Freeman-Sheldon syndrome: A case report
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Freeman-Sheldon syndrome, also called "whistling-face syndrome," is a very rare genetic condition, occurring both sporadically and by transmission through autosomal dominant or recessive mode, which affects primarily the face and skeleton. Characteristics include microstomia of the mouth, which gives the person a whistling appearance, a flat face, club feet, contracted joint muscles of the fingers and hands, and underdeveloped nose cartilage. This article describes a case of Freeman-Sheldon syndrome in a 10-year-old male. The most apparent deformities are associated with orofacial and skeletal development.

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CASE REPORT
A 10-year-old male was referred to the Department of Pedodontics, Faculty of Dentistry, Istanbul University, by his father on June 8, 2001, for management. The patient was unable to open his mouth wide enough to take solid foods and had severe dental caries. The patient is the first child of a mother aged 28 years and father aged 30 years. The patient has a healthy 8-year-old brother. The patient's birth weight was 3 kg, length was 58 cm, and head circumference was 37 cm. The facies were characteristic of Freeman-Sheldon syndrome with sloping forehead, prominent supraorbital ridge, sunken eyes, telecanthus (inner canthal distance was 2.2 cm), a flattened face, short nose, a broad nasal bridge, colobomata of the nostrils, long philtrum (1.7 cm), high arched palate, small tongue, microstomia with a characteristic whistling mouth, and H-shaped.
The face showed microstomia with protruding "whistling" lips (Fig 1). The patient's neck was short, and the hands had symmetrical clenched fingers and camptodactyly of the second, third, fourth, and fifth fingers at the metacarpophalangeal joint with contracture of the thumb (Fig 2). Both feet showed mild talipes equinovarus (Fig 3). Radiographic skeletal survey showed that scoliosis and postural deformities were present, and the subject also had inguinal hernia, kryptorchidism, and renal shape (horseshoe-shaped) anomalies. Despite his nasal speech, he communicated easily and appeared to be of normal intelligence.

The oral examination revealed malocclusion, severe dental caries, and gingival inflammation. Lateral skull findings showed a mixed dentition and delayed eruption, but otherwise findings were not within normal limits (sella-nasion-point A angle [SNA] = 76 degrees, sella-nasion-point B angle [SNB] = 68 degrees, point A-nasion-point B angle [ANB] = 8 degrees, sella-nasion angle [SN] = 57 degrees, gonion-menton angle [Go-Me] = 60 degrees) (Figs 4 and 5). Because of his severe scoliotic body shape, panoramic radiography could not be used. The long-term prognosis for the natural dentition was poor. Therefore, the primary maxillary central and lateral incisors and mandibular central incisors were extracted under general anesthesia. The extractions, even under general anesthesia, were quite difficult because of severe loss of elasticity of the lips and severe microstomia. Tissue healing was normal. His maxillary and mandibular right and left first molars were present and noncarious. Daily fluoride mouthrinse was advised for appropriate fluoride therapy, because it was too difficult for the patient to brush his teeth. Thus, if oral access is a problem, it may not be possible to use a topical fluoride. This patient also was administered antireflux medication, and a less cariogenic diet (no sugary foods or acidic drinks) was advised. The patient was advised to seek consultation from a plastic surgeon a few years following treatment.
The authors report a case with an unusual combination of multiple deformities, one of which, the "whistling face" deformity, is extremely rare. The number of reported patients diagnosed with Freeman-Sheldon syndrome is now more than 70. Freeman-Sheldon syndrome can follow either a dominant or recessive inheritance pattern. Zampino et al. described a sporadic case of the whistling-face syndrome in a male child. Similar to the current case patient, the subject was born to normal, nonconsanguineous parents.

The authors noted that it might be appropriate to speak about the Freeman-Sheldon spectrum rather than syndrome because of the different pathogenic mechanisms (muscular, skeletal, and neurologic), the wide range of clinical manifestations, and the genetic heterogeneity. Lev et al. proposed that there are several distinct syndromes with whistling face and joint contractures, one autosomal dominant with no neurologic involvement, and others recessive with varying degrees of central nervous system involvement.

The facial and skeletal features of the current case patient exhibit the characteristic features of the Freeman-Sheldon syndrome. The skull and facial bone changes include hypoplastic frontal bones with a steep floor of anterior cranial fossa and decreased anteroposterior diameter of the face, giving the appearance of a flattened midface. The obvious facial features include microstomia, hypoplastic nose, and deeply sunken eyes, which were present in the case patient. The microstomia and whistling-like position of the lips have been associated with contracture of the hypoplastic orbicularis oris muscle. Most of the features of this syndrome are due to muscle weakness. Vomiting and feeding problems also have been noted during infancy but improve with age. Severe dental caries, like in the present case, may be due to these vomiting and feeding problems.

An abnormal tongue, and almost horizontal tympanic membranes with bilateral conductive hearing loss have also been reported. In this syndrome, the tongue may be small, and the limited movement of the soft palate may cause nasal speech. Often there is an H- or Y-shaped dimpling of the skin over the chin. The authors also observed this defect in the case patient, who had an H-shaped defect of his chin. Speech therapy has been shown to be beneficial to improve tongue movement for speech and swallowing. Ohyama et al. described the care of a patient with Freeman-Sheldon syndrome over a period of 10 years. Microstomia was treated with a mouth expander for 2 to 3 hours per day before active orthodontic treatment. Depending upon the degree to which various muscles are affected, adaptive devices may be very helpful.

The most familiar congenital form of microstomia occurs in the cleft palate patient who may have a small mandible. Less common congenital conditions that may also be associated with microstomia are Freeman-Sheldon syndrome, calcinosis cutis, Raynaud's phenomenon, esophageal dysfunction sclerodactyly, and telangiectasia (CREST) syndrome, and epidermolysis bullosa.
Other conditions, with an obscure etiology, that may result in microstomia, are the collagen group of diseases, including generalized scleroderma, and submucous fibrosis. Acquired microstomia is often a consequence of burns, either by electricity, or by corrosive liquids, to the circumoral region. It may also be a consequence of surgery to the lips and facial tissues or a result of scarring following radiotherapy.

A dietary assessment should be made and a reduction in the cariogenic potential of the patient's diet should be advised. This will entail the use of a diet diary to monitor accurately the consumption of fermentable carbohydrates and acidic (fruit) drinks. For children up to 10 years of age, fluoride tablets or drops should be prescribed; older patients should use a daily fluoride mouthrinse. Because oral access is a problem, it may not be possible to use a topical fluoride, with the exception of a fluoride varnish, which can be placed on specific areas of demineralization. Clearly the reduced access to the oral cavity will make treatment difficult, especially because any form of restorative treatment requires a clear field of vision. Where access is difficult, less-bulky retracting instruments and the use of a fiber-optic light source are of great value to the operator. Similarly, small-headed mirrors, pedodontic microhandpieces, and burs should improve the field of vision and will help reduce the possibility of patient discomfort. If more advanced restorative therapy is planned, sectional trays may be used to help prepare provisional crowns and/or fixed partial dentures, thus, reducing the duration of the appointment. Endodontic therapy is difficult in these patients.

Children with this syndrome often require orthopedic or plastic surgery to correct the problems with their hands, club feet, and tight mouth. Cosmetic facial surgery can improve both function and appearance. Craniofacial surgery will also reshape the frontal bone and increase eyelid openings. But anesthesia is quite difficult in these cases. There are many reports about the anesthetic and airway management of the patients with Freeman-Sheldon syndrome.

**CONCLUSION**

This article presented a case report of a 10-year-old boy with Freeman-Sheldon syndrome. The principles involved in the treatment of patients with limited oral opening have been outlined. It is clear that oral preventive care is important, but, in those patients who require clinical care, a great deal of patience and some ingenuity is required to achieve successful results. It is essential to assess each case individually because some patients may actually suffer if any restorative or preventive treatments are attempted. This clear information on the underlying cause of the microstomia is required before a treatment plan can be formulated.

**REFERENCES**