International Journal of Research in Medical Sciences
William M et al. Int J Res Med Sci. 2024 Oct;12(10):3877-3880
www.msjonline.org
pISSN 2320-6071 | eISSN 2320-6012

DOI: https://dx.doi.org/10.18203/2320-6012.ijrms20242952

Case Report

Type III Klippel-Feil syndrome with basilar invagination, syringomyelia, and spina bifida: a co-existence of rare congenital abnormalities

Munna William[1]*, Noor Fatima[1], Hassan Mehdi[2], Ahmad Shabbir[1], Muhammad Nabeel[2], Abdul Sattar Anjum[1]

[1]Department of diagnostic radiology and imaging, Nishtar Medical University, Multan, Pakistan
[2]Department of neurosurgery, Nishtar Medical University, Multan, Pakistan

Received: 05 August 2024
Accepted: 17 September 2024

***Correspondence:**
Dr. Munna William,
E-mail: williammunna9@gmail.com

ABSTRACT

Klippel-Feil syndrome (KFS) is a rare congenital skeletal anomaly characterized by the fusion of cervical vertebrae, often leading to a shortened neck, a low posterior hairline, and restricted neck movement. Despite its infrequency, KFS can present as a complex multisystem disorder including neurological and non-neurological defects, which rarely occur simultaneously. This report presents the case of a 20-year-old male diagnosed with KFS exhibiting basilar invagination, syringomyelia, and spina bifida, resulting in myelopathy and radiculopathy. Diagnostic imaging confirmed multiple vertebral fusions and associated neural tissue abnormalities. Surgical intervention, including spinal decompression, resulted in significant symptom relief. This case underscores the diverse clinical presentations and diagnostic challenges associated with KFS, necessitating a comprehensive approach with multiple imaging modalities for accurate diagnosis and tailored management. Prompt identification and intervention are crucial, particularly in cases of symptomatic anomalies, to prevent neurological decline and optimize patient outcomes. The presence of symptomatic basilar invagination underscores the importance of timely surgical intervention to prevent future neurological complications.

Keywords: Klippel-Feil syndrome, Basilar invagination, syringomyelia, Spina bifida, Fused vertebrae

INTRODUCTION

Klippel-Feil syndrome (KFS) is a rare congenital bony disorder that arises from abnormalities in the spine's normal development during the first three to eight weeks of intrauterine life.[1] It is estimated to occur in 1 out of 40,000–42,000 live births, slightly more common in females.[2] It is characterized by the fusion of two or more cervical vertebrae, which results in a triad of short neck, low posterior hairline, and restricted neck mobility.[3] The most commonly affected cervical vertebrae are the upper three; the lower ones are rarely affected. Furthermore, the presence of thoracic or lumbar vertebrae fusion along with cervical vertebrae, a type III KFS, is a very rare incident, seen in only 6-7% of KFS cases.[4] KFS extends beyond just vertebral anomalies, involving multiple other systems and leading to a spectrum of skeletal and non-skeletal abnormalities.[5] Basilar invagination (BI) is one of them in which odontoid process protrudes into foramen magnum, potentially compressing the brainstem and upper cervical spinal cord, causing neurological deficits.[6] Spina bifida and syringomyelia are the other common skeletal and neurological deformities seen in association with KFS.[5] The occurrence of these deformities with KFS in isolation is common. However, their occurrence with KFS as a combined is uncommon and rarely reported. Here we present a case of KFS with

BI, syringomyelia, and spina bifida leading to myelopathy and radiculopathy.

CASE REPORT

A 20-year-old man presented to the outpatient department with complaints of progressive right upper limb weakness since the age of 12. He also mentioned that for the last two months, he had on-and-off constipation and low back pain. The pain was severe, radiating to the left leg, with pins and needle sensations in the left foot. He had also started experiencing weakness in the postural muscles, causing difficulty standing after sitting for some time. He had no significant past medical or surgical history and had achieved developmental milestones at an appropriate age. Any family history of congenital spine abnormalities was denied.

General physical examination revealed short stature, short neck, low posterior hairline, and limited neck movements, more pronounced in the rotational direction (Figure 1). On neurological examination, the power in the right limb was 3/5, with no sensory deficit. The rest of the examinations were normal. With high suspicion of KF syndrome, multiple imaging modalities were adopted, including X-ray, computed tomography (CT), and magnetic resonance imaging (MRI). X-ray showed multiple block vertebrae involving the cervical spines on the lateral view. CT spine showed multiple fused vertebral bodies and their posterior elements in the lower cervical-upper dorsal (C5-D3), lower dorsal (D8, D9), and lumbar (L2, L3) regions (Figure 2). It also revealed a bifid spine at the C4 level with no evidence of lipo/lipomyelocele or myelo/meningomyelocele on MRI (Figure 3). Furthermore, MRI revealed basilar invagination along with syrinx formation in the cervical and dorsal spinal cord. The basilar invagination was displacing the medulla oblongata and cervico medullary junction posteriorly (Figure 4). Based on the clinical and radiological findings, a final diagnosis of KF syndrome with basilar invagination, syringomyelia, and spina bifida occulta was made. KF syndrome was further categorized as type III based on the presence of both cervical and lumbar vertebrae fusion.

The patient was referred to the neurosurgery department, where he underwent surgery for basilar invagination. Spinal decompression was performed to release the tension on the spinal cord and resolve the syrinx. Via posterior instrumentation, the posterior arch of C1 was removed, and the dura was opened. Following resection, C1-C2 fixation was performed to stabilize the spine. Vertebral fusions at different levels were managed conservatively. It was decided not to treat spina bifida as it was asymptomatic in the current scenario. Non-steroidal anti-inflammatory drugs were given for pain. Following surgery, there was a significant improvement in radiculopathy and constipation. Right upper limb weakness gradually improved over six months. MRI was done at the end of the sixth month post-surgery which showed the resolution of syrinx.

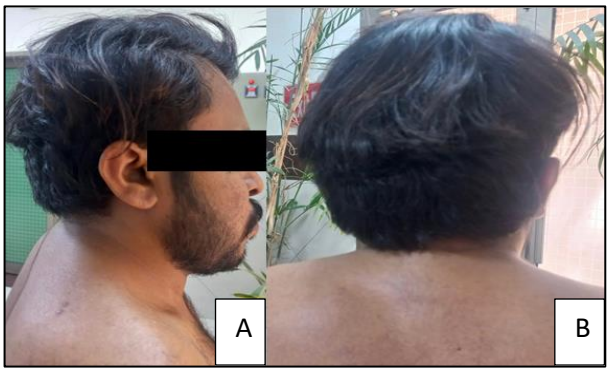

Figure 1 (A and B): Patient showing short neck and low posterior hairline.

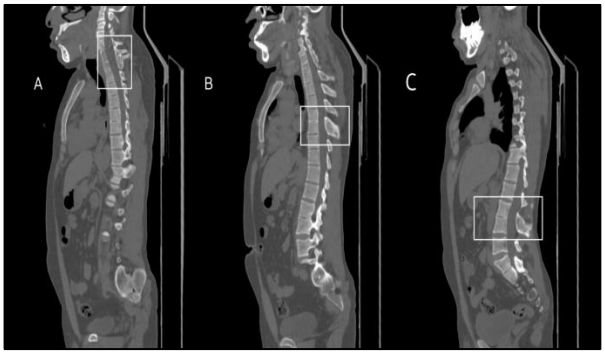

Figure 2: Plain CT whole spine bone window (Sagittal sections) showing multiple fused vertebral bodies and their posterior elements in lower cervical, upper dorsal, lower dorsal and in lumbar spine (white boxes). (A) Fused vertebral bodies and posterior elements of C5 till D3. (B)Partially fused vertebral bodies and fused posterior elements of D8, D9. (C) Partially fused vertebral bodies and fused posterior elements of L2, L3.

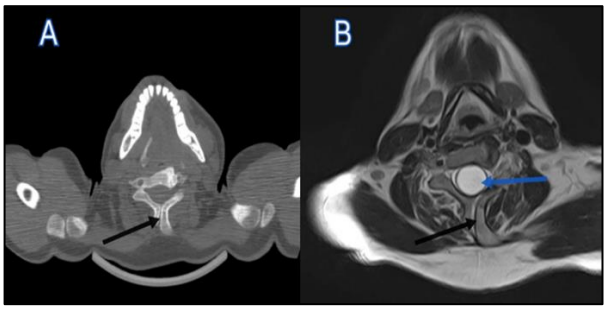

Figure 3: (A) Plain CT bone window (axial sections) showing bifid spine at c4 (black arrow). (B) MRI cervical spine t2w images (axial sections) showing bifid spine at c4 (black arrow), with no associated lipo/lipomyelocele or myelo/meningomyelocele, and syrinx formation (blue arrow) in the cervical cord.

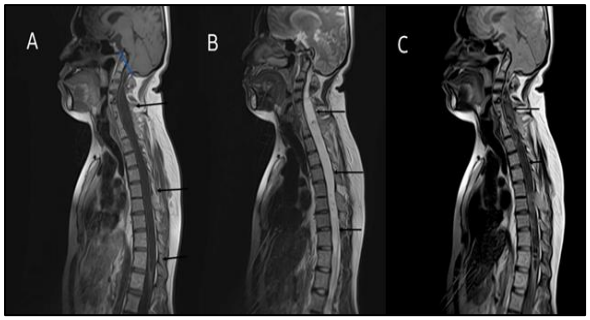

Figure 4: MRI cervical spine (sagittal sections): (A) T1W (B) T2W and (C) Flair sequence.

T1W image shows the tip of the odontoid projecting above the line joining the basion with opisthion on the mid-sagittal plane (blue line), representing basilar invagination. This is causing narrowing of the foramen magnum, with resultant displacement of the medulla oblongata and cervicomedullary junction posteriorly; however, no abnormal MR signals were seen returning from these (Figure 4).

Black arrows in A, B and C show the synrix formation in cervical and dorsal spinal cord that is hypointesnse on T1W, hyperintense on T2W, with low signal on fluid supressed sequences (FLAIR images), confirming synrix formation (Figure 4).

DISCUSSION

The case presented here highlights the rarity of KFS in conjunction with BI, syringomyelia, and spina bifida occulta. KFS itself is an uncommon skeletal congenital disorder, estimated to occur in 1 out of 40,000 to 42,000 live births globally, with a slight female predominance2. The pathophysiology of KFS involves the failure of mesodermal somites to segment properly during the third to eighth week of gestational development, leading to congenital fusion of cervical vertebrae. While most cases of KFS occur sporadically, there have been reports of autosomal dominant or recessive inheritance patterns, with potential involvement of genes such as growth differentiation factor (GDF) 6 or 3 or mesenchyme homeobox (MEOX). In the presented case, the fusion of multiple cervical, dorsal, and lumbar vertebrae, along with the presence of basilar invagination and syringomyelia, suggests a complex interplay of genetic and developmental factors contributing to the observed rare coexistence of congenital abnormalities.

Of the patients, only 50% exhibit the classic clinical trial of KFS, which consists of a short neck, a low posterior hairline, and limited neck mobility.[3] However, it was evident in our patient. Although KFS is congenital, mild form may go unnoticed at birth due to lack of symptoms and are detected incidentally or when symptoms develop in the later decades of life.[8,9] However, extensive vertebral fusion as seen in type III KFS, and the presence of BI and syringomyelia alongside may result in earlier symptoms of myelopathy and radiculopathy due to spinal cord and nerve root compression, as seen in our case.

KFS encompasses three recognized types, each characterized by distinct patterns of vertebral fusion. Type I involves the fusion of multiple cervical vertebrae into a single block, while Type II manifests as incomplete segmentation of one or two cervical levels, sometimes extending to the occipito-atlantal region. Type III, as observed in our case, represents a combination of cervical spine fusion seen in Types I or II, along with concurrent segmentation abnormalities in the lumbar or lower dorsal spine.[1] The classification of our case as Type III KFS underscores the extensive nature of vertebral fusion and associated anomalies observed in imaging studies.

KFS is frequently associated with a spectrum of congenital anomalies affecting various organ systems. These include congenital heart malformations, renal abnormalities, hearing impairment, scoliosis, and sprengel deformity, among others.[5] Notably, neural tissue defects such as syringomyelia, diastematomyelia, and spina bifida occulta are commonly observed in conjunction with KFS.[5,10] In our case, the presence of basilar invagination and syringomyelia highlights the association of KFS with craniovertebral junction abnormalities and spinal cord pathology, further emphasizing the complexity of the patient's presentation.

The diagnosis of KFS involves a combination of clinical and imaging studies. Clinical examination shows a classic triad in half of the cases. Imaging involves multiple modalities, including X-ray, CT, and MRI. X-rays are the initial imaging modality that can reveal vertebral fusion. CT provides more detailed images, helping to precisely evaluate the extent and pattern of vertebral fusion.[7,11] With X-rays and CT, multiple fused vertebral bodies and their posterior elements at different levels, as well as spina bifida, were accurately identified in our case. Furthermore, MRI helped to assess the spinal cord and neural structures, aiding in identifying complications such as brainstem compression and syringomyelia.

Asymptomatic or mild KFS is managed conservatively with medication, activity modification, physical therapy, or bracing and traction. However, surgery is preferred when patients develop myelopathy or when there is co-existing basilar invagination compressing the brain stem with or without resultant syringomyelia. The primary goal of surgery is to decompress the brainstem, restore normal anatomy, and improve neurological functions.[11,12] In this case, following spinal decompression, the patient's symptoms gradually improved with the resolution of syrinx.

CONCLUSION

Although KFS is rare, there should be high suspicion in individuals presenting with neurological symptoms along with the classic clinical trial of a short neck, a low

posterior hairline, and limited neck mobility. Immediate imaging with X-ray, CT, and MRI is crucial to confirm the diagnosis, identify the associated skeletal and non-skeletal disorders, and plan the treatment. The co-existence of symptomatic basilar invagination requires prompt surgical decompression of the brainstem to prevent further deterioration of neurological symptoms.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Vaidyanathan S, Hughes PL, Soni BM, Singh G, Sett P. Klippel-Feil syndrome-the risk of cervical spinal cord injury: a case report. BMC Fam. 2002;22:-3-6
2. Frikha R. Klippel-Feil syndrome: a review of the literature. Clin Dysmorphol. 2020;29(1):35-7.
3. Samartzis D, Kalluri P, Herman J, Lubicky JP, Shen FH. "Clinical triad" findings in pediatric Klippel-Feil patients. Scoliosis Spinal Disord. 2016;11:15.
4. Ding L, Wang X, Sun Y. Prevalence and Risk Factors of Surgical Treatment for Klippel-Feil Syndrome. Front Surg. 2022;9:885989.
5. Naikmasur VG, Sattur AP, Kirty RN, Thakur AR. Type III Klippel-Feil syndrome: case report and review of associated craniofacial anomalies. Odontology. 2011;99(2):197-202.
6. Donnally III CJ, Munakomi S, Varacallo M. Basilar Invagination. In: StatPearls. Treasure Island (FL): StatPearls. Available at: https://pubmed.ncbi.nlm.ni. Accessed on 17 May 2024.
7. Menger RP, Rayi A, Notarianni C. Klippel Feil Syndrome. In: StatPearls [Internet]. Treasure Island (FL): StatPearls. Available at: https://www.ncbi.nlm.nih.gov. Accessed on 17 May 2024.
8. 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;16(4):705-7.
9. 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;6(4):448-53.
10. Pang D, Dias MS. Cervical myelomeningoceles. Neurosurgery. 1993;33(3):363-373.
11. Litrenta J, Bi AS, Dryer JW. Klippel-Feil Syndrome: Pathogenesis, Diagnosis, and Management. J Am Acad Orthop Surg. 2021;29(22):951-60.
12. Brokinkel B, Wiebe K, Hesselmann V. Surgical treatment in a patient with Klippel-Feil syndrome and anterior cervical meningomyelocele: a case report and review of literature. Eur Spine J. 2013;22(3):517-20.

Cite this article as: William M, Fatima N, Mehdi H, Shabbir A, Nabeel M, Anjum AS. Type III Klippel-Feil syndrome with basilar invagination, syringomyelia, and spina bifida: a co-existence of rare congenital abnormalities. Int J Res Med Sci 2024;12:3877-80.