Focal Dermal Hypoplasia: A Case with Minor Clinical Manifestations

Sir,

Since Goltz (1) first coined the term "focal dermal hypoplasia" in the 1960s, over 200 cases have been described worldwide. The syndrome may involve the skin, the skeleton, the ocular system, or, less frequently, the renal, gastroesophageal, auditory and nervous systems. Here we report the case of a patient who presented with less definite clinical features than commonly described.

CASE REPORT

Our patient, a 5-year-old girl in good general condition, was a first child, and her mother had not had any miscarriages. Family history was negative for skin diseases. In the third month of life she had shown mild papulocrusty lesions on the lower limbs that worsened during the summer and became itchy. The lesions were regarded as atopic dermatitis. When first observed by us, the lesions were affecting the flexor surface of the right lower limb and partially of the left one, up to the root of both limbs. They involved a band of skin appearing as atrophic-hypochromic, partly reticulated areas interspersed with reddish papules and small squamocrusts (Fig. 1). The nail plate of the right hallux showed a longitudinal dystrophic fissure in the middle. The lower lateral incisor teeth were cone-shaped and hypoplastic. Clinical chemistry tests (in particular total IgE, ANA, antiENA antibodies, total urinary porphyrins and blood protoporphyrins) were normal. Histologic examination of a papule from an atrophic patch revealed mild irregular acanthosis with focal parakeratosis in the central portion of the lesion. The dermis was thinned and edematous. The fat was located abnormally high (Fig. 2).

X-rays of the upper limbs showed small calcareous deposits and radiopaque symmetrical, horizontal striae in correspondence with the distal metaphyses of humerus. A phototest revealed a higher than normal and long-lasting UV-B skin reactivity. Eye and neurological examinations were negative. The chromosome map showed a 46 XX karyotype with no structural changes of the chromosomes.

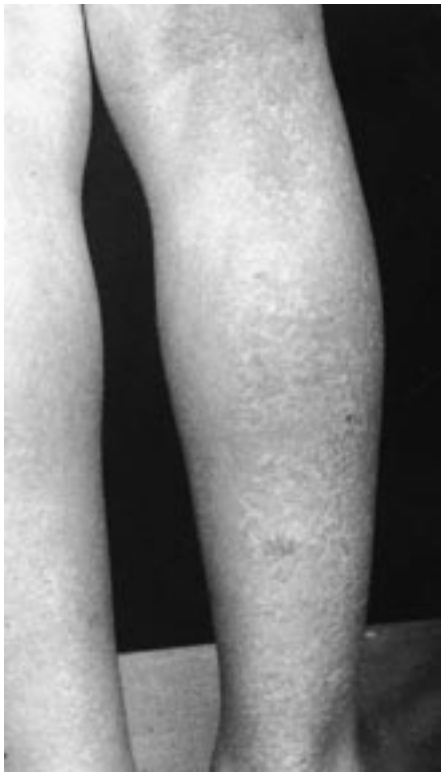

Fig. 1. Band of poikilodermatous skin with papules and squamocrusts lesions.

DISCUSSION

Our patient failed to show the most typical features of the Goltz syndrome, i.e. extroversion of the adipose tissue and eye involvement. Histology and bone X-rays were therefore conclusive for a correct diagnosis. Histological examination revealed a typical picture, but the radiological picture of our patient lacked the classic longitudinal distribution. Normally, the striated osteopathy appears as a net of condensed striae parallel to the longitudinal axis of the bone. The striae usually originate at metaphysis level in correspondence with the osteogenesis areas, where they look thicker and then gradually thin down as they go up along the diaphysis. They are usually bilateral and symmetrical, mainly involving the long bones and the sacral bone, while the vertebrae and the iliac bones are spared (2). Cases showing a less evident

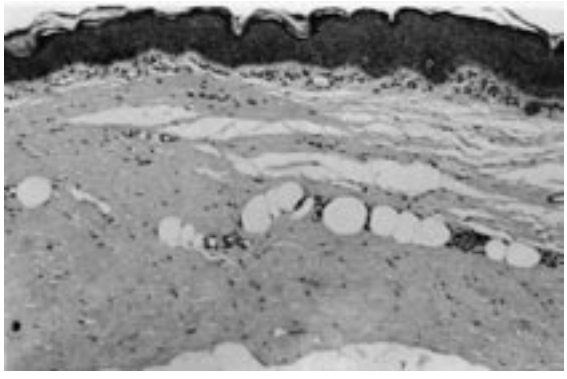

Fig. 2. Dermis thinned an edematous; fat located abnormally high (hematoxylin-eosin stain, 25 ×).

striated osteopathy and often malformation are reported in the literature (3).

Focal dermal hypoplasia can present with negligible clinical features, as described previously (4, 5). In one of the two patients reported by Pujol the skin features were as in our patient, but in the other the diagnosis was made only after the women had undergone several miscarriages and had given birth to a baby girl with quite evident signs of Goltz syndrome.

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