Short reports

Forehead plaque: a presenting skin sign in tuberous sclerosis

A E FRYER, J P OSBORNE, AND W SCHUTT

Department of Paediatrics, Royal United Hospital, Bath, and Department of Neuropaediatrics, Bristol Royal Hospital for Sick Children, Bristol

SUMMARY The forehead plaque can be the earliest skin manifestation of tuberous sclerosis and its presence may lead to early diagnosis and appropriate genetic counselling.

Plaques, usually found on the forehead, are a feature of tuberous sclerosis. They are smooth patches of slightly raised skin with a reddish or yellowish discoloration and are now regarded by Gomez as pathognomonic of the condition (Gomez MR. Personal communication.) The earliest skin manifestations of tuberous sclerosis are usually hypomelanotic macules¹ and, rarely, depigmented hair patches.² Hypomelanotic macules are present in 90% of patients with tuberous sclerosis but may be late appearing.³ We present two patients in whom a forehead plaque appeared before any of these more recognised skin signs.

Case reports

Case 1. This boy was born in 1978. He had no family history of tuberous sclerosis. At 3 months of age he presented to his general practitioner with infantile spasms, though these were not initially recognised as such. By 7 months he was referred to a paediatric clinic where an electroencephalogram showed hypsarrhythmia. He was begun on treatment with steroids. A large raised lesion of reddish coloration was present on the right side of his forehead, which was initially diagnosed as a haemangioma. It had been first noticed by his parents at 4 months. No other skin signs were present on examination at this time. By 2 years hypomelanotic macules had appeared and he had a cranial computed tomogram taken, which showed an area of increased density (not calcified) in the head of the right caudate nucleus with mild ventricular dilatation. Later that year, a retinal phakoma and a cardiac rhabdomyoma were discovered. By 4 years, facial angiofibromas and shagreen patches had appeared (Figure).

Case 2. This girl was born in 1984. She had no family history of tuberous sclerosis. She presented with infantile spasms at 5 months and a computed tomogram taken at that time showed periventricular calcification consistent with a diagnosis of tuberous sclerosis. She had no skin manifestations, and a Wood's light examination yielded negative results. At 9 months two small papules appeared on her

Figure Case 1 at 8 years of age, showing forehead plaque and facial angiofibromas.

forehead, and over the next six months these grew and coalesced into a small reddish coloured plaque. This lesion has subsequently remained the same size. At 2 years two small hypomelanotic macules were visible under Wood's light, but no other skin signs have appeared.

Discussion

Seizures in the first year of life, especially infantile spasms, are an important presenting feature of tuberous sclerosis. Early diagnosis is important for genetic counselling and predicting prognosis. In infants with infantile spasms a careful search for the signs of tuberous sclerosis should be performed, in particular, skin examination (including Wood's light), retinal examination, and cranial computed tomography. Diagnostic skin signs (facial angiofibromas, ungual fibromas, and shagreen patches) are rarely present at this age, and a family history of tuberous sclerosis is only occasionally obtained. Much emphasis has thus been placed on finding depigmented patches of skin or hair. These lesions occur in 90% of cases eventually but are present in only 60% at presentation with tuberous sclerosis. Furthermore, the presence of a single white patch is difficult to interpret, as an examination of normal newborns' skin has shown that 0.8% have hypopigmented naevi 1–2 cm in diameter, which may subsequently disappear.⁴

Attention has only recently been drawn to the diagnostic importance of the forehead plaque. Histologically, these lesions are similar to facial angiofibromas, except that they have a less prominent vascular component.⁵ The two cases presented

here illustrate the importance of the forehead plaque as an early sign of tuberous sclerosis. Its recognition in case 1 would have led to the diagnosis being made at the time of presentation. In his case other signs appeared much later and even his computed tomogram in isolation was not diagnostic, having only a single non-calcified paraventricular lesion. The plaque was initially misdiagnosed as a strawberry naevus, which should appear shortly after birth. In case 2 the plaque remains the clearest sign, though the diagnosis was established early by computed tomography.

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Correspondence to Dr J P Osborne, Department of Paediatrics, Royal United Hospital, Combe Park, Bath BA1 3NG, England.

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Mercury as a health hazard

H A CURTIS, S D FERGUSON, R L KELL, AND A H SAMUEL

Departments of Child Health and Community Medicine, University of Wales, College of Medicine, Cardiff, Wales

SUMMARY Pink disease has virtually disappeared since teething powders were withdrawn.¹ We describe a case in a boy who was exposed to metallic mercury vapour. We discuss the potential health hazard of spilled elemental mercury in the house and the difficulties of removing it from the environment.

Case report

The patient was the only child of healthy unrelated parents. There were no perinatal problems and his early developmental milestones were normal. His general health was good until the family moved house when he was 18 months old. One month later he became irritable and anorexic. He developed a cough and began dribbling saliva. His hands and feet