International Journal of Paediatric Dentistry 2006; **16:** 440–443 DOI: 10.1111/j.1365-263x.2006.00742.x

Freeman–Sheldon syndrome: a case report

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Summary. *Background.* Freeman-Sheldon Syndrome is a rare craniofacial syndrome that has not been described in the dental literature to date. The main feature of relevance is severe microstomia, which limits access for routine dental care.
Case Report. Dental treatment was carried out successfully under local anaesthetic for a young child with Freeman-Sheldon syndrome. A novel coloured compomer material was helpful in the management of the case.
Conclusion. The importance of early referral of children with rare craniofacial anomalies to Specialist Paediatric Dental services is highlighted.

Introduction

Freeman–Sheldon syndrome (distal arthrogryposis, type 2a; OMIM #193700) was first described by E.A. Freeman and J.H. Sheldon in 1938 [1]. The term 'whistling face syndrome' was introduced in 1975 by R.M. Antley *et al.* [2].

Freeman–Sheldon is an uncommon, morphologically well-defined syndrome. A distinctive facial appearance of microstomia, microglossia, a short nose, long philtrum, H-shaped chin dimple, and sunken eyes is described. Bone anomalies, scoliosis, hand abnormalities, and joint contractures are found in association with the characteristic facies. The syndrome has also been termed 'Windmill–Vane hand' and although rare, is one of the commonest causes of multiple inherited congenital joint contractures [3]. Intelligence is usually normal, although some cases have been reported in association with mental disability [4–6]. Congenital respiratory system abnormalities and feeding problems have been documented in a number of cases [1,2,7]. Presentation of complications in adolescence has also been reported. Song *et al.* described a 13-year-old with Freeman–Sheldon syndrome who developed late-onset dysphagia and subsequent weight loss [7].

Freeman–Sheldon syndrome is a heterogeneous condition both in its presentation and in its mode of transmission and both sexes are equally affected. Early diagnosis of the condition is usually made based on the clinical appearance at birth; however, prenatal diagnosis using ultrasound has been reported [8].

Case reports of children with Freeman–Sheldon syndrome are mainly found in the anaesthetic and plastic surgery literature and only one of these documents intraoral findings [9]. To date, there are no reports in the dental literature that describe the oral aspects of Freeman–Sheldon syndrome and the associated problems in providing dental treatment.

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Case report

A 5-year-old girl was referred by her general dental practitioner to the Department of Paediatric and Preventive Dentistry, School of Clinical Dentistry, Belfast, for routine restorative care. Freeman–Sheldon syndrome was diagnosed at birth. She was the youngest of five children and the only affected sibling. There was no positive family history, and a spontaneous gene mutation was implicated in the aetiology of her condition. Her mother also reported that she had a 'hole in the heart' that had closed spontaneously and asthma for which she required daily inhalers.

The child presented with the typical facial features of Freeman–Sheldon syndrome (Fig. 1).

Her upper face was characterized by telecanthus and deep-set eyes with epicanthic folds. She had strabismus and ptosis on the right side. Her nose had a flattened nasal bridge, a wide, up-turned nasal rim, and unusual nostril morphology. She displayed marked microstomia and a prominent chin dimple. The philtrum of her upper lip was long and the

Fig. 1. Typical facial features of Freeman–Sheldon syndrome.

Cupid's bow was wide. In addition to these facial anomalies, she had finger joint abnormalities and a history of knee dislocation.

Intraorally, the upper and lower lip musculature was deeply folded, resulting in bilateral ridges of the labial mucosa (Fig. 2). The extent of mouth opening was limited and access for clinical examination was restricted. Despite this, oral hygiene was good and all primary teeth were fully erupted. Upper and lower dental arches were narrow and a high vaulted palate was noted. The tongue appeared to be of normal size. A temporary glass-ionomer restoration was present in the lower right first primary molar, and occlusal staining was evident in both the upper left primary molars and the lower left second primary molar. No other cavitation was noted clinically.

An orthopantomograph (OPT) indicated the presence of interproximal caries in the lower left and right first and second primary molars. All permanent teeth were visible on the radiograph (Fig. 3).

Management

Preventive advice was of paramount importance. Targeted diet advice was developed by means of a diet diary and oral hygiene advice and tooth brushing instruction given. Fluoride supplements had already been instituted by the referring GDP.

Restorative treatment was carried out following a period of acclimatization and introduction to topical and local anaesthetic. A full mouth prophylactic polish was carried out and fissure sealants were placed on the upper primary molar teeth, as an introduction to operative treatment. In addition, this procedure enabled ease of access and compliance for restorative care to be assessed. Minihead rotary hand-pieces and miniature burs were used to prepare minimal cavities and remove caries. Isolation was achieved with Dry Tips™ (Mölnycke, Sweden) and a slow aspirator.

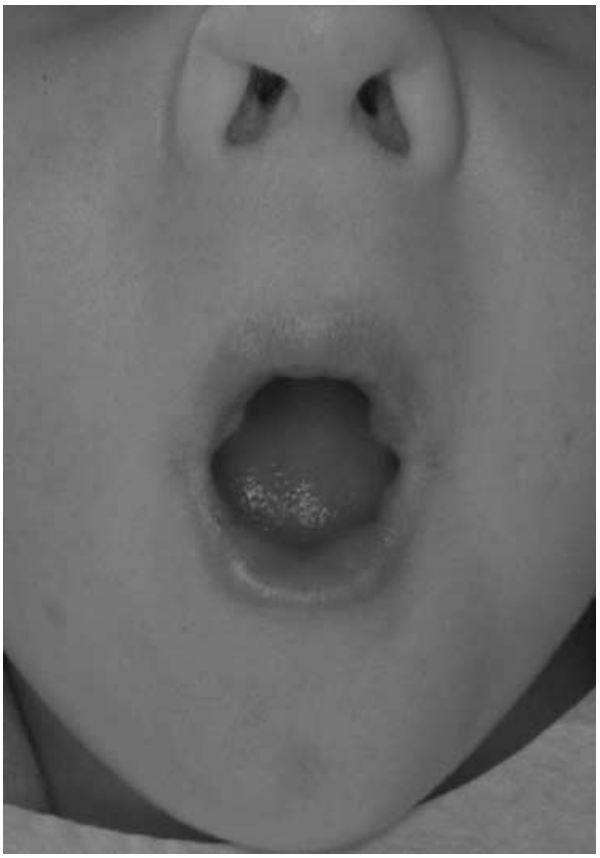

Fig. 2. Unusual nostril morphology and profound microstomia. Note ridged lip musculature, wide Cupid's bow and long philtrum.

The primary molars were restored using coloured Twinky Star™ (VOCO, Germany) compomer material with the aid of a clear matrix band. The child requested 'gold glitter' and 'orange' fillings. Treatment was completed in four visits.

The child was reviewed at 4 months and preventive advice was reinforced. At 8-month review, oral health was good and all restorations were intact. The lower left primary central incisor had exfoliated and the lower left permanent central incisor was partially erupted in the midline in a lingual position. The potential for severe crowding was evident.

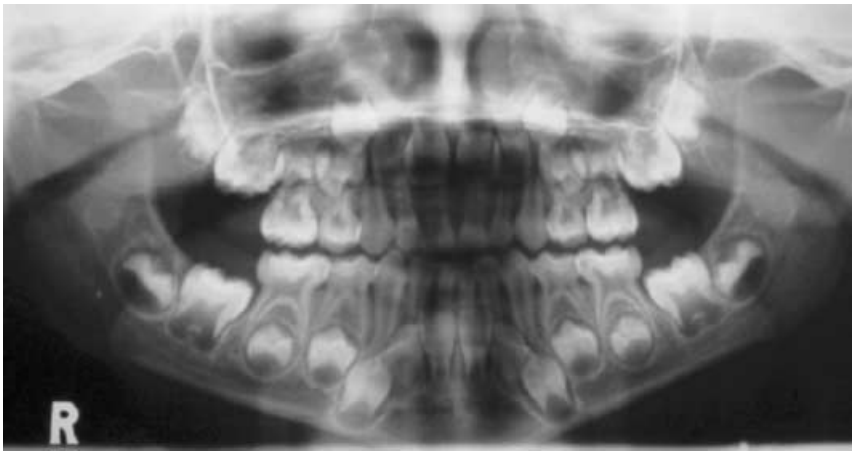

Fig. 3. Orthopantomograph (OPT) showing dental development and caries 74, 75, and 84.

Discussion

This is the first description of Freeman–Sheldon syndrome in dental literature. This report describes a spontaneous gene mutation in the aetiology of the condition. However, in most cases inheritance shows genetic heterogeneity, suggesting autosomal or X-linked recessive genes [10,11].

The individual with Freeman–Sheldon syndrome presents with a unique facial appearance and physical aspects that create a challenge when providing dental treatment. The child presented with microstomia and a high vaulted palate, as described in the criteria for diagnosis of the condition. Microglossia has also been reported as a feature [12], but this was not particularly apparent in her case.

The small mouth, normal-sized tongue, and ridged lip musculature presented a barrier for both diagnosis and treatment. The child was unable to tolerate bite-wing radiographs and so an orthopantomograph was necessary. Although this is not the ideal radiograph for detecting dental caries in young children, it allowed full assessment of dental development and demonstrated interproximal carious lesions.

Provision of restorative treatment was a challenge due to the lack of space to manoeuvre and the muscle ridges that tended to obscure visualization of the teeth both directly and with the dental mirror. Rubber dam placement was not possible.

A minihead hand-piece and mini burs were used to achieve operative access. There was no room for a high-speed aspiration tip when the hand-piece was being used, so cavity preparation was carried out in short episodes, with frequent stops to allow evacuation of saliva and water. Small size Dry Tips™ and slow suction were essential to maintain isolation for placement of the restorations.

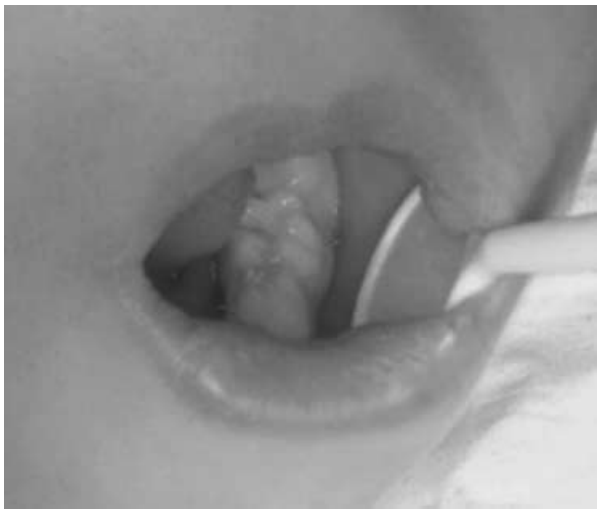

Fig. 4. 'Orange' Twinky Star compomer restorations 74 and 75 at 8-month review.

A coloured restorative material was used in this case to restore the primary molar teeth. Twinky Star™ is a compomer and is available in seven bright colours. The product is marketed as an adjunct for improving young patient cooperation, by offering novel colour choice or a 'glitter' effect filling. It is suitable for class I and II restorations in primary teeth and is light cured [13]. Compomers have been shown to have comparable longevity to amalgam in primary teeth and are superior to conventional glass ionomer materials [14,15]. In addition to the benefits afforded by an adhesive material in terms of minimal cavity preparation and fluoride release on setting, the bright colour made it easier to place where visual access was limited. The integrity of the restoration margins was easy to clinically assess at reviews appointments. In a microstomic oral cavity,

this was a particularly useful property. As clinical data on the durability of the Twinky Star™ material itself were not available, this was discussed with her parent and consent gained before proceeding. The child was delighted with her 'coloured fillings' and cooperated well at each visit.

Data do not exist on the long-term dental health of individuals with Freeman–Sheldon syndrome or the effect of their microstomia on facial development. Ohyama *et al*. presented a case in which a mouth expander was used as a nonsurgical method of correcting microstomia, prior to orthodontic treatment [9]. The authors claimed that this therapy produced an increase in mouth width. It is debatable whether this change was actually induced by mouth expander use or whether it was simply a result of normal facial growth. No other interventions to improve microstomia have been reported in relation to Freeman–Sheldon syndrome. The child already shows signs of marked crowding and her dental development and facial growth will require close monitoring.

Other congenital disorders associated with microstomia include Hutchinson–Gilford progeria, Hallerman–Streiff syndrome, Burton skeletal dysplasia, Fine–Lubinsky syndrome, and LEOPARD syndrome. Yam *et al*. highlighted the need for a multidisciplinary approach between dentists and paediatricians to improve the management and follow up of patients in whom microstomia is a prominent feature [16].

Individuals with Freeman–Sheldon syndrome and other microstomia conditions would benefit from early referral to specialist paediatric dentistry services. Regular follow-up with targeted preventive advice is essential, in view of the potential for disruption of facial growth and the anatomical limitations faced in providing oral care and restorative treatment in children with microstomia. Thorough, long-term documentation of such cases and multicentre audit will enhance our understanding and improve our future management of similar rare and interesting craniofacial anomalies.

What this paper adds

- Severe microstomia associated with Freeman-Sheldon syndrome has not been previously reported in the dental literature.

Why this paper is important to paediatric dentists

- Highlights the skills of the paediatric dentist and their pivotal role in the long-term documentation and oral care of children with rare syndromes.
- Children with microstomia present a challenge for conventional dentistry.

References

1 Freeman EA, Sheldon EN. Cranio-carpo-tarsal dystrophy: an undescribed congenital malformation. *Archives of Disability in Childhood* 1938; **13**: 277–283.

2 Antley RM, Uga N, Burzynski NJ, Baum RS, Bixler D. Diagnostic criteria for the whistling face syndrome. *Birth Defects Origin Article Series* 1975; **11**: 161–168.

3 Krakowiak PA, O'Quinn JR, Bohnsack JF, *et al*. A variant of Freeman–Sheldon syndrome maps to 11p15·5-pter. *American Journal of Human Genetics* 1997; **60**: 426–432.

4 Zampino G, Conti G, Balducci F, Moschini M, Macchiaiolo M, Mastroiacovo P. Severe form of Freeman–Sheldon syndrome associated with brain anomalies and hearing loss. *American Journal of Medical Genetics* 1996; **62**: 293–296.

5 Schrander-Stumpel C, Fryns JP, Beemer FA, Rive FA. Association of distal arthrogryposis, mental retardation, whistling face and Pierre–Robin sequence: evidence for nosologic heterogeneity. *American Journal of Medical Genetics* 1991; **38**: 557–561.

6 Hennekam RCM, Rotteveel JJ, Hall JG. Fetel hypokinesia sequence caused by maternal autoimmune disorder? (Letter). *American Journal of Medical Genetics* 1992; **43**: 1047–1048.

7 Song HR, Sarwark JF, Sauntry S, Grant J. Freeman–Sheldon syndrome (whistling face syndrome) and cranio-vertebral junction malformation producing dysphagia and weight loss. *Pediatric Neurosurgery* 1996; **24**: 272–274.

8 Robbins-Furman P, Hecht JT, Rocklin M, *et al*. Prenatal diagnosis of Freeman–Sheldon syndrome (whistling face). *Prenatal Diagnosis* 1995; **15**: 179–182.

9 Ohyama K, Susami T, Kato Y, Amano H, Kuroda T. Freeman–Sheldon syndrome: case management from 6 to 16 years. *Cleft Palate-Craniofacial Journal* 1997; **34**: 151–153.

10 Wang TS, Lin SJ. Further evidence for genetic heterogeneity of whistling face of Freeman–Sheldon syndrome in a Chinese family. *American Journal of Medical Genetics* 1987; **28**: 471–475.

11 Carakushansky G, Paiva IS, Kahn E, Ribeiro MG. Recessive type of Freeman–Sheldon syndrome – report of two affected siblings. *Journal of Pediatrics* 2001; **77**: 425–430.

12 Manji Karim P. Mbise, Roger L Generalised muscle hypertonia with mask-like face (Freeman–Sheldon syndrome) in a Tanzanian girl. *Clinical Genetics* 1998; **54**: 252–253.

13 Vanhe T, Vanden Abbeele A, Shayegan A. Twinky Star: dear parents, let's have a brand new coloured smile. *International Journal of Paediatric Dentistry* 2003; **13 (Suppl. 1)**: 5–68.

14 Marks LA, Weerheijm KL, van Amerongen WE, Groen HJ, Martens LC. Dyract versus Tytin class II restorations in primary molars: 36 months evaluation. *Caries Research* 1999; **33**: 387–392.

15 Welbury RR, Shaw AJ, Murray JJ, Gordon P, McCabe JF. Clinical evaluation of paired compomer and glass ionomer restorations in primary molars: final results after 42 months. *British Dental Journal* 2000; **189**: 93–97.

16 Yam AA, Faye M, Kane A, *et al*. Oro-dental and craniofacial anomalies in LEOPARD syndrome. *Oral Diseases* 2001; **7**: 200–202.