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Freeman-Sheldon syndrome: a case report

ABSTRACT

Background *Freeman-Sheldon syndrome is a rare hereditary disorder characterised by three basic abnormalities, namely microstomia, camptodactyly with ulnar deviation of the fingers, and clubfoot. The majority of these patients have microstomia and dental crowding, making oral hygiene difficult and increasing the risk of caries. Treatment of these patients requires a coordinated effort by a team of specialists, including a paediatrician, an anaesthesiologist, a plastic surgeon, a paediatric dentist, and an orthodontist. Herein, we describe dental problems of a child with Freeman-Sheldon syndrome and the treatment procedures performed.*

Keywords *Dental treatment; Freeman-Sheldon syndrome; Whistling face syndrome.*

Introduction

Freeman-Sheldon syndrome is a rare, hereditary musculoskeletal disorder with less than 100 cases reported in the literature. It usually occurs sporadically but as autosomal dominant, and rarely autosomal recessive cases have also been reported. Both sexes are equally affected [Ferrari et al., 2008; Bijumon and John, 2013]. This syndrome has pathognomonic skeletal and facial features. It was previously known as the whistling face syndrome due to facial muscle contraction or fibrosis and microstomia [Stevenson et al., 2006]. In patients with this syndrome, three basic abnormalities are seen in the face, hands and feet. Microstomia due to the atrophy of the buccinators muscles, camptodactyly with ulnar deviation, and bilateral club foot are the major abnormalities seen in these patients [Bosenberg, 2003]. Skeletal anomalies, scoliosis, dysphagia, growth retardation, and life threatening respiratory complications due to oropharyngeal and upper airway anomalies are other commonly encountered abnormalities. Prominent supraorbital ridges, sunken eyes, telecanthus, flat nasal bridge, long philtrum, H-shaped dimpling of the skin over the chin, and relatively flat faces are among facial characteristics of these patients. Microstomia, microglossia, narrow deep palate, and dental crowding are among the main oral manifestations [Bosenberg, 2003; Ferrari et al., 2008; Stevenson et al., 2006]. The majority of these patients have hearing limitations, delayed speech, and nasal speech due to microstomia and limited movement of soft palate [Stevenson et al., 2006]. These patients are intellectually normal but some cases of mental retardation due to central nervous system involvement have also been reported [Ferrari et al., 2008]. This syndrome is usually diagnosed at birth. In case of a positive familial history, it can be detected prepartum by ultrasonography at 20 weeks of gestation [Bijumon and John, 2013].

Overall, dental literature is scarce regarding this syndrome and the majority of published articles have discussed genetic aspects, surgical management, and the post-anaesthetic complications.

This case report describes the dental complications associated with Freeman-Sheldon syndrome, along with their management.

Case report

A 4-year-old boy was referred to the Paediatric Dentistry Department of the Shahid Beheshti School of Dentistry in January 2010, complaining of an abscess and pain in the maxillary right central incisor (Fig. 1).

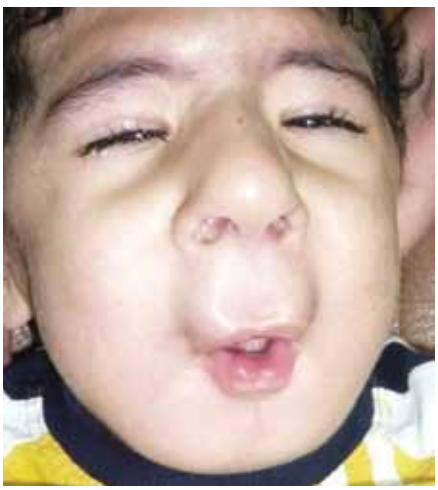

FIG. 1 Frontal view of the patient: severe microstomia, a long philtrum, telecanthus, H-shaped dimpling of the skin over the chin, sunken eyes, flat nasal bridge, abnormal nostrils and prominent supraorbital ridge.

He manifested the typical features of Freeman-Sheldon syndrome. His condition had been diagnosed at birth and he was intellectually normal. He was the second child of the family and had an older brother. There were no similar abnormalities among his other family members.

Physical examination was notable for camptodactyly with ulnar deviation (Fig. 2), clubfoot, and scoliosis. The child had recently undergone orthopaedic surgery and his legs were splinted. On facial examination, significant microstomia, a long philtrum (Fig. 1), telecanthus, sunken eyes, strabismus, a flat nasal bridge, colobomata of the nostrils, and dimpling of the chin were evident (Fig. 1).

Intraoral examination revealed a deep palate and narrow maxillary and mandibular dental arches associated with severe crowding of the mandibular teeth (Fig. 3, 4). The tongue had a normal size but upper and lower lips had deep wrinkles. Soft food regimen and limited mouth opening had led to poor oral hygiene. All primary teeth were fully erupted except for the second primary molars. The primary maxillary right first molar and the primary mandibular left first molar had occlusal and proximal caries. The maxillary central incisors and the maxillary left canine had deep carious lesions as well.

The patient had mouth breathing and nasal speech but reported no oral habits such as finger sucking or tongue thrusting.

FIG. 2 Hand deformity, camptodactyly and ulnar deviation.

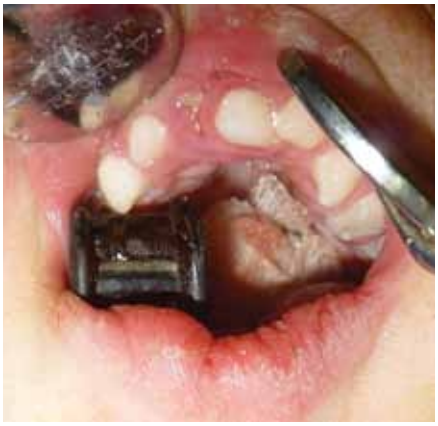

FIG. 3 Intraoral view: maxilla, deep palate, narrow dental arch.

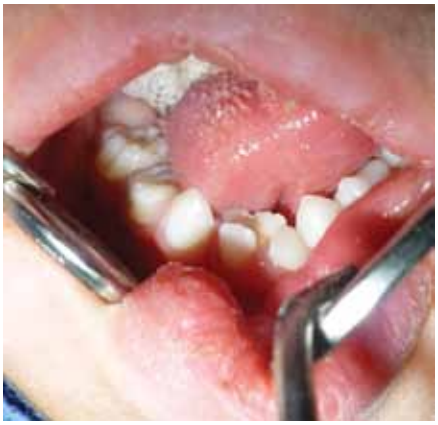

FIG. 4 Intraoral view, mandible, narrow arch, severe crowding.

Treatment

Due to the lack of cooperation and continuous head movement by the patient, we could not obtain a complete set of diagnostic radiographs and could only obtain a periapical radiograph from the maxillary anterior teeth. Other treatments were planned based on the clinical examination results alone.

Although conduction of orthopaedic surgery and fixation of the legs had significantly limited the patient's mobility, it was difficult to control him. Due to limited access to the oral cavity, cavity preparations and restorations of the decayed teeth were performed in several short visits. Pulpectomy and composite restoration of the maxillary central incisors, composite restoration of the primary maxillary left canine and pulpotomy and class II composite restoration of the maxillary right first molar and the mandibular left first molar were performed in three sessions. Due to limited mouth opening and not being able to use rubber dam for isolation, Filtek Z250 composite (3M-ESPE, St. Paul, MN, USA) along with AdheSE two-step, self-etch adhesive (Clearfil, SE Bond) were used. After restorations, fluoride therapy, and oral hygiene instruction, the patient was scheduled for a three-month follow up.

Despite precise oral health instructions and fluoride therapy, the patient returned eight months later with extensive caries in the primary maxillary right canine, primary mandibular right first molar, primary maxillary left first molar and the maxillary lateral incisors. Considering the lack of cooperation from the patient, dental treatments were performed in the operating room under general anesthesia (GA). Before undergoing dental treatment under GA, orthodontic and maxillofacial consultations were obtained to assess the need for making an impression for fabrication of an expander or a functional device. However, due to the young age of the patient, lack of cooperation, and severe muscle contractions, these treatments were postponed until after completion of

corrective surgeries on facial muscles.

Considering the risk of possible difficult intubation, malignant hyperthermia (MH), and pulmonary complications following inhalation anaesthesia in these patients, total intravenous anesthesia (TIVA) was induced using propofol. The required preventive and therapeutic procedures including composite restorations of the primary maxillary left molar, the mandibular right and left first molars, the maxillary right canine, the primary maxillary lateral incisors, as well as the fissure sealants of the primary maxillary and mandibular second molars were performed (Fig. 3, 4).

Despite great emphasis on showing up for the six-month follow up, the patient presented again after 24 months complaining of lingual eruption of permanent central incisors. At this time, the patient was using Milwaukee brace for scoliosis. Fortunately, the rate of tooth decay and caries formation had significantly slowed down due to the patient's improved oral hygiene. Only a small carious lesion in the buccal pit of the primary mandibular left second molar was seen. All restorations had an acceptable quality. Fluoride therapy was performed with fluoride gel, the decayed tooth was restored, orthodontic consultation was requested, and the primary maxillary left and the mandibular right central incisors were extracted because they were interfering with the eruption of the permanent teeth. Oral hygiene instructions were emphasised again and the patient was scheduled for a follow-up within three months.

Discussion

Freeman and Sheldon were the first to report two children with craniofacial dystrophy and the typical associated features in 1938. This anomaly was termed the Freeman-Sheldon syndrome thereafter [Al Kaissi et al., 2011], also known as cranio-carpo-tarsal syndrome, distal arthrogryposis, and whistling face syndrome [Corrigan et al., 2006]. This syndrome has sporadic, autosomal dominant, and rare autosomal recessive traits. Recent studies have shown that mutations in one or several genes that code for muscle contraction proteins play a role in the occurrence of this syndrome. However, the exact underlying mechanisms of these musculoskeletal disorders have yet to be clearly understood [Stevenson et al., 2006; Sung et al., 2003; Carakushansky et al., 2001]. Due to the lack of a positive family history, a gene mutation was suggested to have caused this disease in our patient. According to some researchers, correct diagnosis of this syndrome is difficult due to its similarity to several other distal arthrogryposis syndromes, lack of a family history, and its variable manifestations. Assessment of the genetic base of the patient may help with correct diagnosis. Other studies have reported that characteristic facial, hand, and foot symptoms are pathognomonic for this syndrome. Schwartz Jampel syndrome and chondrodysplasia myotonica are also considered in the list of differential diagnoses for this syndrome [Ferrari et al., 2008; Stevenson et al., 2006].

Due to microstomia, dental crowding, and soft food regimen, these patients have a high risk of developing caries, and dental treatments are difficult because of the small size of the oral cavity. Therefore, it is imperative to educate the parents and seek their cooperation with the child's diet and oral hygiene. Use of electric toothbrushes, fluoride mouthwashes, and dental floss is also recommended. In-office professional preventive measures including fluoride therapy, fissure sealant application, and carbon dioxide laser treatment should also be performed regularly. Regular follow-ups should be scheduled for the patient as well. Due to limited intraoral access, use of chemomechanical caries removal method and mini head handpiece can also be of great help (Bijumon and John, 2013].

Despite precise oral hygiene instructions, the patient presented with new caries at the eight-month follow-up, indicating his lack of compliance with oral hygiene. At the 24-month follow-up after treatment under GA, rate of caries development had decreased significantly. Improved patient cooperation and compliance with oral hygiene were probably due to increased age and fear of future dental treatments.

In terms of restorative treatments in these patients, restoration of posterior carious teeth with stainless steel crown is preferred over other treatment modalities. However, in our case, due to poor patient cooperation and fear of aspiration of SSC, posterior teeth were restored with composite resin (Filtek Z250, 3M-ESPE, St. Paul, MN, USA) along with a two-step self-etch adhesive system (AdheSE, Clearfil SE Bond). This adhesive system includes a primer and a self-etch bonding agent and does not require a separate etch and rinse step. It eliminates the smear layer without decalcifying dentin collagen and creates a high-strength bond and a perfect seal. The advantage of this system over the total etch system is the formation of a uniform hybrid layer, less post-operative hypersensitivity, less technical difficulty, easy application, short working time, and no need for rinsing [Xie et al., 2010]. Therefore, due to the microstomia, limited access and difficult isolation of the area, this adhesive system was used.

General anaesthesia in patients with Freeman-Sheldon syndrome is associated with increased risk of difficult intubation due to microstomia and cervical rigidity, as well as oropharyngeal and upper airway abnormalities. Similarly, these patients are at increased risk of malignant hyperthermia, post-operative pulmonary complications due to muscular disorders, risk of facial muscle rigidity in case of using halothane, and difficult intravenous (IV) access due to thickening of subcutaneous tissue [Lulu et al., 2012; Madijebara et al., 2007].

Considering the aforementioned risks, TIVA was induced with propofol, as propofol provides a deep plane of anaesthesia and has a short duration of action. Additionally, avoiding volatile anaesthetics and depolarising muscle relaxants reduce the risk malignant hyperthermia or pulmonary complications [Richa and Yazbeck, 2007]. In our case, IV access was obtained with no difficulty.

Surgical correction of microstomia in these patients is aesthetically and functionally important [Vyas et al., 2013]. In a study by Corrigan et al. [2006] expander and functional appliances were used for non-surgical correction of microstomia, leading to improved oral hygiene and easy conduction of dental procedures. More recently, Sadrimanesh et al. [2013] introduced a new treatment approach consisting of commissuroplasty and use of a retractor. We did not place an expander in our patient, even temporarily, due to his young age and lack of cooperation, and this treatment was postponed until after the surgical correction of his perioral muscles. However, patients may not respond well to conservative treatments due to muscle rigidity [Ferrari et al., 2008].

Conclusion

Freeman-Sheldon syndrome is a rare myopathic disorder with various manifestations. There is no standard treatment protocol for oral and dental problems in patients with this condition due to the variability of symptoms. Severe limitations in these patients compromise their quality of life and they need several corrective surgical procedures to overcome these limitations. Due to microstomia, and dental crowding, which make oral hygiene difficult, these patients are at increased risk of developing caries. General anaesthesia in these patients may be associated with serious complications. Treatment of patients with Freeman-Sheldon syndrome requires a multidisciplinary team including paediatricians, anesthesiologists, plastic surgeons, paediatric dentists and orthodontists.

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