Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

When the Craniovertebral Junction Fuses: A Rare Case of Klippel-Feil Syndrome with Anterior Atlanto-Occipital Assimilation and Basilar Invagination

A. Bouelhaz[1*], M.K. Diakité[1], S. Aouaq[1], Y. Bouktib[1], A. El Hajjami[1], B. Boutakioute[1], M. Ouali Idrisi[1], N Cherif Idrissi El Ganouni[1]

[1]Department of Radiology, Arrazi Hospital, University Hospital Center Mohamed VI, Marrakech, Morocco

DOI: https://doi.org/10.36347/sjmcr.2025.v13i10.090 | **Received:** 28.08.2025 | **Accepted:** 23.10.2025 | **Published:** 25.10.2025

***Corresponding author:** A. Bouelhaz
Department of Radiology, Arrazi Hospital, University Hospital Center Mohamed VI, Marrakech, Morocco

Abstract | **Original Research Article**

Klippel-Feil syndrome (KFS) is a rare congenital disorder caused by faulty segmentation of the cervical vertebrae during early embryonic development. It is often associated with other craniovertebral anomalies such as atlanto-occipital assimilation and basilar invagination, which may remain asymptomatic or lead to neurological complications. We report the case of a 61-year-old male who presented following craniofacial trauma. While the cerebral CT was unremarkable, cervical imaging incidentally revealed C2–C3 vertebral body fusion, a posterior arch defect of C1, complete anterior assimilation of the atlas to the occiput, and basilar invagination. These findings were consistent with type II Klippel-Feil syndrome. The patient remained neurologically intact. This case highlights the importance of systematic imaging of the craniovertebral junction, even in asymptomatic individuals, as congenital anomalies may predispose to instability or neurological compromise, particularly in trauma settings. Early recognition is essential for proper management and risk assessment.

Keywords: Craniovertebral junction anomalies, Klippel-Feil syndrome, Atlanto-occipital assimilation, Basilar invagination, CT Scan.

INTRODUCTION

The craniovertebral junction is a complex anatomical region whose integrity is essential for the stability and function of the head and neck. [1] Klippel-Feil syndrome (KF) is a congenital condition characterized by the fusion of two or more cervical vertebrae, resulting from a segmentation failure during embryonic development. First described by Klippel and Feil in the early 20th century, [2] it classically presents with a triad of a short neck, low posterior hairline and limited cervical range of motion; associated anomalies may include Sprengel deformity, scoliosis, urinary tract malformations, congenital heart defects and auditory impairment. The overall incidence is estimated at approximately 1 in 42,000 births, with a female predominance of around 60 %. [3]

Atlanto-occipital assimilation is a congenital anomaly in which the atlas (C1) is partially or completely fused to the occipital bone, potentially involving the anterior arch, the posterior arch, or both, and may remain asymptomatic until nerve or vascular compression arises. [4] Basilar invagination, by contrast, is characterized by an abnormal upward displacement of the odontoid process through the foramen magnum, narrowing the space available for the spinal cord and causing neck pain, neurological deficits, and in severe cases complications such as syringomyelia or hydrocephalus. [5]

We report a rare case combining Klippel-Feil syndrome with C1 arch defects, complete anterior atlanto-occipital assimilation, and basilar invagination. The aim of this report is to illustrate the complexity of such malformations and emphasize the critical role of imaging in their diagnosis and management.

PATIENTS AND METHODS

Observations:

Mr. H. Z., a 61-year-old male with no notable medical history, presented to the emergency department following a road traffic accident with a craniofacial point of impact. The initial cerebral CT scan showed no intracranial abnormalities. However, the same scan revealed hematoma-related soft tissue infiltration of the

right frontal and zygomatic regions, containing spontaneously hyperdense materials suggestive of foreign bodies.

Incidentally, on cervical slices, several congenital vertebral anomalies were identified. These included a posterior arch defect of the atlas (C1) and a complete fusion of the anterior arch of C1 with the occipital bone, consistent with anterior atlanto-occipital assimilation. In addition, there was a complete fusion of the vertebral bodies of C2 and C3, corresponding to Klippel-Feil syndrome type II. A basilar invagination was also noted, along with a left-convex scoliotic attitude of the cervical spine. (Figure 1)

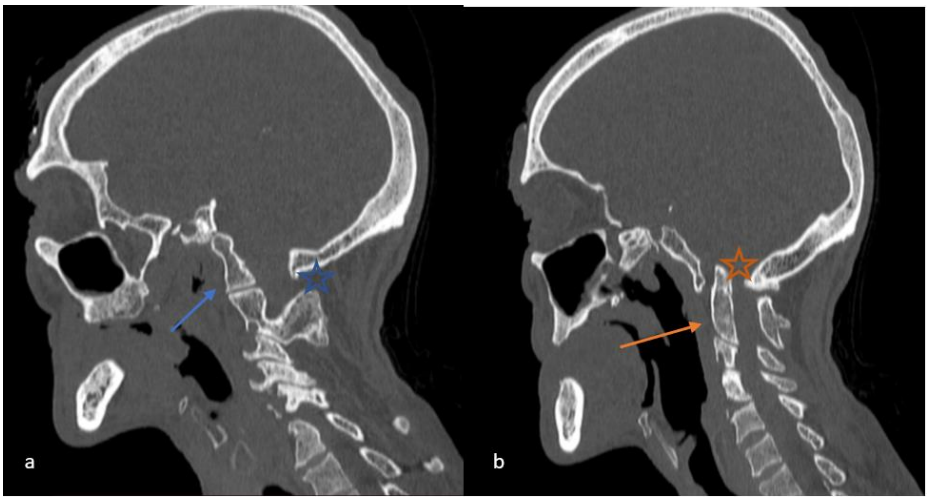

Figure 1: Sagittal CT bone window images (a, b) showing a posterior arch defect of the atlas (C1) (a ; blue asterisk), complete fusion of the anterior arch of C1 with the occipital bone (a ; blue arrow) indicating anterior atlanto-occipital assimilation, complete fusion of the vertebral bodies of C2 and C3 (b orrange arrow) consistent with Klippel-Feil syndrome type II and basilar invagination (b ; orrange asterisk).

The diagnosis of a complex craniovertebral junction malformation was made based on these findings, discovered incidentally during trauma assessment. The patient remained neurologically intact and did not report any chronic cervical symptoms prior to the incident.

DISCUSSION

Klippel-Feil syndrome (KFS) is a rare and heterogeneous congenital condition characterized by the fusion of two or more cervical vertebrae due to faulty segmentation during the 3rd to 8th weeks of embryonic development. [2] The syndrome is classically associated with a triad of clinical signs—short neck, low posterior hairline, and limited cervical mobility—although this complete presentation is seen in fewer than 50% of patients. [6] The estimated incidence is approximately 1 in 40,000 to 42,000 births, with a recognized female predominance. [3]

Our patient presented with a type II Klippel-Feil pattern, defined by the fusion of two adjacent cervical vertebrae (C2–C3), associated with additional anomalies at the craniovertebral junction. These include a posterior arch defect of C1 and a complete anterior assimilation of the atlas with the occiput—findings consistent with atlanto-occipital assimilation, a condition reported in 0.08% to 3% of the population. This anomaly results from a failure of segmentation between the fourth occipital sclerotome and the first cervical sclerotome, and is frequently asymptomatic, although in some cases it may lead to neural or vascular compression due to altered biomechanics at the craniovertebral junction. [7]

In our case, the atlanto-occipital assimilation was associated with basilar invagination, an anomaly characterized by upward migration of the odontoid process through the foramen magnum. This condition, congenital or acquired, may remain clinically silent until adulthood, but can also present with variable neurological symptoms including neck pain, limb weakness, paresthesia, and in severe cases, brainstem compression or syringomyelia.[8] The presence of basilar invagination in association with Klippel-Feil and atlanto-occipital assimilation has been described in the literature and likely reflects the spectrum of segmentation defects affecting this region. [5]

Imaging plays a fundamental role in the diagnosis, characterization, and classification of craniovertebral junction anomalies. Computed tomography (CT) is the modality of choice for evaluating bone architecture with high spatial resolution. It allows accurate assessment of vertebral body fusion, arch defects, and craniovertebral relationships.[9] In our patient, cervical CT clearly demonstrated a complete anterior assimilation of C1 with the occiput, posterior arch agenesis, and vertebral body fusion of C2–C3, consistent with Klippel-Feil syndrome type II. Basilar invagination was also identified on midsagittal reformatted images, with superior migration of the

odontoid process above the Chamberlain line, a hallmark of this condition.[4]

When neurological symptoms are present or suspected, magnetic resonance imaging (MRI) becomes indispensable. It offers superior soft tissue contrast and enables direct visualization of the spinal cord, brainstem, and surrounding structures. MRI is particularly useful in detecting spinal cord compression, myelomalacia, or associated anomalies such as Chiari malformation or syringomyelia, which may coexist in up to 30% of cases of basilar invagination. Even in asymptomatic individuals, MRI may be warranted to assess subclinical compression, especially when congenital fusion alters the mechanics and alignment of the cervical spine.[10]

Two main classification systems have been proposed for Klippel-Feil syndrome:
The original classification described by Klippel and Feil, based on the extent and location of vertebral fusion: [11]
Type I: extensive fusion of multiple cervical and upper thoracic vertebrae
Type II: fusion of one or two cervical vertebrae, often with associated anomalies such as hemivertebrae or occipito-atlantal fusion
Type III: cervical fusion associated with lower thoracic or lumbar vertebral fusion

The updated classification by Clarke *et al.,* which incorporates patterns of inheritance, associated anomalies, and the axial level of the most anterior fusion, providing a broader clinical and genetic framework. [12]

Although the findings in our patient were discovered incidentally following trauma, it is important to recognize these anomalies, as they may predispose to instability or neurological compromise, particularly in the setting of high-energy trauma or degenerative changes. In asymptomatic patients, conservative management and clinical monitoring are often sufficient. However, the presence of progressive symptoms or radiological signs of instability may justify surgical intervention. [13]

Conclusion

Klippel-Feil syndrome is a complex congenital condition that may coexist with other craniovertebral anomalies such as atlanto-occipital assimilation and basilar invagination. Although often asymptomatic, these malformations can have significant clinical implications, particularly in trauma settings. Early recognition through imaging is essential for proper diagnosis, risk assessment, and management planning. This case highlights the importance of a systematic evaluation of the cervical spine, even in incidental findings, to avoid overlooking potentially unstable or neurologically significant anomalies.

Conflicts of Interest: The authors declare no conflicts of interest.

Contributions of the Authors: All authors contributed to the conduct of this work. They have read and approved the final version of the manuscript.

References

1. Smoker WRK. Craniovertebral junction: normal anatomy, craniometry, and congenital anomalies. Radiographics. 1994 ;14(2):255–277.
2. Jones KL. Smith's recognizable pattern of human malformation. 5th ed. Philadelphia: WB Saunders Company ;1997.
3. Stephen R Pledger. Cervical Spine Disease. Ortho Secrets, 3rd Ed,2003.
4. Tubbs RS, Wellons JC, Blount JP, Grabb PA, Oakes WJ. Atlantooccipital assimilation with basilar invagination in a child: case report and review of the literature. Childs Nerv Syst. 2004 ;20(8–9):582–586.
5. Menezes AH. Craniovertebral junction anomalies: diagnosis and management. Semin Pediatr Neurol. 1997 ;4(4):209–223.
6. Prabhu VC, Batjer HH. Klippel-Feil syndrome. In : Youmans Neurological Surgery. 6th ed. Elsevier ; 2011.
7. Lins CCSA, Cardoso F, Santos T. Occipitalization of the atlas associated with basilar impression : case report and review. Arq Neuropsiquiatr. 2011 ;69(2B):408–409.
8. Goel A. Basilar invagination : Instability is the cause and stabilization is the treatment. Neurospine. 2020 ;17(4):624–629.
9. Avadhani A, Bommaraju S, Goel A. Imaging of congenital craniovertebral junction anomalies. World Neurosurg. 2018 ;113 :348–359.
10. Karasick D, Schweitzer ME, Vaccaro AR. The traumatized cervical spine in Klippel-Feil syndrome : imaging features. AJR Am J Roentgenol. 1998 ;170 (1): 85-8. doi :10.2214/ajr.170.1.9423605 - Pubmed citation
11. Yuksel M, Karabiber H, Yuksel KZ et-al. Diagnostic importance of 3D CT images in Klippel-Feil Syndrome with multiple skeletal anomalies : a case report. Korean J Radiol. 2006 ;6 (4): 278-81. Free text at pubmed - Pubmed citation
12. Clarke RA, Catalan G, Diwan AD, Kearsley JH. Heterogeneity in Klippel-Feil syndrome : a new classification. (1998) Pediatric radiology. 28 (12): 967-74. doi :10.1007/s002470050511 – Pubmed
13. Goel A. Atlantoaxial instability: redefining craniovertebral junction pathologies. Neurosurgery. 2015 ;76(1):1–6.