Focal dermal hypoplasia (Goltz) syndrome with multiple family member involvement: A case report

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Goltz syndrome or focal dermal hypoplasia is a rare syndrome with mesoectodermal hypoplasia. This syndrome is an X-linked dominant disorder with involvement of the cutaneous, ocular, dental and skeletal systems. The most significant feature of this disease is connective tissue dysplasia. Here, we report a 30-year old woman who presented with congenital unilateral linear atrophic areas on her trunk, back, and upper and lower extremities. She has 3 daughters, 2 of whom had the same skin lesions that were consistent with Goltz syndrome. Skin lesions in the younger daughter were more severe than her older sister and mother. The younger daughter had multiple bone deformities in the form of clinodactyly and lobster-claw malformation. She also had nail dystrophy of her fingers and umbilical herniation. The older daughter only had syndactyly and skin atrophic lesions.

Keywords: Goltz syndrome, clinodactyly, hypoplasia

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INTRODUCTION

Goltz syndrome (focal dermal hypoplasia: FDH) is a rare syndrome with multisystem involvement that presents with thinning of the dermis and fat herniation [1,2]. FDH is an X-linked dominant disorder, which is lethal in homozygous males [2,3]. The causative gene in this disorder has been identified as PORCN on chromosome X p11-23 [2]. Variations in clinical severity are partly attributed to lionization of the X-chromosome and postzygomatic genomic mosaicism, which is also responsible for the sporadic cases [3]. Over 200 cases have been reported worldwide, although the incidence is likely to be underestimated, as mildly affected subjects may go unrecognized [2].

Case1: Mother

A 30-year old woman with no positive family history of skin disease presented to the Dermatology Department of Kerman Afzalipour Hospital with unilateral linear atrophic areas on her trunk, back, and upper and lower extremities, which were congenital (Figure 1). On physical examination, she had no developmental delay or other organ involvement. She has 3 daughters, 2 of whom had the same skin lesions.

Case 2: First child of the family

The second case was an 8-year-old girl, produced from a non-consanguineous marriage. The case had

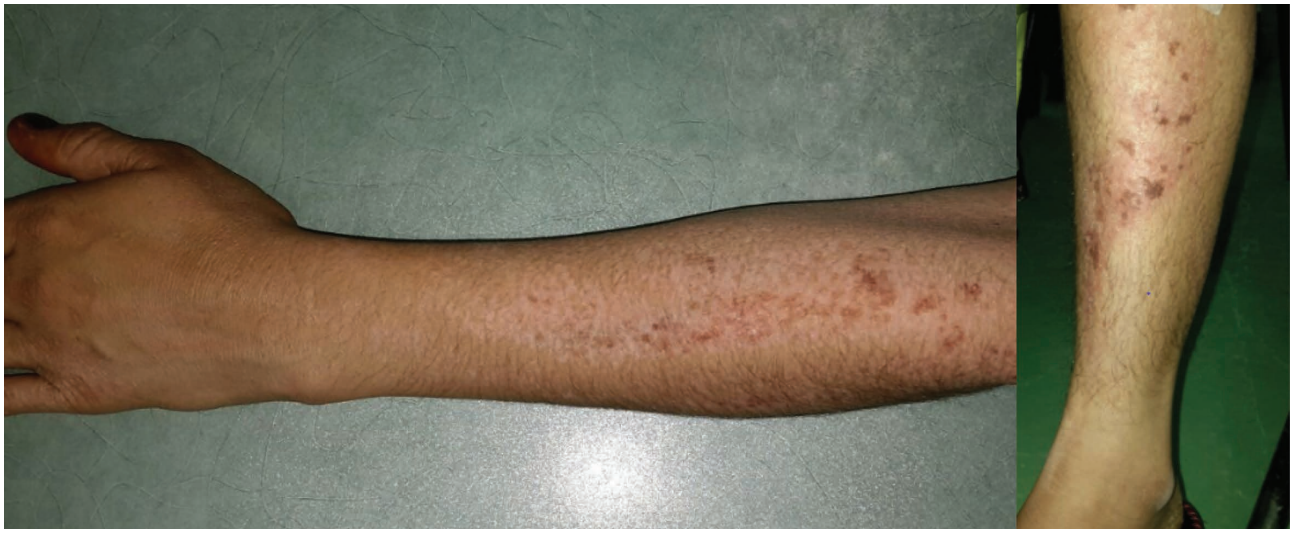

Figure 1. Unilateral linear atrophic areas on the upper and lower extremities.

yellowish atrophic skin along the Blaschko lines that were located on the flexural side of her right arm and axilla, left side of the trunk, and lower extremities. She had syndactyly in the second and third toes of her left foot (Figure 2). Other fingers and toes were normal. She had no developmental delays. Dental, hair, nail, and ocular examinations were normal.

Case 3: Third child of the family

The third case was a 2-year-old girl who had the most severe presentation of the disease (genetic anticipation) in the family. She had multiple atrophic areas with yellow-brown color along the Blaschko lines on her trunk, axillary, and upper and lower extremities (Figure 3). She had multiple bone deformities in the form of clinodactyly in her left hand. She had one finger on her right hand and an accessory finger on the forearm (lobster-claw malformation; Figure 4). The toes were normal. She had nail dystrophy of the fingers with nail shedding in both hands. She also had an umbilical hernia. Her hair, dental, and ocular

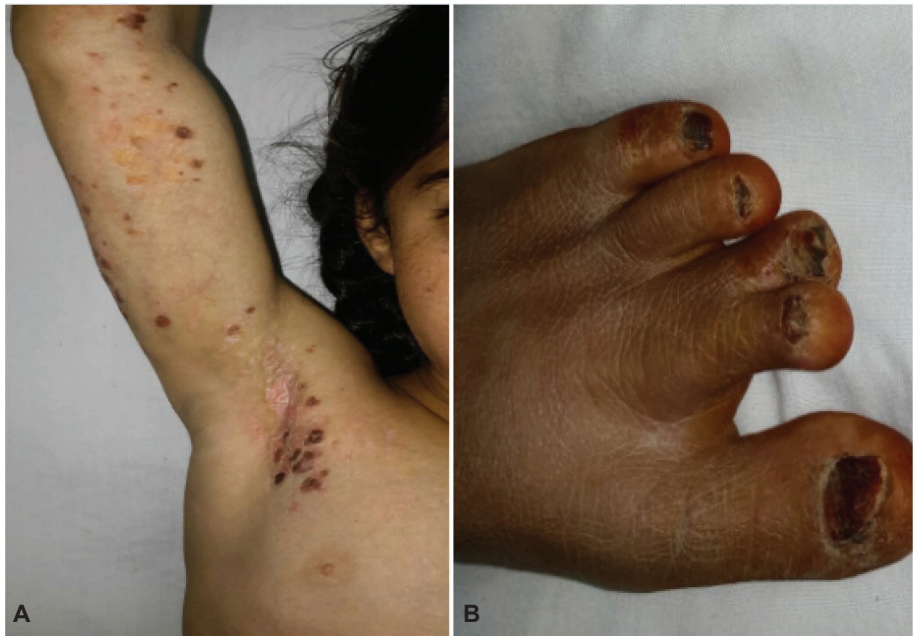

Figure 2. (A) Yellowish atrophic skin along the Blaschko lines on flexural side of right arm and axilla and (B) syndactyly.

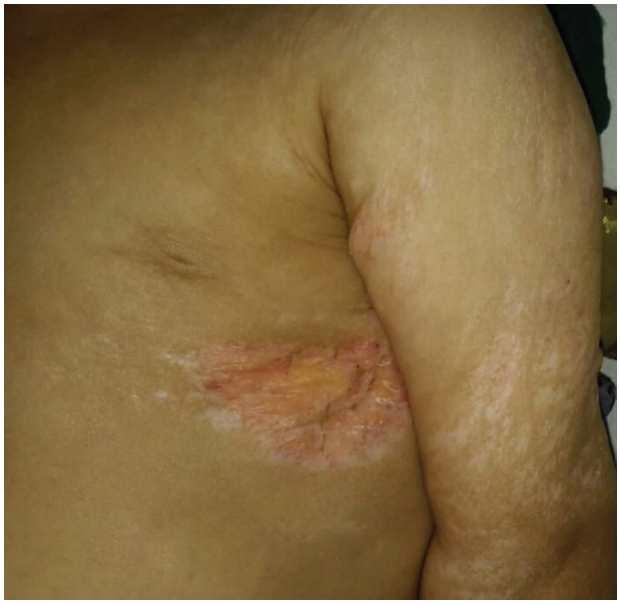

Figure 3. Multiple atrophic, yellow-brown areas along the Blaschko lines on her trunk, axillary, and upper extremities.

examinations were normal and she had no facial asymmetry.

DISCUSSION

The characteristic manifestation of FDH is connective tissue dysplasia, particularly in the skin and skeletal system. Histopathology shows atrophy and thinning of the dermis and collagen fibers as well as herniation of fatty tissue to the dermis [4-6].

Skin involvement is necessary for the diagnosis of FDH [1]. The classic skin lesions include linear atrophic areas with telangiectasia along Blaschko's lines that appear at birth [2]. Hypo- and hyperpigmentation, in addition to fat herniation can appear later [7]. Red colored verrucous papilloma can occur in periorificial areas, fingers, toes, buccal mucosa, and the esophagus [8]. Nails may be absent or dystrophic. The hair is usually sparse and brittle, and patchy alopecia of the scalp or pubic area can be seen [2].

Abnormal teeth and dimorphic fancies such as notched nasal alae, pointed chin, and large malformed ears may be observed. Bony reduction deformities of the hands are common, particularly ectrodactyly (split-hand/foot or lobster-claw malformation) [7]. Other skeletal anomalies include short stature, scoliosis, absence or hypoplasia of digits, and absence of an extremity [2,4].

Mental development is sometimes delayed [2]. Other systems such as gastrointestinal and renal systems can be commonly involved. There are a few reports about the presence of ectopic, hypoplastic or total absence of a kidney [3].

There are different surgical procedures for the treatment of the skin lesions. Vascular pulsed dye laser (PDL) can be used for the treatment of telangiectatic skin lesions [4]. Verrucous papilloma can be treated with curettage or photodynamic therapy [7]. Airway papilloma can be treated with endoscopic laser treatment [2]. Orthopedic and plastic surgical advice should be sought early with regard to limb deformities. Dental management is important and education regarding caries is necessary [4].

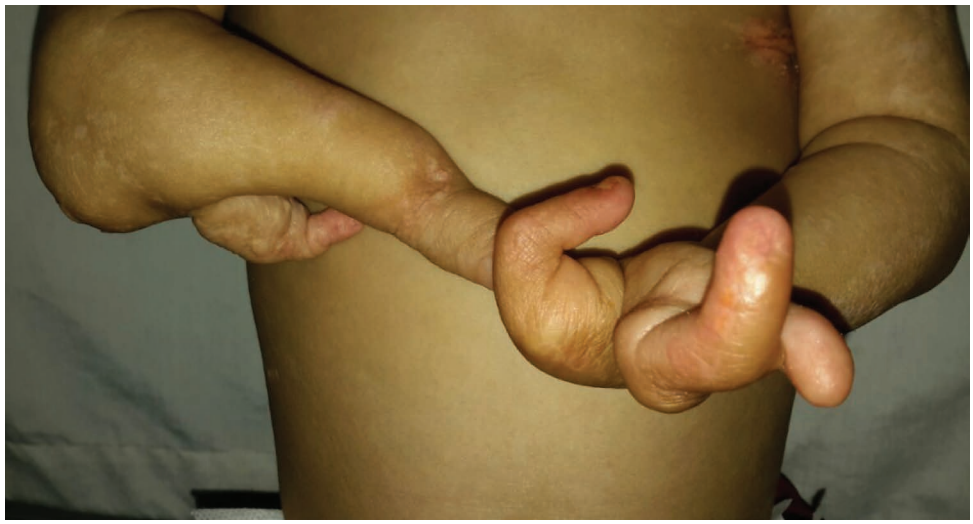

Figure 4. Lobster-claw malformation and nail dystrophy with nail shedding in the upper extremities.

CONCLUSION

Early diagnosis and referral to a related specialist for treatment of associated anomalies is essential and can help patients to have a normal life. Genetic counseling has a major role in preventing new cases.

Conflict of Interest: None declared.

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