Case Report

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Paediatrics Section

Dermal and Ophthalmological Manifestations of Goltz Syndrome in a Four-month-old Female Child: A Case Report

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ABSTRACT

Goltz syndrome, also known as Focal Dermal Hypoplasia (FDH), is a rare X-linked dominant genodermatosis characterised by multisystem involvement affecting the skin, skeletal system, eyes, and craniofacial structures. Mutations in the Porcupine O-acyltransferase (PORCN) gene are responsible for the multisystem involvement. The authors report a case of a four-month-old female who was brought with linear hypopigmented atrophic skin lesions, patchy alopecia, facial dysmorphism, and bilateral iris colobomas. She also had a partial cleft lip and palate. Radiological imaging revealed anterior rib notching and fused pelvic kidneys. Genetic testing confirmed a pathogenic variant in the PORCN gene (c.374-2del), hence, establishing the diagnosis of Goltz syndrome. The child was managed conservatively with topical medication and nutritional supplementation. A multidisciplinary approach was planned for long-term follow-up. The present case highlights the importance of early recognition of the characteristic cutaneous and extracutaneous features of Goltz syndrome, which often present in the neonatal or early infantile period, and can be further supported by genetic testing. Prompt diagnosis enables comprehensive care and improved quality of life through timely multidisciplinary interventions.

Keywords: Ectodermal dysplasia, Gene mutation, Genetics, Microphthalmia, Ophthalmic cutaneous syndrome

CASE REPORT

A four-month-old female child was brought to the Paediatric Outpatient Department with complaints of multiple light coloured skin lesions present all over the body. They appeared as linear lesions at birth which healed gradually after a few days with scarring. There were no complaints of bleeding from the lesion or any other site, fever, any abnormal body movements. Antenatal history was unremarkable. There was no history of any drug intake by mother, any illness during the antenatal period. She was second in birth order, born at 36 weeks of gestation, out of non-consanguineous marriage, via caesarean section in view of oligohydramnios. Postnatal course was uneventful and she was discharged on day two of life on haemodynamically stable conditions. There was no history suggestive of neonatal jaundice, respiratory distress or requirement of oxygen support. Child was immunised for age. She was diagnosed with partial cleft palate at birth and was due for surgical clearance. There were no members in the family having similar looking lesions.

On general physical examination, vitals were stable. She did not attain visual fixation and did not recognise her mother's voice, however social smile was present at the time of presentation. Head to toe examination revealed facial dysmorphism, patchy alopecia, hypopigmented hair, bilateral iris colobomas, enlarged forehead, depressed nasal bridge, prominent philtrum, thin upper lip, papillomatous tongue, partial cleft of lip and palate and a narrow skull transversely. At the 18-month follow-up, these clinical features persisted and were documented photographically, demonstrating bilateral iris colobomas, facial dysmorphism with enlarged forehead, depressed nasal bridge, prominent philtrum and thin upper lip, patchy alopecia with hypopigmented hair, and multiple hypopigmented atrophic linear streaks and macules of varying sizes over the thighs and gluteal region [Table/Fig-1]. The child had almost no perception of light. Cutaneous examination revealed multiple hypopigmented atrophic linear streaks, and macules of varying sizes present on the forehead, neck, trunk, thighs, gluteal region, genitalia, and upper and lower limbs along the lines of Blaschko. There was extrusion of fat at the hypopigmented patch area. Nails of all four limbs were dystrophic. Oral mucosa was however, normal. Cardiovascular system examination, respiratory system and central nervous system examination revealed no

[Table/Fig-1]: An 18-month follow-up images of the child depicting: a) Bilateral iris colobomas; b) Facial dysmorphism, enlarged forehead, depressed nasal bridge, prominent philtrum, thin upper lip; c) Patchy alopecia and hypopigmented hair; d) Multiple hypopigmented atrophic linear streaks, and macules of varying sizes present on the thighs, gluteal region.

abnormality. Based on the clinical presentation and characteristic skeletal abnormalities, a probable diagnosis of FDH was made.

On investigations, complete haemogram, liver and renal functions were normal. Thyroid functions were normal. Ultrasonographic evaluation of the kidneys demonstrated bilaterally empty renal fossae. Two renal structures were identified within the pelvic cavity, lying in close apposition and appearing fused, consistent with a fused pelvic kidney, likely representing a variant of crossed fused renal ectopia. The renal parenchyma showed preserved corticomedullary differentiation, with no evidence of hydronephrosis or pelvicalyceal dilatation. An infantogram revealed anterior rib notching, suggestive of an underlying skeletal or vascular anomaly. Echocardiography was formulated to evaluate for associated congenital cardiac anomalies and demonstrated normal cardiac anatomy and function, with no structural defects identified.

Based on the multisystem involvement, differential diagnosis including incontinentia pigmenti, MIDAS (Microphthalmia, Dermal Aplasia, and Sclerocornea) syndrome, Rothmund-Thomson syndrome, and Adams-Oliver syndrome. Incontinentia pigmenti was excluded due to the absence of staged pigmentary changes and eosinophilic spongiosis, along with the presence of subcutaneous fat herniation. MIDAS syndrome lacks the characteristic Blaschkoid atrophic lesions and skeletal anomalies seen in FDH. Rothmund-Thomson syndrome and Adams-Oliver syndrome were ruled out based on differing inheritance patterns and the absence of hallmark clinical features. The presence of Blaschkoid atrophic lesions with fat herniation, multisystem involvement was more in favour of Goltz syndrome. Genetic analysis later revealed a heterozygous 3' splice site variation in intron 4 of the PORCN gene affecting the invariant AG acceptor splice site of exon 4 which has been classified as a pathogenic variant of the Goltz Syndrome.

The parents were counselled regarding the prognosis of the child and the requirement of a multidisciplinary approach for management. Application of clobetasone was advised for two weeks which binds to dermal and intradermal cells and induce the inhibitory proteins, leading to decreased activity of prostaglandins, kinins and histamine, oral zinc supplementation, and vitamin supplementation and advised regular follow-up.

The child subsequently underwent surgical correction of the cleft palate at six months of age. A detailed pre-anaesthetic evaluation was performed prior to surgery which included airway management, intraoperative monitoring, fluid therapy, and postoperative pain control, was formulated considering the infant's age and weight. Cardiovascular evaluation, including echocardiography, was normal, and baseline laboratory investigations were within normal limits for age. The procedure and associated risks were explained to the parents, and written informed consent for anaesthesia was obtained. After comprehensive evaluation and necessary optimisation, the infant was deemed fit to undergo the planned surgical procedure under appropriate anaesthetic care. The surgical procedure and postoperative period were uneventful. On 18-month follow-up, she had developmental delay and has not been able to achieve the milestones expected for her age with failure to achieve age-appropriate motor, language, and social milestones. The parents were counselled in detail regarding the nature of developmental delay, its possible association with underlying structural or genetic factors, and the importance of early intervention. They were informed that developmental outcomes can vary widely, and early stimulation plays a critical role in maximising functional potential. A multidisciplinary approach was emphasised, including regular developmental assessment, physiotherapy for gross motor delay, speech and language therapy for communication skills, and occupational therapy if required. The need for periodic ophthalmologic and dermatologic evaluations was reinforced, given the possibility of associated systemic involvement. Parents were educated about home-based stimulation techniques and the importance of structured follow-up to monitor progress and modify therapy plans accordingly. Psychological support and reassurance were provided.

DISCUSSION

Goltz syndrome, also known as FDH, is a rare, X-linked dominant condition genetic disorder characterised by distinctive skin manifestations and has a multisystem involvement including the skeletal system, eyes, and face [1]. Recognition of the phenotypic presentation of the syndrome is essential to make the diagnosis. Although very rare, early recognition of familial dermal hypoplasia is important to ensure reduced morbidity and timely effective intervention [2]. It is caused due to mutations in PORCN gene, mosaicism for mutations has been implicated as the genetic basis for FDH. This PORCN gene encodes an endoplasmic reticulum transmembrane protein that is implicated in processing the Wingless-related integration site (WNT) signalling proteins, which is a vital protein for skin and bone development. Mosaicism for mutations in the PORCN gene on chromosome Xp11.23 has been implicated as the genetic basis for FDH [3].

Paller AS et al., hypothesized the pattern of distribution indicates that the WNT proteins stimulation lead to epidermal-dermal signals which induce changes in the dermis. The hallmark of FDH is thinning of the dermal layer, which eventually results in depressed linear lesions and soft, reddish-yellow outpouchings of the skin caused by herniation of subcutaneous fat as was present in the index of present case [4]. At birth, the lesions may present as blisters or erosions that leave behind atrophic scars that can mimic incontinentia pigmenti. Red, hypopigmented, or depigmented atrophic macules, arranged in a linear or blaschkoid pattern or in a reticulate grouping can be found on any part of the body, but commonly involve the trunk, thighs and buttocks [5]. The second most common extracutaneous abnormality is skeletal defects. These include syndactyly, polydactyly, ectrodactyly, hypoplasia of the digits, and vertebral abnormalities like scoliosis, kyphosis, vertebral body fusions, and spina bifida. Lobster claw deformity (split hand or split foot) is a major distinct feature of this condition. In few cases, there is osteopathia striata, which are longitudinal linear striations in the metaphyses of the long bones seen on radiography [6].

Several case reports and series have expanded the clinical spectrum of Goltz syndrome (FDH). Tenkir A and Teshome S described a four-year-old girl with unilateral ocular, cutaneous, and skeletal abnormalities, including microphthalmos, iris coloboma, hypopigmented atrophic macules following Blaschko lines, lobster-claw hand deformity, and oligodactyly, highlighting an unusual asymmetric presentation of FDH that typically affects both sides of the body [7]. A similar unilateral pattern was reported in a 16-year-old girl exhibiting reticulated atrophic macules, hypoplasia of the right breast, dental enamel abnormalities, and partial alopecia confined to the right side of the face and arm, reinforcing the importance of considering mosaicism in FDH [8]. Genetic insights were provided by a case reporting a novel PORCN frameshift mutation (c.854-855insACCTGAC) in a 17-year-old with almost unilateral FDH manifestations, illustrating how X-chromosome inactivation patterns may influence phenotype [9]. Rare multisystem involvement was also documented in a preterm neonate with anophthalmia, microform cleft lip, subcutaneous fat herniation, split foot, solitary kidney, and patent ductus arteriosus, underlining the extensive variability of anomalies in FDH. Other reports have described unusual associations such as concurrent cardiac anomalies (hemitruncus, severe aortic hypoplasia, and atrial septal defect) in patients with PORCN variants, further broadening the clinical phenotype and emphasising the need for comprehensive evaluation [10].

Associated ophthalmological findings included chorioretinal colobomas (61%), iris colobomas (50%), microphthalmia (44%), anophthalmia (11%), cataracts (11%), and conjunctival and eyelid papillomas. [11]. Visual acuity ranged from 20/20 to no perception

of light. Structural abnormalities of kidneys and urinary system may include, hypoplastic kidney, cystic renal dysplasia, hydronephrosis, and fused/horseshoe kidney. The most common differential diagnosis for FDH is incontinentia pigmenti, Rothmund-Thomson syndrome, and Microphthalmia, Dermal Aplasia, and Sclerocornea Syndrome (MIDAS). However, the linear pattern, stages of pigmentary changes, eosinophilic spongiosis, and absence of fat herniation rules out incontinentia pigmenti. Other differential diagnosis to be considered is epidermal nevus, Bart syndrome, Adams-Oliver syndrome, ectodermal dysplasia, and congenital erosive and vesicular dermatoses. Genetic confirmation via molecular analysis of the PORCN gene- as demonstrated in this case (c.374-2del pathogenic variant)- is essential for definitive diagnosis, particularly in patients with overlapping phenotypes [12].

In the present index case, the child had cutaneous involvement which was concerning to parents, along with further investigations led to discovery of involvement of the renal system and ocular involvement. The findings were confirmed upon genetic analysis. There is currently no definitive treatment for Goltz syndrome; management is supportive and multidisciplinary, focusing on symptomatic relief and surgical correction of anomalies whenever required. Dermatologic care involves emollients, topical corticosteroids, and management of secondary infections. Nutritional support, including zinc and vitamin supplementation, is often recommended. Surgical correction of orofacial clefts and regular follow-up with ophthalmology, dermatology, genetics, and developmental paediatrics are critical to improve quality of life and developmental outcomes.

CONCLUSION(S)

The present case highlights the different typical as well as unfamiliar features of Goltz syndrome and underscores the importance of recognising the classic cutaneous and extracutaneous features of Goltz syndrome, which often present in the neonatal or early infantile period. Timely clinical recognition supported by genetic confirmation allows for appropriate counselling, multidisciplinary management, and anticipatory guidance. Although rare, early diagnosis of FDH is essential to mitigate complications and optimise long-term outcomes in affected children. Being an X-linked disorder emphasises on the importance of family genetic counselling for the family.

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