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Focal Dermal Hypoplasia Syndrome in a Male

Sir,

Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genodermatosis affecting tissue derived from embryonic mesoderm and ectoderm. Goltz et al. (1) first reported 3 cases of FDH in 1962 and reviewed 20 probable cases from the literature. The abnormalities seen in this multisystem condition include linear and reticular pigmentation, telangiectasias and papillomas, as well as skeletal, dental and ocular defects (1, 2). More than 200 cases of FDH have been reported or have been otherwise known (3). Most cases are seen in females and X-linked dominant inheritance fatal to male infants has been proposed as the likely mode of inheritance (4). Rare cases reflecting half chromatin mutation have been reported in male subjects (4–6). An additional case of FDH in a male with cystic bone defects is described herein.

CASE REPORT

An 18-year-old man was first seen in our department for evaluation of multiple congenital anomalies. Since birth he had disseminated areas of slightly erythematous and telangiectatic macules with atrophic areas in linear pattern on his extremities (Fig. 1). The lesions were asymptomatic but slowly progressed to involve his 4 extremities and trunk. Numerous papillomas were noted in the perianal area, on the penis, inguinal region, right nasal fold and upper lip (Fig. 2).

Skeletal abnormalities included absence of the third and fourth digits of the right hand. The left foot had a lobster claw deformity with syndactyly of the second and third toes. Microcephaly, triangular face, asymmetric limbs and dorsolumbar scoliosis were also present. All his nails were thinned and many of them showed linear streaking. His ears, although fully formed, were asymmetric. The rest of the physical examination was normal.

Results of the ophthalmologic examination revealed microphthalmia, strabismus, nystagmus and coloboma of the left choroid. Odontologic examination disclosed irregularly arranged, deformed teeth with enamel hypoplasia. The patient's intellectual and motor development was markedly retarded.

Radiologic survey revealed cystic defects involving the right tibia. No evidence of osteopathia striata was noted in the patient's long bones. There was no family history of any cutaneous or other associated findings recorded in FDH. Chromosomal analysis showed a 46, XY karyotype.

Two skin biopsies were performed. The first specimen from one of the papillomas revealed epidermal hyperplasia with pronounced papillomatosis, parakeratosis and spongiform pustules in the epidermal invagination and moderate inflammatory infiltrate in the papillary dermis. A biopsy specimen from the atrophic lesions showed irregular mild acanthosis, oedematous papillary dermis with scanty lymphocytic infiltrate and thinning of the dermis.

Cutaneous and extracutaneous features permitted us to establish the diagnosis of FDH.



Fig. 1. Multiple linear, atrophic, erythematous macules of the legs.

DISCUSSION

Goltz et al. (1) and Gorlin et al. (2) coined the term FDH for this rare genodermatosis on the basis of the histologically apparent areas of connective tissue hypoplasia. They summarized cutaneous, skeletal, ocular, dental and soft tissue defects associated with this syndrome. The clinical features of FDH have marked heterogeneity, with widely varying presentations (7). Stalder et al. (6) suggested that clinical spectrum of the disease is similar in both sexes.

Cutaneous findings are the most common manifestations of FDH (6). The present case showed the most prominent dermatologic manifestations including atrophic skin lesions, linear and reticular areas of telangiectasias, hypopigmentation, hyperpigmentation and multiple papillomas. Linear



Fig. 2. Wart-like lesions on the penis.

arrangement of the skin lesions following the course of Blaschko lines in our patient was found in 51% of the previously reported cases (5). Herniations or hamartomas of fat were not present at the time of examination.

The hair of patients with FDH is often sparse and brittle and 30% of them have alopecia (5). The nails may be absent or may show atrophy, dystrophy, grooving and spooning. Teeth may be defective in size, shape or formation. Enamel hypoplasia, irregular spacing and deformed teeth, which were observed in the present case, are often reported (7). The most common ophthalmologic findings such as coloboma, microphthalmia, strabismus and nystagmus (8) were also present in our case.

Nearly all cases of FDH demonstrate skeletal defects in addition to the cutaneous and ocular findings. Musculoskeletal defects most commonly include syndactily (60%), hypoplasia and absence of digits (33%), asymmetry of the body (31%), scoliosis (17%), and anomalies in hand or foot bones (13%) (7). A lobster-claw deformity is a unique feature of FDH.

Osteopathia striata, osteoporosis, and findings consistent with the above-mentioned skeletal changes may be revealed by radiologic evaluation. Osteopathia striata, the longitudinal striation of the long bones, crossing the epiphyses, is not present in all cases. This finding is not pathognomonic but it is found in approximately 20% of gene carriers (9).

There are several reports of cystic lesions occurring in the metatarsals, fibula, tibia, maxilla, ilium, and ischium (10–14). Selzer et al. (10) found giant cell tumour-like histologic features of one of these cystic lesions. Joannides et al. (11) described another giant cell tumour of the bone in FDH. Lynch et al. (12) reported on a patient with expanding lesions in the ilium and Cox & Paterson (13) on another patient with an aneurysmal bone cyst in the pelvis. There is also a report of an osteochondroma of the humerus (14).

In differential diagnosis of FDH, aplasia cutis congenita, Rothmund-Thomson syndrome and anetoderma should be considered. The typical cutaneous manifestations with musculoskeletal, ocular, dental, ear defects and mental deficiency confirmed the diagnosis of FDH in our case.

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