Case Report

Klippel–Feil syndrome: A rare case and its management

ABSTRACT

A relatively smaller number of orthodontic patients are affected by known craniofacial anomalies that affect oral and dental tissues. The greatest advantage in knowing that a patient has a particular syndrome is that it allows a much better prediction of future development that might happen in such individual and necessary precautions to be taken while management of these cases. This case report provides features of a patient with Klippel–Feil syndrome and orthosurgical management of an adult male to effectively improve patient esthetics and function.

Keywords: Cervical vertebrae, Class III malocclusion, Klippel–Feil syndrome

INTRODUCTION

Cervical vertebral segmentation anomalies are referred to as Klippel–Feil anomaly where there is an involvement of fusion of two segments or the entire cervical spine. Klippel–Feil syndrome (KFS) was first reported by Maurice Klippel and Andre Feil in 1912,[1,2] with characteristic features of congenital fusion of at least two of seven cervical vertebrae in the cervical spine, with a short neck, limitations to lateral movement of the head or neck, and a low posterior hairline.[3] However, in many cases, fusion or anomaly of the vertebrae may be apparent in the thoracic or lumbar spine. Additional anomalies include deafness, elevation of the scapula, and congenital heart defects. The incidence of KFS is approximately 1:40,000–42,000, and 60% of cases occur in females.[4-6] When the symptoms of a patient with KFS are minor, these symptoms may be unnoticed. Thus, the condition remains undiagnosed, and the person may lead a normal life. Conversely, dentists may incidentally confront an anomaly of the cervical spine associated with KFS on routine radiological examinations. This case report describes an anomaly of the cervical vertebrae that was first seen on orthodontic examination of a 21-year-old adult male with skeletal Class III malocclusion.

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CASE REPORT

A 21-year-old adult male reported to the department of orthodontics and dentofacial orthopedics with a chief complaint of inability to eat food as his upper front teeth were not meeting with lower front teeth.

During evaluation, he had a history of jaundice at the age of 10 years, and he was operated on for inguinal hernia 2 years back. On general physical examination, he had a good built with normal overall physical and mental development.

He exhibited dolichocephalic head, leptoprosopic facial form, a concave profile with anterior divergence, and lip competency at rest. On smiling, he displayed less than full crown of the upper incisor and a nonconsonant smile. Intraoral examination revealed Angle Class III molar relationship on both sides with a reverse overjet of 3 mm and open bite of

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4 mm. Mandibular dental midline is shifted to right by 2 mm and crowding in upper and lower dentition is present. The patient had severe fluorosis in his complete dentition and restorations in 17, 27, 36, 46, and 47 [Figures 1 and 2].

On panoramic radiograph, missing 18 and impacted 38 and 48 were found. On lateral cephalometric examination, along with skeletal Class III relationship and open bite, we also reported fused cervical vertebrae C4, C5, C6, and C7 [Figure 3].

The literature reports on several severe abnormalities of vertebral column including changes of number of vertebrae and their segmentation and morphology. Changes of number and morphology of vertebrae as well as fusion of some of them result in an asymmetry of posture.

On re-examination of the patient, we found low posterior hairline and mild restriction of lateral neck movement. This significant finding instigated us for complete systemic examination of the patient. He was referred to medicine, cardiology, ENT, and orthopedics, while three-dimensional reconstructions from computed tomography verified the diagnosis of KFS along with limitations of neck movements

Table 1: Cephalometric analysis pretreatment, presurgical, and posttreatment

Parameter	Normal	Pretreatment	Presurgical	Posttreatment
Maxilla				
SNA	82°	78°	78°	81°
Na per Pt A	0 mm	−6 mm	−7 mm	−2 mm
Co to A		75 mm	75 mm	80 mm
Mandible				
SNB	80°	83°	83°	79°
Na per Pog	2 mm	4.5 mm	5 mm	1.5 mm
CO-Gn		120 mm	120 mm	114 mm
Maxilla-mandible relation				
ANB	2°	−5°	−5°	2°
WITS		−8 mm	−11 mm	−1 mm
Vertical				
FMA	25°	29°	30°	31°
SN to GoGn	32°	35°	36°	37°
Bjork Sum	396°	387°	388°	389°
Jarabak ratio	62%-65%	68%	67.5%	67%
Dental				
1 to NA	22°, 4 mm	31°, 7 mm	24°, 4 mm	24°,4 mm
1 to SN	102°	108°	102°	102°
1 to NB	25°, 4 mm	23°, 4 mm	22°, 4 mm	22°,4 mm
Interincisal angle	131°	122°	131°	131°
IMPA	90°	87°	90°	91°
S line to upper lip	−2-2	−1 mm	−3 mm	1 mm
S line to lower lip	−2-2	1 mm	3 mm	−1 mm

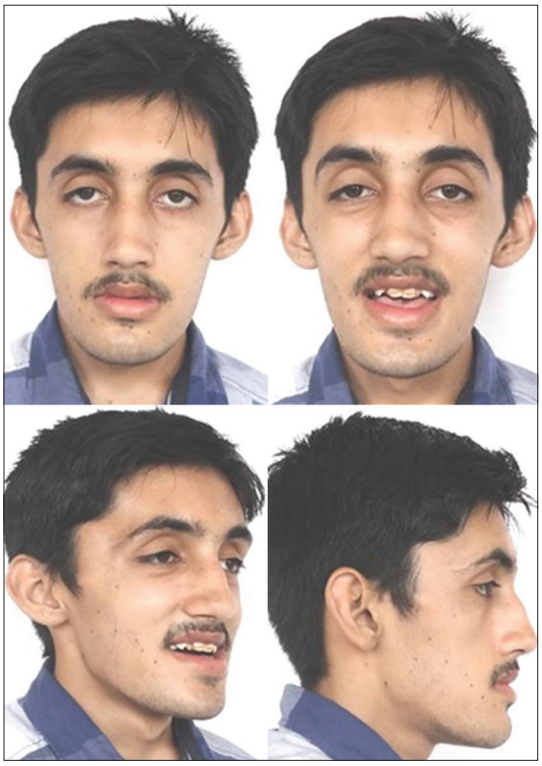

Figure 1: Pretreatment extraoral photographs

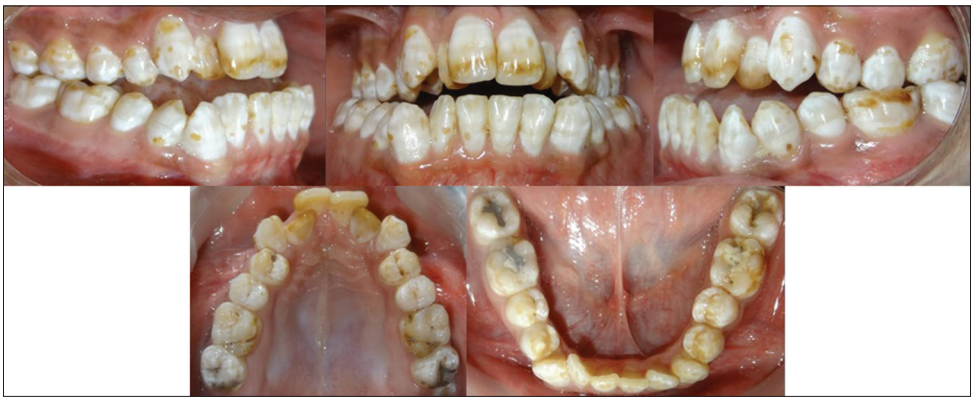

Figure 2: Pretreatment intraoral photographs

and low posterior hairline. Although no treatment was necessary at this point, he was advised to do regular follow-ups for possible future neurological deficits.

Cephalometric analysis indicated Class III jaw bases on the account of the retrognathic maxilla and prognathic mandible with underlying average growth pattern. The upper incisors were proclined, whereas the lower incisors were normal.

Treatment objectives were to achieve normal and pleasant profile, to achieve ideal overjet and overbite, Class I molar relationship on both sides, and to achieve normal inclination of the upper incisors.

Treatment plan

Keeping the above objectives in mind, treatment was planned to carry out in three phases. The first phase was decompensation of maxillary and mandibular dentition by extraction of maxillary first premolars and lower incisor extraction. The second phase was orthognathic bi-jaw surgery including Le Fort advancement of the maxilla and bilateral sagittal split osteotomy (BSSO) setback of the mandible. This was followed by postsurgical orthodontics including settling of occlusion.

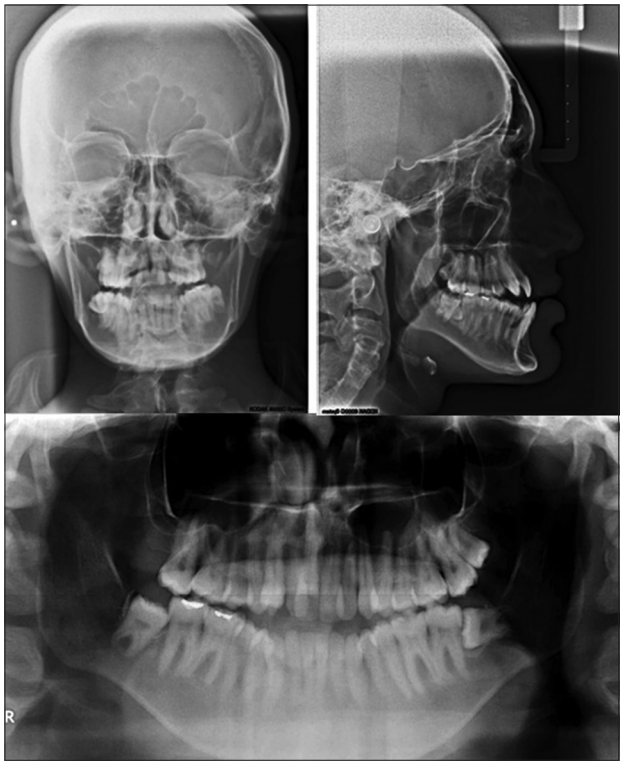

Figure 3: Pretreatment radiographs

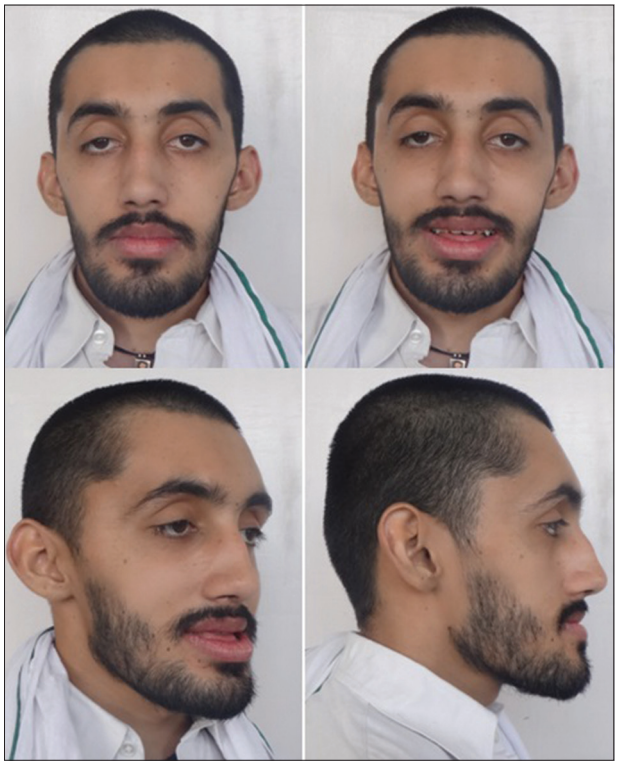

Figure 4: Presurgical extraoral photographs

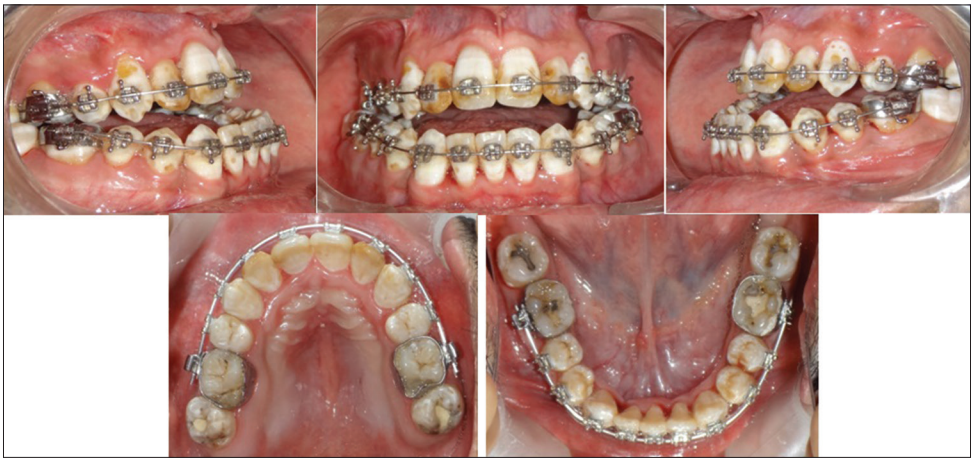

Figure 5: Presurgical intraoral photographs

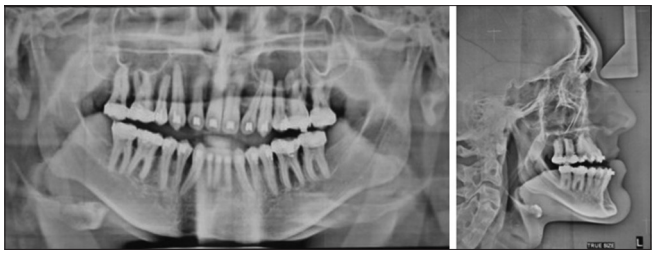

Figure 6: Presurgical radiographs

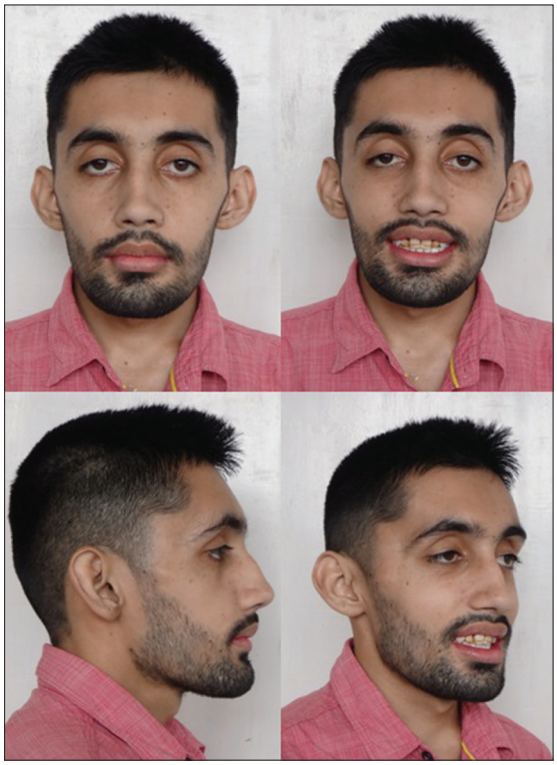

Figure 7: Postsurgical extraoral photographs

Treatment progress

After taking informed and written consent from the patient, the treatment was started. Extractions of the upper first premolars, the lower incisors, and all the third molars (28, 38, and 48) were carried out before starting fixed mechanotherapy. After 11 months of treatment, an 8-mm reverse overjet was present [Figures 4 and 5]. Normal inclination of the upper incisors and lower incisors was achieved [Figure 6 and Table 1].

Mock surgery was accomplished following facebow transfer, and surgical splints were fabricated. Surgical phase included Le Fort advancement of 5 mm and use of intermediate splint, and BSSO setback of 6 mm was done as planned to achieve the final overjet of 3 mm. This was followed by rigid maxillomandibular fixation.

Postsurgical orthodontics was started 4 weeks after the surgery. The objectives of this phase of active orthodontic treatment were to achieve a good occlusal relationship by settling the occlusion. Short intermaxillary elastics were given. Appliances were removed 7 months after surgical procedure [Figures 7-9]. Postdeboned radiographic and photographic records were taken. The upper and lower fixed retainers were placed in the retention phase. Superimpositions were done to assess treatment changes. The total treatment time was 18 months.

DISCUSSION

Gunderson *et al.*[7] distinguished three types of cervical vertebral fusion defect related to Klippel–Feil anomalies: type I – massive fusion of many cervical and upper thoracic

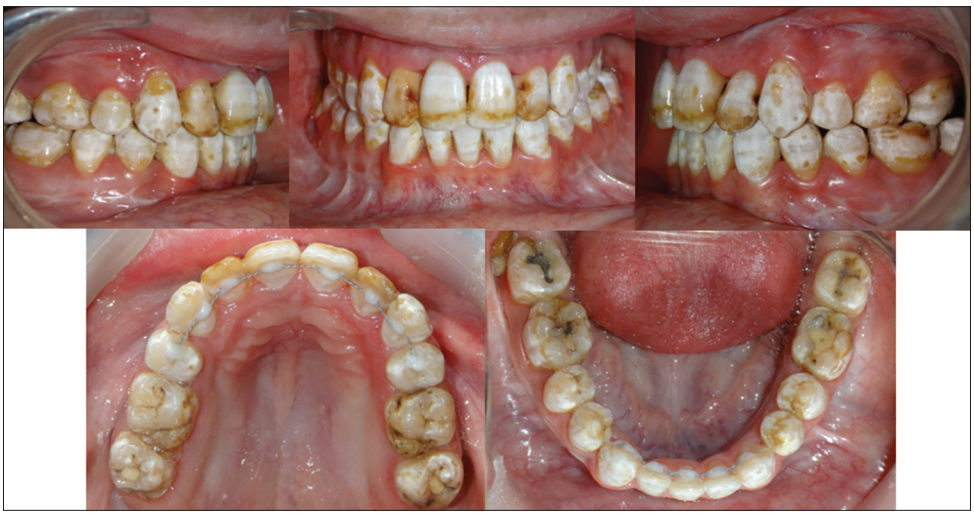

Figure 8: Postsurgical intraoral photographs

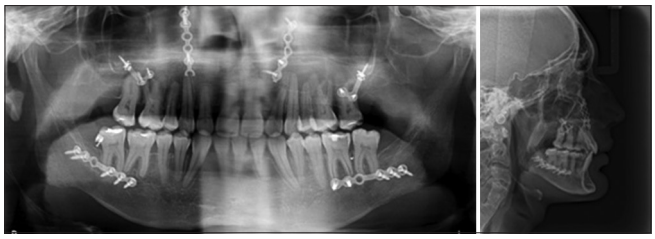

Figure 9: Postsurgical radiographs

vertebrae into bony blocks; Type II – fusion of only one or two interspaces, usually C2–C3 or C5–C6, but there can be intrafamilial variability; Type III – both cervical fusion and lower thoracic or lumbar fusion often associated with multiple organ anomalies and subsequent neurologic compromise.

Our patient presented with a short neck, limited neck movements, and a low-set posterior hairline. His symptoms included fusion of the C4, C5, C6, and C7 vertebrae without elevation of the scapula. With these features, our patient fits the Type II category of KFS. Complete systemic examination along with radiographic evaluation aids in more accurate diagnosis and treatment planning of such cases. Even in the literature, the patient with KFS usually has additional jaw deformities, and skeletal Class III malocclusion is more common in such cases. Complete management of such cases was found to significantly improve the psychological status of the patient.

CONCLUSION

The teamwork of orthodontist and oral and maxillofacial surgeon improves not only the patient's oral health but also the quality of life and self-esteem also. It is a good example of role of careful and detailed examination that leads to better diagnosis and treatment planning and necessary precautions required during treatment.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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