

Focal dermal hypoplasia syndrome (Goltz syndrome): the first dental case report

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Abstract

The first dental case report of a patient with focal dermal hypoplasia syndrome (FDHS) is presented. A review of signs and symptoms of FDHS is presented along with newly reported oral findings. This patient apparently is the first person with FDHS to have an absent sternum and the fifteenth person reported with confirmed osteopathia striata.

Carious lesions can be difficult to restore in FDHS due to a severe enamel hypoplasia and large pulp horns associated with this syndrome. Orofacial swellings in FDHS can be either odontogenic in origin or related to an infected skin lesion as presented in this case report.

Focal dermal hypoplasia syndrome (FDHS), also known as Goltz syndrome, is a rare congenital mesoectodermal disorder which was first described in 1962.¹ This syndrome is characterized by cutaneous defects consisting of thinning of the skin, herniations of adipose tissue in the form of yellowish papules and abnormal skin pigmentations. Patients with FDHS also may present with skeletal, dental, ocular, hair, and nail anomalies.²⁻⁶

The purpose of this report is to: (1) present the first case report of FDHS in the dental literature; (2) review histopathology of the dermal lesions; (3) document the oral manifestations of FDHS in the literature; and (4) present new oral findings associated with this syndrome.

Literature Review

In 1983 Hall⁴ conducted a review of the literature and documented 125 reported cases of FDHS in the United States, Europe, Japan, and Mexico. Although FDHS generally has been considered incompatible with survival of the male fetus,³ 12% (15/125) of Hall's reported cases were living males. Some familial relationships have been reported, yet the etiology and transmission methods still are unknown.

In 1970 Goltz suggested that FDHS is an inherited disease since 5 of 33 reported cases of FDHS had relatives with anomalies associated with the syndrome.²

The most favored genetic hypothesis is that FDHS is caused by an X-linked dominant gene of variable expressivity with a pronounced lethality in males.^{2,3,6,7-10} Alternately, an X-linked autosomal translocation or environmental teratogen might be responsible.¹¹ To date, karyotype examinations have provided no indication of chromosome alterations.^{4,12}

The first case reports in the literature of what appear to be FDHS were reported by Jessner in 1921 and 1928.^{13,14} The first patient, however, who was histologically confirmed to have what is now known as FDHS was reported by Lieberman in 1935.¹⁵ He described a 14-year-old female with hypoplasia of the teeth, wart-like lesions on the maxillary mucosa and anal orifice, papillomas on the gingiva, generalized eruptions of macular, annular, and linear areas of cutaneous atrophy, telangiectasia, and pigmentation of the skin. The patterns of cutaneous atrophy can be reticular, cribriform, punctate, or linear in shape, with reddish to tan pigmentations or areas of hypopigmentation.^{1,2,14} Microscopic evaluation of these skin lesions revealed normal epidermis with varying areas of partial to complete replacement of the connective tissue in the dermis by adipose tissue. Lieberman called this finding "lipomatosis" of the cutis.¹⁵

In 1941 Cole et al.¹⁶ reported a case of a 26-year-old female whose clinical and histologic presentation was consistent with FDHS. This patient exhibited microdontia and hypoplasia of the upper right molars and premolars, syndactyly of several toes and fingers, and a missing toe with an aplastic metatarsal bone. The patient's skin lesions were consistent with FDHS both histologically and clinically. Histologic

evaluation of patients with FDHS reveals a hypoplastic dermis with thin, sparse collagen bundles and complete absence of the elastic fibers with vascular hyperplasia and ectasia,^{5,17} which clinically produce telangiectatic atrophy. The dermis exhibits abundant quantities of adipose tissue which herniates and produces yellowish and brown papules, nodules, and plaques of the skin.¹⁸ Several authors have questioned whether adipose tissue near the epidermis is the result of the overgrowth of fat or the underdevelopment of collagen. Either or both of these could result from the profound mesodermal or ectodermal dysplasia which these patients manifest in a variety of ways.^{2,19}

In 1978 Happle²⁰ suggested that the migration of 2 populations of dermal cells might explain the linear streaking of the hypoplastic areas of skin. According to the Lyon hypothesis,²¹ only 1 X-chromosome is active in any mammalian cell. Since this is a matter of chance in each cell, 2 populations of dermal cells could arise, resulting in pigmented areas being mixed with normal areas.⁷

Osteopathia striata is a recent documented finding which may be a diagnostic feature of FDHS. This radiographic presentation, unique to this syndrome, consists of fine linear areas of dense bone arranged parallel to the long axes of bones originating at articular surfaces and extending for short distances into the diaphyses.^{6,22-24} This finding first was reported by Larregue et al. in 1972²⁵ who found a longitudinal striation in the metaphysis of long bones in 9 of 11 patients. This finding has been confirmed since by several authors, and in 1979 Knockaert and Dequeker⁶ described in the literature the fourteenth known case of osteopathia striata. They stressed the importance of a radiological survey of the skeleton for differential diagnosis of FDHS and related syndromes. This morphologically variable presentation also may be explained by the Lyon hypothesis.^{7,23,26}

In 1983 Hall noted 34 of his 125 cases presented with oral manifestations.⁴ The most commonly reported oral manifestations enumerated by Hall are dysplasia of teeth, papillomas, enamel defects with caries, oligodontia and malocclusion with irregular spacing, microdontia, gingival hypertrophy, high arched palate, hypoplasia of the mandible, defect in the alveolar ridge, notching of incisors, and median cleft of tongue. In addition to these findings, early loss of teeth,^{6,8,9,12,27-30} high frenum attachment,^{14,17,28} midline deviation,^{17,31} and ectopic eruption³² also have been reported.

Case Report

Medical History

JB is a Caucasian female born August 15, 1978, to nonconsanguineous parents. She was the product of



FIG 1. Hand and wrist radiograph illustrating congenitally missing radius, 4 metacarpal bones, and absent thumb on the left upper limb, and 2 metacarpal bones with thumb present on the right hand.

a 41-week gestation of a gravida 5/para 5 mother and weighed 2.6 kg. The pregnancy was normal and the mother denied having taken medications during her pregnancy. The family history was negative for any congenital anomalies.

The child was noted at birth to have atrophic skin lesions with an erythematous and bullous-type dermatitis producing streaking of the face, arms, and abdomen. Multiple congenital anomalies were present and consisted of low-set ears, midline cleft of the chest, dermal hypoplasia, nail dystrophy, bilateral syndactyly and oligodactyly forming a lobster claw deformity of both hands and feet, shortened extremities, and spina bifida. In addition, the left eye was microphthalmic with a purulent discharge and presented with an iris coloboma. The right eyelid also had a coloboma appearing as a punched out lesion and without lashes in its upper-middle portion. The oral cavity was remarkable for a slightly high-arched palate and a long pointed tongue which deviated to the right. There was a visible cardiac pulsation in the midline, but the heart had a normal s1 and s2 split without a murmur or gallop. A neurologic examination, including a CAT scan and electroencephalogram revealed no neurologic abnormalities. Her mental status exam was age appropriate with no sign of mental deficiency. A roentgenographic survey revealed vertebral anomalies, abnormalities of both upper and lower limbs, as well as the absence of a sternum. The left upper limb had a congenitally missing radius, short forearm, 4 metacarpal bones, and an absent thumb. The right upper limb had a normal radius and ulna with only 2 metacarpal bones and a thumb (Fig 1).

Shortly after birth JB underwent surgical closure of a supraumbilical defect of the abdomen and tho-

racic closure of the anterior chest wall defect at The Children's Hospital, Boston. JB was discharged at age 3 weeks with a head circumference and weight in the tenth percentile.



FIG 2. Radiograph of distal femur with arrow demarcating radiopaque lines of calcifications extending from the articulating surfaces suggesting osteopathia striata.

At age 2, calcified striations in the distal femoral metaphysis compatible with osteopathia striata were noted (Fig 2). She was admitted for surgical placement of a pin in her left hand and correction of the



FIG 3. Extraoral photograph of JB illustrating diffuse erythematous cribriform scar tissue over her cheeks and chin and the triangular appearance of the mandible. Note the coloboma of the right eyelid and the scarring about the left microphthalmic eye which results in a closed eyelid at rest.

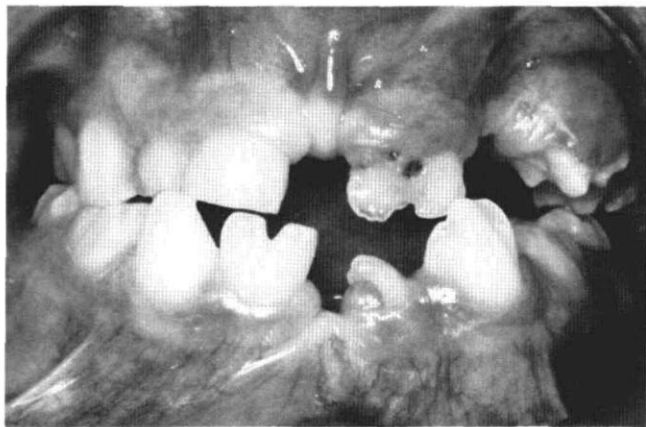


FIG 4. Intraoral photograph illustrating significant notching of the alveolar ridge in the maxillary left lateral incisor area and the mandibular midline. Minor alveolar notching is evident in the maxillary midline and mandibular left lateral incisor. Note the presence of gemination of the mandibular right primary incisor and fusion of the maxillary right primary central and lateral incisors.

syndactyly between her fingers to allow for better grasping of objects and self-feeding. At age 3, she underwent a cystoscopy with a bilateral ureteral reimplantation to correct pyelonephritic changes in both kidneys resulting from chronic urinary tract infections.

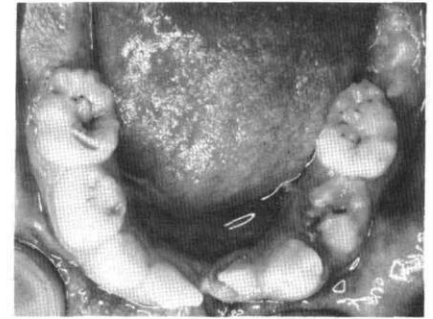
Dental Findings

JB presented to the dental clinic at The Children's Hospital at age 5 years for an initial clinical and radiographic evaluation. The extraoral examination revealed diffuse erythematous cribriform scar tissue over the cheeks and chin. Her chin was pointed, giving the mandible a triangular appearance (Fig 3). Intraorally, she presented with significant notching of the alveolar ridge in the maxillary left lateral incisor area and the mandibular midline. Minor alveolar notching was evident in the maxillary midline and the mandibular left lateral incisor area. She had a high maxillary and mandibular labial frenum attachment and a generalized gingivitis (Fig 4).

The masticatory oral mucosa was normal except for an unusual striated pattern of the palatal mucosa. The primary dentition exhibited several morphological abnormalities: microdontia of the mandibular left primary central incisor, gemination of the mandibular right primary incisor, fusion of the maxillary right primary central and lateral incisors, and mulberry-like molars and maxillary left primary canine (Figs 5a, b). The dentition exhibited numerous small to moderate carious lesions. Multiple chronic draining fistulas were noted. The radiographic survey revealed agenesis of the following permanent teeth: maxillary right first and second molars, second pre-



FIGS 5a, b. Photographs of the maxillary (*left*) and mandibular dentitions illustrating prominent caries with morphologic abnormalities: microdontia of the mandibular left primary central incisor and mulberry-like molars including the maxillary left primary canine. Note the unusual striated character of the palatal mucosa.



molars, left lateral incisor; mandibular right lateral incisor, left central, and lateral incisors. The patient also presented with an ectopically erupting mandibular left first permanent molar (Fig 6).

The patient returned to the dental department for routine restorative dentistry consisting of amalgam restorations and stainless steel crowns. The maxillary right and left primary canines, maxillary incisors, and mandibular left primary molar were extracted due to the inability to restore effectively the large carious lesions and because of pulpal involvement. JB was placed on oral penicillin (250 mg q.i.d. for 10 days) to ensure resolution of the chronic alveolar abscesses. Histological evaluation of the extracted teeth revealed large pulp chambers and decreased quantities of dentin. These findings were consistent with the diagnosis of FDHS. The decalcified sections revealed normal dentin, cementum, and pulp tissue with focal areas of carious involvement.

Four days after the extractions were performed, she presented with a low grade fever. Examination revealed an indurated erythematous swelling of the mucobuccal fold extending from the mandibular right

primary canine to the angle of the mandible. The right mandibular primary molars and first permanent molar were all sensitive to percussion, but were not mobile. Slight trismus was evident. A periapical radiograph of these teeth did not show any periapical pathology. After careful periodontal probing, a purulent exudate was noted from the buccal gingiva sulcus of the first permanent molar. Subsequent curettage resulted in approximately 5 cc of purulent exudate. The patient returned again that evening with increased swelling. At that time a resistant organism was suspected and J.B.'s antibiotic regimen was changed to oral Cephalexin (500 mg q.i.d. for 10 days).

Five days later the swelling was markedly reduced and only a small indurated 5 mm nodule just anterior to the masseter muscle was noted. One month later J.B. again developed swelling over the right mandibular region and buccal vestibule. Examination revealed skin eruptions with small pustules over her cheek. Intraorally, there was a fluctuant abscess in the sulcus opposite the mandibular right first permanent molar. Radiographic and clinical examinations did not show any pathology of odontogenic



FIG 6. Panoramic radiograph illustrates agenesis of the maxillary right first and second molars, second premolars, maxillary left lateral incisor, mandibular right lateral incisor, and left central and lateral incisors. Note the ectopic eruption of the mandibular left first permanent molar.

origin. An incision and drainage was performed and the area was decompressed. A culture was taken and sent to the lab for identification. The patient was again placed on oral Cephalexin since this was an effective antibiotic during her previous infective episode.

The laboratory culture was positive for staphylococcus aureus, streptococcus viridans, alpha hemolytic streptococcus, and haemophilus para influenza suggesting skin and respiratory flora. On follow-up examination, the intraoral swelling and the skin eruptions over her cheek had completely resolved.

Discussion

Although FDHS has not been reported previously in the dental literature, this mesoectodermal disorder has many significant dental anomalies. After reviewing the literature, a retabulation of Hall's study along with cases not included in that report which the author has collated, reveals that the occurrence of oral anomalies associated with FDHS is more frequent than previously reported. JB presented with many of these oral anomalies. Her pointed mandible, ectopically erupting molars, oligodontia of the maxillary right molars, notching of the alveolar ridge, irregular spacing of the dentition, high frenum attachments, and midline deviation all contributed to her dental malocclusion. The morphological abnormalities in the size and shape of her dentition and poor dexterity of her deformed hands made plaque control difficult, resulting in a generalized gingivitis and high caries rate.

Although "notched incisors" is an anomaly frequently associated with FDHS, according to the literature no further clinical delineation of this anomaly has been made, i.e., a Hutchinson's incisor, fusion, or gemination. JB presented with fusion of the maxillary left primary central and lateral incisor. This finding is based on the appearance of a bifid crown, 2 pulp canals, and a congenitally missing tooth confirmed by radiographic analysis.³³ The clinical and radiographic findings of the bifid crown and 1 pulp canal suggested that the mandibular right primary incisor is geminated and that agenesis of the mandibular left incisor exists. Although missing and malformed teeth occur in FDHS, other conditions, such as Down's syndrome, anhidrotic ectodermal dysplasia, chondroectodermal dysplasia, incontinentia pigmenti, and Rieger's syndrome should be considered when making a differential diagnosis.²⁸

The notching of the alveolus appears to mimic anomalies often associated with the cleft lip and palate patients. In the areas where notching occurred in the alveolar ridge, JB exhibited an obvious disturbance of the dental lamina resulting in the anomalies of agenesis, fusion, gemination, and/or microdontia

of the primary and permanent dentition. It is also interesting to note that the palatal mucosa is remarkable for a striated character which is similar in appearance to the linear streaking of the hypoplastic areas of the skin. Orofacial infections in FDHS can be either odontogenic in origin or related to an infected skin lesion. One can only speculate that the recurrent buccal space swellings noted in J.B. might have originated as a staphylococcus aureus skin infection which left a scarred residual lymph node in the anterior masseteric region. Presumably, the masseteric node became reinfected during dental manipulation and the tract led to the buccal sulcus.

Summary

A report of focal dermal hypoplasia syndrome (FDHS) is presented with a review of the literature, signs and symptoms and newly reported oral findings of fusion, and a striated character of the palatal mucosa. JB is the first person with FDHS to have an absent sternum, and is now the fifteenth person with confirmed osteopathia striata.

Although there is often a complicated medical history associated with children who present with FDHS, there is no contraindication to dental treatment. Due to the severe enamel hypoplasia and large pulp horns, carious lesions can be difficult to restore. Differential diagnosis of facial swellings should include infections of both dental origin and facial dermal lesions. The restorability of the teeth is dependent on tooth morphology and the extent of pulpal involvement. Prevention should be emphasized through dietary control, fluoride supplementation, and special consideration in plaque control. Since many of these children are of normal intelligence, esthetics becomes an important consideration in masking their physical handicap. Further considerations in dental treatment should emphasize space maintenance and/or replacement of missing teeth for esthetics and guidance of the developing occlusion which eventually will require full corrective orthodontics.

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