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Case Report

Cervical Klippel-Feil syndrome progressing to myelopathy following minor trauma

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ABSTRACT

Klippel-Feil syndrome (KFS) is a rare disease with a clinical triad of low posterior hairline, short neck, and limited neck motion. Frequent fusion of two or more cervical vertebrae resulting from a congenital segmentation defect can lead to adjacent level hypermobility, instability, and even neurologic symptoms that require surgical intervention. However, surgical results in adults with KFS with concomitant atlantoaxial subluxation and cervical spinal stenosis have not been reported. We report a 58-year-old man with complaints of an unsteady gait, general weakness, and clumsiness in both hands for 6 months. Deep tendon reflexes in both knee joints were increased, with a positive Babinski sign. Bladder and sphincter function were intact. Radiographic findings included C2–C7 congenital fusion with atlantoaxial subluxation and spinal cord compression. He was treated with posterior occipitocervicothoracic fusion, instrumentation, and posterior decompression with a partial craniectomy under the diagnosis of cervical myelopathy. Postoperatively, the neurologic deficits improved without any complications, although bilateral rod breakage was noted at consecutive outpatient department (OPD) follow-ups. He recovered well with residual left hand numbness.

1. Introduction

Klippel-Feil syndrome (KFS), an uncommon disease, is now recognized in some clinical and radiographic retrospective studies [1–3]. Most studies reported no patients with cervical spine-related symptoms. Some symptomatic patients had neck pain, radiculopathy, or myelopathy since childhood and adolescence and the risk of cervical stenosis in these patients was high [4]. However, there are no reports of symptomatic adult patients with KFS presenting with concomitant atlantoaxial subluxation and cervical spinal stenosis. We report a 58-year-old man who sustained cervical myelopathy because of KFS with concomitant atlantoaxial subluxation and cervical stenosis.

2. Case report

A 58-year-old man complained of unsteady gait with clumsiness in both hands for 6 months. He had no other systemic diseases. The symptoms exacerbated in the previous 2 weeks after he fell against a bathroom washstand, resulting in a head contusion with a hyperflexion injury to the cervical spine. He reported bruising over his forehead. He had limited neck extension and flexion movement. On physical examination, he was noted to have a short neck and low posterior hairline. He also complained of general weakness with a sensation of tightness over his body. The deep tendon reflexes in both knee joints were increased, with a positive Babinski sign. Sphincter function was intact. Plain radiography (Fig. 1) showed C2–C7 congenital fusion with atlantoaxial subluxation. The anterior atlantodens interval (AADI) was 8.7 mm and 4.6 mm in the flexion and extension views, respectively. Magnetic resonance imaging (MRI) of the cervical spine showed C1–C5 spinal stenosis with a posterior ossifying enlargement at the C3 and C4 levels (Fig. 2). The space available for the cord was 6 mm at the

Conflicts of interest: none.

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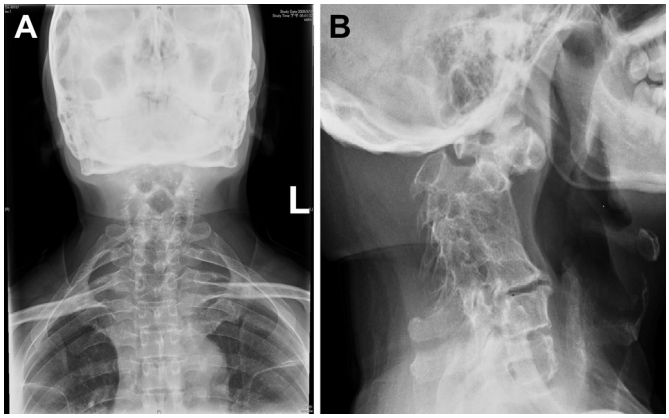

Fig. 1. (A) Anteroposterior and (B) lateral radiographs of the cervical spine of a 58-year-old man with type III Klippel-Feil syndrome with atlantoaxial subluxation.

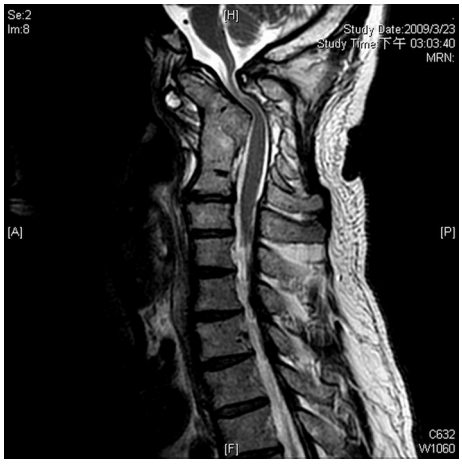

Fig. 2. Sagittal T2-weighted MRI scan shows extensive C2–C7 fusion with ossifying enlargement and C1–C5 spinal stenosis. There is no increased signal intensity within the spinal cord.

C1 level. Computed tomography (CT) showed occipitocervical junction instability (Fig. 3).

After the diagnosis of KFS with cervical myelopathy was made, the patient underwent a two-stage operation. First, he received halo vest external fixation under local anesthesia, assisted by fluoroscopy. The aim was to maintain correct craniocervical alignment and help the patient adapt to postoperative conditions without further neurologic deterioration (Fig. 4). After being observed for 2 days, he was treated with posterior decompression with a laminectomy from C1 to C5 and a partial craniectomy of the occiput. Good pulsation of the dura was observed immediately after decompression. The altered anatomy of the congenitally fused segments made it too difficult to apply lateral mass screws. Occipitocervicothoracic fusion was extended to the T3 level using a titanium screw-rod fixation system (Summit occipitocervicothoracic spinal fixation system, DePuySpine, Rayham, MA, USA). Occipital screws were bicortically purchased and secured with the plates on the midline. Then the occipital screws and plate were connected to the thoracic pedicle screws with rods, followed by assembly of the crosslinks. The exposed dura was covered with absorbable gelatin sponge. A humerus strut allograft plate was placed posteriorly over the decompressed segments between the cranium and C6 for occipitocervicothoracic fusion (Fig. 5). There was no worse change of the somatosensory-evoked potential intraoperatively. After the surgery, the patient reported subjective improvement. The Nurick score [5] improved from 3 preoperatively to 0 at 6 months postoperatively. The halo vest was removed 1 month postoperatively under stable conditions.

The patient tolerated the rehabilitation program well. The muscle power in his four limbs recovered well except for residual

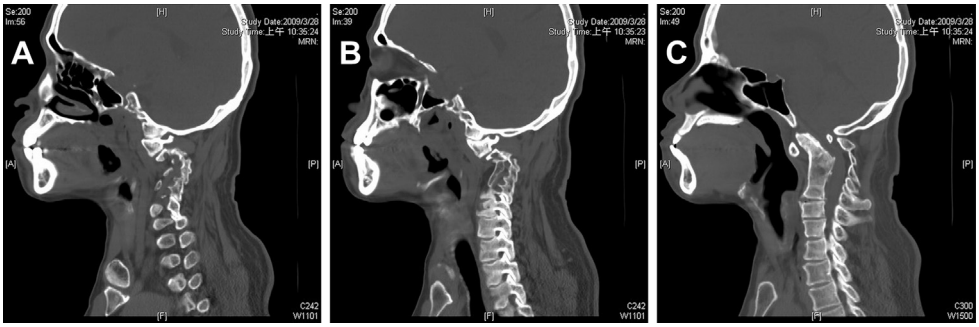

Fig. 3. (A, B, and C) Sagittal CT scans showing superior odontoid migration without occipitalization. Angular motion has caused an increase in the atlantodens interval, indicating instability.

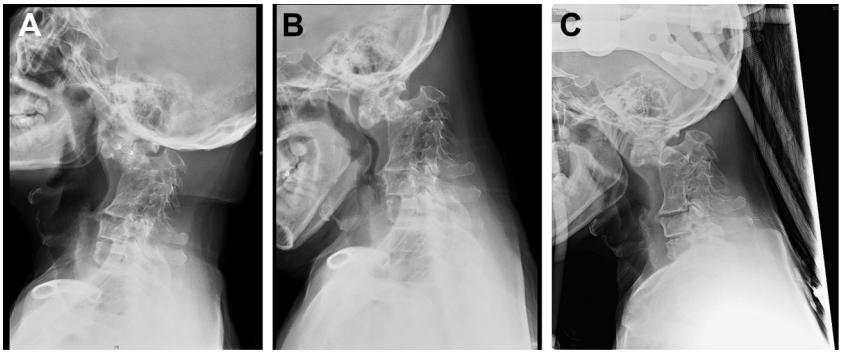

Fig. 4. (A and B) Instability of the occipitocervical junction preoperatively. (C) After application of a halo vest. Atlantodens interval = 4 mm, upper hard palate-upper edge of T1 angle: 0°.

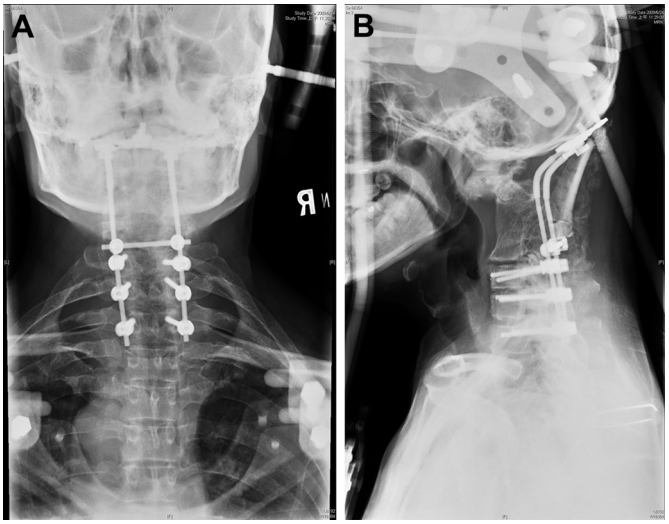

Fig. 5. (A) Postoperative cervical spine anteroposterior and (B) lateral radiographs showing good positioning of the allograft bone plate and good occipitocervicothoracic fixation alignment. Atlantodens interval: 4 mm, upper hard palate-upper edge of T1 angle: 0°.

left upper limb numbness. He was followed up at 4 weeks, 3 months, 6 months, 9 months, 12 months, 18 months, 24 months, and 36 months postoperatively. The atlantodens interval (ADI) was 4.0 mm. The angle of the upper hard palate and upper edge of T1 was 0°. Plain radiographs 3 months postoperatively showed the ADI had increased to 5.8 mm and the angle between the upper hard palate and upper border of T1 was 5° kyphosis. His condition kept improving without recurrent neurologic deficits or neck pain. Left rod breakage was noted 6 months postoperatively without complaints of axial pain. The ADI and hard palate-upper T1 angle were the same as before, and the dynamic view showed no instability. Consecutive follow-up plain radiographs revealed no further change in alignment, with good stability and a fusion callus over C7–T1. Although breakage of both rods was noted 18 months postoperatively on plain radiography, good fusion was noted over the craniocervicothoracic junction with stability and recovery of function. MRI 24 months postoperatively revealed good decompression of the spinal cord (Fig. 6). Stability and alignment of the fused spine was noted 36 months postoperatively, although bilateral rods were broken (Fig. 7). Follow-up cervical CT showed no subluxation or dislocation over the C1–2 bilateral facet joints. Good fusion callus formation was noted over the C7–T1 junction and the

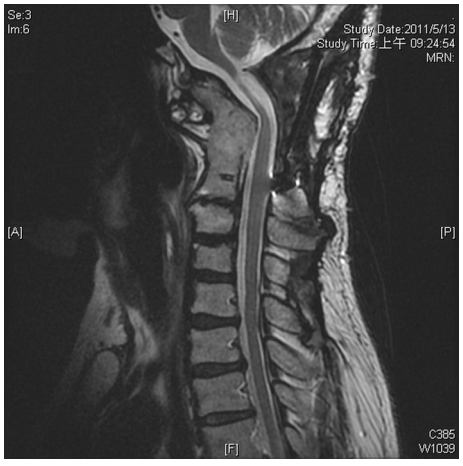

Fig. 6. Eighteen-month postoperative MRI shows a patent spinal cord.

contact area between the allograft bone plate, cranium, and C6, indicating that stability of the bony fusion structure was solid without intact rod protection (Fig. 8). The patient was able to work as a taxi driver postoperatively.

3. Discussion

The classification of KFS was first described in 1912 based on fusion patterns [1]. A better classification of the symptoms associated with radiographic patterns was proposed in 2006 by Samartzis et al (Table 1) [2]. According to this classification, associated symptoms can be predicted and treatment suggested in these individuals [2]. In this case, plain radiography showed type III multiple contiguous fused segments from C2 to C7. Patient complaints included an unsteady gait and hand clumsiness, findings that were consistent with cervical myelopathy. Although most adult patients with KFS are free of symptoms, 35–40% develop at least one cervical-related symptom. These symptoms can be classified into three groups: axial pain only, radiculopathy, and myelopathy [6]. There have been no reports of an adult with KFS with coexisting atlantoaxial subluxation and cervical stenosis. Atlantoaxial subluxation resulting from loss of the ligamentous stability between the atlas and axis has also been associated with other disorders, such as rheumatoid arthritis and Down syndrome [7]. The associated conditions of spinal stenosis and degenerative arthritis or subluxation of the adjacent segments are thought to be major contributing factors to the neurologic symptoms [8]. Patients with a fused cervical spine also have a greater chance of sustaining minor injuries than those with a healthy spine. However, it is unclear whether the subluxation developed gradually prior to the accident in our patient. A greater Pavlov ratio in KFS, either cephalad or caudal to the fusion segments, has been reported and was also noted in our patient. Altered cervical anatomy during the process with a small-diameter cord and large canal may delay the neurologic symptoms in asymptomatic individuals with KFS. Although they have a large space available for the cord, neural compromise might be affected by segmental instability such as atlantoaxial subluxation [9].

Imaging evaluation with plain radiography, CT, and MRI are necessary before surgery. Craniometric evaluation of the Chamberlain line, Wackenheim clivus line, and Welcker basal angle could be used to assess the stenosis but they are sometimes not easily drawn on plain radiography because of blurred and overlapping bony margins [10]. Widening of the AADI of more than 5 mm in the flexion–extension view indicates atlantoaxial instability. In patients with KFS, sagittal CT images are useful for the evaluation of craniocervical abnormalities such as superior odontoid migration and occipitalization [11]. Atlantoaxial subluxation was impressed by widening of the AADI of more than 5 mm in this patient with superior odontoid migration without occipitalization noted in the CT scan [12] (Fig. 3). Occipitocervical junction instability is also a surgical indication [13].

In this case, the ADI increased and the Cobb angle between the palate and upper thoracic spine changed after the halo vest was removed. However the patient's condition has remained unchanged since then. He has had no recurrent neck pain or neurologic deficits. Postoperative CT showed no subluxation or dislocation of the C1–2 facet joints. This may result from varying morphology of the occipitocervical junction in patients with KPS because angular motion could cause an increase in the ADI without joint disruption.

Rod breakage was noted in our patient 6 months postoperatively. Failed fusion was suspected. The patient's condition continued to improve, with no neck pain. Postoperative plain radiography and CT revealed good fusion callus formation over the C7–T1 segment and the contact ends between the allograft bone

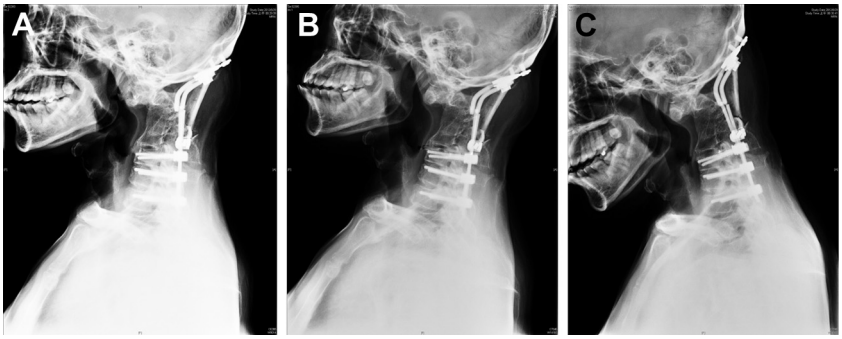

Fig. 7. (A, B, and C) Thirty-six-month postoperative radiographs. Atlantodens interval: 5.8 mm, upper hard palate-T1 upper edge angle: 5° kyphosis. There is no instability over the dynamic view.

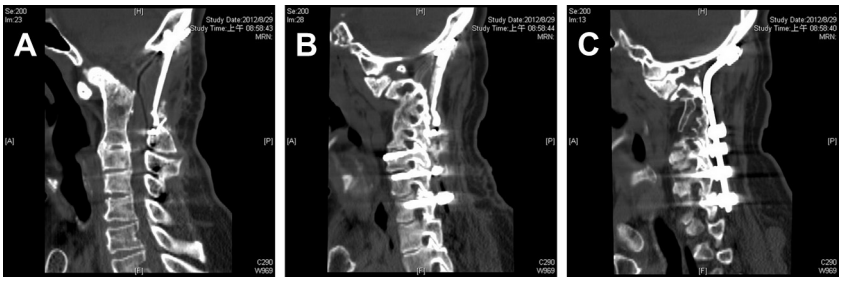

Fig. 8. (A, B, and C) Forty-two-month postoperative sagittal computed tomography reveals a good fusion callus over C7–T1 and the contact regions between the allograft bone plate, cranium, and C6. No subluxation or dislocation is noted over the cranium–C1–C2 joint.

Table 1
Classification of Klippel-Feil syndrome [2].

Classification	Radiographic presentation
Type I	Single fused segment
Type II	Multiple noncontiguous fused segments
Type III	Multiple contiguous fused segments

plate, cranium, and C6. The allograft bone plate functioned successfully as a roof to enhance stability, protect the decompressed dural sac, and increase the fusion bed. With these aspects, good fusion stability was obtained in this case. The screw and rod system is the most rigid fixation system for occipitocervicothoracic fusion. The bilateral rods broke at the same point as the prebent segment, which might have been due to stress concentration.

Prophylactic surgical fixation in asymptomatic patients is not well defined. However, minor trauma may have resulted in the severe neurologic deficits reported in case reports. These patients should be informed of potential neurologic deficits and avoid contact sports. Patients with KFS and axial pain or radiculopathy may not require surgical intervention [14]. Samartzis et al reported only an 11% surgical rate in symptomatic patients with KFS who presented with myelopathy or persistent radiculopathy after conservative treatment [3]. However, symptomatic patients are at risk of progressive neurologic symptoms [15,16]. Our patient was treated successfully with posterior decompression and instrumented fusion to address his myelopathy and instability. The altered anatomy in his cervical vertebrae made it difficult to apply lateral mass screws, so occipitocervicothoracic fusion from the occiput to T3 was done.

This is a case of KFS with concomitant atlantoaxial subluxation and cervical stenosis. The patient was successfully treated with posterior decompression and instrumented fusion to address the stenosis and instability. The patient has been followed up for 36 months after surgery and has had a good clinical and radiographic outcome.

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