

Intermittent Transient Quadriparesis in an Adolescent with Klippel-Feil Syndrome

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Abstract

Background: Klippel-Feil syndrome (KFS) is a rare congenital condition characterised by segmentation failure of the cervical spine, resulting in vertebral fusion and altered biomechanics. Patients may remain asymptomatic or present with neurological complications. The classic clinical triad (short neck, low posterior hairline, restricted cervical range of motion) is historically associated with KFS, but many individuals do not exhibit all three features and diagnosis is therefore frequently missed or delayed. **Case Presentation:** We report a 17-year-old male with previously undiagnosed KFS who presented with recurrent episodes of transient quadriparesis following minor head trauma. Initial CT imaging demonstrated multilevel congenital cervical fusion. Retrospective history revealed a three-year pattern of transient neurological deficits triggered by cervical extension. Subsequent specialist assessment identified upper motor neuron signs and dynamic instability at C4/5. MRI demonstrated severe canal stenosis with near-complete cerebrospinal fluid effacement and focal intramedullary T2 hyperintensity consistent with cervical myelopathy. The patient underwent single-level anterior cervical discectomy and fusion (ACDF) at C4/5 with instrumentation. Early postoperative recovery was uncomplicated. **Conclusion:** This case highlights the risk of delayed diagnosis of clinically significant instability in adolescents with KFS presenting with transient neurological symptoms. Recognition of congenital segmentation anomalies at initial presentation should prompt continued immobilisation, advanced imaging, and early specialist referral. Timely surgical decompression and stabilisation may prevent progression to irreversible spinal cord injury. Increased awareness of KFS in emergency settings is essential to avoid missed or delayed diagnosis.

Keywords

Klippel-Feil Syndrome, Quadriparesis, Cervical Myelopathy, Spinal Instability, ACDF

1. Introduction

Klippel-Feil syndrome (KFS) is a congenital disorder characterised by fusion of two or more cervical vertebrae arising from abnormal segmentation during early embryological development [1] [2]. The classic clinical triad (short neck, low posterior hairline, restricted cervical range of motion) is historically associated with KFS, but many individuals do not exhibit all three features and diagnosis is therefore frequently made on imaging, sometimes incidentally [3]. Incidence is estimated at 1 in 40 - 42,000 and contemporary classification commonly uses the Samartzis system, which stratifies congenital fusions into single-level, multiple non-contiguous and multiple contiguous fusions, with prognostic implications for symptom patterns including radiculopathy and myelopathy [4] [5].

From a biomechanical perspective, congenital fusion reduces motion at fused segments and may increase stresses and hypermobility at adjacent mobile segments [6] [7]. However, the magnitude and clinical inevitability of adjacent segment disease in KFS remains debated, with limited paediatric longitudinal data and mixed conclusions in small series. Importantly, KFS and congenital cervical stenosis have been associated with increased vulnerability to neurological injury after relatively minor trauma, possibly related to altered biomechanics and hypermobility [8] [9].

In adults with degenerative cervical myelopathy, international clinical practice guidelines recommend surgical decompression for moderate to severe myelopathy and consideration of surgery for mild disease depending on clinical context [10]. While these guideline data are largely derived from adult populations, the general principle of timely decompression and stabilisation for progressive cord compromise informs practice when structural compression and instability are demonstrable in an adolescent.

This case study presents a 17-year-old male with Klippel-Feil syndrome who developed recurrent transient quadriparesis and progressive cervical myelopathy following head trauma at 14 years of age. This case highlights how a diagnosis of clinically significant instability may be delayed in the context of an atypical neurological history on the background of a rare congenital condition. This case also highlights the importance of prompt recognition of cord compromise and raises awareness of KFS management.

2. Case Presentation

In November 2024, a previously fit and well 17-year-old male was brought in by ambulance to the emergency department after a fall while playing football with subsequent transient weakness in all four limbs. While during onsite paramedics' examination he had pins and needles sensation with profound weakness in all of four limbs, the initial emergency department assessment revealed full recovery of sensation with slight weakness (4/5) in all four limbs. Upon arrival to the emergency department, he was immobilised with blocks and collar. A computed to-

mography (CT) scan of the cervical spine reported satisfactory alignment without acute fracture but identified multilevel segmentation anomalies with congenital block vertebrae involving C2 - C4 and C5 - C7, with lack of fusion of posterior elements of C4 keeping with occult dysraphism (**Figure 1**). A posterior disc-related protrusion was also described at C4/5 mildly indenting the anterior thecal sac. Symptoms resolved after approximately one hour in the department and the cervical spine was clinically cleared following the CT.



Figure 1. Initial CT at first presentation.

Within the patient's history, they described experiencing intermittent neurological symptoms beginning after a head impact at age 14 following hyperextension injury to his neck while diving backwards in a swimming pool. Immediately after the Initial incident he reported similar whole-body weakness and numbness and was unable to exit the pool without assistance. He did not attend hospital at the time and the symptoms resolved spontaneously.

Over subsequent years he described recurrent episodes of transient weakness and numbness, variably lasting from approximately one hour to "a couple of days". He reported provocation of symptoms by cervical extension, including limb numbness and a shock-like pain radiating down the spine. He denied fine motor deterioration, gait imbalance, falls or bowel/bladder disturbance. Functionally, he was independent in transfers, mobility and self-care at baseline, living with his mother. An outpatient spinal surgery referral was completed but no inpatient specialist review was made when initially presenting to ED in November 2024.

He was subsequently reviewed in the paediatric spine clinic and clinical examination showed preserved limb power (5/5) but brisk upper-limb reflexes with bilateral Hoffmann's positivity and Lhermitte-type symptoms triggered by neck extension. Romberg was negative but he had bilateral clonus and upward plantar

reflexes on examination. Flexion/extension radiographs demonstrated dynamic malalignment/instability centred at C4/5, between the congenitally fused segments (**Figure 2**).

An urgent whole-spine MRI (**Figure 3**) was compared with prior CT scans and radiographs and demonstrated: (i) multiple congenital segmentation anomalies with a C2 - C4 block vertebra (incompletely fused on the right at C3-4) and a C5 - C7 block vertebra; (ii) at C4/5, severe canal stenosis with near-complete effacement of CSF in the anteroposterior dimension, attributed to a disc-osteophyte complex, abnormal facets, and likely ligamentous laxity in the context of congenital anatomy; and (iii) focal linear intramedullary T2/STIR hyperintensity at C4/5 consistent with myelopathy/myelomalacic change. The cord signal abnormality was interpreted as likely chronic, related to dynamic instability rather than an acute injury.

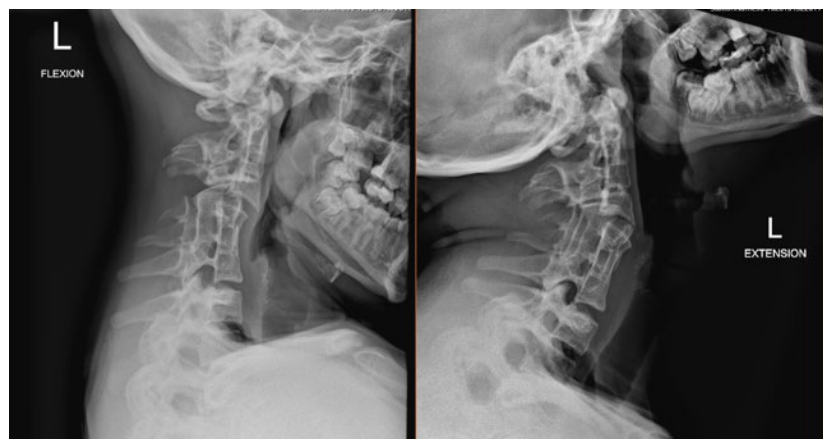


Figure 2. Flexion/extension radiographs.

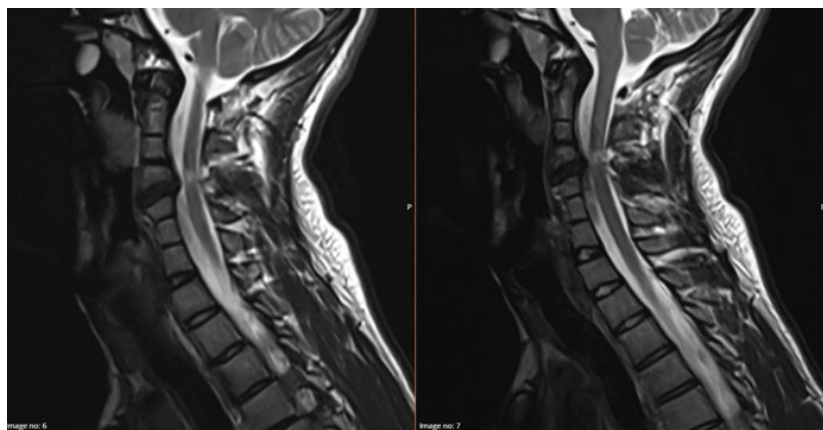


Figure 3. Whole spine MRI. Sagittal cervical spine.

The local spinal multidisciplinary team reviewed the multi-year history of intermittent neck pain and transient four-limb paralysis with the current imaging and Anterior Cervical Discectomy and Fusion surgery was offered. The MDT didn't ad-

vocate posterior approach due to C4 occult dysraphism and risk of spinal cord injury. The MDT found no need to perform any CT angiography prior to the operation.

At the pre-operative clinic visit, the working diagnoses was KFS (congenital fusion at C2/3 and C5 - C7), C4/5 disc prolapse, cervical cord compression with signal change and cervical myelopathy. At pre-operative review, the patient and their family were counselled that surgery aimed to arrest progression and might not reverse established cord dysfunction.

His recurrent episodes were attributed to cervical cord neurapraxia/transient quadriparesis from C4/5 instability based on flexion/extension radiograph as well as CT scan and MRI findings, rather than other causes of transient weakness such as Multiple Sclerosis or Transient Ischemic Attack.

3. Operation and Technical Note

In March, the patient underwent a primary anterior cervical discectomy and fusion (ACDF) with plating at C4/5. The patient was positioned supine and a transverse incision was made along the anterior aspect of the neck on the right-hand side centred over the affected cervical level after confirming with an image intensifier. Spinal monitoring was used throughout.

Following incision, the platysma was divided and the cervical fascia was identified and opened. The anterior cervical spine was exposed and retractors were placed to provide optimal visualisation. Using fluoroscopic guidance, the target intervertebral disc space was identified with subsequent insertion of the Caspar pin into adjacent vertebra.

Under the microscope, a discectomy was performed at C4/5 by carefully removing the intervertebral disc, including herniated or degenerated material, using a combination of Kerrison rongeurs, pituitary rongeurs, curettes and high-speed burr. Partial removal of the posterior longitudinal ligament, osteophytes and bilateral anterior foraminotomy were performed.

Endplate preparation was performed using a high-speed burr and curettes, ensuring a smooth and bleeding bone surface for fusion. The appropriate-sized interbody cage filled with Attrax, (Coalition 8 mm), was then inserted into the C4/5 disc space (**Figure 4**). The cage was stabilised in position with two anchors 13 mm in length.

The Helix plate, 28 mm, was properly positioned and secured to the vertebral bodies using 2 × 13 mm screws ensuring appropriate alignment and stability of the construct. Haemostasis was achieved using bipolar cautery and meticulous irrigation with antibiotic of the surgical site was performed. The surgical field was thoroughly inspected for any bleeding points or complications.

The cervical fascia was closed using vicryl and the subcutaneous tissues and skin were closed in layers using monocryl. Sterile dressings were applied. No intraoperative complications or adverse spinal monitoring events were encountered.

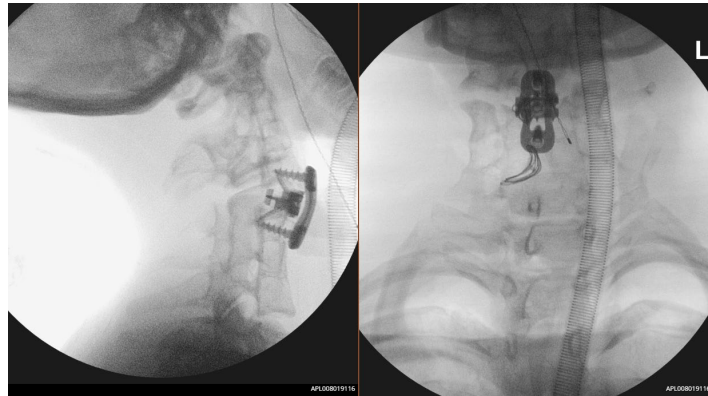


Figure 4. Intra-operative imaging.

4. Follow-Up and Outcomes

Post-operatively the patient recovered well and was discharged 1 day post operatively following repeat X-ray (**Figure 5**). He was discharged with 6 weeks of soft collar use, wound review at 2 weeks and orthopaedic/spinal follow-up at 6 - 8 weeks. At three month-follow up he had normal neurology in all his four limbs with no further episode of transient quadriparesis. His radiograph images were satisfactory (**Figure 6**).

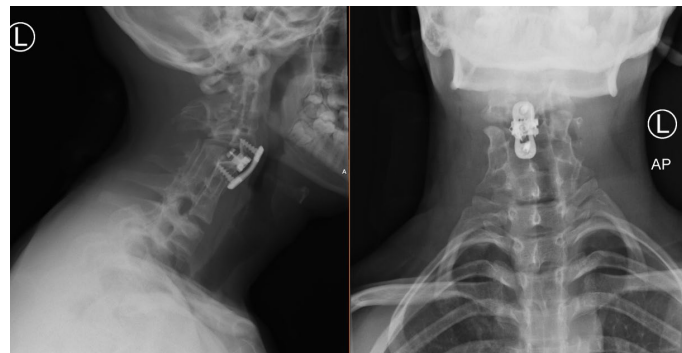


Figure 5. Post-operative radiograph.



Figure 6. Three month follow up.

5. Discussion

This case illustrates an adolescent presentation of FKS in which congenital cervical fusion with an intervening hypermobile segment culminated in severe focal stenosis, dynamic instability and clinical myelopathy manifesting as recurrent transient four-limb weakness after relatively minor sporting trauma.

The patient's episodes of transient weakness and numbness, often after cervical impact, with symptoms provoked by extension, are clinically compatible with cervical cord neurapraxia/transient quadriplegia (CNN/TQ) phenomena described in sports-related cervical injuries [11]-[13].

Current reviews describe cord CNN/TQ as a transient neurological deficit localising to the cervical cord after trauma [12]. Congenital stenosis and canal narrowing are recognised risk factors in mechanisms involving hyperflexion, hyperextension or axial loading [12]. As such, a greater awareness of FKS as a possible diagnosis and its distinct radiographic features along with an appreciation for symptoms typical for cervical cord neuropraxia may have led to a more timely diagnosis and management plan in this case and in similar future presentations [14].

In this patient, CT and flexion/extension radiographs demonstrated frank dynamic malalignment at C4/5 supporting a structural mechanism rather than a radiographically acute injury. Learning to take from this is that reliance on CT reports excluding an acute fracture as a basis for cervical spine clearance may be insufficient in the presence of congenital or degenerative abnormalities. More nuanced but clinically significant findings, such as Klippel-Feil syndrome, can be overlooked if the focus remains solely on excluding acute bony injury. Where such abnormalities are identified, particularly in the context of neurological symptoms, they should prompt heightened caution, maintenance of immobilisation and escalation for specialist spinal opinion during the index presentation rather than deferred outpatient review.

Furthermore, FKS can present peri-operative challenges beyond the index compressive lesion. Associated anomalies may include genitourinary abnormalities, hearing impairment and vertebral artery anatomical variation. Therefore, multi-system screening and specialist anaesthetic planning are recommended especially given likely difficult airway management due to limited cervical mobility [15]-[17]. Although not encountered in this case, awareness of such vascular anomalies is important for operative planning to reduce iatrogenic injury risk [2] [18]. Despite not being the focus of this report, a thorough evaluation extending beyond the cervical spine owing to associations with genitourinary anomalies, hearing impairment and cardiac defects should form part of standard pre-operative work up for FKS patients [16] [17]. While this case focuses on the index cervical pathology, these broader considerations are important for comprehensive care planning [19].

In this case, the anatomical configuration (two fused blocks at C2 - 4 and C5 - 7 separated by a single mobile level at C4/5) likely concentrated loading at C4/5,

where both dynamic instability and severe stenosis with cord signal were demonstrated on imaging. In Samartzis terms, this is compatible with a Samartzis type II pattern, in which radicular and myelopathic symptoms are reportedly more commonly than in single-fusion patterns [5]. This further emphasises the importance of early specialist review at the time of presentation, as such configurations carry a heightened risk of instability and neurological compromise. Greater appreciation within emergency departments of KFS and similar spinal abnormalities and their clinical implications are required as early recognition and escalation may potentially mitigate progression to established cord injury.

This case report parallels previously published KFS reports which have been successfully treated with single-level ACDF [20]. However, long-term surveillance remains an important consideration as adolescents have decades for potential adjacent-level degeneration compared with typical adult degenerative ACDF cohorts [6] [7] [21].

With regards to imaging, MRI demonstrated intramedullary T2/STIR hyperintensity at C4/5. Systematic reviews have identified MRI factors, particularly number of segments involved and combined T1/T2 changes, as negative predictors of post-operative neurological recovery in degenerative cervical myelopathy [22]. Earlier meta-analysis also reports that T2 hyperintensity is associated with poorer post-operative recovery ratios compared with cases without T2 hyperintensity [23] [24]. Contemporary reviews reiterate the importance of MRI signal change for prognosis and reinforce the concept that delayed decompression may reduce reversible recovery potential, though surgery remains important to prevent progression [25]. The evidence base therefore unanimously emphasises the importance of urgent MRI imaging and enables the subsequent MRI findings to form an important foundation when counselling the patient and their family on likely functional morbidity post-operatively [26] [27].

6. Conclusions

This case describes an adolescent with previously unrecognised congenital multi-level cervical fusion consistent with Klippel-Feil syndrome. The patient presented with years of intermittent, trauma-associated transient quadriparesis and Lhermitte-type symptoms. Imaging demonstrated an unstable mobile segment at C4/5 between fused vertebrae segments with severe stenosis accompanied by focal intramedullary cord signal change.

Crucially, this report underscores a systems-level learning point that recognition of segmentation anomalies at the index presentation should prompt immobilisation, early specialist input and consideration of dynamic and advanced imaging at the time of presentation. Early identification may mitigate progression to established myelopathy. Therefore, for emergency clinicians, heightened awareness of KFS and atypical neurological presentations are essential to avoid delayed diagnosis and to enable timely intervention in a potentially preventable pathway

to cord injury.

In the setting of focal compression and instability, single-level ACDF with instrumentation is a literature-supported strategy and showed good short-term outcomes in the present case. However, long-term follow-up is essential.

Acknowledgements

This case report is written according to the CARE reporting framework. Explicit informed consent was obtained.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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