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Short Report

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Focal Dermal Hypoplasia (Goltz Syndrome) in a Male Infant

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Focal dermal hypoplasia (FDH) is a rare mesoectodermal hypoplasia which was described by Goltz[1] in 1962. This syndrome is predominantly seen in females and it is possibly an X-linked dominant disorder with lethality in males (2). Although FDH is generally considered incompatible with survival of the male fetus, 12% of the reported cases were living males (3-11).

It is characterized by linear areas of thinning and protrusions of fat through surrounding skin. Atrophic scars, aplasia cutis, telengiectasias, skeletal, ocular, dental, oral and mental defects may be present (2).

The rarity of this syndrome in males prompted us to report an additional male case with Goltz syndrome who is still alive and neurodevelopmentally healthy.

A two-day-old male infant was referred for evaluation of multiple anomalies. He was born at term to a primagravid 26-year-old mother with normal growth parameters. He reportedly had a five-minute Apgar score of 9. There was no parental consanguinity and there were no miscarriages or birth defects reported in the family.

Physical examination at our nursery revealed facial asymmetry, an atrophic and scaly skin lesion beneath the lower lid of the right eye and striations over the chin. There were circumscribed, depressed, atrophic, telangiectatic skin areas, grouped in a linear pattern on the limbs and trunk, prominent on the extensor aspect of the leg, on the abdomen and on the left side of the back (Fig. 1). The scalp hair had a normal appearance and no papillomatous lesions were observed. The pinnae appeared misshapen and pinpoint depressions were noted. There was also depression on the root of the nose. The gingivae had an irregular hypertrophic appearance. A soft mass with indistinct borders and normal overlying skin was palpated on the left shoulder. There was a sacral dimple with no cutaneous opening. Both fingernails and toenails were dystrophic with longitudinal ridges.

Other findings included underdevelopment of the left side of the face and fusion of the second, third and fourth toes of the right foot. (Fig. 2). The eyes were normal, except for blockage of the lacrimal ducts.

Except for hypocalcemia, laboratory studies revealed normal hematology and blood chemistry.

A skeletal survey showed neither osteopathia striata nor osteoporosis. There was no fusion or sacralization of the vertebrae, pelvic abnormalities or scoliosis. Abdominal ultrasonography showed ectopic localization of the left kidney in the left inguinal region, which was also confirmed by excretory urography. A CT scan showed cavum septum pellucidum and asymmetry in the Sylvian fissures.

The biopsy specimen from atrophic lesions revealed a normal epidermis and markedly narrow hypoplastic dermis with hypoplastic appendageal structures and blood vessels (Fig. 3).

A chromosome analysis of the patient revealed a normal karyotype (46 XY).

Auditory brain stem responses were within the normal ranges.

Goltz syndrome, or FDH, is primarily a disorder of the skin, eyes and digits. Although the literature includes a male patient from as far back as 1928, until recently the disorder was considered incompatible with survival of the male fetus (2). However, there have since been reports of

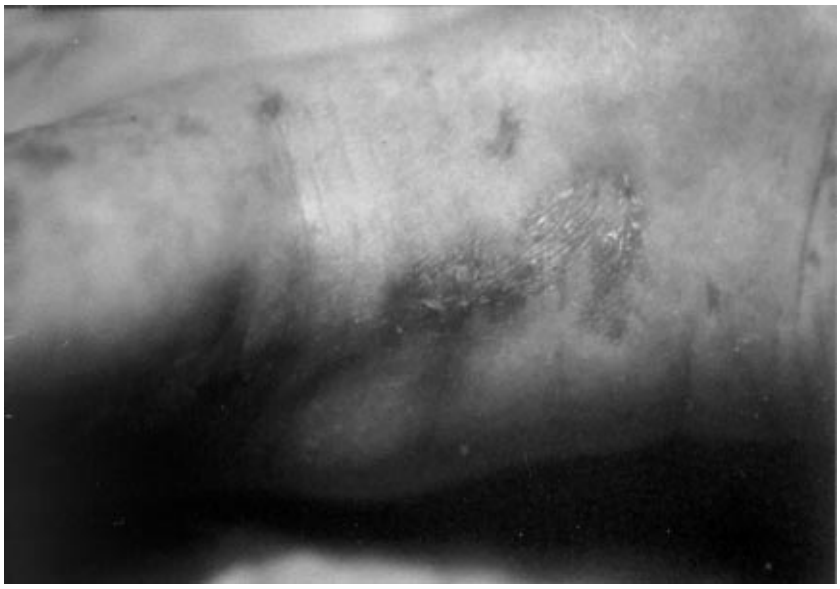

Figure 1. Atrophic telangiectatic skin of the limb.

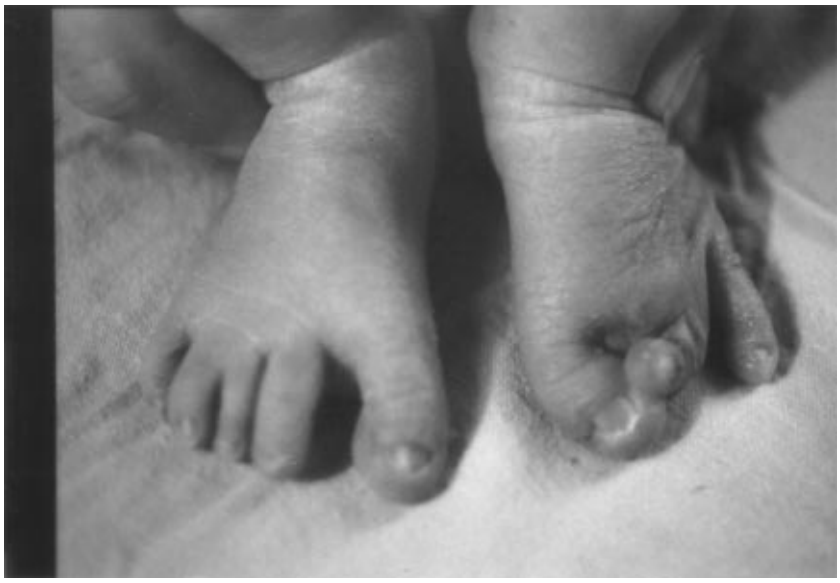

Figure 2. Abrormally placed toes.

surviving male patients (3-11). The inheritance pattern has not been determined but it has been postulated that an X-linked dominant gene usually causing lethality in hemizygous males or an autosomal-dominant sex-limited mode of inheritance may account for the sex distribution (3). Cases of father-daughter transmission, evidence of an autosomal locus, 9q32-qter, and the unusually high proportion (10%) of males with this disorder, however, argue against X-linked dominance with lethality in males (7, 12). Affected males may have an early half-chromatid mutation or autosomal dominant inheritance affecting the germ line (13). Somatic cell mosaicism has been suggested to explain the skin and bone lesions in males (14). Others suspect X-autosomal translocation or environmental teratogens (15). The patient in the present study is now four years old and was considered neurodevelopmentally healthy in follow-up examinations.

The most common cutaneous abnormalities of FDH are dermal hypoplasia (61.6%) and fat herniations (46.4%). Papillomas (46.4%) of the lips, gums, perianal and perioral areas may develop within the first few months of life (3). The patient had only atrophic, depressed skin lesions with telangiectasias in a linear

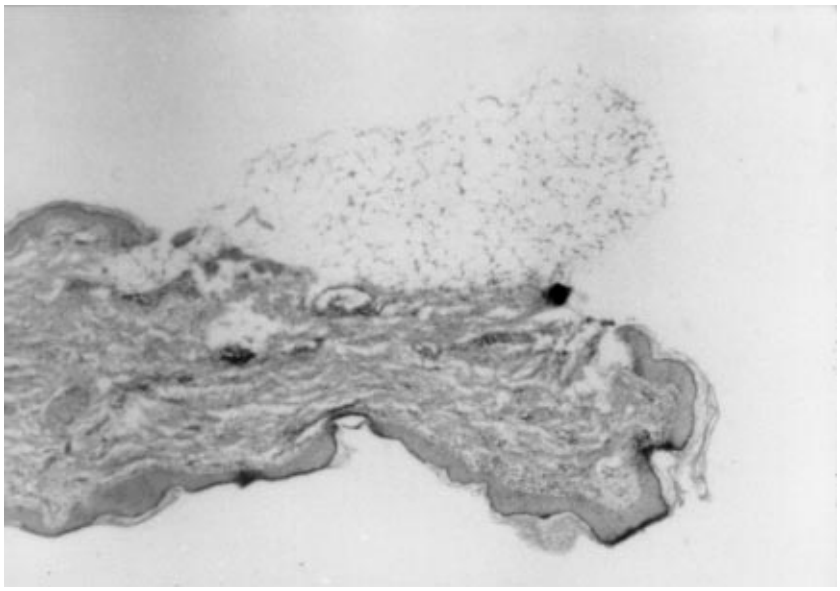

Figure 3. Histologic features of atrophic skin lesions displaying hypoplastic dermis and skin appendages (HEx10).

pattern. Nail dystrophy, which was observed in this patient, was present in 41% of other reported cases (3). Scalp-hair abnormalities, another common feature, were absent. Other reported cutaneous lesions include total absence of skin at various sites, with blistering and crusting, hyperkeratosis of the palms and soles, radial folds around the mouth, hyperhidrosis, hypohidrosis, urticaria, photosensitivity and local poliosis (4), of which only crusting was observed in this patient. A variety of soft tissue defects are associated with FDH (1, 3). The patient had a soft tissue mass located on the left shoulder and asymmetry of the face was also observed. The ears may be abnormal in shape, as in this case. Skeletal defects are the second most common abnormalities with a frequency of 80%. The most common skeletal abnormalities are hypoplasia or absence of fingers or toes, syndactyly, polydactyly, asymmetric development of the trunk and extremities, vertebral abnormalities, micrognathia and deformities of the long bones (3, 4, 14). The patient displayed syndactyly of the toes. Osteopathia striata, which is a common finding in Goltz syndrome (14), was absent in this case.

Oral and dental defects were found in more han half of the previously reported cases. Enamel may be defective, leading to early caries. Cleft lip and/or palate, cleft tongue or high arched palate may be present. Hemihypoplasia of the tongue, intraoral papillomas and gingival hypertrophy may also be observed (16). Only the last of one these was found in this case.

Ocular abnormalities are: coloboma or congenital defects of the iris, retina, choroid and optic nerve, strabismus, nystagmus, small eyes, cloudy cornea or even anotphthalmia (17). The tear ducts may be blocked (4), as was observed in this patient.

Ectopic kidney, the incidence of which is reported as 9% in the literature (3), was determined in this case by abdominal ultrasonography and confirmed by excretory urography. A review of the literature, revealed no previous cases of abronmal CT signs as observed in this patient.

As indicated in most of the cases in the literature (2, 3), the patient did not show any chromosomal abnormalitiy.

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