A Case of Segmental Macular Neurofibromatosis

Sir,

The term segmental neurofibromatosis indicates a peculiar form of the disease, characterized by the unilateral localization of café au lait spots along one or more dermatomes, in patients with no familiarity for neurofibromatosis (NF). It corresponds to the fifth type in Riccardi's classification (1).

A 10-year-old girl presented with several café au lait macules on her left hemibody, already visible at birth. The skin lesions, round or oval-shaped, were 1 to 4 cm in diameter and spread from the left axilla to the left side of the abdomen and left lumbar region down to the left lower limb (Fig. 1); no lesion crossed the midline. The café au lait spots had enlarged and increased in number slightly over the years. There was no family antecedent of NF; only a sister showed a single café au lait macule on the anterior aspect of the trunk. No disturbances of the central nervous system (CNS) were found in the family history. At the age of 10, the the girl had begun to complain of headache. An EEG revealed an aspecific dysrhythmia. An encephalic computerized tomography and a psychological profile were normal. A biopsy of a skin lesion was performed: the histological examination revealed a considerable amount of melanic pigment in the basal layer of the epidermis and a normal number of melanocytes. The electron microscopy observation revealed numerous subepidermal nerves just below the dermoepidermal junction and intraepidermal macromelanosomes.

About twenty cases of segmental NF are described in the literature (2). Only some of them fit the features defined by Riccardi. Some authors have reported bilateral clinical pictures (3); others have described patients with visceral involvement (4, 5) and cases with a familiar history of NF type I (6). Riccardi himself (1) recorded a case affected by NF lesions, initially with segmental distribution, which evolved into a diffuse pattern. Only the case of an 11-year-old girl (4) was characterized solely by café au lait macules with thoracolumbar localization. The peculiarity of our case lies not only in the exclusive macular skin manifestations, but also in the presence of CNS disturbances, the lack of which, on the contrary, seems to be a typical feature of segmental NF (7).

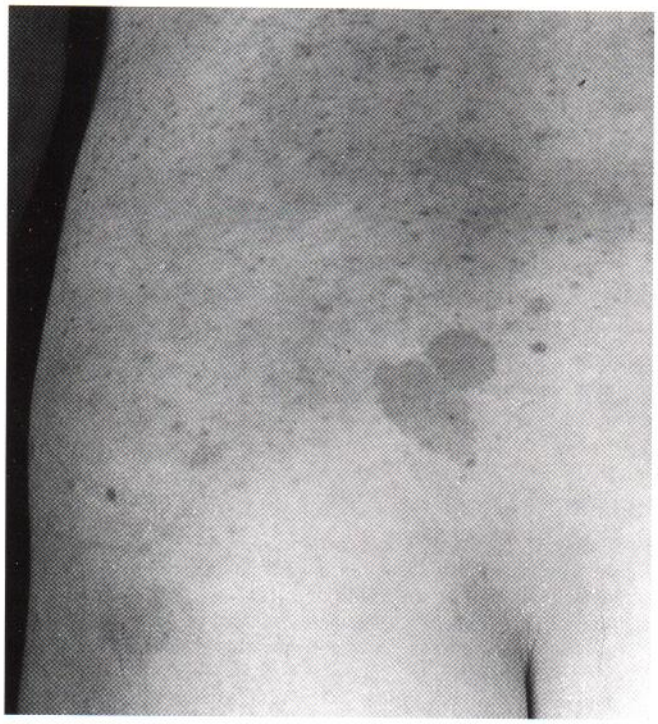

Fig. 1. Café au lait macules on the patient's left hemibody.

Riccardi (8) emphasizes that it is not necessary to perform a biopsy to confirm the diagnosis of NF, but to diagnose segmental NF is not always easy. The differential diagnosis may be a problem with unilateral or segmental lentiginosis. In this rare picture the histological observation may be decisive; in fact, elongated rete ridges and an increased number of solitary melanocytes are present. Also the naevus spilus may be sometimes differentiated only by the observation of microscopic features similar to those of unilateral lentiginosis in correspondence to the light-brown macules and the presence of melanocyte nests at the dermoepidermal junction in the dark brown spots. The electron microscopy examination has proved significative: the presence of many macromelanosomes in the basal cells of epidermis may itself orient the diagnosis towards a segmental NF rather than to a McCune-Albright syndrome, where macromelanosomes are rare. The presence of a considerable number of nerves next to the dermoepidermal junction may be regarded as a characteristic of the café au lait macules in NF, as other authors have hypothesized on finding subepidermal and intraepidermal nerves in the café au lait macules with white halo in children affected by NF (9).

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