SAS Journal of Medicine
Abbreviated Key Title: SAS J Med
ISSN 2454-5112
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Klippel-Feil Syndrome Associated with Sprengel's Syndrome: A Rare Case Report

O. Kanali[1*], H. Chenter[1], K. Outaghyame[1], A. El Hajjami[1], Y. Bouktib[1], B. Boutakioute[1], M. Ouali Idrissi[1], N. Cherif Idrissi Ganouni[1]

[1]Department of Radiology, CHU Mohammed VI, University of Cadi Ayyad, Marrakech, Morocco

DOI: https://doi.org/10.36347/sasjm.2024.v10i10.011 | **Received:** 25.08.2024 | **Accepted:** 02.10.2024 | **Published:** 05.10.2024

***Corresponding author:** O. Kanali
Department of Radiology, CHU Mohammed VI, University of Cadi Ayyad, Marrakech, Morocco

Abstract **Case Report**

Klippel-Feil Syndrome (KFS) is a rare congenital disorder characterized by the fusion of cervical vertebrae, often associated with other anomalies such as Sprengel's deformity. We report the case of a 61-year-old female patient presenting with chronic neck pain and paresthesia in the lower limbs, worsened after a minor domestic accident. Cervical CT imaging revealed multisegmental vertebral fusion (C2-C3, C7-T1) and Sprengel's deformity with an omovertebral bone. This case highlights the importance of early diagnosis and comprehensive imaging, including X-rays, CT scans, and MRIs, for the optimal management of KFS and its associated complications. Awaiting the correction before proceeding with the payment.

Keywords: Klippel-Feil syndrome, Sprengel deformity, Cervical vertebral fusion, Omovertebral bone, Imaging.

INTRODUCTION

Klippel-Feil Syndrome (KFS) is a rare congenital disorder first described by Maurice Klippel and André Feil in 1912, characterized by the fusion of cervical vertebrae [1], with a notable prevalence in females, accounting for about 60% of cases [2].

The classical presentation includes a triad of features: a short neck, a low posterior hairline, and a limited range of neck movements, particularly lateral bending; however, fewer than 50% of patients exhibit all three elements [3].

KFS frequently coexists with other congenital anomalies, notably Sprengel's Syndrome, which is defined by the abnormal positioning of the scapula. In approximately 30% of cases, the scapula is tethered to the cervical spine by fibrous tissue, cartilage, or an omovertebral bone, restricting shoulder abduction beyond 90 degrees [4].

The rare co-occurrence of these syndromes presents unique challenges for diagnosis and management.

We present the case of a 61-year-old woman who experienced neck pain and paresthesia, ultimately leading to a diagnosis of Klippel-Feil Syndrome associated with Sprengel's Syndrome.

CASE REPORT

We present the case of a 61-year-old female with a longstanding history of chronic cervicalgia and paresthesia in both lower limbs. Her symptoms had persisted for over three years despite conservative treatment. She reported a progressive decrease in her ambulatory capacity, with her walking distance reduced to less than 10 meters. Recently, she presented to the emergency department following an aggravation of her symptoms after a minor domestic trauma.

Upon clinical examination, the patient exhibited classical phenotypic features of Klippel-Feil Syndrome (KFS), including a short, webbed neck, limited cervical range of motion, and a low posterior hairline. Neurologically, there was evidence of pyramidal tetraparesis, with spasticity and hyperreflexia in both upper and lower limbs. The patient also reported persistent lower limb weakness and numbness, further raising concerns for spinal cord involvement.

An emergent cervical computed tomography (CT) scan was performed, which revealed complex vertebral anomalies. There was a left-sided hemivertebra

at C3, which was congenitally fused with C2, and a further fusion was noted between C7 and the first thoracic vertebra (D1). The scan also demonstrated multiple posterior elements' closure defects at C1, C4, C5, C6, C7, and D3. Additionally, the posterior left lamina of C6 was elongated and hypertrophic. Importantly, the imaging identified a high-riding left scapula, consistent with Sprengel's deformity, and an omovertebral bone extending from the scapula to the cervical spine.

Based on these findings, the diagnosis of Klippel-Feil Syndrome type II was confirmed, complicated by Sprengel's deformity and posterior vertebral element closure defects. The fusion of vertebrae, particularly at C2-C3 and C7-D1, contributed to altered spinal biomechanics, increasing the risk for myelopathy. This explained the patient's progressive neurological decline, which was exacerbated following her minor trauma.

In this patient, the associated Sprengel deformity, a congenital elevation of the scapula, was notable for the presence of an omovertebral bone, which often restricts shoulder mobility. This congenital anomaly further complicates the clinical management, as it is frequently associated with other musculoskeletal malformations.

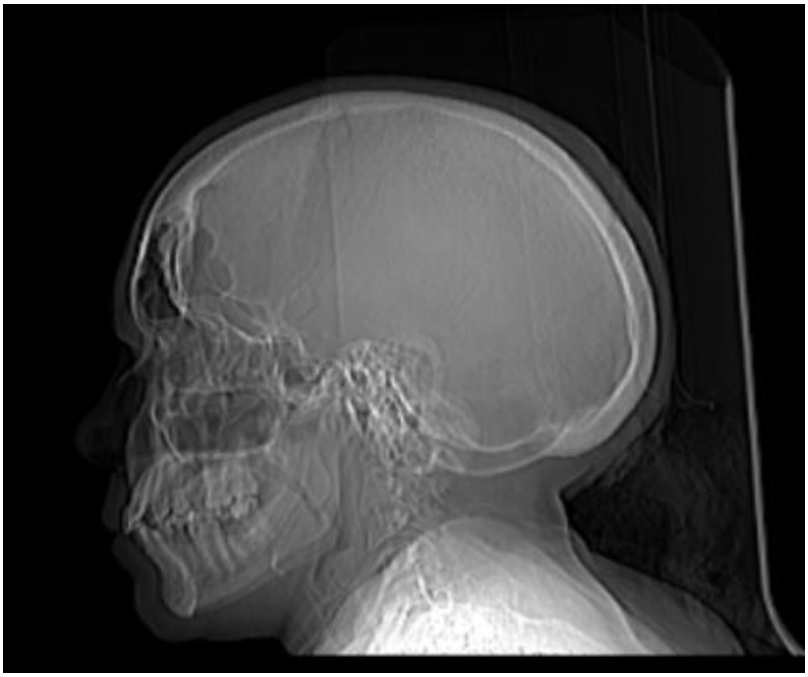

Figure 1: Scout view of the cervical spine CT scan showing a short neck

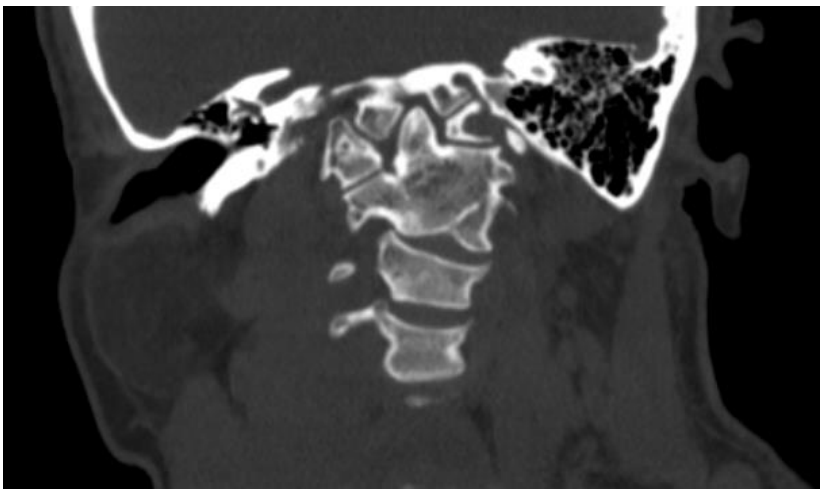

Figure 2: Coronal CT scan with bone window shows a hemivertebra at C3 fused with C2

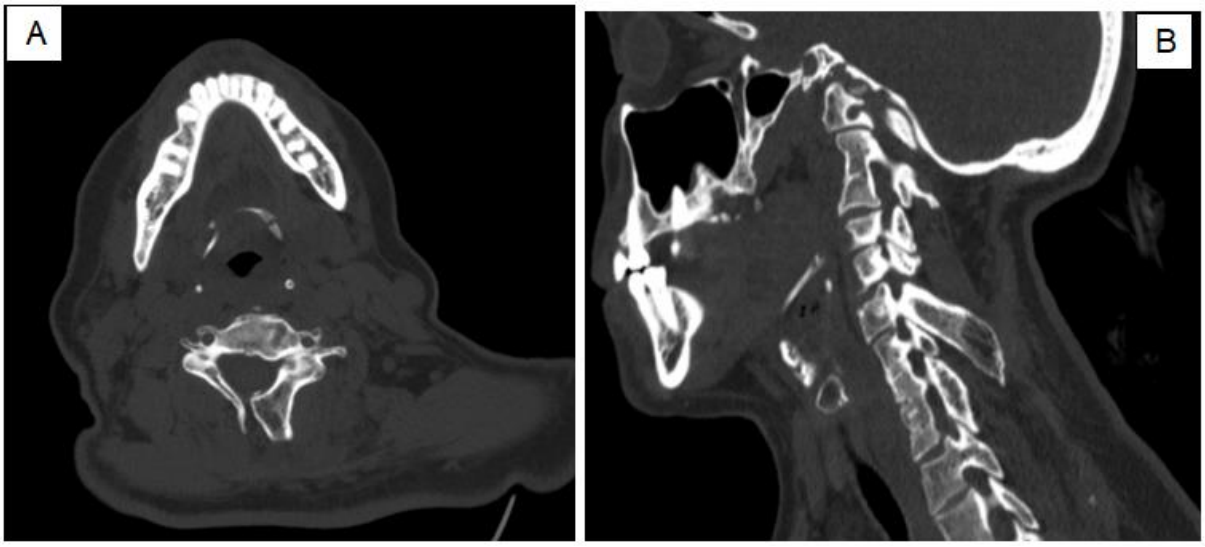

Figure 3: CT scan of the cervical spine with bone window (A) axial and (B) sagittal images shows a posterior vertebral element closure defect at the level of C6, along with elongation and hypertrophy of the left posterior lamina of C6

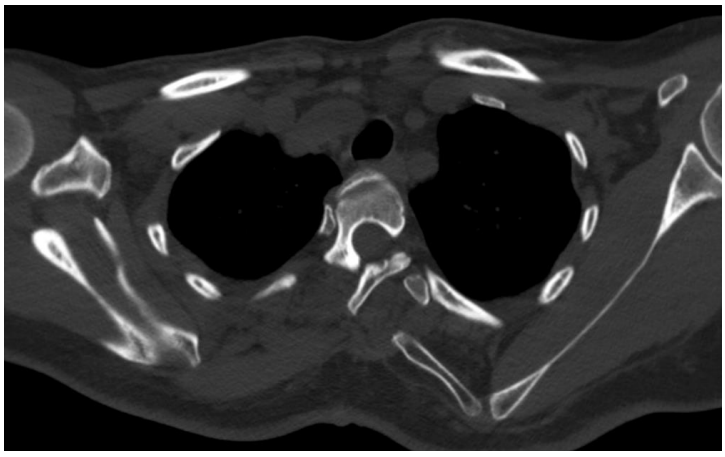

Figure 4: CT scan of the cervical spine with bone window shows elevation of the left scapula with an omovertebral bone on its superomedial surface

DISCUSSION

Klippel-Feil Syndrome (KFS) is a rare disorder characterized by the abnormal fusion of cervical vertebrae, leading to a triad of symptoms: a short neck, low posterior hairline, and limited neck mobility. Fewer than 50% of patients exhibit all three features, while others are classified as having Klippel-Feil variants [3]. This fusion increases the risk of neurological injuries due to altered spinal biomechanics, raising the likelihood of hypermobility, spondylolisthesis, and stenosis.

The exact cause of KFS remains unclear but is believed to arise from genetic mutations [5] related to failures in normal vertebral segmentation during the fourth week of gestation [6]. KFS is frequently associated with anomalies in the skeletal system and other congenital defects. In our case, we observed spinal deformity along with abnormal scapular positioning.

Regarding its prevalence, KFS occurs in approximately 1 in 42,000 live births, with a female predominance of 60%. Fusion typically occurs at the C2-C3 level in most major studies [7], while multilevel fusion is rare, accounting for less than 10% of cases. Our patient was a woman who exhibited multilevel fusion at C2-C3 and C7-D1, leading to paresthesia and tetra pyramidal syndrome following minor trauma.

In addition to its prevalence, KFS is often linked to other congenital lesions, including congenital scoliosis or kyphosis (60%), renal disease (35%), synkinesis or mirror movements (20%), and Sprengel deformity (30%) [8, 9]. In our case, we found Sprengel deformity but no clinical evidence of renal or congenital heart disease.

The patient presented with the classical triad of low posterior hairline, short neck, and limited cervical range of motion.

Diagnosis of KFS is established radiologically, starting with a preliminary evaluation by neck X-ray,

followed by CT and MRI for detailed imaging. KFS is classified into three types based on the extent and location of vertebral fusion, as originally described by Klippel and Feil [10]:

- **Type I**: Fusion of multiple cervical and upper thoracic vertebrae.
- **Type II**: Fusion of two or three vertebrae with associated hemivertebrae, occipitocervical fusion, or other anomalies.
- **Type III**: Cervical fusion with fusion of lower thoracic or lumbar vertebrae.

Plain radiographs can reveal features such as fused facets and spinous processes, anteroposterior narrowing of the vertebral bodies, hemivertebrae, omovertebral bone, spina bifida, associated scoliosis, and Sprengel deformity. CT and MRI are essential for comprehensive assessment [11], with CT providing detailed imaging and potential demonstration of canal stenosis, while MRI is particularly recommended for patients exhibiting neurological deficits [10].

In summary, early diagnosis and thorough imaging are crucial for effective management of KFS and its associated complications.

CONCLUSION

Klippel-Feil Syndrome (KFS) is a rare congenital disorder characterized by abnormal cervical vertebrae fusion, which can lead to significant complications. It is often associated with other anomalies, such as Sprengel Syndrome. Radiological imaging—particularly X-ray, CT, and MRI—is crucial for confirming the diagnosis, assessing vertebral fusion, and identifying associated defects. Timely and accurate imaging is essential for guiding treatment decisions and improving patient outcomes in KFS.

REFERENCES

1. Jones, K. L. 5th ed. Philadelphia: WB Saunders Company; 1997. Smith's recognizable pattern of human malformation.
2. Stephen, R. P. *Ortho Secrets.* 3rd Ed 2003. Cervical Spine Disease.
3. Youmans, J. R., & Winn, H. R. Philadelphia, PA: Saunders; 2011. Youmans Neurological Surgery.
4. Paul, W. E., & Brain, E. B. *Ortho Secrets.* 3rd Ed 2003. Miscellaneus Congenital Disorders.
5. Nauñay, V. G., & Carvajal, I. M. (2019). Klippel-Feil autosomal dominant syndrome: a malformation of vertebral segmentation. *Rev Chil Pediatr*, *90*(2), 194-201.
6. Valdés, A., Pérez, H., García, R., & López, A. (2010). Embriología humana. *La Habana: Editorial Ciencias Médicas*, 8, 103-108. https://www.academia.edu/14839535/Embriolog%C3%ADa_humana
7. Moses, J. T., Williams, D. M., Rubery, P. T., & Mesfin, A. (2019). The prevalence of Klippel-Feil syndrome in pediatric patients: analysis of 831 CT scans. *Journal of Spine Surgery*, *5*(1), 66-71.
8. Van Kerckhoven, M. F., & Fabry, G. (1989). The Klippel-Feil syndrome: a constellation of deformities. *Acta orthopaedica belgica*, *55*(2), 107-118.
9. Sudhakar, A. S., Nguyen, V. T., & Chang, J. B. (2008). Klippel-Feil syndrome and supra-aortic arch anomaly: A case report. *Int J Angiol*, *17*(3), 118.
10. Weerakkody, Y., Sharma, R., & Wahab, R. Klippel-Feil syndrome. Reference article, Radiopaedia.org (Accessed on 22 Sep 2024)
11. Nagib, M. G., Maxwell, R. E., & Chou, S. N. (1985). Klippel-Feil syndrome in children: clinical features and management. *Child's Nervous system*, *1*(5), 255-263.