Case report

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Occult Klippel-Feil syndrome unmasked by cervical spine trauma: A case report and literature review

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Background: Klippel-Feil syndrome is a rare congenital condition characterized by the fusion of two or more cervical vertebrae. Half of the patients present with the triad of short neck, low posterior hairline and limited neck motion. Patients are at increased risk of cervical spine injury due to cervical canal stenosis. The syndrome is often associated with other congenital anomalies.

Case presentation: We report the case of a 60-year-old male who presented with a 6-hour history of neck pain following a rider motorcycle-motor vehicle road traffic crash. Cervical spine X-ray imaging showed fusion of the entire subaxial cervical spine. However, the patient had no neurologic deficit. He was managed non-operatively and discharged with a rigid cervical collar after resolution of neck pain.

Conclusion: This case report highlights an incidental discovery of Klippel-Feil syndrome during evaluation and management of cervical spine injury.

Keywords: *Klippel-Feil syndrome; cervical spine injury; cervical canal stenosis; case report*

Background

Klippel-Feil syndrome (KFS) is a rare congenital condition characterized by the non-segmentation or fusion of two or more cervical vertebrae [1]. It is classically characterized by the triad of short neck, low posterior hairline, and restricted neck movement [1].

The syndrome, first described in 1912 by Maurice Klippel and Andre Feil, results from an embryonic failure of normal segmentation of the cervical somites between the third and eighth weeks of gestation [2].

Although vertebral fusion is the typical feature, KFS is a complex disorder often involving multi-systemic anomalies, particularly involving the renal, cardiac, and central nervous systems. It may also be associated with spinal deformities such as scoliosis and Sprengel's deformity [3]. The prevalence is estimated to be around 1 in 40,000 to 42,000 live births; however, many cases likely remain undiagnosed or present later in life with secondary complications, including chronic pain, neurological deficits, or osteoarthritis due to biomechanical stress on adjacent hypermobile segments [4].

The diagnosis of KFS remains primarily clinical and radiological, with management focused on early identification of associated anomalies and prevention strategies of spinal cord injury [5]. Despite advances in understanding the genetic heterogeneity of KFS, the highly variable clinical presentation and potential for severe, life-altering comorbidities mean that each case offers valuable insights into its pathogenesis, optimal management strategies, and long-term outcomes [6]. This report details an incidental case of KFS, a rare spine condition that is often associated with other systemic anomalies, diagnosed during clinical and radiological evaluation for traumatic cervical spine injury.

Case presentation

A 60-year-old male presented to our facility six hours after a road traffic crash with posterior midline neck pain which radiated to both arms, was aggravated by movement and relieved by immobilization. He was an unhelmeted motorcycle rider who was hit from the rear by a fast moving vehicle. He fell off the motorcycle with immediate loss of consciousness which he fully regained about five minutes later. Upon regaining consciousness, he noted the onset of the neck pain. There were, however no motor, sensory or autonomic dysfunction. He had no headache, vomiting, seizures or history to suggest injuries to any other parts of the body. He was transported from the scene of the crash to our facility seated in a car without any form of neck immobilization.

Physical examination revealed a fully conscious middle-aged man with normal vital signs and an essentially normal neurological examination. Examination of the head and neck, however, showed brevicollis, a low posterior hair line and a restricted range of neck motion in all directions. He also had diffuse posterior midline neck tenderness. Both scapulae were normal in location and other regional examinations were unremarkable. A clinical diagnosis of mild traumatic brain

injury (concussion) with ASIA E cervical spine injury and suspected background Klippel-Feil syndrome was made. Further probing revealed a long-standing history of recurrent neck pain dating back to his teenage years. The patient, however, adduced the neck pain to repeated carrying of heavy objects on the head since childhood; hence he never sought medical attention.

Cervical spine X-ray showed a block cervical spine with complete fusion of the third to seventh cervical vertebrae with degenerative changes at the adjacent C2/C3 level. These changes included marginal osteophytes formation involving the inferior half of C2 and superior half of C3 in addition to a widened C2/C3 intervertebral disc space. The wasp-waist sign typically described in KFS, was also appreciated on the cervical spine X-ray – the sign refers to a reduced and smaller anteroposterior diameter at the affected levels relative to the diameter of the adjacent uninvolved discs and vertebrae (***Fig. 1A&B***).

The patient was managed non-operatively with analgesics and a rigid cervical spine collar. Further investigations, including a cervical spine CT scan or MRI and abdominopelvic ultrasound scan for a possible occult renal anomaly, were not done due to financial constraints. He was discharged home one week after the trauma with a rigid neck collar following resolution of neck pain and appropriate counseling regarding his condition, including the need to avoid high risk activities such as contact sports and possible sequelae i.e. neurologic deficits in the event of a recurrent cervical trauma. He has since been followed up in the outpatient clinic and remains neurologically intact.

Discussion

Also described as cervical vertebral fusion syndrome, Klippel-Feil syndrome (KFS) is a rare congenital condition with its hallmark being the non-segmentation or fusion of two or more cervical vertebrae [1]. Embryologically, the anomaly results from a failure of normal segmentation and differentiation of the cervical somites during the third to eighth weeks of embryogenesis [7]. Although mutations in the GDF6, GDF3, MEOX1, MYO18B, and RIPPLY2 genes have been implicated, the genetic basis remains uncertain [1]. Recent genetic analysis identified five rare variants (BAZ1B, FREM2, VANGL1, SUFU, and KMT2D) associated with cervical fusion in patients with KFS [1, 8]. The estimated incidence of KFS is about 1 in 40,000 live births, with a slight female preponderance [9]. However, the condition is often asymptomatic and may remain undiagnosed until adulthood [1]; this situation is mirrored in our case: a previously asymptomatic 60-year-old male incidentally diagnosed with KFS following a road traffic crash. Consequently, some literature suggests a significantly higher prevalence of this condition [10, 11].

The classic clinical triad of KFS - brevicollis, a low posterior hairline, and a limited cervical range of motion which was present in our patient is however only observed in about 50% of cases [12]. The absence of these overt physical signs in many patients contributes to the underdiagnosis in routine clinical practice. The clinical significance of KFS lies not only in the fusion itself but in the many other possible associated skeletal and non-skeletal anomalies, as well as the

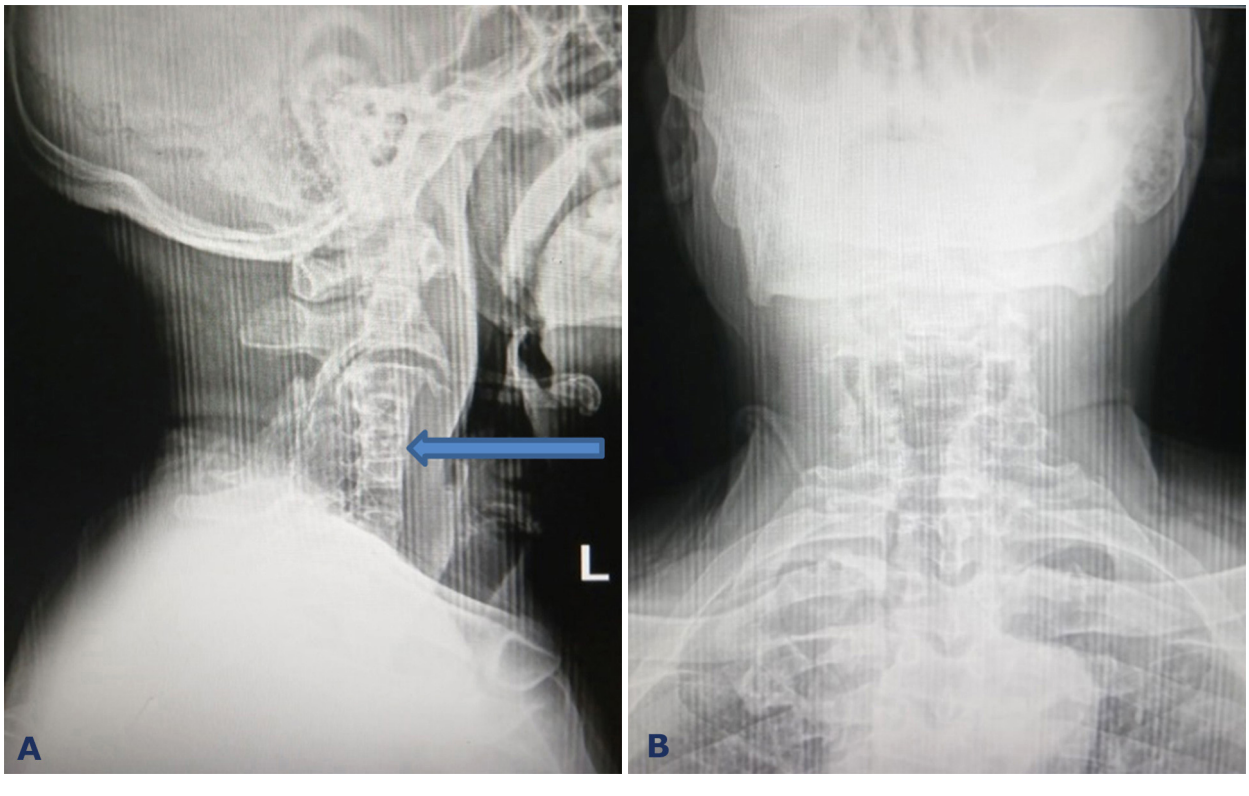

Fig. 1A & B. Cervical spine X-ray, lateral and antero-posterior views, showing a block cervical spine involving the entire subaxial cervical spine and the wasp-waist sign (blue arrow) as well as degenerative changes at the adjacent C2/3 level.

altered biomechanics of the spine [11]. The adjacent, non-fused segments are subjected to increased stress and compensatory hypermobility, leading to accelerated degenerative changes, disc herniation, spinal canal stenosis, and potential neurological deficits [13]. Some of these compensatory adjacent segment degenerative changes were observed on the cervical spine X-ray of our patient. These findings could have been evaluated in greater details using cervical spine MRI or CT scan; however finance was a major constraint in the full evaluation of this patient. Based on the pattern and number of fused segments on imaging, KFS can be classified into types I, II or III according to Samartzis *et al.* [14]. Type I KFS involves a single fused segment; type II KFS involves multiple non-contiguous fused segments, while type III KFS involves multiple contiguous fused segments [11]. The index case is a type III variant. This classification has implication on the clinical symptoms of patients with KFS. A single level cervical fusion (type I) does not increase the risk of developing limited cervical mobility, while patients with type II or III KFS are more likely to develop radiculopathy or myelopathy, as shown in a cohort study by Samartzis *et al*. involving 28 KFS patients followed over an eight-year period [14]. The patient's presentation following a road traffic crash underscores a critical risk in individuals with undiagnosed KFS: even a trivial trauma can result in significant neurological injury, such as myelopathy or quadriplegia, due to the underlying spinal abnormalities. This patient was quite fortunate to have remained neurologically intact despite having Samartzis type III KFS. Although our patient was intact neurologically, a cervical spine X-ray was performed as part of the standard trauma workup, leading to the incidental discovery of the condition. This emphasizes the importance of maintaining a high index of clinical suspicion for underlying congenital conditions in trauma cases, regardless of the patient's age or the apparent severity of the initial injury. For patients diagnosed with KFS, a comprehensive evaluation beyond the cervical spine is essential, as the syndrome may be associated with a wide spectrum of other anomalies [3, 5]. These can include renal agenesis or malformations (occurring in approximately 33% of cases), hearing impairment (around 30%), congenital heart defects (15-30%), Sprengel's deformity, and other craniospinal anomalies like Chiari malformations or spina bifida [3, 5].

While immediate management of this patient focused on the traumatic cervical spine injury, the incidental finding of KFS necessitated a broader workup for other co-existing anomalies, which may have previously gone undetected. This was not feasible due to financial constraints earlier alluded to. The differential diagnosis of KFS include syndromes such as VACTERL, Goldenhar, and Wildervanck which are often associated with multisystem anomalies such as renal, cardiac, auditory, and craniofacial defects in addition to cervical vertebral abnormalities [1].

Management of KFS is mostly conservative in asymptomatic individuals and focuses on lifestyle modifications, such as avoiding contact sports and high-risk activities that could lead to spinal cord injury [1]. The index patient was properly counseled prior to discharge from the hospital. Surgical intervention may be indicated in patients with persistent neurological pain, spinal instability, or progressive neurological deficits [1, 15]. This case highlights that KFS may remain clinically silent until adulthood, and an incidental detection warrants patient education on these inherent risks and a careful, comprehensive diagnostic workup to prevent future complications and optimize long-term outcomes.

Conclusion

This case of incidental KFS diagnosis in a 60-year-old man after a road traffic crash illustrates the hidden burden of this congenital condition. It also serves as a reminder for clinicians that KFS can present at any point during a lifetime and that a holistic evaluation and patient counseling are paramount to avoid future morbidity and mortality associated with the condition and its associated co-existing anomalies.

Disclosure

Ethics approval and consent to participate

Consent was obtained from the patient.

Competing interests

None declared

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References

1. Kadiri SE, Bqaq IE, Qajia H, Izi Z, Chat L, Jiddane M, Touarsa F, Haddad SE. Klippel-Feil syndrome revealed by post-traumatic neck pain: Case report and literature review. Radiol Case Rep. 2025 Aug 23;20(11):5686-5690. doi: 10.1016/j.radcr.2025.07.056
2. Agarwal AK, Goel M, Bajpai J, Shukla S, Sachdeva N. Klippel Feil Syndrome: A Rare Case Report. J Orthop Case Rep. 2014 Jul-Sep;4(3):53-5. doi: 10.13107/jocr.2250-0685.197
3. Yadav D, Bhattarai A, Bhandari P, Danai A, Singh UK. Klippel-Feil Syndrome Associated with Renal and Cardiac Anomalies in an Infant: A Case Report. JNMA J Nepal Med Assoc. 2023 Oct 1;61(266):819-821. doi: 10.31729/jnma.8303
4. Frikha R. Klippel-Feil syndrome: a review of the literature. Clin Dysmorphol. 2020 Jan;29(1):35-37. doi: 10.1097/MCD.0000000000000301
5. Siddiqui F, Talal Ashraf M, Khuzzaim Khan M, Admani B, Sam SJ, Imran M, Hameed M. A Comprehensive Approach to the Diagnosis and Management of Klippel Feil Syndrome. Arch Razi Inst. 2023 Dec 30;78(6):1868-1872. doi: 10.32592/ARI.2023.78.6.1868
6. Tracy MR, Dormans JP, Kusumi K. Klippel-Feil syndrome: clinical features and current understanding of etiology. Clin Orthop Relat Res. 2004 Jul;(424):183-90.
7. Umamaheshwar KL, Sehrawat A, Parashar MK, Mavade K. Two case reports of an unusual association between Klippel-Feil syndrome and amyotrophic lateral sclerosis: Do they share same genetic defect? Ann Indian Acad Neurol. 2013 Oct;16(4):705-7. doi: 10.4103/0972-2327.120456
8. Li ZQ, Geng MZ, Zhao S, Wu ZH, Zhang JG, Wu N, Wang YP. [Clinical Characteristics and Genetic Analysis of Klippel-Feil Syndrome]. Zhongguo Yi Xue Ke Xue Yuan Xue Bao. 2021 Feb 28;43(1):25-31. Chinese. doi: 10.3881/j.issn.1000-503X.12629
9. Nouri A, Tetreault L, Zamorano JJ, Mohanty CB, Fehlings MG. Prevalence of Klippel-Feil Syndrome in a Surgical Series of Patients with Cervical Spondylotic Myelopathy: Analysis of the Prospective, Multicenter AOSpine North America Study. Global Spine J. 2015 Aug;5(4):294-9. doi: 10.1055/s-0035-1546817
10. Gruber J, Saleh A, Bakhsh W, Rubery PT, Mesfin A. The Prevalence of Klippel-Feil Syndrome: A Computed Tomography-Based Analysis of 2,917 Patients. Spine Deform. 2018 Jul-Aug;6(4):448-453. doi: 10.1016/j.jspd.2017.12.002

11. Moses JT, Williams DM, Rubery PT, Mesfin A. The prevalence of Klippel-Feil syndrome in pediatric patients: analysis of 831 CT scans. J Spine Surg. 2019 Mar;5(1):66-71. doi: 10.21037/jss.2019.01.02
12. Litrenta J, Bi AS, Dryer JW. Klippel-Feil Syndrome: Pathogenesis, Diagnosis, and Management. J Am Acad Orthop Surg. 2021 Nov 15;29(22):951-960. doi: 10.5435/JAAOS-D-21-00190
13. Reyes-Sánchez A, Zárate-Kalfópulos B, Rosales-Olivares LM. Adjacent segment disease in a patient with klippel-feil syndrome and radiculopathy: surgical treatment with two-level disc replacement. SAS J. 2007 Nov 1;1(4):131-4. doi: 10.1016/SASJ-2007-0114-CR
14. Samartzis DD, Herman J, Lubicky JP, Shen FH. Classification of congenitally fused cervical patterns in Klippel-Feil patients: epidemiology and role in the development of cervical spine-related symptoms. Spine (Phila Pa 1976). 2006 Oct 1;31(21):E798-804. doi: 10.1097/01.brs.0000239222.36505.46
15. Ding L, Wang X, Sun Y, Zhang F, Pan S, Chen X, Diao Y, Zhao Y, Xia T, Li W, Zhou F. Prevalence and Risk Factors of Surgical Treatment for Klippel-Feil Syndrome. Front Surg. 2022 Jun 7;9:885989. doi: 10.3389/fsurg.2022.885989