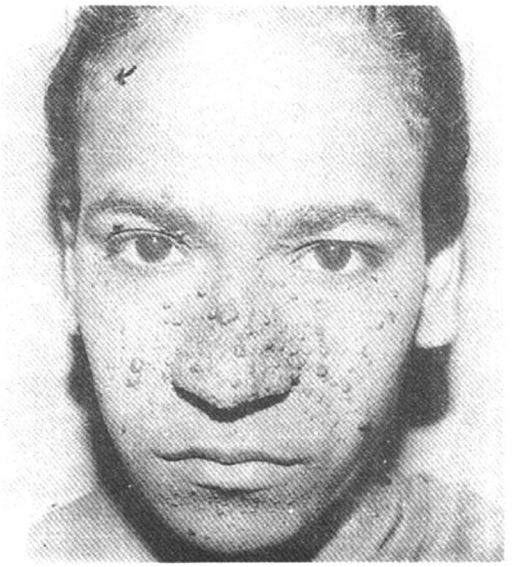

Fig. 1. Multiple angiofibromas over the face mainly in centrofacial distribution. Arrow indicates a fibrous plaque over the forehead near the hairy margin.

seen in the lumbosacral region. Cutaneous tags were seen around the neck. The scalp hair showed premature canities. Two hypomelanotic macules of size 3mm - 5mm were present over the extremities. There were no ash leaf macules. Fundoscopy revealed mulberry like retinal phakomas in the left eye. Psychiatric assessment revealed subnormal intelligence. Other systems were clinically normal. Routine haematological, biochemical tests, skiagrams of the chest, skull, hands and feet, ECG, echocardiogram, EEG and ultrasonogram of the abdomen and pelvis were normal. CT scan of the brain showed multiple periventricular calcifications (Fig.2). Skin biopsy from one of the lesions on face showed features compatible with angiofibroma. A definite diagnosis of tuberous sclerosis was made as she had one primary (facial angiofibromas) and four secondary features (retinal phakoma,

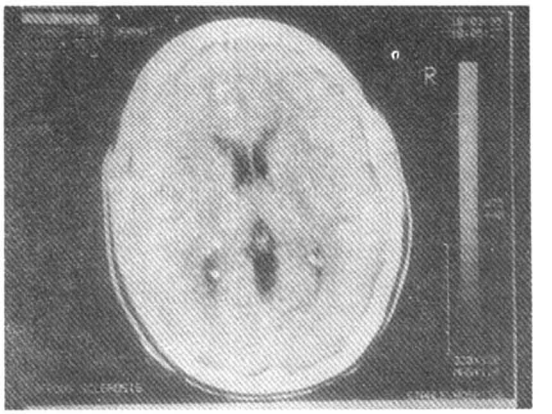

Fig. 2. CT scan of the brain showing multiple periventricular calcified spots.

cerebral tubers, shagreen patch and forehead plaque).

Electrocautery was done for the skin tags and small angiofibromas. Cryosurgery and plastic surgery were planned for larger lesions. The patient is being followed up regularly for development of systemic symptoms.

Discussion

This case is reported for the following rare interesting features. In spite of the presence of multiple intracranial calcifications and mild mental retardation, the patient did not give history of epilepsy since birth. Ash leaf macule though considered to be the most characteristic manifestation of tuberous sclerosis was absent in this case. Visual symptoms were not present despite the presence of retinal phakomas. To our knowledge, premature canities has not yet been reported in literature though poliosis has been recorded under pigmentary changes in tuberous sclerosis.

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