Goltz Syndrome (Focal Dermal Hypoplasia) with Unilateral Ocular, Cutaneous and Skeletal Features: A Case Report

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ABSTRACT Goltz syndrome is a rare genetic X-linked dominant condition seen commonly in females in which ectodermal and mesodermal structures – primarily skin, bones, teeth and eyes – are affected in a mosaic pattern. Around 300 cases have been reported worldwide and very few cases from India.[1],[2],[3],[4] Mutation in the PORCN gene is said to cause these defects. A 12 month old male born with hyperpigmented plaques with areas of atrophy and deformities of the digits (syndactyly and claw-like deformities) consulted for right undescended testis

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Case Report

An year old male born out of non-consanguineous marriage presented with right undescended testes and multiple generalized skin lesions since birth. He had right-sided facial deformed pinna with low set ears, patchy alopecia, multiple linear atrophied hypo- and hyper pigmented macules, multiple papilloma, syndactyly of right hand (3rd and 4th digits), ectrodactyly in right foot, hemi vertebrae, right undescended testes with hypospadiasis. Initially patient underwent right orchiopexy and skin biopsy from a nodule which was suggestive of focal dermal hypoplasia, then ophthalmic examination revealed microophthalmos with coloboma iris of right eye, hearing assessment was within normal limits. Patient then followed up for limb deformities and underwent repair of the Lobster foot.

Conclusion
Fig 3: Follow-up image after 6 months
Discussion

Goltz syndrome is a X-linked inherited disease with a probable locus at Xp11.23. The gene in this disorder has been identified as PORCN. It was first identified by Goltz et al. in 1962 with a report of three affected girls.[5] It has a multitude of clinical features having developmental skin defects with ocular, dental and skeletal anomalies. Skin involvement is considered as essential for the diagnosis of Goltz syndrome, and most of the cutaneous manifestations were seen in this case.

Skeletal defects are second most common extracutaneous abnormalities, seen in 80% of the patients that include asymmetrical involvement of hands and feet with syndactyly, polydactyly or clinodactyly.[6] This patient had limb deformities in the form of syndactyly of right hand (3rd and 4th digits), ectrodactyly in right foot, and features of hemivertebrae. Vertebral anomalies may be seen as scoliosis, kyphosis, vertebral body fusions and spina bifida. Many patients have short stature, and one side of the face or body may be underdeveloped. In the present case, facial asymmetry with right-sided hypoplasia, deformed pinna and low set ears were observed. Ocular anomalies include coloboma of iris, retina and globe; microphthalmia, anophthalmia, lacrimal duct anomalies as well as ocular muscles and corneal anomalies. Microphthalmos with coloboma of iris was observed on ophthalmic examination.

Diagnosis was clinical, as genetic study to pinpoint the exact nature of the defect could not be done due to lack of the facility. Though this condition is X-linked dominant and lethal in males, some males have been reported to survive till adulthood. This condition has to be differentiated from incontinentiapigmenti, epidermal naevi, aplasia cutis congenita and MIDAS syndrome (Microphthalmia, Dermal Aplasia and Sclerocornea) on the basis of clinical features. Early recognition of Goltz syndrome may lead to more effective.

In an Indian setup where patients' accessibility to super speciality services is poor it plays a major role for general surgeons to diagnose such rare conditions and bridge the gap.

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