Freeman-Sheldon Syndrome: A Rare Case Report with Dental Perspective

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Abstract: Freeman-Sheldon syndrome is a rare form of multiple congenital contracture syndromes (arthrogryposes) and is the most severe form of distal arthrogryposis. The main skeletal malformations include camptodactyly with ulnar deviation and talipes equinovarus while facial characteristics include deep-sunken eyes with hypertelorism, increased philtrum length, small nose and nostrils, and a small mouth. Here we report a rare case of Freeman-Sheldon syndrome (FSS) in an 8-years-old patient giving emphasis on the dental management of FSS.

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Introduction

Freeman-Sheldon syndrome (FSS) formerly known as whistling face syndrome is often autosomal dominant is a genetic disorder. Distal arthrogryposis (DA) includes a consistent pattern of distal joint (i.e. hands and feet) involvement, limited proximal joint involvement, autosomal dominant inheritance, reduced penetrance, and variable expressivity. DA syndromes were classified into 10 hierarchically related disorders (i.e. DA1–DA10). The prototypic DA is called DA type 1 which is characterized by contractures of the distal joints of the hands and feet, usually camptodactyly and clubfoot, respectively, without involvement of the facial muscles or other organ systems (Freeman and Sheldon, 1938; Bijumon and Johns, 2013). Freeman-Sheldon syndrome, also termed as distal arthrogryposis type 2A (DA2A), craniocarpotarsal dysplasia, cranio-carpo-tarsal syndrome, windmill vane hand syndrome, or whistling face syndrome, was originally described by Freeman and Sheldon in 1938 (Millner, 1991).

FSS is phenotypically similar to DA1. In addition to contractures of the hands and feet, FSS is characterized by oropharyngeal abnormalities, scoliosis, and a distinctive face that includes a very small oral orifice (often only a few millimeters in diameter at birth), puckered lips, and an H-shaped dimple of the chin; hence, FSS has been called "whistling face syndrome". The limb phenotypes of DA1 and FSS may be so similar that they can only be distinguished by the differences in facial morphology (Sung et al., 2003).

Here we report a rare case of Freeman-Sheldon syndrome with all the characteristic features in an 8-years-old patient. Here, we have also given emphasis on the dental management of FSS. The presence of decreased mouth opening and crowding of dentition makes oral hygiene maintenance difficult.

Case report

An 8-years-old male patient reported to the Department of Oral Medicine and Radiology with a chief complaint of multiple decayed teeth and difficulty in mouth opening. During the course of history taking his family history was non-contributory with no abnormalities observed in parents and younger brother. His height was 147 cm and weight was 35 kg. His face was expressionless (Figure 1), and his mouth puckered as he was whistling. His neck movement was limited in flexion and extension. General physical examination revealed hand deformities including ulnar deviation of fingers, campylodactyly (Figure 2). The patient had scoliosis (Figure 3), skin over his wrists was smooth with stiff elbows and ankles.

Oral examination revealed high narrow palate, microsomia and microglossia (Figure 4). Due to small tongue and limited movement of soft palate had caused nasal speech. Hard tissue examination revealed multiple carious teeth and poor oral hygiene. Panoramic radiograph was taken which revealed mixed dentition and confirmed the clinical dental findings (Figure 5).

On the basis of clinical and radiological examination, patient was diagnosed as a case of Freeman-Sheldon syndrome. Patient parents were counselled regarding the features and complications of this disorder. Genetic studies were then performed which confirmed the genetic association. Genetic testing of the *MYH3* gene can confirm the diagnosis. Patient had no known

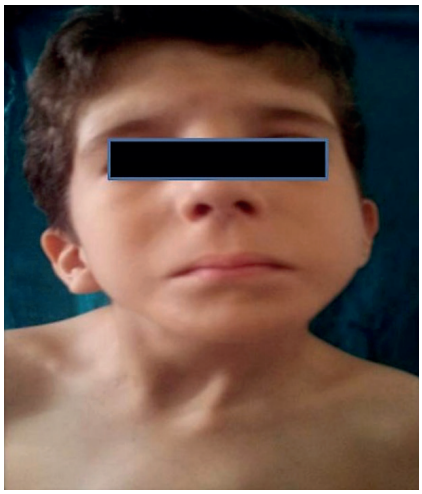

Figure 1: Expressionless face of the patient.

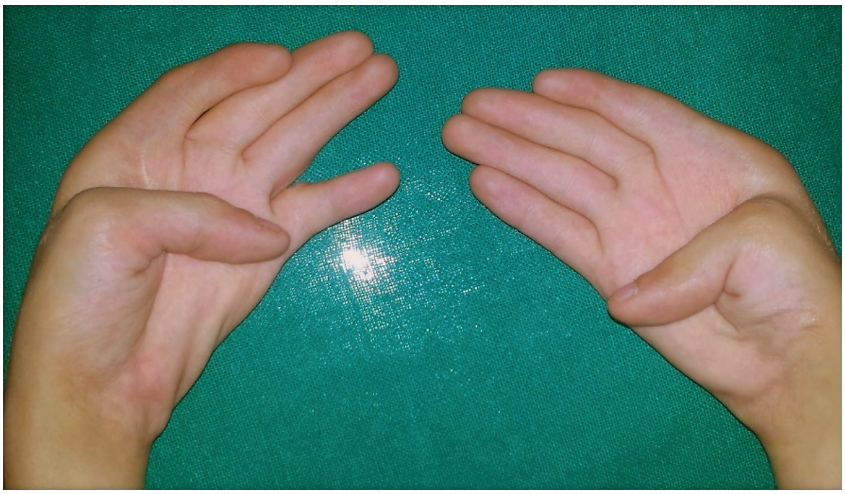

Figure 2: Revealing hand deformities including ulnar deviation of fingers, campylodactyly.

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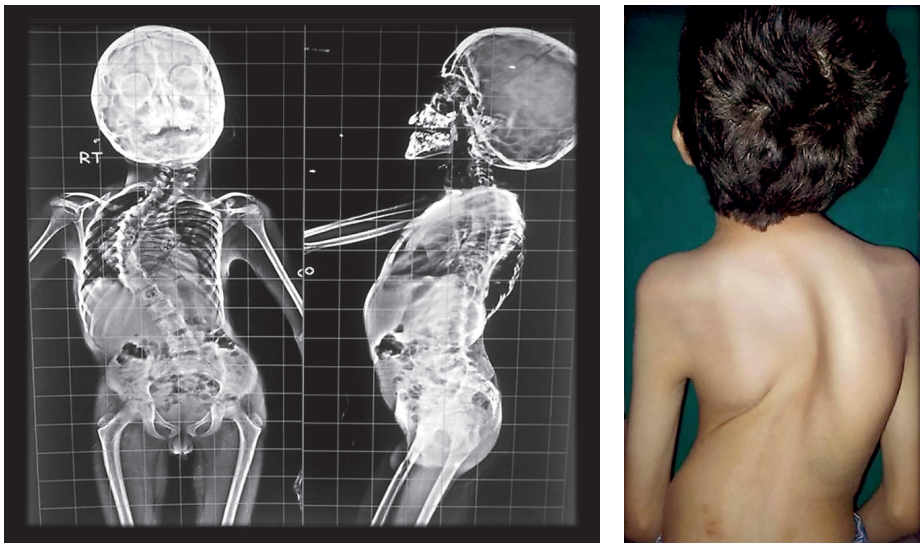

Figure 3: Presence of scoliosis.

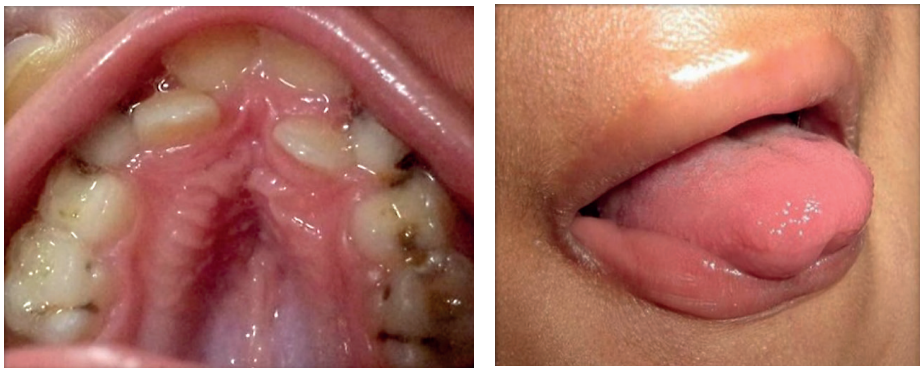

Figure 4: Oral examination revealing high narrow palate, microsomia and macroglossia.

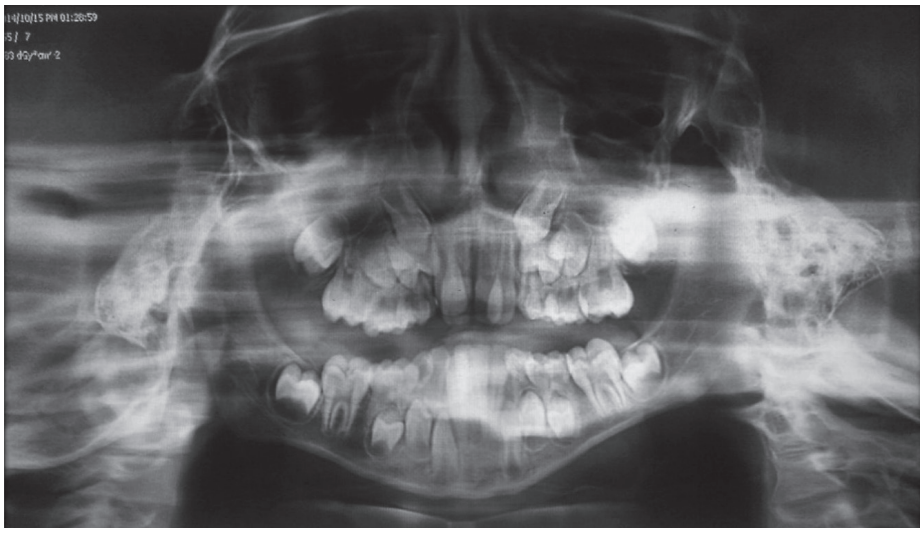

Figure 5: Panoramic radiograph showing crowding of teeth and multiple carious teeth.

family history of congenital abnormalities or consanguinity.

A thorough oral prophylaxis was performed, and rehabilitation of all carious lesions was done by composite tooth colour restorative material. Antimicrobial therapy and topical fluoride varnish was applied. Diet modifications and instructions were given to improve dental health. Emphasis was given for a recall visit of 3 months.

Discussion

The incidence of the disease is rare, and less than 100 cases have been reported in literature. In FSS, inheritance may be either autosomal dominant, most often demonstrated or autosomal recessive (MIM 277720). Males and females are affected in equal numbers. Some individuals present with minimal malformation; rarely patients have died during infancy as a result of severe central nervous systemic involvement or respiratory complications. Several syndromes are related to the Freeman-Sheldon syndrome spectrum, but more information is required before undertaking such nosological delineation (Buyukavci et al., 2005).

Diagnosis is made by clinical examination. Typical facial features, camptodactyly with ulnar deviation of fingers, and bilateral clubfeet are the fundamental findings in this syndrome. The other variable features could be scoliosis, articular stiffness, supra-orbital swelling, hypotrophic (myopathic) musculature, delayed milestones and short stature, hypertelorism, epicanthus, telecanthus, prominence of the supra-ciliary arches, hypoplasia of the nasal alae, low-set ears, contracture of the thumb in adduction, spina bifida occulta, and dysplasia or congenital dislocation of the hip (Mayhew, 1993). The orofacial findings include long philtrum, mouth puckering, microstomia, H-shaped chin dimple and microglossia (Laishley and Roy, 1986).

The diagnostic criteria for classical FSS comprise: the presence of >/2 of the major clinical manifestations of DA (ulnar deviation of the wrists and fingers, camptodactyly, hypoplastic and/or absent flexion creases, and/or overriding fingers at birth, talipes equinovarus and calcaneovalgus deformities, a vertical talus, and/or metatarsus varus) plus the presence of a small pinched mouth, prominent nasolabial folds, H-shaped dimpling of the chin (Poling and Dufresne, 2022). The presented patient fulfilled criteria for FSS.

For treatment, patients must have early consultation with craniofacial and orthopaedic surgeons, when craniofacial; clubfoot or hand correction is indicated to improve function or aesthetics. When operative measures are to be undertaken, they should be planned early in life to minimize developmental delays and negate the necessity of relearning basic functions. Due to the abnormal muscle physiology in Freeman-Sheldon syndrome, therapeutic measures may have unfavourable outcomes (Sehrawat et al., 2021). Difficult endotracheal intubations and vein access complicate operative decisions in many DA2A patients, and malignant hyperthermia (MH) may affect individuals with FSS, as well. Reports have been published about spina bifida occulta in anaesthesia management and cervical kyphoscoliosis in intubations (Poling and Dufresne, 2018).

Dental crowding is a universal finding in FSS patients. Several individuals required extraction of multiple teeth because of crowding and/or treatment for malocclusion. The presence of microstomia further exacerbated dental care, because access to the oral cavity for routine hygiene and treatment procedures becomes more difficult (Sen et al., 2021). Small oral openings necessitate the use of powered tooth brushes. The patient and the patient's parents should be made aware of the importance of home-based preventive measures such as diet control, oral hygiene maintenance, fluoride mouth rinse, and fluoride dentifrices. Professional care such as pit and fissure sealants, oral prophylaxis, and topical fluoride application must be provided (Biria et al., 2015).

Conclusion

FSS syndrome is a rare disorder which not only causes physical abnormalities but also social and psychological problems. Children with this syndrome may grow up to feel alone and "defective". Parental education is required early in the child's life. Treatment is supportive and in accordance with symptoms. 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. Much more research is warranted to evaluate this apparent relationship of idiopathic hyperpyrexia, MH, and stress. Further research is wanted to determine epidemiology of psychopathology in FSS and refine therapy protocols.

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