

CASE REPORT

Focal dermal hypoplasia (Goltz syndrome)

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ABSTRACT

Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genodermatosis, characterized by multiple abnormalities of ectodermal and mesodermal origin. We present an infant with focal dermal hypoplasia who, besides having a constellation of anomalies commonly encountered in patients with this syndrome, manifested additional unusual features such as cleft lip and palate.

INTRODUCTION

Focal dermal hypoplasia (FDH) or Goltz syndrome is a rare genodermatosis, characterized by multiple abnormalities of ectodermal and mesodermal origin.¹ Resulting anomalies involve a wide range of body structures and organs including the skin and its appendages, the ophthalmologic, oro-dental, skeletal, urinary, gastrointestinal, cardiovascular and central nervous systems.² We report an FDH patient with unusual cutaneous manifestations.

CASE REPORT

A 7-day-old female was referred to us for evaluation of multiple cutaneous, skeletal and ocular abnormalities present since birth. She was born to a 17-year-old primi mother. Family history was noncontributory. Cutaneous findings were erythematous reticulate atrophic lesions on the face and trunk following the lines of Blaschko (Fig. 1). The hair was sparse with areas of alopecia (Fig. 2). She had unilateral notching of the left ala nasi

and left-sided cleft lip and palate and ophthalmologic examination revealed microphthalmia (Fig. 1). The pinna was malformed (Fig. 3). There was syndactyly of the 3rd and 4th digits of the left hand and ectrodactyly (lobster-claw deformity) of the left foot (Fig 4,5). Abdominal ultrasound reported bilateral hydronephrosis. MRI of the orbit showed microphthalmia with a colobomatous cyst and MRI of the brain was normal.



Fig. 1 Erythematous reticulate atrophic lesions, cleft lip, microphthalmia and sparse hair.

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Fig. 2 Sparse hair with patchy alopecia.



Fig. 4 Syndactyly of the 3rd and 4th digits of the left hand.



Fig. 3 Malformed pinna.



Fig. 5 Lobster-claw deformity of the left foot.

DISCUSSION

Focal dermal hypoplasia (FDH) or Goltz syndrome was first described by Goltz et al in 1962.¹ Since then, more than 200 FHD patients have been reported in the literature.^{3,4} Although the genetic background remains unclear, a high preponderance of affected females, approximately 88%, and high lethality in homozygous males strongly support X-linked dominance.⁵⁻⁷ The diagnosis of FDH relies in a constellation of clinical findings. The characteristic features include atrophic linear hypo or hyperpigmented patches, fat herniation

through dermal defects, and multiple papillomas of the mucous membranes or skin. Histopathologically, these lesions are characterized by the presence of fat cells and lobules in the superficial atrophic dermis. Dental anomalies include hypodontia, oligodontia, microdontia, enamel fragility and dysplasia, retarded eruption and malocclusion. Warburg observed microphthalmia with bilateral coloboma of the iris and ectopia lentis. Other ocular lesions described are strabismus and anophthalmia.⁸ Skeletal anomalies include 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.^{8,10} Management includes genetic counseling and reconstructive surgery. In conclusion, this typical FDH patient's presentation was considered valuable in light of the "lobster claw" deformity and the cleft lip and palate.

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