Focal Dermal Hypoplasia (Goltz Syndrome): A Rare Case

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Sir,

Goltz syndrome, described first by Liebermann in 1935 as "atrophoderma linearis maculosa et papillomatosis congenitalis", is a rare mesoectodermal 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 Blaschkoid 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 microophthalmia of the right eye, iris coloboma, and inferonasal retinal detachment of left eye. A 1 × 1 cm^2 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 too institutional unavailability. On clinical findings, we considered a differential diagnosis of FDH and incontinentia pigmenti. There was no history of cutaneous vesiculation and verrucous lesions, which ruled out incontinentia pigmenti. 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.[3] 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 chromosomal 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 cribriform atrophic lesions with telangiectasias and associated hyper or hypopigmentation in linear Blaschkoid distribution usually involving the trunk and extremities but may be present on any part of the body.[1]

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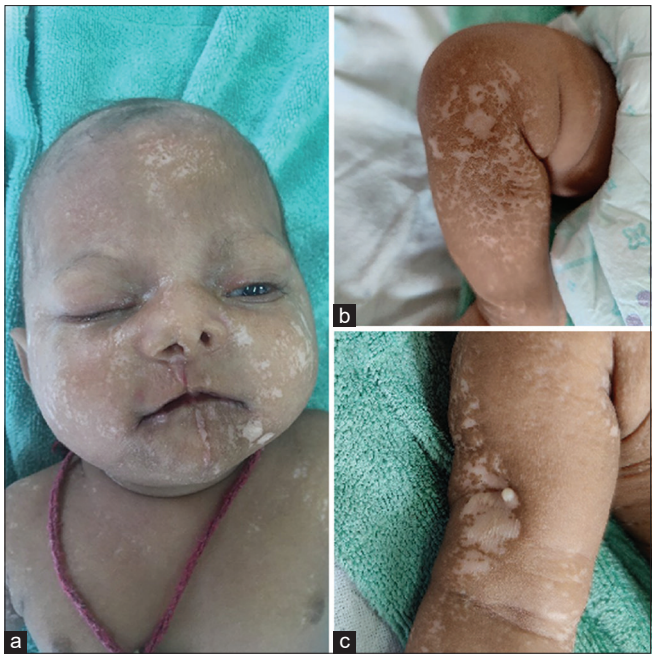

Figure 1: (a) Multiple atrophic, depigmented macules over face with healthy surgical scar of cleft lip repair. (b) Multiple atrophic, depigmented macules seen over the right leg. (c) Multiple atrophic, depigmented macules with fat herniation seen over the right arm

Figure 2: Nonscarring alopecia of scalp

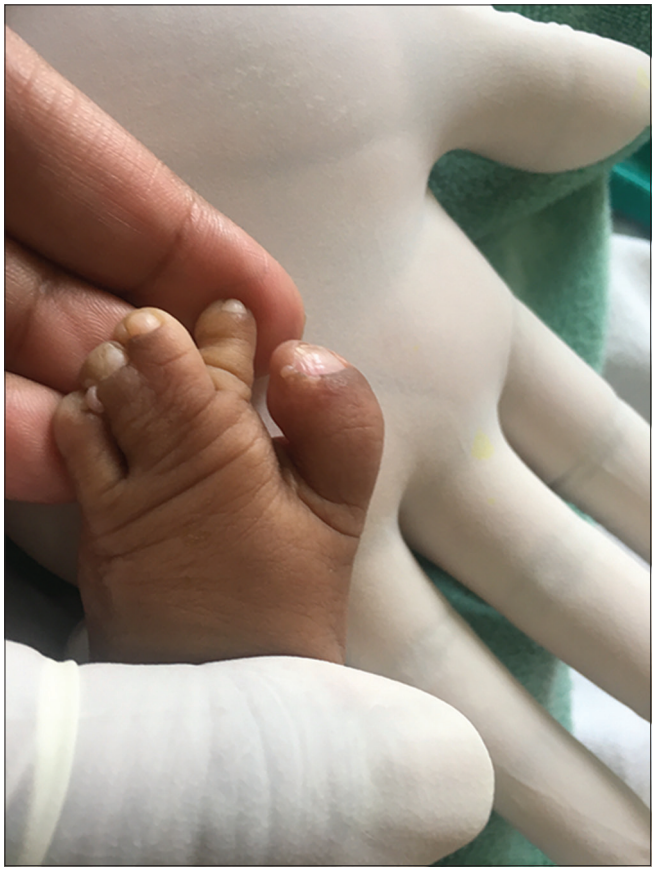

Figure 3: Lobster claw deformity of the left foot

Atrophic areas have thinned to absent dermis resulting in fat herniations appearing as depressive yellow-pink excrescences. 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.

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Conflicts of interest

There are no conflicts of interest.

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