Focal dermal hypoplasia: management of complex dental features

L.X.G. Stephen* / N. Behardien** / P. Beighton***

A 17 year-old female who presented for treatment of grossly carious lower first molar teeth had multiple features of Focal Dermal Hypoplasia (FDH). These included enamel pitting and hypoplasia, anomalies of shape, size and positioning of teeth, as well as soft tissue papillomas and telangiectasis of tongue. A case report and a synopsis of the syndrome (FDH) are presented, the oral aspects of the condition are reviewed and the management of the complex dental anomalies is discussed.
J Clin Pediatr Dent 25(4): 259-261, 2001

INTRODUCTION

Focal Dermal Hypoplasia (FDH) (MIM 305600) is a rare genodermatosis characterised by widespread linear dermal hypoplasia and hyperpigmentation, with herniation of subcutaneous adipose tissue.[1] Telangectasia and multiple papillomas are additional features.[2] (Radiographs reveal striations in the tubular bones. Several authors have reviewed the phenotypic features of FDH.[3-5] [Table 1]. Alternative designations include Goltz syndrome, Gorlin-Goltz syndrome and congenital ectodermal and mesodermal dysplasia. The condition is inherited as an X-linked dominant trait with potential male lethality and for this reason, the vast majority of affected persons are female.

Dental involvement is an important but under-recognised component of FDH. In this article we document the dental manifestations in an affected woman, and discuss holistic management of these complications.

CASE REPORT

A female aged 17 years presented for treatment of grossly carious lower first molar teeth. She also had enamel hypoplasia, anomalies in shape, size and positioning of the teeth, intra-oral papillomas and telangiectasis of the palate and gingiva (Figure 1). She had previously received treatment at a Children's Hospital at a few months of age, and the Department of Human Genetics, University of Cape Town had established a diagnosis of FDH. Although the main reason for her attendance at the dental clinic was pain associated with carious teeth, it soon became apparent that this sensitive young female was keen to improve her dental aesthetics and overall appearance.

She had facial asymmetry, with abnormalities of the alae nasi, ear, eye and lips. Fatty herniation and cutaneous dysplasia, including telangiectasis of the skin and palate, was marked (Figures 1 and 2); linear hypopigmented areas of skin of upper limbs were also present. Squamous papillomas were evident on the palate, gingiva and perioral regions. Clinodactyly of the fifth finger of the right hand was the only obvious skeletal feature.

The patient had the following dental anomalies:

- Enamel dysplasia that was more prominent in the upper arch and also involved the first and second lower molars.
- Microdontia and abnormalities of the crowns resulting in generalised spacing and malalignment of the upper arch. (Figure 3).
- Severe periodontal angular bone loss in the distal components of the upper and lower arches, with mild crestal bone loss in upper anterior region.
- An exaggerated marginal gingival response, which was associated with the teeth that had enamel dysplasia (Figures 1 and 3).

Aims and Objectives of Dental Treatment

A comprehensive treatment plan, which included the chief complaint of painful carious teeth #36 and #46, and for the management of other carious teeth, was made. This plan included conventional oral hygiene

* L.X.G. Stephen, BCh.D, PhD, Faculty of Dentistry, University of the Western Cape.
** N. Behardien, BCh.D, Faculty of Dentistry, University of the Western Cape.
*** P. Beighton, MD, PhD, FRCP, Department of Human Genetics, University of Cape Town.
From: UWC Special Dental Clinic for Genetic Disorders, Red Cross Children's Hospital, Rondebosch, Cape Town.

Send all correspondence to Professor L. Stephen, Faculty of Dentistry, University of the Western Cape, Private Bag X 17, Mitchells Plain, 7785, Cape Town, South Africa.

Tel: +27-21-328116
Fax: +27-21-323250

Table 1. Clinical anomalies recorded as syndromic components or associated features.

1. Fatty herniation and cutaneous dysplasia.
2. Linear hyperpigmentation or hypopigmentation.
3. Focal absence of skin.
4. Telangiectasis of the skin.
5. Multiple papillomas.
6. Facial asymmetry including abnormalities of the alae nasi, ear, eye and lips.
7. Ocular abnormalities such as chorio-retinal and iris colobomata; strabismus, nystagmus, blocked tear ducts and microphthalmia.
8. Skeletal abnormalities resulting in short asymmetric stature and scoliosis. Digital abnormalities of the extremities, notably clinodactyly.
9. Oral abnormalities including papillomas, malocclusion, hypodontia and hypoplasias.

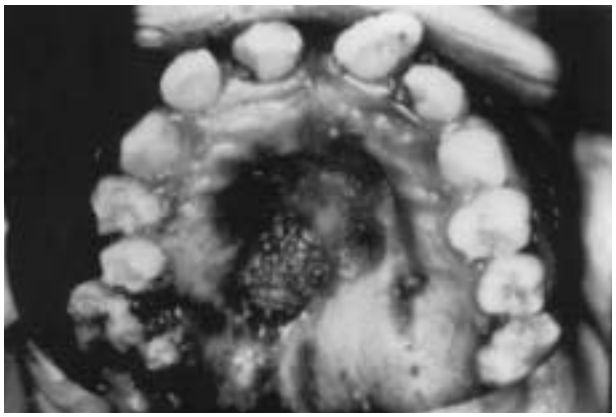

Figure 1. Enamel hypoplasia; anomalies in size, shape and number of teeth; papilloma of the palate.

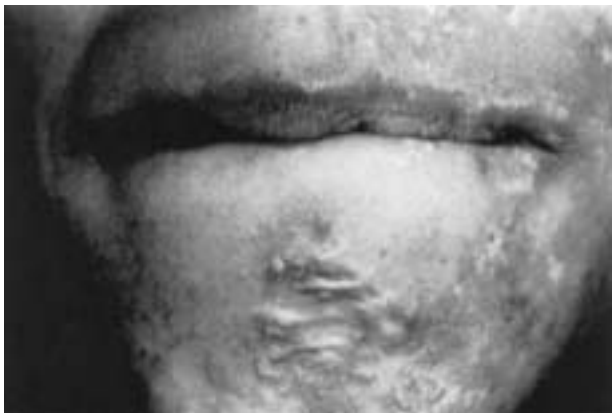

Figure 2. Fatty herniation and cutaneous dysplasia.

instruction, caries treatment, periodontal and orthodontic therapy as well as the aesthetic reconstruction of the teeth.

Initial phase of treatment

Aesthetic reconstruction of 11, 12, 21, 22, with direct composite veneers was done (Figure 4 and 5). Oral hygiene instructions and motivation, scaling and polishing were performed. Minor oral surgery: extraction of teeth 46 and 36, as well as the excisional biopsy of palatal papilloma was done. The characteristic histological features of a squamous papilloma were evident in the palatal tissue that was biopsied (Figure 6).

Secondary phase of treatment

Orthodontic treatment, viz. the de-rotation of tooth 11 and alignment of anterior teeth was done. Correction of malocclusion and stabilisation of the gingival hypoplasia was performed.

Final phase of treatment

Orthodontic maintenance was accomplished with an upper removable appliance. Placement of permanent porcelain veneers on upper anterior teeth, and stabilisation of the occlusion was accomplished as well as maintenance of the oral hygiene.

DISCUSSION

The soft tissue, skeletal and dental anomalies in this patient are in keeping with the diagnosis of FDH. Periodontal bone loss, which may or may not be a syndrome component, was present; to date no comment has appeared in the literature in this regard. Seemingly, the enamel hypoplasia had not made the teeth more susceptible to caries, but may in fact have attenuated the gingival response.

The squamous papilloma of the palate had not recurred at six months and at one year follow up; the site tended to be hemorrhagic and it was similar in clinical appearance to the interdental tissue on the buccal aspect of the 12 (Figures 4 and 5). Subsequently, this site developed into a papilloma. It is possible that areas

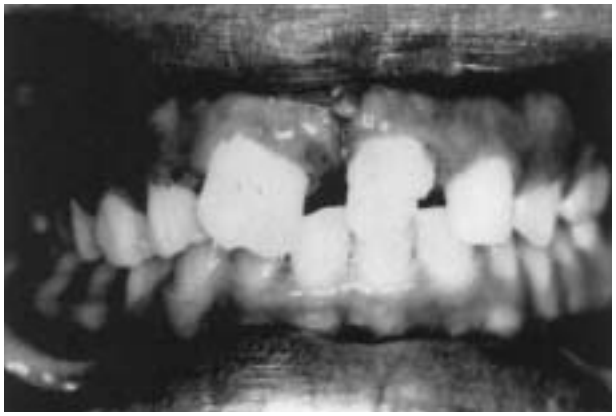

Figure 3. Dysmorphic clinical crowns and spacing of the teeth.

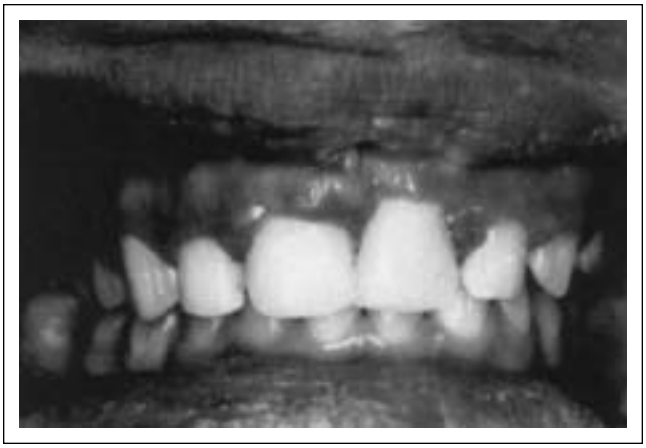

Figure 4. Well-aligned upper anterior teeth with direct composite veneers on incisors.

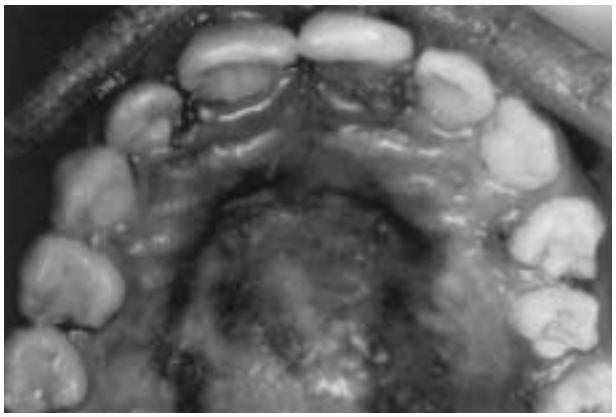

Figure 5. Well-aligned dentition with final porcelain veneers on the anterior incisor teeth. A hemorrhagic lesion is present on the palate.

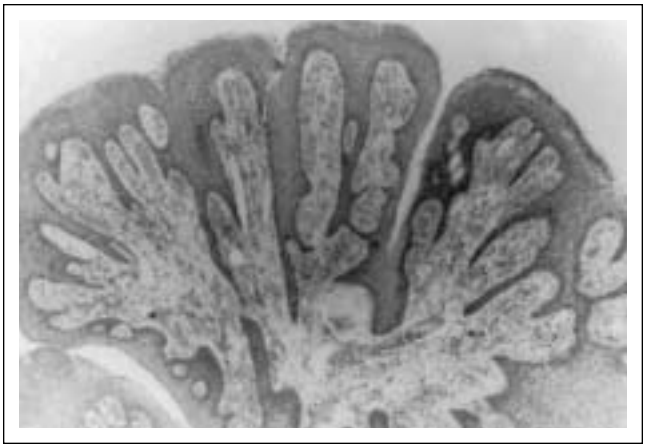

Figure 6. Histological features of a squamous papilloma - tissue biopsied from the palate.

of papillomatous outgrowths are preceded by hemorrhagic and mildly hyperplastic tissue.

Persons diagnosed with FDH at a young age need dental evaluation. Early detection of dental defects such as enamel dysplasia, microdontia and soft tissue lesions could facilitate timely preventative and/or corrective treatment planning. It is apparent that there are periods of exacerbation during the course of this syndrome. Regular surveillance from an early age with the frequency of visits increasing as needed during and after adolescence is indicated.

The psychological aspects of this syndrome emphasise the role of the dentist in intervention and improvement of aesthetics and function. This approach enables the patient to develop an enhanced sense of self-acceptance and promotes integration into society.

ACKNOWLEDGEMENTS

We are grateful to Yolanda Erasmus and Greta Beighton for preparing the manuscript with enthusiasm and diligence.

Deon Valentine of the University of the Western Cape undertook the laboratory procedures and manufacturing of the porcelain veneers; his contributions are acknowledged with gratitude.

We appreciate support received from the World Health Organisation – University of the Western Cape Collaborating Centre research fund.

REFERENCES

1. Goltz FW, Peterson WC, Gorlin RJ, Ravits HG. Focal dermal hypoplasia. Arch Derm 86: 708-717, 1962.
2. Gorlin RJ, Meskin LH, Peterson WC, Goltz RW. Focal Dermal Hypoplasia syndrome. Arch Derm Venereol 43: 421-440, 1963.
3. Goltz RW, Henderson RR, Hitch JM, Ott JE. Focal dermal hypoplasia syndrome. A review of the literature and report of two cases. Arch Derm 101: 1-11, 1970.
4. Goltz RW. Focal dermal hypoplasia, an update (editorial). Arch Derm 128: 1108-1111, 1992.
5. McNamara T, Trotman CA, Hahessy AM, Kavangh P. Focal dermal hypoplasia (Goltz-Gorlin) syndrome with taurodontism. Spec Care Dentist 16: 26-8, 1996.