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Klippel-Feil Syndrome: Oral and maxillofacial rehabilitation: Rare case report

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Abstract
Klippel-Feil Syndrome is a complex condition due to aberrant fusion of two or more cervical vertebrae characterised by short neck, low hairline, facial asymmetry and limited neck movement. Mandibular hypoplasia, submucous cleft, congenitally missing teeth, facial asymmetries and mouth breathing are frequently observed in patients with Klippel-Feil Syndrome. Management is symptomatic and supportive which necessitates a multidisciplinary expertise. This case report describes the dental management and prosthetic ear rehabilitation executed on a 12-year-old female patient with Klippel-Feil Syndrome.

Keywords: Klippel-Feil Syndrome (KFS), dental management, prosthetic ear rehabilitation

Introduction
Klippel-Feil Syndrome (KFS) is a rare congenital anomaly which is characterized by the abnormal fusion of two or more cervical vertebrae. It a complex bone disorder caused by the fusion of two or more cervical vertebrae and is the result of defective segmentation along the axis of the developing embryo (sclerotomes of somites) from the 2nd-8th week of gestation [1]. Classic triad of short webbed neck, limitation of neck motion and low posterior hairline are the clinical diagnostic features of a typical KFS [2]. In 1912 the first complete clinical description of this syndrome was given by Maurice Klippel and Andre Feil independently. KFS is also known by other synonyms such as Cervical Fusion Syndrome or Congenital Brevicollis [3]. Prevalence of KFS is approximately 1 out of 40,000 to 42,000 new-borns with a slight predisposition for females [4]. This syndrome was initially believed to be mostly sporadic [5], but both autosomal dominant and recessive forms have recently been reported [6]. KFS can present alone or with other anomalies, such as Goldenhar Syndrome, renal ectopia and/or agenesis, Fetal Alcohol Syndrome and other abnormalities of the extremities [6-7]. We present the case report of a Klippel-Feil syndrome with oral & maxillofacial facial rehabilitation.

Case Report
A 12-year-old girl was referred to the department of Pediatric and Preventive dentistry for treatment of multiple decayed teeth and for management of unesthetic facial appearance due to abnormal right ear. She was having a known history of KFS. She was one of the monochorionic diamniotic twins (MCDA). She and her twin sister are the second daughters of a non-consanguious marriage. The twin sister and the elder sister did not have any signs and symptoms of KFS. Natal history revealed that gestation period around 37 weeks, normal delivery and birth weight was 2.25 kg suggesting low birth weight (LBW).

Medical history
She gave history of small ASD on echocardiography which got rectified eventually. Her Appearance, Pulse, Grimace, Activity and Respiration (APGAR) score was 9.

Extraoral examination

She was conscious and oriented. There was no signs of pallor, icterus, cyanosis, clubbing, lymphadenopathy and oedema. On clinical examination; she was presenting with short neck, limited neck movement, slight torticollis, Sprengel's deformity (Figure 1a and b) where the scapulae (shoulder blades) are hypoplastic, undescended and positioned high on one side of body causing weakness of the shoulders, facial asymmetry, a low posterior hairline. She was presenting with deformed dysplastic right ear which was grade III microtia with no external auditory canal (Figure 1b), hearing issues, mandibular hypoplasia, restricted mouth opening.

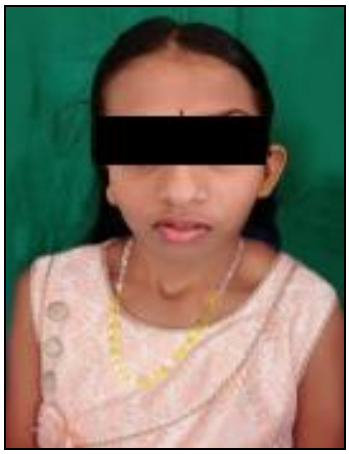

Fig 1a: Extra oral image

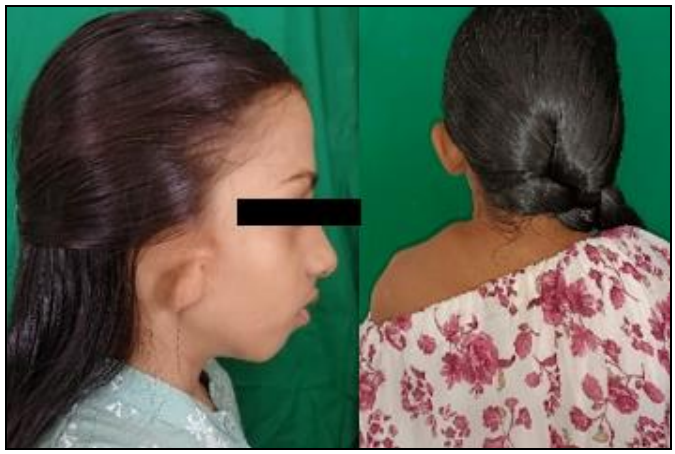

Fig 1b: Grade III microtia and Sprengel's deformity

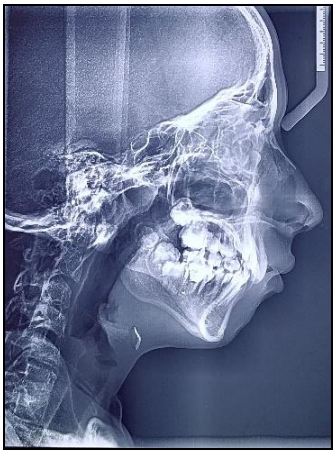

Fig 1c: Lateral cephalogram showing cervical vertebrae fusion

Intraoral examination

Patient was in late phases of second transitional mixed dentition period. On intraoral examination, she was having poor oral hygiene, multiple carious teeth, root stumps and pulp exposed permanent mandibular molars. She was presented with crossbite of upper left lateral incisor, high arched palate and severe dental crowding. She was also presenting with commissural lip pit on left side (Figure 3).

Radiographic findings

OPG showed reduced height of body of mandible, pulp exposed permanent mandibular molars with open apices (Figure 2).

General management

Management is symptomatic and may include surgery to correct craniocervical instability and spinal cord constriction. She is undergoing physiotherapy for shoulder muscle strengthening.

Dental management

The treatment plan focused on the prevention of dental caries and periodontal disease. Grossly decayed teeth which are not restorable and root stumps were extracted under local anaesthesia. Pulp exposed permanent mandibular molars managed by apexification treatment using calcium hydroxide iodoform paste. Multiple carious teeth were restored with GIC due to its fluoride releasing property. As a part of preventive treatment measures, supragingival oral prophylaxis and topical fluoride applications were carried out and proper brushing technique was demonstrated. Diet counselling and appropriate home care instructions with special emphasis on importance of oral hygiene were also given. Patient was kept on regular maintenance phase with special emphasis given for orthodontic treatment. (Figure 4).

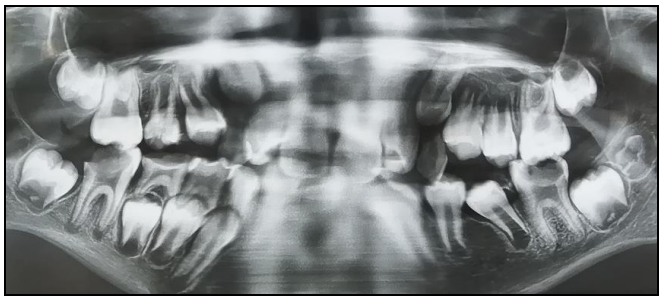

Fig 2: OPG

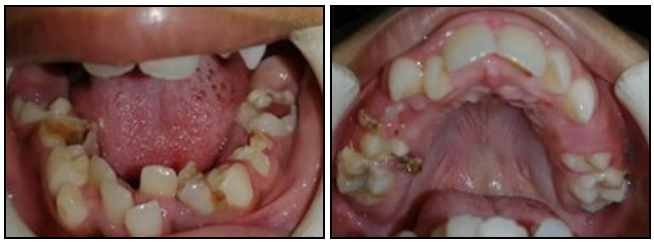

Fig 3: Pre-operative intraoral image

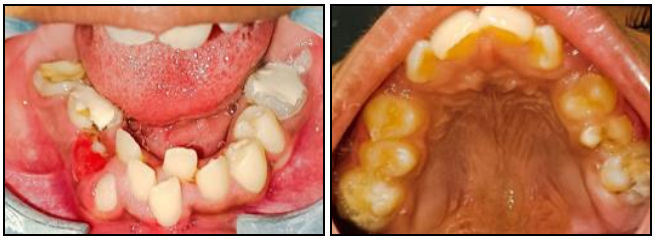

Fig 4: Post-operative intraoral image

Prosthetic rehabilitation of ear

Phase 1-Impression phase

Impression of the residual tissue was made by injecting irreversible hydrocolloid material. To support the alginate impression material, plaster of Paris was mixed and spread on the impression immediately (Figure 5). The impression was poured immediately with type III gypsum. Impression of the patient's twin's right ear was also made after blocking

external auditory meatus with cotton. Impression was made by injecting light body around helical portions of the ear and supported by heavy body putty.

Phase 2-Modeling and wax trial

Wax pattern (Hindustan modelling wax no. 2) of the right ear was done using the template ear model and adapted to the working cast. The pattern was tried on the patient (Figure 6). Sculpted ears were tried for verification of dimension, esthetics, angulations and orientation, wax patterns of ears were attached firmly onto the working cast and sealed.

Phase 3-Investing and de-waxing phase

The wax patterns sealed in the master cast were then invested using type III gypsum material. The entire mould was boxed.

Phase 4-Shade matching and Packing

Shade matching was done using trial and error basis. Once the patient and parents were satisfied with shade, room temperature vulcanization silicone (Factor II) was mixed in appropriate proportions with the selected shade and loaded into the mould.

Phase 5-Fabrication& insertion of final prosthesis

The prosthesis was allowed to vulcanize for 24 hours in room temperature. The vulcanized prosthesis was retrieved, trimmed and polished using silicone trimming bur. It was tried on the patient for colour, translucency and an appropriate location and position was marked to attach with the hairband. Extrinsic stains were used to give the prostheses a close to natural look, thus improving esthetics. After reassuring the final shade, position and angulation it was bonded to the hairband using silicone platinum primer. Retention was adequate in final stage (Figure 7).

Phase 6-Maintaince phase

The patient and her parents were educated about the maintenance and use of the prostheses. The patient was recalled for review after 1 week. She was very comfortable. She was then periodically reviewed and asked to report back in case of any problems.

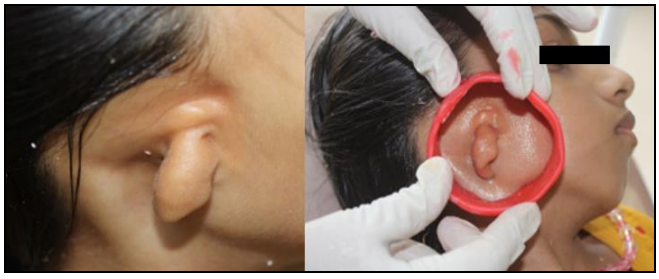

Fig 5: Impression phase

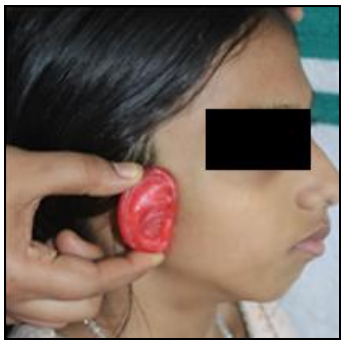

Fig 6: Wax trial phase

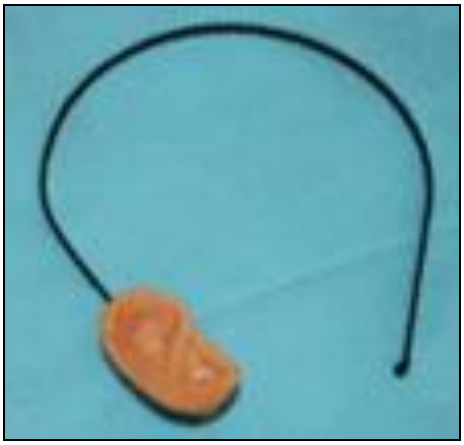

Fig 7: Prosthesis bonded to hairband

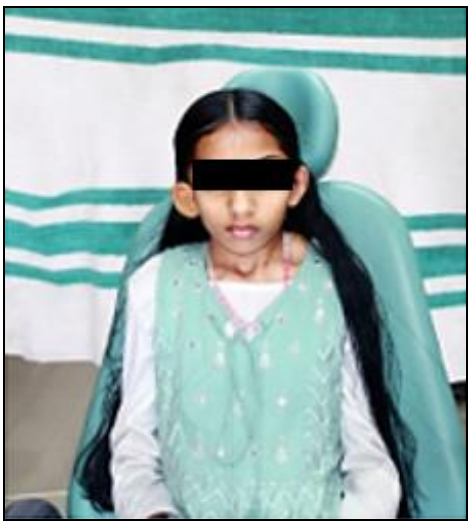

Fig 8: Post-operative image with prosthetic ear

Discussion

Klippel-Feil Syndrome (KFS) is defined as a congenital aberrant fusion of two or more cervical vertebrae and characterized by the presence of classical triad including short webbed-neck, restricted movement and low hairline. Feil classified this syndrome into 3 types: Type 1 cases present with fusion of cervical vertebrae; Type 2 cases present with fusion of cervical and thoracic vertebrae, along with an associated cervical hemi vertebrae anomaly and abnormal fusion of the atlanto-axial joint with the occiput. Type 3 includes cases with fusion of cervical, thoracic or lumbar vertebrae with associated rib anomalies [2]. Clarke *et al*. further updated this classification by associated anomalies, grouped patterns of inheritance, and the axial level of the most anterior fusion.

Mutations in the genes *GDF6*, *GDF* 3, and *MEOX1* can cause KFS and can be inherited either by autosomal dominant or recessive type [8]. Mutations in GDF6 and GDF3 genes are autosomal dominant, and are classified as KFS types 1 and 3, while mutations in MEOX1 and MYO18B genes are autosomal recessive and are called KFS types 2 and 4 respectively [9].

About 50% of KFS patients have a combination of a short neck, restricted neck movement and a low posterior hairline. Most common skeletal abnormalities include kyphosis or scoliosis (60%), torticollis (20%), Sprengel deformity (20%), and facial asymmetry (20%). Hearing loss (30%), genitourinary system anomalies (35-65%) and congenital heart defects (5-15%) are among the most common systemic

abnormalities coexisting with KFS [10]. Common heart problems are ventricular septal defects and aortic arch abnormalities. In our case, patient was diagnosed with ASD which got rectified eventually. Mandibular hypoplasia occurs in a small number of patients with KFS and may contribute to upper airway obstruction, resulting in obstructive sleep apnea. Submucous cleft, congenitally missing teeth, facial asymmetries and mouth breathing are frequently observed in patients with KFS. Moore *et al*. reported the association of many renal diseases to this syndrome and showed that 64% of KFS patients had significant genitourinary system anomalies. But in our case, patient was free from genitourinary anomalies.

Management of KFS is symptomatic and may include surgery to correct craniocervical instability and spinal cord constriction. Physiotherapy can also be useful. In accordance with previous studies, severe mandibular retrusion can be treated with orthognathic surgery or distraction osteogenesis. In this patient prosthetic rehabilitation of right ear with microtia was done. Other treatment options available are the surgical reconstruction with Costal Cartilage Graft, a safe approach that has been practised since the 1920's. Medpor graft surgery which uses a synthetic prefabricated porous polyethylene scaffold, was also an alternative method.

Conclusion

KFS may be noticed in childhood itself, but some of them are also identified in adulthood stage. Modern medicine along with multidisciplinary team work can improve the quality of life among these patients. The present case report highlights the conservative management of oral & maxillofacial facial problems by interdisciplinary approach, thus improving functional, aesthetic and psychological wellbeing of the patient.

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