Goltz syndrome: a case report from Iran

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Focal dermal hypoplasia or Goltz syndrome is a rare genodermatosis involving all three embryonic layers. Herein, the first case of this syndrome from Iran will be reported. The main clinical features were fat herniation, reticulate pigmentations, telangiectasia, and skeletal defects.

Keywords: focal dermal hypoplasia, genodermatosis, Goltz syndrome

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INTRODUCTION

Goltz syndrome, also known as Focal Dermal Hypoplasia (FDH), is a rare X-linked dominant disorder with involvement of all three embryonic layers. There have been approximately 200 reported cases, 10% of which are male [1]. As an X-linked dominant disorder, FDH is usually lethal in men [2].

FDH is a rare multisystemic disorder and can affect the skin, hair, nails, teeth, bones and eyes. Cutaneous manifestations include hypoplasia of the skin, verrucoid papillomas of the skin and mucous membranes, and fat herniation, often in a Blashkoid distribution [1]. Sparse or brittle hair and ridged, hypoplastic or dysplastic nails are also characteristic of FDH [1]. Skeletal defects including digital anomalies such as syndactyly, polydactyly, and camptodactyly and limb reduction deformities such as split hand or foot have been noted [1,3,4]. Associated ocular abnormalities include anophthalmia, microphthalmia, iris and chorioretinal coloboma, and lacrimal duct abnormalities [4]. Systemic effects, such as neurological, cardiovascular and renal disorders, have also been observed.

CASE REPORT

We present a 22-year-old woman with clinical features consistent with FDH. Her chief complaint was a breast mass since two years ago. On examination, she exhibited a large soft boggy yellow swelling suggestive of fat herniation on her right breast (Figure 1). Reticulate pigmentations

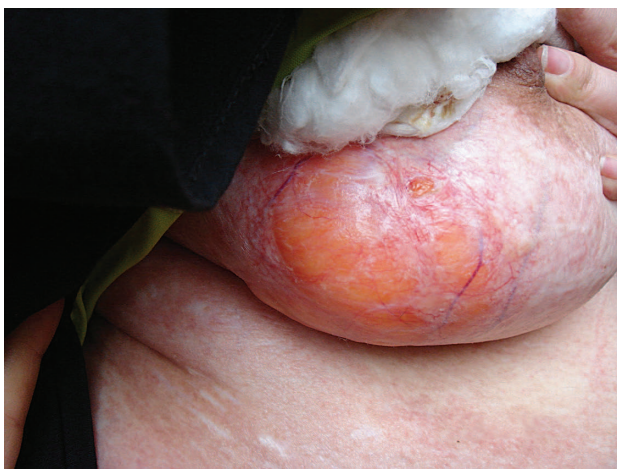

Figure 1. Outpouching of fat tissues on the breast.

and telangiectasis distributed on Blashko lines on her back were also evident (Figure 2).

She had a short stature (142 cm height) and multiple skeletal defects, including syndactyly in the right foot and deformities in other digits (Figure 3). Also, the nails were dystrophic with longitudinal ridging. Dental anomalies in different shapes were further noted (Figure 4). Neurological and ophthalmological examinations revealed no significant pathology. There was no osteopathia striata on radiological examinations. The patient refused skin biopsy and surgical repair of her breast fat herniation.

Based on the clinical features, a diagnosis of focal dermal hypoplasis was made for this patient. None of the patient's siblings or family members showed any dermatological, skeletal, or ophthalmological abnormalities reminiscent of FDH.

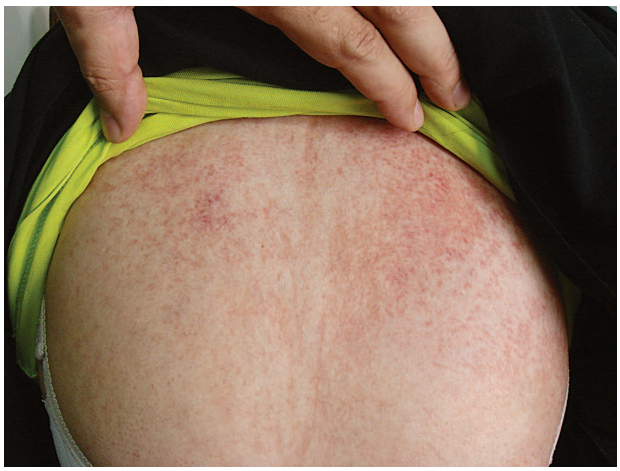

Figure 2. Streaks of pigmentations and telangiectasias.

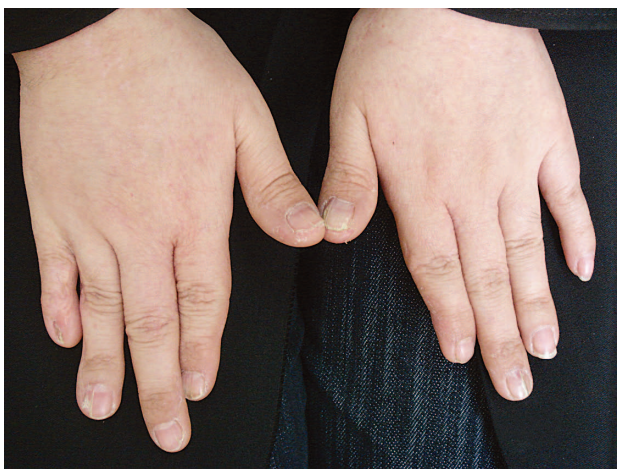

Figure 3. Digital anomalies in association with nail dystrophy.

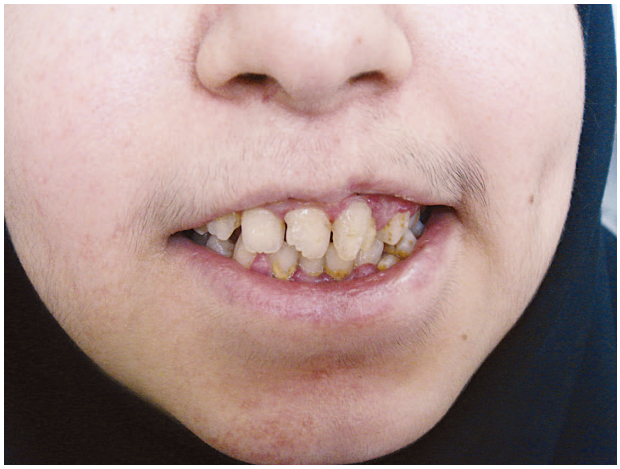

Figure 4. Various forms of dental anomalies.

DISCUSSION

FDH was first described by Goltz in 1962 [5]. It is an X-linked dominant disorder involving the ectoderm (skin and teeth), mesoderm (dermis and bone) ad endoderm (mucosa of the mouth and larynx) in a mosaic pattern. It mainly affects females and is characterized by asymmetrical streaks of skin atrophy, linear pigmentations, small red-yellow nodules and a wide variety of defects that affect the eyes, teeth, and skeletal, urinary, gastrointestinal, cardiovascular, and central nervous systems. Musculoskeletal abnormalities are present in 80% of the cases of FDH. These include short stature, syndactyly (60%), hypoplastic or absent digits (33%) scoliosis (17%) and asymmetry of the face [6].

The diagnosis of FDH is usually made based on clinical findings. Affected individuals are often recognized at birth or occasionally prenatally, but cases involving a minor expression of the syndrome may be diagnosed later in life. FDH has been associated with mutations and deletions in the *PORCN* gene (Xp11.23), which codes for proteins that regulate embryonic development, on the X chromosome [6,7].

The mnemonic FOCAL can be used to remember some of the key features of this syndrome: female sex; osteopathia striata; coloboma; absent ectodermis-, mesodermis-, and endodermis-derived elements; and lobster claw deformity.

Management includes genetic counseling and reconstructive surgery. Moreover, flashlamp-pumped pulsed-dye lasers have been shown to reduce the pruritus and erythema, characteristic

of FDH [8]. Mallipeddi et al, discussed the use of Photodynamic Therapy (PDT), a method of treating neovascularization through selective photosensitization and destruction of proliferating vessels, in treating FDH. They provided preliminary evidence that PDT was effective in treating FDH [9].

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