# FOCAL DERMAL HYPOPLASIA SYNDROME

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A case of focal dermal hypoplasia syndrome is being reported in a young girl

*Key Words: Focal dermal hypoplasia, Golz Syndrome*

## Introduction

Focal dermal hypoplasia (FDH) - although the term is a misnomer (because here the defect not only involves the dermis but the epidermis, the subcutis and even the underlying bones in combinations) was first reported by Libermann in 1935.[1] Golz coined the term 'Focal dermal hypoplasia' for the rare genodermatosis on the basis of histologically apparent areas of connective tissue hypoplasia.[2] This rare mesoectodermal disorder is characterised by linear depressed lesions in conjunction with fat nodules, dysmelanosis, wart-like excresences and variable bone, eye, tooth, hair and nail abnormalities. Presence of fine parallel vertical striations in the metaphysis of long bones - referred as osteopathia striata-possibly due to mesodermal mosaic dysfunction is a reliable marker of this syndrome. This is a very rare disorder and so far only 200 cases have been reported in the world literature.[3] Here is a report of such an extremely rare case.

## Case Report

A one year and seven months old girl was brought to us for her inborn skin disorder. She was born in full term uneventfully. At birth there were superficial ulcerations on left leg and diffusely scattered erythematous patches on buttocks, back and axillae. The ulcer healed with scar formation in a course of three to six months. The erythematous patches also led to atrophic scars. The left foot was deformed since birth. The baby was otherwise alright. She was the only offspring and none in the family was known to have any musculo-cutaneous disorder. The mother was not given any medication antenatally. There was no parental consanguinity.

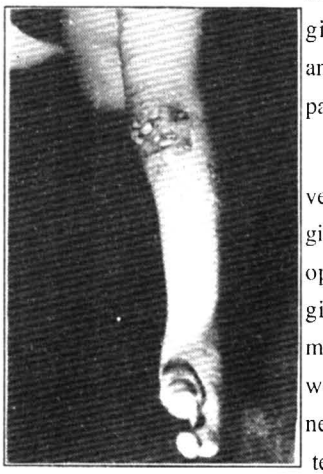

Fig.1. Focal dermal hypoplasia

Examination revealed a well developed girl with no cardiological, ophthalmic or neurological abnormality. The most remarkable changes were detected in the cutaneous and skeletal systems. Skin showed widespread scattered atrophic areas of varying sizes especially over the buttocks, left leg, thigh, and back. The pattern of atrophy was strikingly linear at some places, particularly over the left lower extremity within the atrophic areas. There were numerous soft yellowish nodules often in linear arrangements in the line of Blaschko (Fig.1). The skin showed mottled hypopigmentation over the lower limb and abdomen. The

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scalp, face, palms and soles were normal. There was no abnormality in nails and teeth.

The webbing between middle and ring fingers on left side was increased. Few fingers were hyper-extensible. Few digits were lacking on left foot and there was associated syndactyly. This deformity along with the generalised thinning out of left leg resulted in the limping gait of the baby. This deformity of left leg was the characteristic 'lobster claw deformity' (Fig.2). Routine blood, urine and stool examinations were normal. The X-ray of the left leg showed osteoporosis and absence of two digits. Histology from the atrophic patches showed diminished collagen density leading to diminished dermal thickness. Histology of the soft nodules showed adipose tissue largely replacing the dermis.

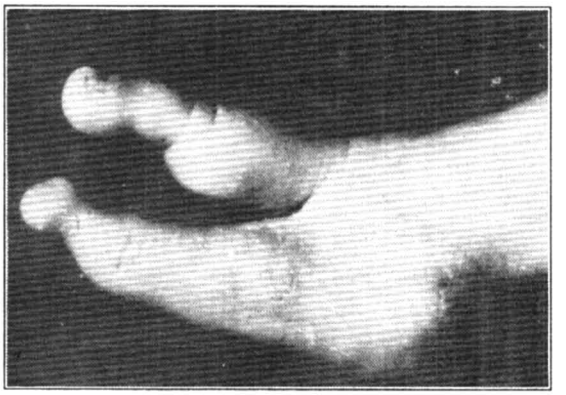

Fig.2 'Lobster claw' deformity of left leg

## Discussion

Cutaneous findings in FDH include hypoplastic skin, linear and reticulate areas of hypo and hyper- pigmentation, lipomatous lesions, periorificial and mucous membrane papillomas and telangiectases. It was previously presumed that hypoplastic connective tissue allows herniation of adipose tissue into the dermis. Howell and Freeman showed that the fat tumors of FDH was the result from the dysplasia which leads to lipomatous hamartoma of fat nevus.[4] Nail changes in FDH include atrophy, dystrophy, spooning and even anonychia. Hair may be sparse or brittle. There may be alopecia.In addition to these cutaneous changes there are a variety of extracutaneous manifestations which include musculo-skeletal, ocular and rarely oral involvements. Musculo-skeletal defects include syndactyly, hypoplasia or absence of digits, asymmetry of body, scoliosis and abnormality in hand and foot bones. Ocular lesions include coloboma, microphthalmia, strabismus and lens subluxation. Oral lesions include agenesis of teeth, papilloma of oral mucosa, microdontia and high arched palate.

FDH is presumed to be an x-linked dominant condition with lethality in males. The few reported cases of males with this syndrome are explained on the basis of mosaicism, as described by Happle [5] or by new mutation.

## References

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