An unusual presentation of tuberous sclesosis complex

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Abstract

We report a child with tuberous sclerosis complex with an unusual cutaneous manifestation with underlying bony involvement. The child also had several typical cutaneous findings of the disease.

Introduction

Tuberous sclerosis complex(TSC) first described in 1880 by Bourneville represents a neuro cutaneous disorder characterized by harmatoma formation in many organs, particularly in the skin, eyes, and the brain. It can occur either as a autosomal dominantly inherited disease or as a result of a spontaneous mutation. Although previous studies had greatly underestimated its prevalence now TSC is considered as one of the more common single gene disorders.

The disease expressivity has a very wide spectrum of features, and therefore the classically described triad of angiofibromas, seizures and mental retardation is not always seen. In this report we present a young girl with TSC who had an unusually large plaque of segmental angiofibromas on the left side of the face and extending down to the neck, with underlying maxillary involvement of the left side.

Case

A two and a half year old girl born to non consanguineous parents was first referred to the dermatology clinic of the Lady ridgeway Hospital for children at the age of 10 months. A large reddish purple plaque had been present on the left side of the face involving the lower eye lid since birth. Apart from the facial lesion multiple, skin coloured nodules arranged in groups had been noted on the back of the trunk since early age, together with several hypopigmented lesions on the trunk and limbs.

The facial lesion had grown in proportion to the growth of the child but hat remained limited to the area over the maxilla and upper neck. There was no history of fits and normal motor mile stones were noted but a delay in speech development was noted later.

There was no family history of features suggestive of TSC.

A biopsy was taken from the facial plaque during the first presentation which was reported as a nerve sheath tumor. After this the child was lost for follow up for next one and half years. Examination in May 2007 revealed a large (10 × 7cm) brownish firm plaque over the left maxillary area extending down to the neck. The plaque was made of reddish brown papules suggestive of angiofibromas. Facial bones underlying the lesion were irregular on palpation, and has resulted in disfiguration of facial features. In addition to the plaque smaller number of angiofibromas were found on both nasolabial folds and surprisingly few lesions were found in the upper lip on the affected side as well. A few fibrous plaques were seen adjacent to the large plaque.

7 ash leaf macules were noted over the trunk and lower limbs including one giant lesion over the right thigh.

A shagreen patch was seen on the right lumbosacral region. No periungual or gingival fibromas were found.

A second deep biopsy was taken from the nodule at the angle of the mandible as well as from the lower eye lid. Histology of the former was reported as in keeping with angiofibroma:

Ophthalmic examination under anesthesia did not reveal any retinal phakomas or any major abnormalities.

CT Scan of the brain was performed and it demonstrated multiple lesions characteristic of TSC including

1. Calcified subependymal tubers
2. Cortical harmatomas
3. Early Giant cell astrocytoma in the region of foramen of monro

Bony changes noted on the CT scan included thickening of underlying maxilla associated with narrowing of the cavity of maxillary sinus.

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Discussion

Our patient fulfills criteria for the diagnosis of Tuberous scleroses complex. But the very large segmental type plaque of angiofibroma is a very rare presentation of this common entity. Furthermore the usual time of appearance of angiofibromas is between 2 – 5 yrs and the presence of angiofibromas at birth as in this case is very rare.

Also the commonly described skeletal abnormalities in TCS include bony cysts found on hands, feet and more rarely sclerotic lesions usually on long bones, vertebrae ect. Our patient was noted to have significant thickening and sclerosis of facial bones to the extent of compressing the left maxillary sinus.

Typical cutaneous features of Tuberous sclerosis complex include ash leaf spots, facial angiofibromas usually in a symmetric dirtibution over the cheeks, fibrous papules/plaques of face, shagreen patch, café au lait macules and periungual fibromas later in life. Unusual cutaneous manifestations include Molluscum fibrosum pendulum, poliosis, Unilateral facial involvement and occipital angiofibromas. Occurance of giant plaque type angiofibromas are described in literature. However the association of such a lesion with bony involvement has not been described.

References

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