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Focal Dermal Hypoplasia (Goltz Syndrome): A Case Report and Review of Literature

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Summary

An 18 years old girl presented suffering from asymmetrical linear streaks of hypo & hyper-pigmented atrophic lesions with telangiectasia over her trunk and extremities, she had multiple bone deformities especially of the limbs with facial asymmetry which were confirmed by multiple X-rays. Abdominal examination was normal and eye examination revealed myopia. Histopathological examination revealed total dermal absence.

Introduction

Focal dermal hypoplasia (FDH) is a rare meso-ectodermal disorder characterized by focal total dermal absence with the epidermis directly overlying the subcutaneous tissue. It is inherited by an X-linked dominant gene, which is lethal in homozygous males. Occasional occurrence in males is due to fresh mutations. It was first described by Goltz in 1962 [1].

Case Presentation

An 18 years old female presented to our outpatient clinic at Al-Haud Al-Marsoud Hospital suffering from asymmetrical linear streaks of hypo & hyper-pigmented atrophic lesions with telangiectasia evident over her trunk (chest, abdomen, back & axillae) and extremities (upper and lower limbs) (**fig. 1-3**).



Fig 1: Asymmetrical linear streaks of hypo & hyper-pigmented atrophic lesions with telangiectasia on face and extremities.



Fig 2: Same lesions on lower extremities.

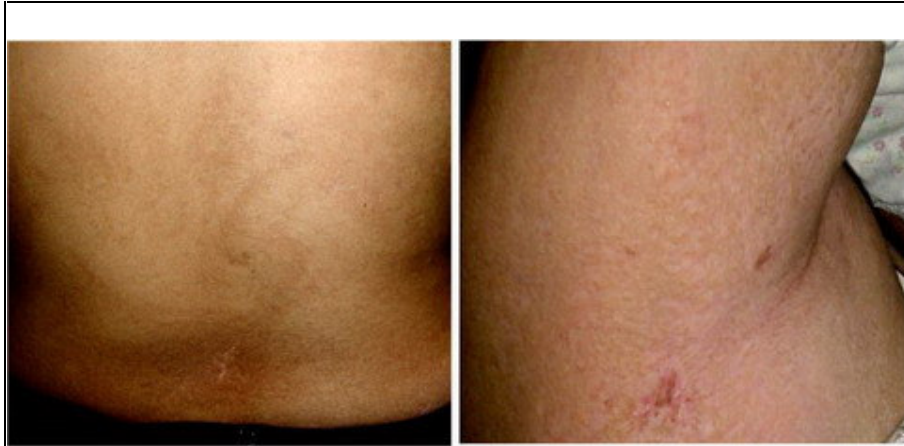


Fig 3: Lesions on trunk (back and axillae).

On examination, she had multiple skeletal anomalies in the form of multiple deformities of fingers and toes with syndactyly of her left third and fourth toes. Asymmetry of all toes was evident with nail dystrophy (**fig. 4**).



Fig 4: Multiple deformities of fingers and toes.

Facial asymmetry was observed in the form of a small rounded skull with triangular facial outline, asymmetry of the ala nasi, pointed chin, prognathism, hypertrophy of the gums, irregular teeth spacing and anomalous tooth form (**fig. 5**).



Fig 5: Small rounded skull, asymmetry of the ala nasi, pointed chin, prognathism, hypertrophy of the gums, irregular teeth spacing and teeth anomalies.

General abdominal examination was done and detected no abnormal findings and ocular examination diagnosed a myopic eye.

Our clinical differential diagnoses included Incontinentia pigmenti, MIDAS syndrome (microphthalmia, dermal aplasia, and sclerocornea), lichen sclerosis et atrophicus and focal dermal hypoplasia.

Investigations included multiple X-rays which showed multiple bone deformities affecting both upper and lower limbs including hands and feet (**fig. 6**).



Histopathological examination showed total absence of the dermis so that the adipose tissue lies directly beneath the epidermis (**fig. 7**).



Fig 7: H&E stained sections showing total absence of the dermis.

Based on the previous findings, a diagnosis of focal dermal hypoplasia (Goltz syndrome) was made.

Discussion

Focal dermal hypoplasia (Goltz Syndrome) is an X-linked dominant mesoectodermal hypoplasia. It usually occurs in a sporadic fashion and the affected persons are females, however it has been reported occurring simultaneously with giant aplasia cutis congenita in a newborn black male [2]. More than 200 cases have been reported, 90% of them are females who are heterozygous or mosaic for mutations in *PORCN*; 10% are live-born affected males who are mosaic for mutations in *PORCN* which has been mapped to locus Xp11.23 [3]. It is usually bilateral but unilateral cases have been also described [4]. Our patient had most of the typical features of Goltz syndrome.

Goltz syndrome starts at birth and has a multitude of clinical features including cutaneous, ocular, dental & skeletal abnormalities. Skin involvement is essential for diagnosis in the form of asymmetrical linear streaks of atrophy & telangiectasia which follows Blaschko's lines. In racially pigmented skin, the lesions may be hypo or hyper-pigmented. There is generalized dryness and pruritus with soft reddish-yellow nodules (fat herniation). Raspberry-like papillomas occur on the lips, perineum, ears, fingers, toes, buccal mucosa and oesophagus. Nail absences or dystrophy with sparse & brittle hair are evident. Characteristic facial features include the presence of a small rounded skull, triangular facial

outline, pointed chin, protruding ears and asymmetrical ala nasi [5]. Our patient suffered from all the previous signs except for the skin herniations and papillomas.

Oral and dental anomalies include prognathism, agenesis, hypodontia, oligodontia, microdontia, enamel fragility and dysplasia, retarded eruption, irregular teeth spacing, enamel defects and malocclusion. Hypertrophy of the gums, high-arched palate; cleft lip, palate, papillomas of the gums, tongue, palate & buccal mucosa may all take place [6]. Warburg observed microphthalmia with bilateral coloboma of the iris and ectopia lentis [7]. Other ocular lesions described are strabismus, anophthalmia, keratoconus and corneal opacification [8]. Most of those findings were found to be consistent with our patient.

Skeletal anomalies include short stature with asymmetric involvement of the hands and feet in 60% of patients, including syndactyly, ectrodactyly, polydactyly, absence or hypoplasia of digits and even absence of an extremity. Cervical rib has been reported [9]. Scoliosis occurs in 20% of cases. Skeletal asymmetry, clavicular dysplasia and spina bifida occulta can occur. The characteristic radiological change is osteopathia striata of the long bones [10]. Our patient had multiple skeletal anomalies like osteopathia striata, syndactyly and asymmetrical fingers and toes.

Goltz reviewed this disorder in 1992 [11]. Patients with areas of total absence of skin at birth have been reported. Apocrine gland anomalies and hydrocystomas near the eyes also have been described. Fibrovascular papillomas, especially in the perianal and vulvar regions, are sometimes mistaken for condylomas. Rarely, laryngeal and esophageal papillomas are seen. Osteopathia striata is a frequent finding and 'lobster-claw' hand is a striking feature of Goltz syndrome. Occasional anomalies include short stature, joint hypermobility, mental retardation (15%), hearing defects, microcephaly, horse-shoe kidneys, umbilical, inguinal, epigastric, or diaphragmatic hernias [12]. Cardiac anomalies include cardiac tumors [13] and congenital heart diseases like truncus arteriosus [14]. Other systems affection includes central nervous, gastrointestinal and genitourinary systems affection [15].

Concerning management, skin lesions are not a major therapeutic problem as the redness of early lesions tends to fade with age, but facial lesions may be a cosmetic worry. Pruritus can be troublesome and should not be overlooked. Constructive surgery and vascular pulsed dye laser for telangiectatic skin lesions had been tried [16]. Papillomata, particularly around the mouth, may be unsightly and can be excised or ablated with cautery or cryotherapy; unfortunately they may recur. Orthopaedic and plastic surgical advice should be sought early with regard to limb deformities. Dental management is important and education regarding caries is imperative. Although developmental delay is more likely in more severely affected children, the degree cannot accurately be predicted and is independent of any of the other features of FDH. Regarding the prognosis, the majority of patients can lead a normal life. Where reported, menses

have occurred at a normal age [8].

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