Focal Dermal Hypoplasia (Goltz Syndrome): A Rare Case

Sir,

Goltz syndrome, described first by Liebermann in 1935 as “atrophoderma linearis maculosa et papillomatosis congenitalis”, is a rare mesodermal dysplasia with X-linked dominant inheritance mainly affecting females and lethal in males.[1] The term focal dermal hypoplasia (FDH) was coined by Goltz in 1962.[2] Around 200–300 cases have been described in literature so far.[3] It is characterized by the classical cutaneous, skeletal, ocular, and dental defects. We describe a case of 2-month-old female presenting with the same.

A 2-month-old female, preterm (32 weeks) with low birth weight of 1.5 kg born out of nonconsanguineous marriage, presented with characteristic linear atrophic depigmented macules in Blaschko distribution over whole body with a history of surgery for cleft lip and palate 1 month back [Figure 1a]. On examination, atrophic macules [Figure 1b] were associated with fat herniation over the right arm [Figure 1c]. Also, there was nonscarring alopecia of scalp [Figure 2] with sparse eyebrows and few, small raspberry like papillomas at the tip of the tongue and perioral area. She had typical facies, namely, microcephaly, triangular facial outline, pointed chin, multiple atrophic scars, microphthalmia, and prominent ears. Syndactyly of third and fourth toe with characteristic “Lobster claw deformity” of left foot were seen on skeletal examination [Figure 3]. Ocular examination revealed microphthalmia of the right eye, iris coloboma, and inferonasal retinal detachment of left eye. A 1 × 1 cm² reducible umbilical hernia was seen which was prominent on crying. There was no similar history or history of abortion in the family. The rest of the systemic examination was within normal limit. Ultrasound abdomen revealed an umbilical hernia with a defect of size 1 cm. Chest X-ray and electrocardiography were also advised to rule out systemic involvement. The parents did not consent for skin biopsy, and genetic testing could not be done due to institutional unavailability. On clinical findings, we considered a differential diagnosis of FDH and incontinentia pigmendi. There was no history of cutaneous vesiculation and verrucous lesions, which ruled out incontinentia pigmendi. A diagnosis of Goltz syndrome was made on the basis of clinical presentation and the patient was referred to the respective departments after counselling the parents.

FDH has a very low prevalence of less than 1 in 10,00,000.[5] After an extensive search on PubMed and MEDLINE database with the search terms “FDH,” “Goltz syndrome,” and “India,” we could only find 19 case reports and a single-case series consisting of eight patients. The syndrome has been seen worldwide without any known genetic or racial predisposition. It is associated with mutation in PORCN gene located on X chromosome, which is involved in WNT signaling pathway that helps in embryogenesis, tissue homeostasis, and stem cell maintenance. The 10% affected males survive due to mosaicism of PORCN gene or somatosomal anomalies.[4]

It is an ectomesodermal malformation disorder with multisystem involvement like skin, teeth, skeleton, and CNS. Cutaneous changes are the primary diagnostic features showing considerable variability due to postzygotic somatic mutations in both males and females and random X chromosome inactivation in females.[5] The prominent features are red-yellow to reddish cribiform atrophic lesions with telangiectasias and associated hyper or hypopigmentation in linear Blaschko distribution usually involving the trunk and extremities but may be present on any part of the body.[5]

Isha Gupta,
Neha Dhankar¹,
Surabhi Dayal,
Meha Tyagi²

Department of Dermatology,
Venerology and Leprosy, Pandit B. D. Sharma PGIMS, Rohtak, Haryana, ¹Department of Dermatology, Venerology and Leprosy, Deen Dayal Upadhyay Hospital, New Delhi, ²Department of Dermatology, Venerology and Leprosy, Guru Teg Bahadur Hospital, Delhi, India

Address for correspondence:
Dr. Neha Dhankar,
Department of Dermatology, Venerology and Leprosy, Deen Dayal Upadhyay Hospital, New Delhi - 110064, India.
E-mail: dhankarneha21@gmail.com

How to cite this article: Gupta I, Dhankar N, Dayal S, Tyagi M. Focal dermal hypoplasia (Goltz syndrome): A rare case. Indian Dermatol Online J 2022;13:502-4.

Received: 29-Oct-2021. Revised: 16-Nov-2021. Accepted: 03-Dec-2021. Published: 24-Jun-2022.
Atrophic areas have thinned to absent dermis resulting in fat herniations appearing as depressive yellow-pink excrences. Various oral anomalies found are linear enamel hypoplasia, hypodontia, jaw cysts, clefting, hemihypoglossia, and papillomatosis that can develop throughout life and occur in perigenital, perioral, intertriginous, and mucosal surface. Other dermatologic features include patchy alopecia, brittle or sparse hair, nail dystrophy or anonychia, and palmar and plantar hyperkeratosis.\[6\]

Skeletal changes include syndactyly, polydactyly, oligodactyly, ectrodactyly, lobster claw deformity, and osteopathia striata (vertical banding of epiphysis and metaphysis of bones on radiography). Vertebral anomalies may be seen in the form of scoliosis, kyphosis, vertebral body fusions, and spina bifida.\[2\] Ocular anomalies comprise of coloboma, strabismus, microphthalmia, and nystagmus. Intellectual disability have been reported in 15% of cases. Minority may have defects in other organ systems including hearing defects, cardiac defects, abdominal wall defects, malrotation of gut, duodenal atresia, and renal malformations.\[6\] Because of pleomorphism, all the features may not be present in a single case.

It requires multispecialty approach for diagnosis and management. Early recognition may lead to more effective intervention. Frequent evaluation should be done in order to prevent further damage. It is also important to address psychological issues along with the physical and functional problems.

**Financial support and sponsorship**

Nil.
Conflicts of interest

There are no conflicts of interest.

References

1. Kothari D, Nayak CS, Madke B, Giri A. Goltz Gorlin syndrome: A rare genodermatosis. Indian J Paediatr Dermatol 2014;15:133-6.
2. Bharani S, Thakkar S. A case report of focal dermal hypoplasia- Goltz syndrome. Indian Dermatol Online J 2013;4:241-3.
3. Itin P. Ectodermal dysplasias. In: Griffiths C, Barker J, Bleiker T, Chalmers R, Creamer D, editors. Rook’s Textbook of Dermatology. 9th ed. Oxford: Wiley-Blackwell; 2016. p. 67.1-27.
4. Leoyklang P, Suphapeetiporn K, Wananuck S, Shotelersuk V. Three novel mutations in the PORCN gene underlying focal dermal hypoplasia. Clin Genet 2008;73:373-9.
5. Wang L, Jin X, Zhao X, Liu D, Hu T, Li W, et al. Focal dermal hypoplasia: Updates. Oral Dis 2014;20:17-24.
6. Riyaz N, Riyaz A, Chandran R, Rakesh SV. Focal dermal hypoplasia (Goltz syndrome). Indian J Dermatol Venereol Leprol 2005;71:279-81.