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The Unilateral Focal Dermal Hypoplasia - A Case Report and Literature Review

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Abstract: ***Introduction: Focal dermal hypoplasia (FDH), formerly referred to as Goltz syndrome, is a rare multisystem condition characterised by an atypical genodermatosis which involves tissues derived from both ectoderm and mesoderm. It also affects various other tissues and organs, with the greatest impact on the connective tissue of the bones, skin, eyes, and oral cavity. Case presentation: A 7 - year - old female patient presented with a history of FDH exhibited a diverse range of systemic and oral manifestations. Extra and Intraoral examination showed Cutaneous changes along with facial asymmetry and few other oral manifestations including increase in incidence of caries and radiographic interpretation shows congenitally missing permanent teeth. Conclusion: This paper will enlighten the distinctive general and oral observations which will help the pediatric dentist to understand this rare clinical anomaly, formulating plans, and providing preventive and rehabilitative management.***

Keywords: Focal dermal hypoplasia (*FDH),* Goltz syndrome. oral manifestation, polydactyly

1. Introduction

Focal dermal hypoplasia (FDH), frequently referred to as Goltz syndrome (Goltz Gorlin Syndrome/ Congenital Ectodermal/ Mesodermal Dysplasia atrophoderma/ Linearis Maculosa/ Papillomatosis Congenitalis), was first documented in the year 1921 by Jessner. The initial instance, of histologically shown Linearis Maculosa and Papillomatosis Congenitalis condition, was documented by Lieberman in 1935.1 The documentation of this disorder, known as focal dermal hypoplasia, was given by Goltz and his coworkers in 1962.[2]

Genetic Background

FDH is an X - linked or an autosomal dominant with an X linked inherited disorder that is caused by mutations in a single gene the PORCN (porcupine homolog—Drosophila) precisely at the Xp11.23 region which is responsible for the synthesis of protein involved in the process of Wnt signalling during embryogenesis involving both ectodermal and mesodermal tissues.[3]

The prevalence of Focal dermal hypoplasia was reported to be very low (i. e) being less than 1 in 10, 00, 000, in which only 300 cases have been documented in the literature since the first report of focal dermal hypoplasia was published. Approximately 19 cases have been reported among the Indian population, with no genetic or racial predisposition identified.[4] PORCN gene, which is located on the X chromosome, explaining the higher occurrence in females (9: 1 ratio).[5] Only 10% of the published occurrences are observed in males, probably as a result of half chromatid mutations.[6] Around 95 percent have a de novo mutation, and 5 percent inherit the gene defect from their parents. The variance in severity observed among affected females can be attributed to mosaicism, which is an outcome of the Lyon hypothesis. The gene dosage imbalance between male and female was due to the presence of two X chromosomes in female (XX) whereas male have only one X chromosome (XY), This is compensated by random inactivation of one of the X chromosomes in the somatic cells of female.[7] It is also been reported to occur mostly in parents who are consanguineously married (3rd degree of consanguineous marriage).[8]

This disorder being ecto - mesodermal in origin mostly affect multiple systems in the body, namely the skin, skeletal system, eyes, and face.[3] Initially, skin involvement is characterized by erythematous lesions spread across the body. Over the years, these progress to asymmetrical Blaschko - linear patterns, pigmentary changes, reticular atrophy, and telangiectasia. The majority of patients have also reported lipomatous alterations and papillomas.[8, 9] Skeletal abnormalities are the second most prevalent extracutaneous anomaly, observed in 60 - 70% of patients.[10] Patients with FDH also exhibit dysmorphic features like facial asymmetry, a pointed chin, a large nasal tip with a narrow bridge, and maxillary hypoplasia mostly being bilateral form of FDH.[11] There have been only a limited number of previously published cases with unilateral FDH.[12]

In this case study, A 5 - year - old female pediatric patient is identified with a range of systemic and oral symptoms associated with FDH mostly appearing to be unilateral focal dermal hypoplasia.

2. Case Report

A mother of five - year - old female pediatric patient reported to the Department of Pedodontics with a chief complaint of missing upper front teeth in her child for the past 2 years. On examining the medical history it was noted that the patient was diagnosed with Focal Dermal Hypoplasia. at birth. Upon reviewing the medical history further it is found that the child was delivered prematurely (at 36 weeks) by a caesarean section exhibiting major skin abnormalities, including wrinkled skin with erythematous lesion all over the body. The patient is under regular dermatological and genetic monitoring since childhood. The Patient also gives a history of microphthalmia of left eye with corneal ulceration for which the patient has undergone ophthalmic surgery before 3 years. The family history of the patient's parents was consanguineous married (3rd degree) and had one younger brother who is apparently healthy.

The patient looked malnourished, and on physical examination the patient was underweight with a low body mass index. The dermatological condition identified was the hypochromic, type of lesions which was predominantly observed on the head, upper limbs, trunk, and the abdomen regions. Additionally, it was also observed that the patient has Nonscarring alopecia of scalp, Dystrophic nails and Polydactyly involving the left feet (Figure 1).

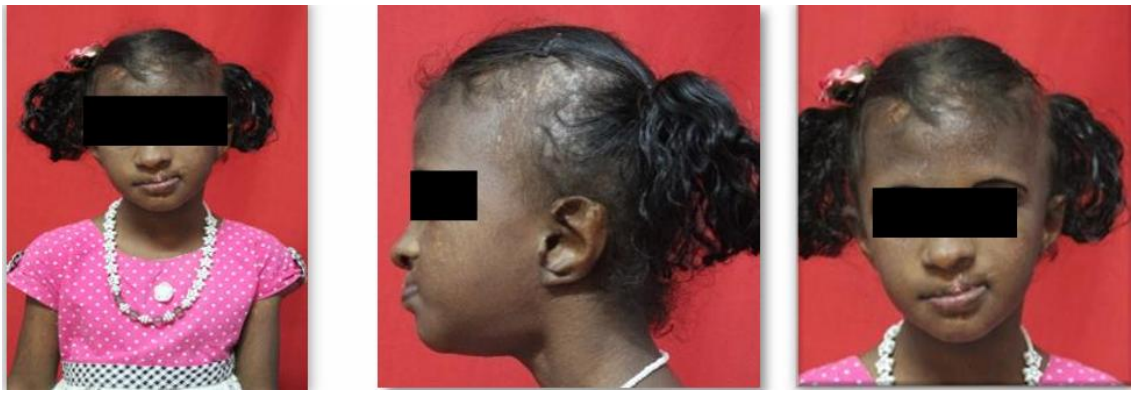

Figure 1: (A) Facial features in Goltz syndrome; **(B)** Congenitally low set left ear; **(C)** left eye blindness

On Extraoral examination of the patient revealed that patient had Facial asymmetry with convex profile, chin and nasal tip was broad and the ala of the nose was notched towards the left side along with blindness of the left eye. Eventually her left ear was congenitally malformed with cryptotia and was set low compared to the right ear, which states to be a first - degree dysplasia of the ear according to the Weerda's combined classification of auricular defects (Figure 2).

Intraoral examination revealed varies oral anomalies, like enamel hypoplasia, hypodontia, dental caries in 52, 54, 55, 63, 65, 73, 74, 75, 84, 85 (FDI system), Missing 21, fusion of 31, 32 with mild cleft on the left side in the alveolar region of 21 which was fused improperly. On soft tissue examination, a raspberry - like squamous papillomatous lesion measuring 6x6 mm on the upper lip.

On Radiographic interpretation patient has deviation of nasal septum to the right side, thin maxillary bone, prominent ante gonial notch on right and left side, generalised enamel defects and Odontodysplastic appearance of unerupted teeth. On detailed examination of each tooth shows different morphological abnormalities exhibiting Peg shaped teeth in 14, 15, 24, 2534, 35, 44 with Nolla stage 7 in 14 and 5 in 15, blunderbuss canal in 16 with Nolla stage 8, Congenitally missing 22, 23, 26, 46 Horizontally positioned 21, fused 31, 32. Radiolucency involving enamel dentin and approximating pulp in 74, 75 and also taurodontism in relation to 84, 85 (figure 3).

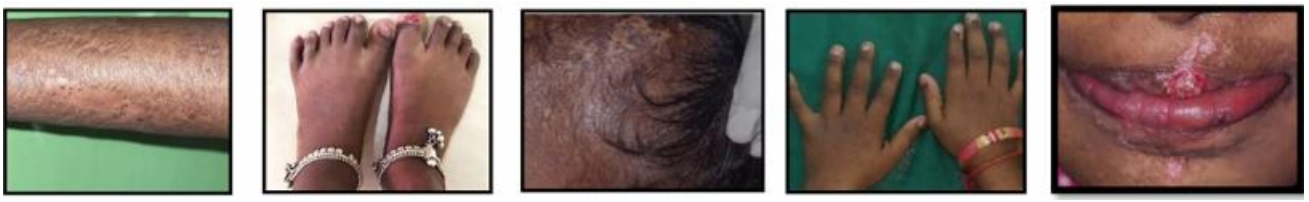

Figure 2: (A) Atrophic lesions on the hands, legs, the trunk and abdomen, **(B)** Polydactyly in the left feet, **(C)** Dystrophic nails noted, (D)Nonscarring alopecia of scalp, **(E)** Oral (raspberry-like) squamous papilloma on the upper lip

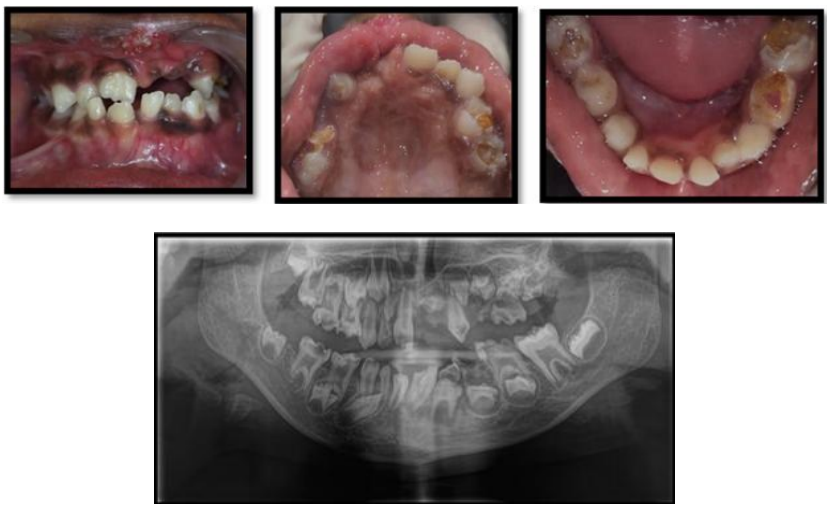

Figure 3: (A) Missing teeth – 21,22. **(B)** Partially fusion of the soft tissue noted in relation to left side of lips. **(C)** fusion of 31,32, **(D)** Orthopantomogram (OPG)

3. Discussion

Focal Dermal Hypoplasia is a characteristic systemic representation of skin, limb, ophthalmic, oral, and dental abnormalities, which are frequently seen among patients with FDH.[13]

Goltz in 1962 described Focal dermal hypoplasia, where he diagnosed 3 females reporting with severe congenital thinning of the skin and yellow papules, with herniation of adipose tissue.[14] The spectrum of severity observed in affected females is a consequence of mosaicism, which is explained by the Lyon hypothesis. Globally, focal dermal hypoplasia has been documented in 300 case reports, amongst only 9 reports had unilateral or almost unilateral type of FDH.[15, 16]

According to the literature, 70% were predominantly affected on the right side of their body, however in our instance, the left side was affected. The presence of skin involvement along with skeletal abnormalities is typical in all documented cases of FDH, hence supporting its clinical diagnosis. In our case, the child had typical unilateral features of Goltz syndrome, which is a rare fact. Although genetic examination wasn't carried out in our case, there was no signs of hereditary factors or comparable instances within their family.

The cutaneous signs of FDH consist of congenital patchy skin aplasia, which is observed in 95% of cases. Most of the patients with FDH demonstrates bullae, erythematous or urticarial lesions at birth along with skin that is atrophic and hypoplastic, typically following the lines of Blaschko which was one of the typical features in patient there were also areas appearing as depressed regions of either pink or white colour, often having a fibrous feel. In the affected skin the connective tissue is attenuated and the collagen fibres are thin and wispy, in some places these collagen and elastin are completely absent showing a transient feature of aplasia cutis congenita.[8] Typically, congenital nodular fat herniation is observed as soft, yellow - pink nodules on the skin, representing fat nodules in the dermis. These nodules are commonly found on the trunk and extremities, accounting for 60 - 70% of cases [(17).] Telangiectasias, which occur in approximately 80% of cases, can be observed on the face, trunk, and extremities. Verrucoid papilloma's, present in about 65% of cases, affect both the skin and mucous membranes.

Additionally, around 58% of individuals may experience a pebbled skin texture, while 40% may exhibit photosensitivity. Common hair manifestations of FDH include localised hair loss on the scalp (80%) and changes in the structure of hair shafts as shown through scanning electron microscopy (80 - 90%). The nail abnormalities observed in FDH consist of congenital ridged, dysplastic, or hypoplastic nails, which are present in 80 - 90% of cases. The patient exhibited hypopigmented - reticulated atrophic macules and patches in a linear pattern scattered along the lines of Blaschko on the right side of the face and right arm. Furthermore, she exhibited a scarcity of hair and experienced partial alopecia on the right side of her body, affecting her scalp, eyebrows, and eyelashes.

Skeletal abnormalities are the second most prevalent extracutaneous anomaly, observed in 60 - 70% of patients. The conditions encompassed are syndactyly, ectrodactyly, polydactyly, hypoplasia of the digits, and vertebral anomalies. [(10)] Ocular involvement often noted in patient reporting with focal dermal hypoplasia presenting microphthalmia, colobomas, and microcornea of the eye.[10]A similar clinical finding of polydactyly was diagnosed in the patient left foot.

Oral manifestations of the reported patient have congenitally missing multiple permanent teeth, raspberry - like papilloma in the upper lip, enamel hypoplasia or oligodontia, taurodontia, abnormal root morphology, and also unilateral cleft lip were noted. The patient had difficulty during mastication and in pronunciation of Linguodental and Labiodental sounds (/f/, /v/, /ph/ and /th/).

Table 1 depicts different author's reporting variant cases of unilateral focal dermal hypoplasia with varied clinical and oral manifestations comparing the similarities in our case report.

Table 1: Previously reported and current cases of unilateral or almost unilateral focal dermal hypoplasia

General consideration	Tenkir et al	Asano et al	Lee et al.	Our case
Gender	Female (4yrs)	Female	Female (19yrs)	Female (7yrs)
Involved side	Left	Right	right	Left
Atrophic patches	+	+	+	+
Alopecic patches	+	-	+	+
Dental abnormality	+	+	+	+
Ocular abnormality	+	-	+	+
Neuropsychiatric problem	-	-	-	-

The differential diagnosis of FDH encompasses several conditions, including incontinentia pigmenti, MIDAS (microphthalmia, dermal aplasia, and sclerocornea), Rothmund - Thomson syndrome, nevus lipomatosus superficialis, Adams - Oliver syndrome, and aplasia cutis.[18]

The management of FDH mostly consists of supportive care, as there is currently no efficacious treatment available. An interdisciplinary approach is essential, including the involvement of expert teams comprising a dermatologist, paediatrician, ophthalmologist, plastic surgeon, and orthopaedic surgeon[19]. On dental examination, patient had multiple caries lesion which was excavated and teeth with deep caries lesion with root resorption were also extracted based on the clinical findings and the patient was recalled for review for assessing the eruption of the other permanent successor and for aesthetic rehabilitation in near future. The patient appeared to be malnourished for which diet counselling and diet modification was advised. The psychological facets of FDH highlight the dentist's responsibilities in intervening and enhancing both the aesthetic and functional elements. This approach facilitates the patient's establishment of an elevated level of self - acceptance and encourages their assimilation into society.[20]

4. Conclusion

This case demonstrates oral manifestations associated with unilateral FDH. Identification of these features can contribute to the early diagnosis of this syndrome. The patients should be facilitated with proper genetic counseling, specific treatment and preventive procedures which involve many fields of dentistry. A multidisciplinary team approach can improve the quality of life of these children.

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