Concomitant Temporomandibular Joint Ankylosis and Maxillomandibular Fusion in a Child with Klippel- Feil Syndrome: A Case Report

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Klippel-Feil syndrome (KFS) is classically characterized by fusion of any of the two of seven cervical vertebrae. It is identified by the presence of a triad of clinical signs including short neck, limitation of head and neck movements and low posterior hairline. Unusual bony malformations leading to facial asymmetry is the most common oral manifestation associated with KFS. Such maxillomandibular fusion can also result in restricted mouth opening in children. It's a challenge to provide complete rehabilitation in such children. This paper presents a report of a type II KFS with both maxillomandibular fusion and temporomandibular joint ankylosis which led to the limited mouth opening in a six-year-old child. Also, the child showed an inadequate development of speech, facial asymmetry and compromised oral health owing to the restricted mouth opening. After thorough investigations, surgery was done which restored limited mouth opening which led to normalizing of speech and oral health.

***Keywords**: Klippel-Feil Syndrome, Maxillomandibular fusion, Temporomandibular joint ankylosis.*

INTRODUCTION

Klippel–Feil syndrome (KFS) is a rare condition classically presented with unusually short neck, lower posterior hair line and limited movement of the head and neck.[1] KFS is characterized by the congenital fusion of any of the two of the seven cervical vertebrae. It occurs in 1in 30,000 to 1 in 40,000 live births with an increased predilection to females.[2] KFS may be associated with anomalies of the genitourinary, musculoskeletal, neurological, respiratory, cardiac and urogenital systems.[3] Since this condition involves multiple systems, the rehabilitation of these patients requires a multidisciplinary approach.

Divergent reports exist in the literature concerning the oral findings in individuals with KFS. While some authors have communicated congenital duplication of the mandible, maxillomandibular fusion has also been reported by others.[4-8] Mandibular prognathism has been reported in few cases of KFS, while other cases of KFS reported micrognathia.[1, 9] Other oral findings include the presence of cleft palate, jaw cysts, impacted canine, bifid tongue, etc.[1, 3, 9-12] There are four different types of KFS.[3] The oral manifestations are not unique to any particular KFS. Therefore the clinician should keep in mind that no classical oral findings will be associated with any particular KFS.

Usually, restricted mouth opening in patients with KFS is commonly attributed to pseudo-ankylosis of the temporomandibular joint (TMJ).[1] The association between true TMJ ankylosis and concomitant maxillomandibular fusion in children with KFS is rarely reported in the literature. This paper presents a report of a type II KFS with both maxillomandibular fusion and

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temporomandibular joint ankylosis which led to the limited mouth opening in a six-year-old child. We discuss the challenges faced in surgically rehabilitating this child due to the osseous defects that were seen in the head and neck region.

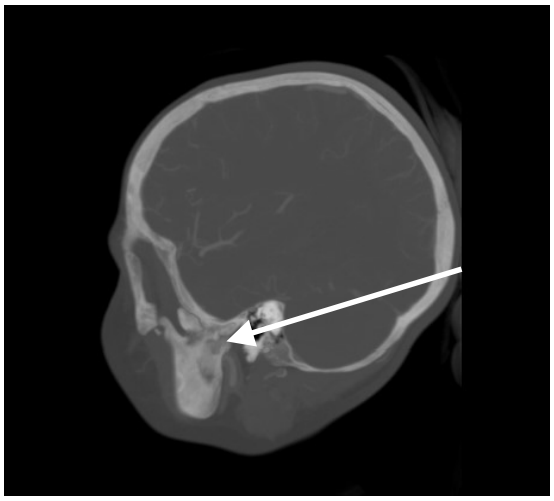
Case Presentation

A six-year-old girl known case of type II KFS was referred from Department of Pediatrics with the chief complaint of restricted ability to open her mouth and eat her food properly since childhood. The parents reported that the limited mouth opening increased progressively since then. The parents did not give any history of trauma to the face during her childhood. There was no significant familial history.

On examination, she presented with a short and twisted neck like torticollis, small palm, and delayed development of speech. She had mild asymmetry affecting the lower part of the face. With an inter-incisal opening of 10mm (Figure 1), a minimal intra-oral examination was possible which revealed multiple decayed primary teeth.

Figure 1: Child patient with inter incisal opening of 10mm

Figure 2: Two-dimensional computerized tomography scan showing abnormal bony deposition in the left TMJ with obliteration of the joint space as well as an elongated coronoid process



Sagittal sections of the two-dimensional computerized tomography (CT) scan of this patient showed abnormal bony deposition in the left TMJ with obliteration of the joint space as well as an elongated coronoid process (Figure 2).The coronal sections showed the unusual bony mass between the left maxillary buttress and the left coronoid region (Figure 3) and the complete obliteration of the TMJ area on the left side as compared to the right side (Figure 4). The unusual fusion present in between the second and third cervical vertebrae is evident in the CT scan (Figure 5).

Figure 3: Two-dimensional computerized tomography scan showing the unusual bony mass between the left maxillary buttress and the left coronoid region.

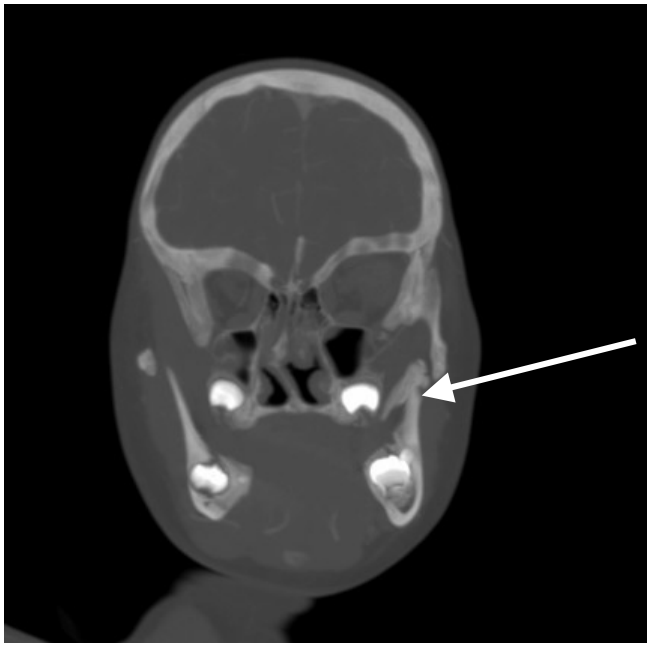
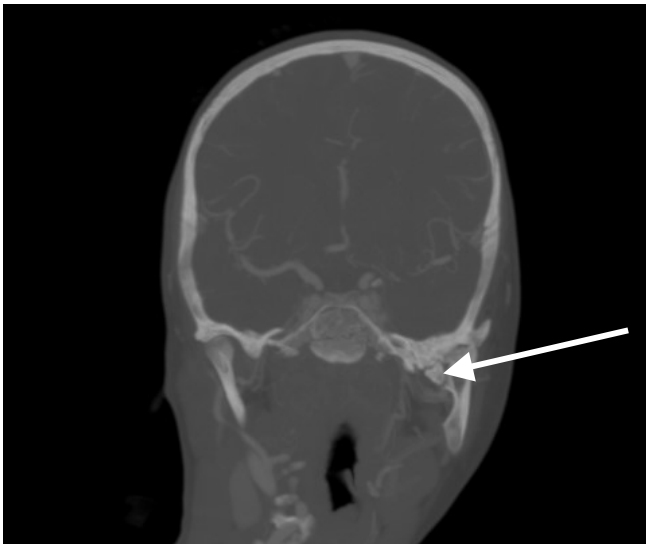


Figure 4: Two-dimensional computerized tomography scan is showing the complete obliteration of the temporomandibular joint region on the left side as compared to the right side.



The three dimensional CT images of head and neck of the patient showed the apparent tilt in the neck with an abnormal fusion of the second and third cervical vertebrae (Figure 6). The left lateral view also revealed abnormal fusion present in the left TMJ area (Figure 6, 7), abnormal fusion of maxillary buttress to duplicated coronoid process and normal appearing right TMJ (Figure 8).

The child was diagnosed of left TMJ ankylosis with the left maxillomandibular fusion and duplication of coronoid process on the left side of the mandible. The child was taken up for surgical resection of the fused masses present in the left TMJ and the left maxillomandibular area. Extensive fusion of TMJ and maxillomandibular segments reduced the mouth opening severely, while the abnormal fusion of second and third cervical vertebrae led to atlantoaxial instability. These two factors posed a significant challenge in intubation of the patient in our case hence a fiberoptic intubation was used to provide general anesthesia.

Figure 5: Two-dimensional computerized tomography scan showing the unusual fusion present in between the second and third cervical vertebrae

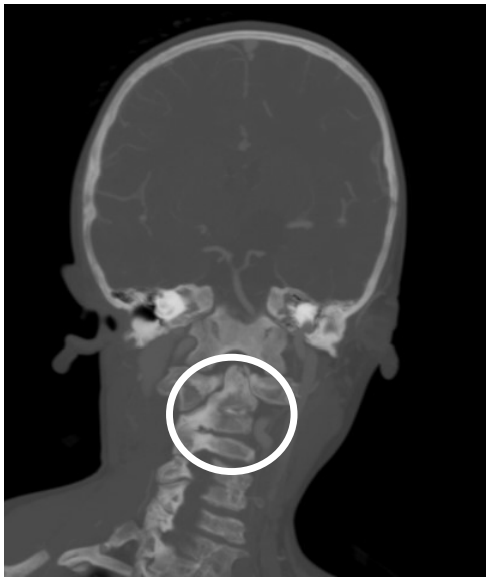
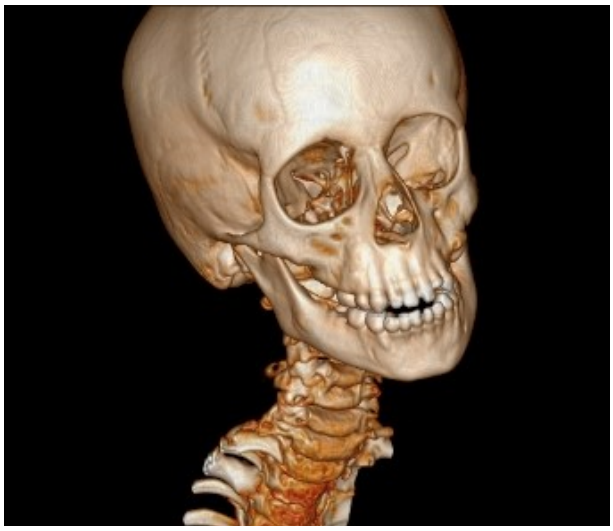


Figure 6: Three-dimensional computerized tomography image showing the obvious tilt in the neck



The access for the ankylotic mass was through a standard pre auricular approach with the Al-Kayat and Bramley modification.[13] The initial incision was carried through the skin, the subcutaneous tissue, superficial temporalis fascia, all to be incorporated in a single unit. The flap was raised exposing the deep temporalis fascia overlying the temporalis muscle. The dissection started initially above the zygomatic arch staying close to the auricular cartilage to avoid injuries to the facial nerve. Once the root of the zygoma was identified an oblique incision was made parallel to temporal branch of facial nerve. A periosteal elevator was inserted beneath the superficial layer of temporalis fascia, and the periosteum was reflected to expose the ankylotic mass.[14] The ankylotic mass was then delivered by sequential osteotomy. The fusion between the left maxillary buttress and the left coronoid process was removed via an intraoral approach (Figure 9). Following the removal of the unusual fusion there was a significant improvement in the mouth opening

Figure 7: Three-dimensional computerized tomography image showing abnormal fusion present in the left temporomandibular joint area.

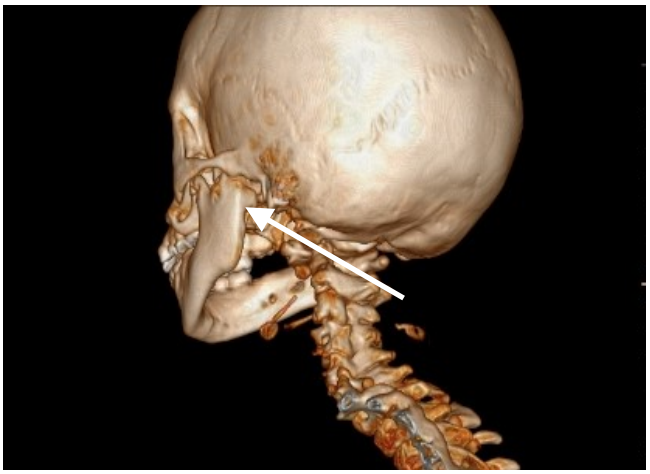
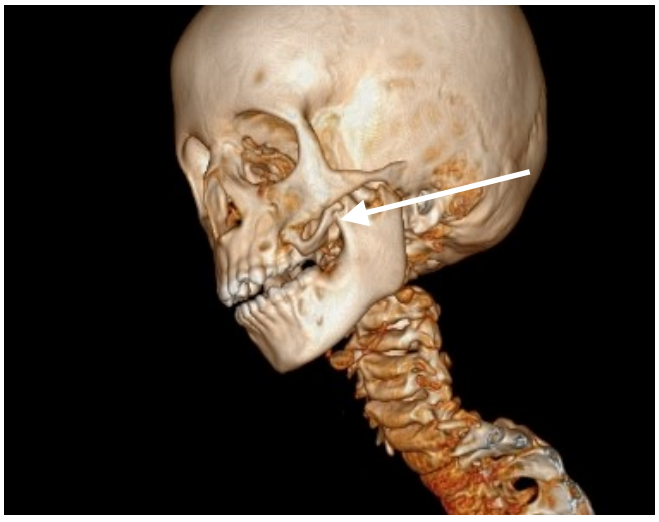


Figure 8: Three-dimensional computerized tomography image showing an unusual fusion of the maxillary buttress region with the left coronoid region.



and intraoperatively the inter-incisal opening measured up to 30mm (Figure 10). A posteriorly based temporalis muscle flap was used to interpose between the resected margins at the left TMJ region of ankylosis. (Figure 11). This was done with the intention to prevent re-ankylosis.

The patient was put on a rigorous regimen of mouth opening physiotherapy in the postoperative phase, and she continued to maintain a mouth opening of 28mm inter incisally. The patient was followed up after one year with a mouth opening of 28mm (Figure 12).

Figure 9: Showing the retrieved unusual mass between the left maxillary buttress region and left coronoid region.

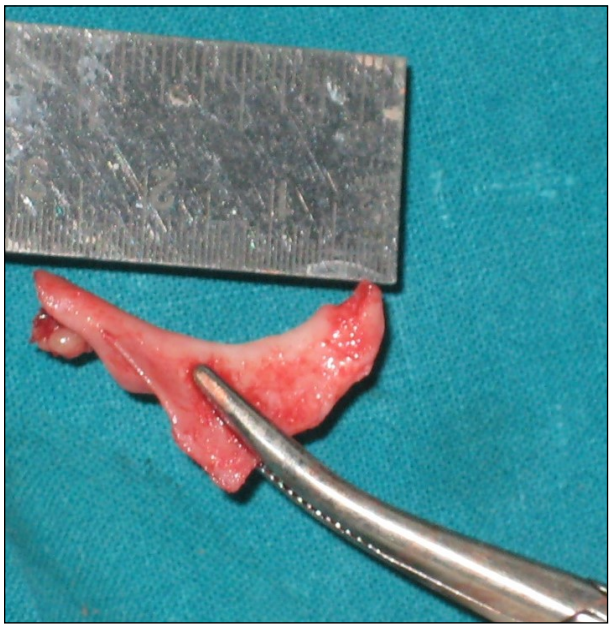


Figure 10: Shows the resultant mouth opening (30mm) achieved after the removal of the unusual bony masses.

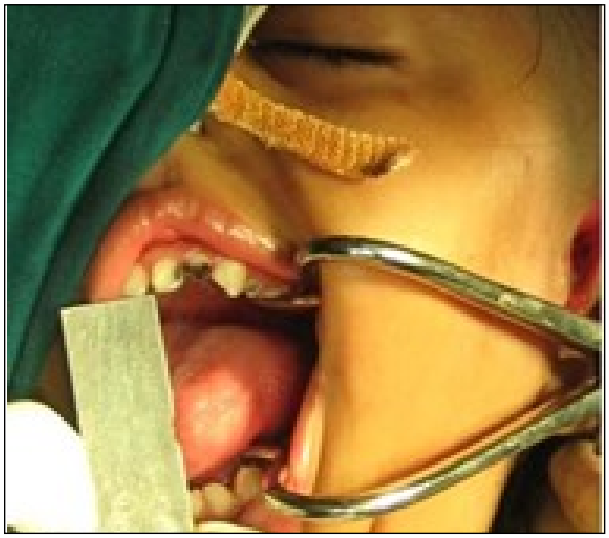


Figure 11: Showing the interposition of the Temporalis Myofascial Flap (TMF) to prevent re-ankylosis

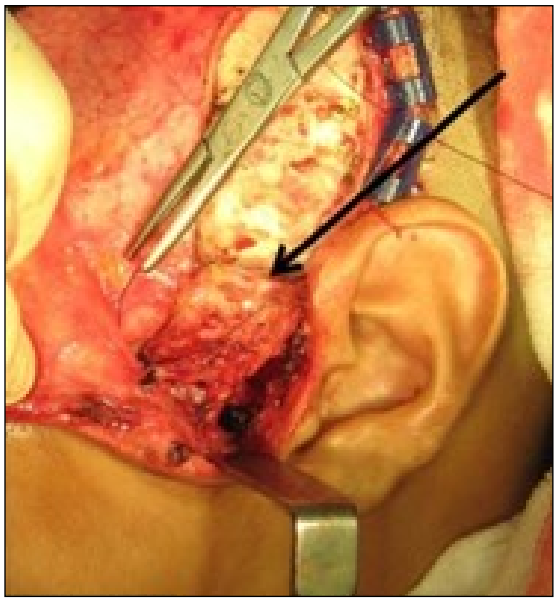
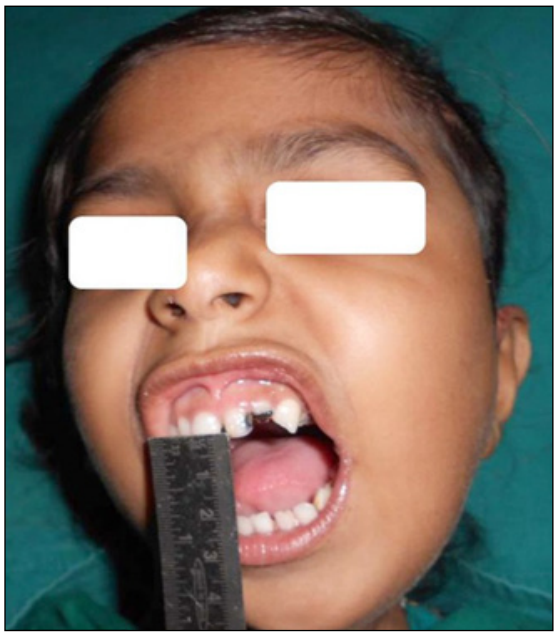


Figure 12: One-year follow-up picture of the inter-incisal opening of 28mm.



DISCUSSION

KFS is named after Maurice Klippel and Andr**é** Feil who first reported this syndrome in a forty-six-year-old tailor from France in 1912. They found that this patient had only four cervical vertebrae, which were fused to a single mass with posterior spina bifida, causing him to have an unusually shortened neck and restricted movement of the neck.[1]

Feil in 1919 categorized KFS into three morphological subtypes, and the fourth subtype was later introduced by Raas-Rothschild *et al* [15] Type I: Fusion of several cervical and upper thoracic vertebrae. Type II: Fusion of one or two interspaces sometimes accompanied

by hemivertebrae, occipito-atlantal fusion, and other vertebral abnormalities. Type II has the lowest risk of scoliosis. Type III: Fusion of cervical vertebrae associated with fusion of lower thoracic and lumbar vertebrae. Type IV has been suggested as being related to sacral agenesis.[15]

Craniofacial abnormalities are the most predominant oral findings associated with KFS.[3] The osseous malformations leading to facial abnormalities commonly include duplication of mandible or fusion of maxilla with mandible by accessory bone formations, which result in restricted mouth opening.[1, 3] The etiology of these osseous defects occurring commonly in KFS is not precisely known. However, the facial abnormalities seen in KFS are suggested to occur due to defect in the development of first branchial arch derivatives.[16]

In our case the mouth opening was decreased due to a true TMJ ankylosis as well as concomitant maxillomandibular fusion unlike in earlier reported cases restriction in mouth opening was due to pseudo-ankylosis of the temporomandibular joint.[1] A restricted mouth opening not only compromises the nutritional status of the patient but also influences the maintenance of oral health. Besides these complaints in our case, the parents reported that the mouth opening decreased progressively. A surgical resection of fused bony units with inter positional arthroplasty was planned with the aim of releasing the abnormal bony union and preventing recurrence of bony fusions. Surgical resection of fused mass improved the mouth opening intraoperatively, and the patient was on active physiotherapy to prevent re-ankylosis.

The aim of treatment of TMJ ankylosis in children is to restore mandibular movements and to allow normal growth to reduce the possibility of facial asymmetry in the future.[17] The success of treatment depends upon prevention of recurrence by interposition and active physiotherapy. The best treatment option suitable in cases of restricted mouth opening due to TMJ ankylosis or maxillomandibular fusion would be surgical resection of fused segments. Various options are available for reestablishing normal joint function that includes gap arthroplasty, inter positional arthroplasty with an allogenic or autogenic material, or total or partial joint replacement techniques. Gap arthroplasty is associated with high recurrence, and joint replacement options are considered only in cases of failed or multi-operated cases.[18] Therefore inter positional arthroplasty with temporalis muscle flap was used. Our patient demonstrated inter-incisal opening of 28mm during one year follow up depicting the success of our treatment in this case.

CONCLUSIONS

KFS is a rare condition with a wide range of oral findings which are not classical or unique to any particular KFS. Although abnormal bony fusions leading craniofacial anomalies are most typical of the oral findings in KFS, they can present with or without any complaint of oral problems. Hence an investigatory radiograph like orthopantomogram and CT scan can discover the probable oral manifestations associated with KFS. Early identification of osseous malformations and appropriate intervention can rehabilitate the patient adequately and limit the facial dysmorphism in these patients.

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