

**Case Report**
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## Almost Unilateral Focal Dermal Hypoplasia in a Young Yemeni Girl: A Case Report

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**ABSTRACT**

Focal dermal hypoplasia (FDH), also known as Goltz syndrome or Goltz–Gorlin syndrome, is an extremely rare X-linked dominant multisystem genodermatosis caused by mutations in the PORCN gene on the X chromosome. These mutations primarily affect ectodermal- and mesodermal-derived tissues, including the skin, skeletal system, and eyes. To date, approximately 300 cases have been reported in the literature, with 95% of cases occurring in females. FDH typically involves both sides of the body; however, only 11 cases of unilateral FDH have been reported. Herein, we discuss the case of a 15-year-old Yemeni girl with an almost unilateral (left-sided) distribution of blaschkoid atrophic lesions accompanied by limb defects. FDH was diagnosed based on the typical clinical findings. To the best of our knowledge, this is the first reported case of FDH in Yemen.

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**Introduction**

Focal dermal hypoplasia (FDH), also known as Goltz syndrome or Goltz–Gorlin syndrome, is an extremely rare X-linked dominant multisystem genodermatosis caused by mutations in the PORCN gene on the X chromosome [1]. These mutations primarily affect tissues derived from the ectoderm, mesoderm, and endoderm, such as those of the skin, skeletal system, teeth, and eyes [2]. Since FDH was first reported by Goltz in 1962, approximately 300 cases have been reported in the literature, with 95% occurring in females [3]. FDH typically presents with bilateral involvement; however, only 11 cases of unilateral FDH have been reported (Table 1). Herein, we report a case involving a nearly unilateral (left-sided) distribution of blaschkoid atrophic lesions accompanied by limb defects in a young girl. The case findings may help raise awareness of this unusual manifestation of an extremely rare disease that can pose a diagnostic challenge.

**Table 1: Reported Cases of Unilateral Focal Dermal Hypoplasia (Goltz Syndrome)**

Publication year	First author	Title	Journal	Country	Age	Sex	No.
2008	Aoyama M	Case of unilateral focal dermal hypoplasia (Goltz syndrome)	J Dermatol	Japan	8 Years	Female	1
2010	Tenkir A	Goltz syndrome (focal dermal hypoplasia) with unilateral ocular, cutaneous and skeletal features: case report	BMC Ophthalmol	Ethiopia	4 Years	Female	1
2012	Asano M	A Case of Almost Unilateral Focal Dermal Hypoplasia Resulting From a Novel Mutation in the PORCN Gene	Acta Derm Venereol	Japan	2 Years	Female	1
2012	Maalouf D	A novel mutation in the PORCN gene underlying a case of almost unilateral focal dermal hypoplasia	Arch Dermatol	Lebanon	17 Years	Female	1

2013	Nakanishi G	Novel and recurrent PORCN gene mutations in almost unilateral and typical focal dermal hypoplasia patients	Eur J Dermatol	Japan	5 Days	Female	1
					2 Months	Male	1
2014	Chandran S	Unilateral labial mass in a neonate: A rare clinical presentation of focal dermal hypoplasia	J Clin Neonatol	Singapore	1 Day	Female	1
2017	Lee S	Almost Unilateral Focal Dermal Hypoplasia	Ann Dermatol	Korea	19 Years	Female	1
2017	Ong JEL	Unilateral Tonsillar Hypertrophy in a 4-Year-Old Girl with Focal Dermal Hypoplasia (Goltz Syndrome)	Philipp J Otorhinolaryngol Head Neck Surg	Philippines	4 Years	Female	1
2018	Alsharif S	Unilateral Focal Dermal Hypoplasia (Goltz Syndrome): Case Report and Literature Review	Case Rep Dermatol	Saudi Arabia	16 Years	Female	1
2020	Lyons D	A Rare Case of Mainly Unilateral Focal Dermal Hypoplasia (Goltz Syndrome) in an Adult Male: A Case Report and Review of the Literature	J Med Cases	Australia	48 Years	Male	1
Total						9 Females, 2 Males	11

### Case Report

A 15-year-old girl presented with progressive skin changes that had been present since birth, mainly affecting the left side of her body. Cutaneous examination revealed linear, streaky, atrophic hypo- and hyperpigmentation in a blaschkoid pattern on the trunk and limbs (Figure 1a–d). Cribriform atrophic scars were observed on the lower left side of the trunk (Figure 1b–d). Syndactyly of the left fourth and fifth toes, hypoplasia of the left thumb and index finger, and micronychia of the thumbnail (Figure 2a–b) were also observed. A longitudinal groove was observed on the right index fingernail (Figure 2c). Linear atrophic lesions resembling striae dystensae were present on the left arm (Figure 2d). No papillomas or asymmetry of the face were evident, and the patient’s hair and teeth appeared normal.



**Figure 1a:** Streaky, Blaschkoid Atrophic Hyperpigmented Patches, More on the left side



**Figure 1b:** Left Side of the Trunk. Streaky, Blaschkoid Hyperpigmentation on the Upper Part, and Cribriform Atrophic Hyperpigmentation on the Lower Part



**Figure 1c:** Cribriform Atrophic Scar on the Lower Left Side of the Trunk., Close-up View of figure 1b



**Figure 1d:** Less Affected Right Side of the Trunk



**Figure 2a:** Syndactyly of the Left 4<sup>th</sup>, and 5<sup>th</sup> Toe



**Figure 2b:** Hypoplasia and Micronychia of the Left Thumb



**Figure 2c:** Longitudinal Groove of the Right Index Finger Nail



**Figure 2d:** Streaky Skin Atrophy Admixed with Hypo-, and Hyperpigmentation, on the Left arm, and Cubital Fossa

According to the diagnostic criteria for FDH proposed by Bostwick et al., a patient must present with at least three skin findings and one limb finding to be diagnosed with FDH (Table 2) [4]. Our patient fulfilled three cutaneous criteria (criteria 1, 2, and 5) and one limb criterion (criterion 3), thereby confirming the diagnosis of FDH. A genetic test for PORCN mutation, which would have further confirmed the diagnosis, was unfortunately not performed due to lack of facilities.

**Table 2: Diagnostic Criteria for Focal Dermal Hypoplasia**

Characteristic skin findings	Present case
1. Congenital patchy skin aplasia	+
2. Congenital nodular fat herniation	-
3. Congenital hyper/hypo-pigmentation in Blaschkolinear distribution	+
4. Telangiectasia	-
5. Congenital ridged dysplastic nails	+
Characteristic limb malformations	
1. Split hand/foot (ectrodactyly)	-
2. Transverse limb defects	-
3. Syndactyly	+
4. Oligodactyly	-
5. Marked long bone reduction	-

**Discussion**

Bree et al. reported the cutaneous manifestations of FDH in 2019 in detail and found that nail changes such as V-nicking, longitudinal ridging of the nail plate, and micronychia occurred in 68%, 84%, and 68% of cases, respectively. Longitudinal ridging and micronychia were identified in our case [5]. Bree et al. also identified a newly recognized skin finding, namely, progressive hyperpigmented freckling within hypopigmented areas, which was found in 68% of patients who also exhibited photosensitivity. This finding was also observed on the volar aspect of the left arm in our case. Bree et al. concluded that characteristic dermatological features can aid in the early diagnosis of FDH and reduce the need for extended investigations. Patchy scalp alopecia was observed in 79% of cases, and hair shaft anomalies of any type in 89%; however, neither was observed in our case. Bree et al. reported skin atrophy in all study patients, while papillomas were found in only 65% of cases. However, papillomas were absent in our case.

This case highlights the importance of recognizing such atypical features and underscores how clinicians’ awareness of such manifestations can facilitate timely diagnosis and minimize unnecessary extensive investigations.

**References**

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