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ADENOMA SEBACEUM IN ENCEPHALOFACIAL ANGIOMATOSIS (STURGE-WEBER SYNDROME)

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Abstract. A case of encephalofacial angiomatosis (Sturge-Weber Syndrome) with unilateral adenoma sebaceum is presented.

The diagnosis of encephalofacial angiomatosis (Sturge-Weber syndrome) usually presents little difficulty. The unilateral angiomatous lesion involving the cutaneous distribution of the ophthalmic, maxillary or facial portion of the trigeminal nerve, associated with intracranial calcifications, epilepsy and glaucoma are usually so characteristic as to present little challenge to the clinician (3). On occasion, however, the diagnosis may be less evident and require a series of diagnostic studies to ascertain its nature. We have recently studied such a patient with encephalofacial angiomatosis and feel it would be of interest to report its unusual clinical features and diagnostic challenge.

CASE REPORT

The patient is a 19-year-old Caucasian boy who developed seizures at age 6 days following a normal full-term pregnancy and uncomplicated forceps delivery. He has had, since then, a recurrent convulsive disorder, rigid quadriplegia and profound mental retardation. A study of the family pedigree showed no other abnormalities in the patients' and two preceding generations. The patient's mother, father, 2 brothers and a sister were all alive and in good health. The mother had had three pregnancies which terminated spontaneously for unknown reasons.

Case history

The patient had measles at age 3 years and since then, several intervening episodes of facial impetigo. There had been congenital "shortness" of the right achilles tendon which was repaired. On physical examination, the patient exhibited rigid quadriplegia and had great difficulty standing or sitting upright. Examination of the patient was otherwise unremarkable except for the skin (Figs. 1 and 2). The face had an eruption of fleshy papules limited to the right cheek and nose areas with sparing of the upper lip typical of adenoma sebaceum. There was a dusky hyperpigmentation of the right side of the forehead, stopping at the midline, and a heavier growth of hair on the right cheek and forehead than on the left. In addition, the entire face, chest, and back were affected by moderately severe acne vulgaris. It is particularly noteworthy that there was no evidence of raised intraocular pressure on the right and no evidence of angiomatous involvement of the right eye.

Laboratory studies: Examination of the formed elements of the blood, serum protein electrophoresis, BUN, serum glucose, urinalysis and chromosomes were all normal. Radiographic examination of the chest was normal. Radiographic examination of the skull showed diffuse intracortical calcifications on the right, there was no deep cerebral calcification. The electroencephalogram showed focal spike wave activity over the right temporal, parietal and occipital areas.

Histopathological examination of sections from punch biopsies taken from several areas of the papular lesions showed a marked increase of dilated capillary-like structures in the dermis. These structures were filled with blood and showed little evidence of new vessel formation. The vessels were lined by a single layer of endothelial cells. There was, in addition, around the vessels an infiltrate composed of lymphocytes, histiocytes, plasma cells and fibroblasts (Fig. 3). The covering epidermis showed some acanthosis and spongiosis. The diagnosis was: angioma consistent with a clinical diagnosis of encephalofacial angiomatosis.

DISCUSSION

Adenoma sebaceum of Pringle (6) is a characteristic facial eruption found as a salient cutaneous feature of tuberous sclerosis (2). The term Adenoma Sebaceum has fallen into disfavor (4) since Nickel & Reed (5) showed that the

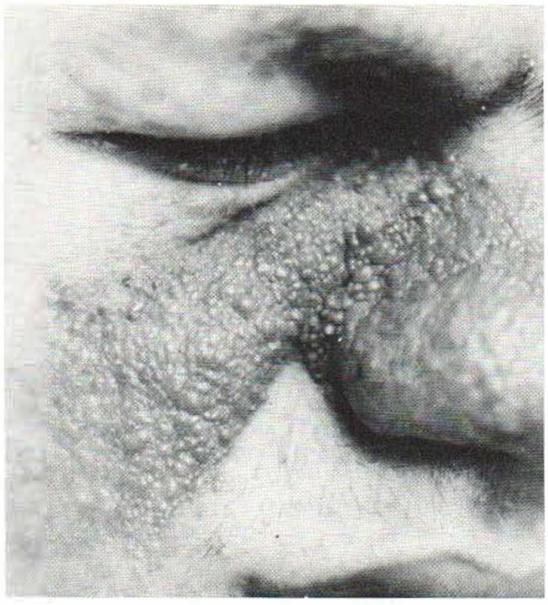

Fig. 1. Dusky, fleshy, clustered papules resembling adenoma sebaceum involve the right paranasal area and nose. The upper lip is spared.

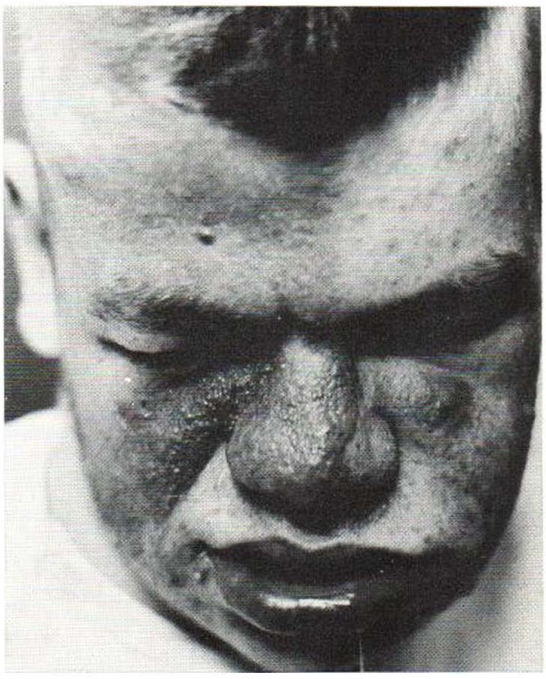

Fig. 2. The papillomatous lesions on the right cheek are accompanied by a hyperpigmented area occupying the distribution area of the ophthalmic and maxillary portion of the trigeminal nerve.

cutaneous lesions in tuberous sclerosis are angiofibromas due to capillary fibrosis and dilatation and not adenomas of the sebaceous glands. At first sight, our patient's skin lesions could best be described as unilateral adenoma sebaceum. Because there was no evidence of hereditary transmission of our patient's disease and because of the presence of epilepsy and cortical calcifications as well as histopathological evidence in the skin of an angioma, we felt the most tenable diagnosis was encephalofacial angiomatosis. A diagnosis of tuberous sclerosis could not be supported in the absence of deep intracerebral lesions, shagreen

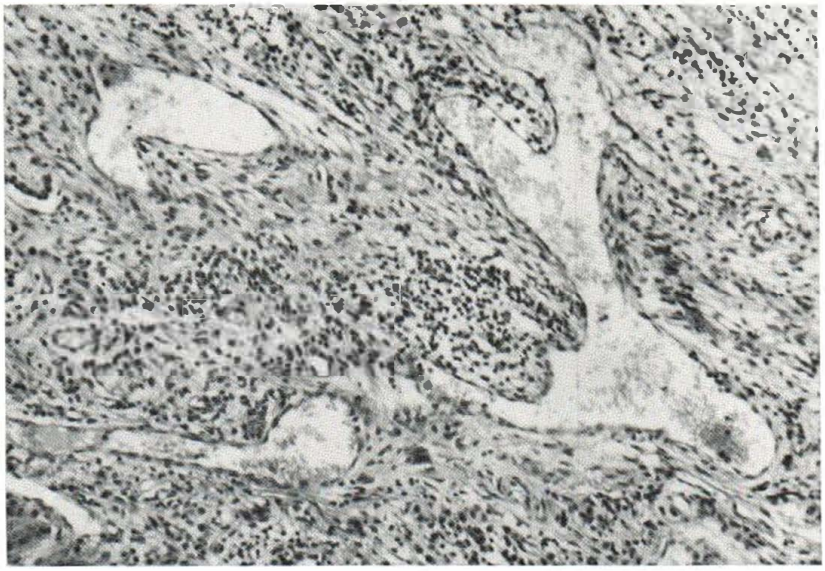

Fig. 3. Photomicrograph of cross-section from one of the papular lesions. Large vascular spaces containing red blood cells are seen. There is no neovascular formation, and the vessels are lined by a single layer of cells. The intervascular spaces contain acute and chronic inflammatory cells. × 27.

patch, periungeal fibromas, white ash leaf macules and a negative family history (1). Ocular involvement with buphthalmos or glaucoma is common but may be absent in encephalofacial angiomatosis. The finding of lesions of adenoma sebaceum as part of Sturge-Weber's syndrome must be extremely unusual, though not entirely surprising, since adenoma sebaceum are angiofibromas.

Solitary angiofibromas have been described as occasionally occurring on the face of normal adults (7) and it is the purpose of this report to point out that similar lesions may also occur in large numbers as a presenting feature of the Sturge-Weber syndrome. The term adenoma sebaceum is admittedly a misleading one but there may be some merit in retaining it to represent a clinical picture not necessarily pathognomonic of tuberous sclerosis.

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