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QUIZ SECTION

Cutaneous Atrophic Guttate Lesions in a Linear and Reticulate Pattern: A Quiz

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A 3-year-old girl presented since birth with reddish atrophic hypopigmented guttate lesions with sparse telangiectasias in a linear and reticulate pattern on the trunk and extremities (Fig. 1), some papillomatous lesions on the lower lip, ocular coloboma, syndactyly toes, plagiocephaly (a malformation of the head), hypertelorism, low-set protruding ears, and an arachnoidal cyst. No family history of syndromic pathologies and/or mental retardation was reported. A punch biopsy from a representative cutaneous lesion showed a normal epidermis over a hypoplastic dermis (Fig. 2a). Collagen bundles were scarce, thin and loosely arranged. Increased numbers of blood vessels were seen subepidermally. The elastic fibres were markedly reduced as elastic tissue stains evidentiated, and islands of mature adipose tissue were found scattered within the thin papillary dermis, often in close proximity to the epidermis (Fig. 2b). Cutaneous appendages were absent.

What is your diagnosis? See next page for answer.

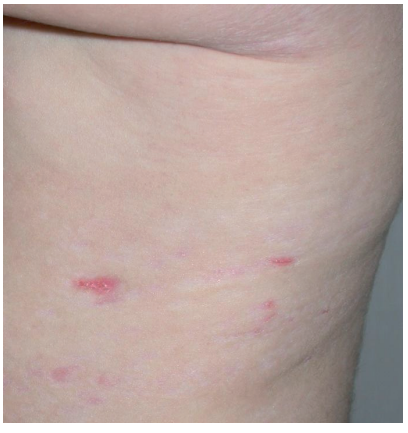

Fig. 1. (a) The skin on the lateral side of patient's trunk shows reddish atrophic guttate lesions in a linear pattern, sometimes depressed and hypopigmented.

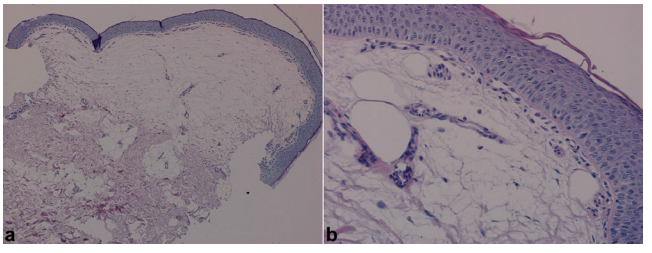

Fig. 2. (a) Low magnification shows loosely arranged collagen fibres in the upper dermis, with scattered islands of mature adipose tissue within the papillary and reticular dermis. The cutaneous appendages are absent and an increased number of small blood vessels are distributed mainly in subepidermal location. (b) High magnification highlights the superficial, abnormal location of adipose tissue enclosed in the looseness of the dermis whose collagen fibers gather to form a very thin layer beneath the basal membrane (Hematoxylin and Eosin: (a) × 4; (b) × 10).

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ANSWERS TO QUIZ

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Diagnosis: Focal dermal hypoplasia (Goltz syndrome)

Focal dermal hypoplasia (FDH) is a rare genodermatosis characterized by lesions involving both ectoderm and mesoderm. It is also known as Goltz syndrome, due to its first description by Goltz et al. in 1962 (1, 2). It is thought to be inherited as an X-linked dominant trait because of its almost exclusive occurrence in females, usually being lethal in males; however, sporadic variants have been reported, as well as a small numbers of affected males probably due to a somatic mosaicism or a sporadic new mutation (3).

The pathogenetic mechanism of this condition has not been clearly determined, although it has been linked to mutations in the gene *PORCN*, a regulator of Wnt signalling, thus implicating a role for Wnt signalling in the development of this disorder (4).

Clinically, FDH is characterized by cutaneous, bone, ocular, and oral lesions (1–3, 5–7).

Cutaneous manifestations include diffuse areas of dermal thinning, fat herniations, and linear streaks of hyper- and hypo-pigmentation of the skin, following Blaschko's lines (5, 7). Telangiectasias, papillomas (7), not always related to human papillomavirus, and sparse hair and nails dystrophy, are seen. Skeletal lesions include syndactyly, polydactyly, short stature, and osteopathia striata (5, 8). Ocular alterations include microphthalmia, coloboma, strabismus, and photophobia. A variety of orofacial and dental manifestations can be observed.

Cutaneous lesions are regarded as an essential component of FDH, although there are a few reports of the syndrome with no skin lesions (9).

The most specific diagnostic technique is the histopathological examination of a skin biopsy. The diagnostic clues are the marked reduction in thickness of the dermis and the extension up to the epidermis of the adipose tissue (3, 10), as shown in our patient's punch biopsy. Immunohistochemical staining for S-100 protein outlined the superficial, abnormal location of adipose tissue in our case, as described previously (11).

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