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Tooth Pits: An Early Sign of Tuberous Sclerosis

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The pit-shaped enamel defects seem to be pathognomonic of tuberous sclerosis and its detection may be an important help in the early diagnosis of this disease, especially in oligosymptomatic cases and when the dermatologic signs are still absent or only slightly evident. (Received February 20, 1987.)

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Sometimes early diagnosis of tuberous sclerosis may be difficult, because the disease does not always appear in its fully developed form. The ash-leaf-shaped lance-ovate hypopigmented macules are particularly valuable early markers of this disorder, because they appear at birth or shortly thereafter, and, although they may enlarge as the infant grows, usually persist throughout life and do not otherwise change in size or shape. These macules are now recognized as the earliest cutaneous markers of tuberous sclerosis (1). Nevertheless, particularly in individuals with light pigmentation, these macules are easily overlooked. The characteristic "adenoma sebaceum" (in fact, angiofibromas), scattered over the cheeks and the bridge of the nose with a bilaterally symmetrical distribution, are pathognomonic cutaneous lesions but these tumours are rarely present at birth and do usually not appear until puberty. Only 13% of children with tuberous sclerosis develop the facial lesions of "adenoma sebaceum" during the first year of life (2).

Diagnosis during life often depends upon the appearance of the characteristic triad of seizures, mental deficiency and skin lesions (hypopigmented macules, angiofibromas, shagreen patches and periungual fibromas). These dermatologic signs can be late, and even absent. In such cases other diagnostic signs must be searched for. In this report we propose a dental sign, tooth pits, as a reliable marker of tuberous sclerosis.

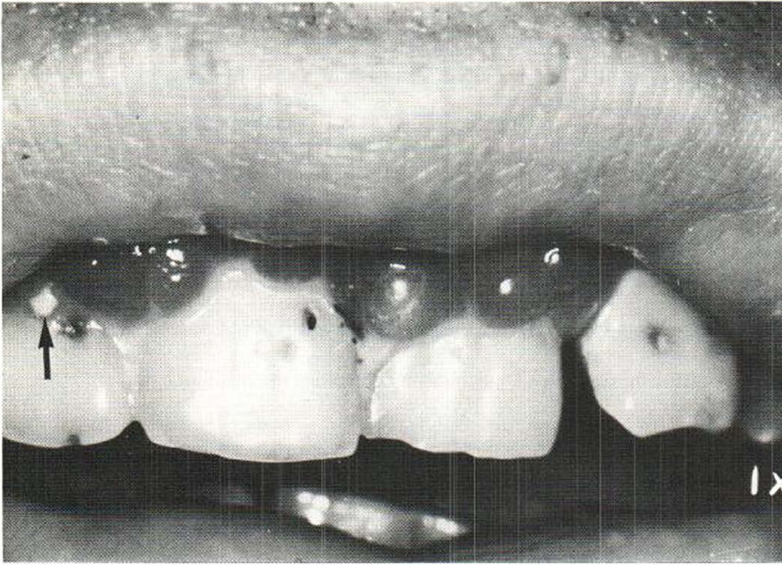


Fig. 1. Hyperplastic gingiva and pit-shaped enamel defects on the labial surface of the incisors and cuspids. The arrow indicates an enamel pearl.

CASE REPORT

An 18-year-old man was referred to the hospital for dermabrasion of facial angiofibromas. He previously had symptoms diagnosed as tuberous sclerosis. The parents of the patient described him as having been normal at birth and until the age of 2 years. At that time grand mal epilepsy seizures began. From this time medications included phenobarbital and diphenylhydantoin, but in spite of these drugs, he had frequent seizures, and there was an evident mental retardation.

With regard to the family history, the patient has two sisters and one brother, none of whom has neurological abnormalities or other dermatologic markers of tuberous sclerosis. The parents of the patient were apparently healthy. No accurate additional family history could be obtained.

Dermatological examination revealed typical "adenoma sebaceum" on the face, shagreen patches on the back and on the left shoulder and numerous periungual fibromes in hands and feet. No evident hypopigmented macules could be observed with natural light, but when illuminating the skin with a Wood's light in a dark room it was possible to discover two irregular-shaped white macules on the abdominal skin. The computerized axial tomography (CAT) of the brain showed periventricular calcifications and an abdominal CAT revealed tumoral and cystic pictures in both kidneys, which were interpreted as hamartomas.

The oral examination showed a gingival hyperplasia, predominantly in the anterior segments of the maxillary and mandibular arches and a pedunculated gingival growth from the second upper left molar level. This tumour was removed and the histopathologic study was consistent with fibrous hyperplasia of the gingiva. A meticulous examination of the tooth surfaces of the patient revealed several pit-shaped enamel defects on the labial surface of the incisors (Fig. 1). Some of these enamel defects were filled up with an enamel pearl of a whitish and brilliant aspect. The X-ray films of these teeth showed small depressions on the enamel surface. The total number of these pits in our patient was twelve. They were more abundant in incisors and cuspids, but could also be seen in premolars and molars.

COMMENTS

This patient virtually had all the skin manifestations of tuberous sclerosis, and therefore the diagnosis was easy. But this does not always occur, and the diagnosis of the disease can be complicated in mildly affected individuals with "formes frustes" or incomplete symptoms of the disorder.

There is little reference to the oral manifestations of tuberous sclerosis in the literature. Both enamel defects and hyperplastic gingiva have been reported in dental literature (3, 4) but they are scarcely known by dermatologists. In dermatologic literature, only one

description of tooth abnormalities has been given (5). Although the presence of fibroma-like lesions on the attached gingiva has been reported as specific stigmata of tuberous sclerosis (4), the diphenylhydantoin-induced gingival hyperplasia is well known (6). Because this therapy was necessary in the medical management of this patient, we have considered the gingival lesions as drug-induced. However, a review of the literature reveals no mention of pit-shaped enamel defects in patients with other neurocutaneous syndromes. So these pits seem to be pathognomonic of the tuberous sclerosis. The scanning-electron-microscope investigations have shown that these enamel defects extended from the tooth surface to the amelodentinal junction and have been interpreted as the result of defective amelogenesis (3). Although these pits are smaller and less frequent in deciduous teeth than in permanent teeth (7), this feature strengthens its usefulness as an early marker of tuberous sclerosis and may help to establish the diagnosis in oligosymptomatic cases.

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