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

H. Tahiri[1*], O. Tounsi[1], S. Ouassil[1], C. Ahmanna[1], B. Zouita[1], D. Basraoui[1], H. Jalal[1]

[1]Service de Radiologie, hôpital Mère et Enfant, CHU MOHAMMED VI Marrakech

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***Corresponding author:** H. Tahiri
Service de Radiologie, hôpital Mère et Enfant, CHU MOHAMMED VI Marrakech

Abstract | **Case Report**

Background: Klippel-Feil syndrome (KFS) is a rare congenital anomaly caused by segmentation failure of cervical somites, leading to vertebral fusion. It is often associated with other malformations, the most frequent musculoskeletal anomaly being Sprengel's deformity. ***Case presentation:*** We describe a 6-year-old boy with shoulder asymmetry and restricted cervical mobility. Clinical assessment revealed elevation of the right scapula. CT imaging demonstrated multiple vertebral fusions involving C2–C3, C7–D1, and D4–D5, with fusion of the spinous processes at C2–C3 and D1–D2. The right scapula was elevated and classified as Rigault grade II Sprengel's deformity. Neurological evaluation showed no deficits, and no visceral abnormalities were detected. ***Conclusion:*** This case highlights the uncommon association between KFS and Sprengel's deformity. Cross-sectional imaging, especially CT, is essential for defining skeletal abnormalities and planning multidisciplinary management.

Keywords: Klippel-Feil syndrome, Sprengel's deformity, vertebral fusion, congenital anomaly, CT imaging.

INTRODUCTION

Klippel–Feil syndrome (KFS), first described by Klippel and Feil in 1912 [1], is a rare congenital disorder resulting from abnormal segmentation of cervical somites between the 3rd and 8th weeks of embryonic development [2]. Its incidence is estimated at approximately 1 in 40,000–42,000 live births [3]. The classical clinical triad of short neck, low posterior hairline, and restricted cervical mobility is present in fewer than 50% of patients [2].

KFS is frequently associated with additional anomalies, including scoliosis, hearing impairment, renal malformations, and congenital heart disease [2,5]. Among musculoskeletal anomalies, Sprengel's deformity is the most common, reported in 7–42% of cases [6,7,9,10]. This anomaly corresponds to congenital elevation of the scapula due to incomplete caudal migration during the 9th–12th weeks of development [6]. In 25–50% of cases, Sprengel's deformity is associated with an omovertebral bone, a fibro-osseous connection between the scapula and cervical spine [9,10].

CASE PRESENTATION

A 6-year-old boy was referred to our department for evaluation of a cervicothoracic deformity and right shoulder asymmetry. Pregnancy and perinatal history were unremarkable, and psychomotor development was age-appropriate. There was no family history of congenital or genetic disorders.

On physical examination, the patient presented with restricted cervical mobility, particularly in rotation, and an elevated right scapula with its medial border reaching the C7 level, producing marked shoulder asymmetry with cosmetic concern. Neurological examination was normal.

A thin-slice cervical and upper thoracic CT scan (bone window) demonstrated a straightened cervical spine and normal spinal canal caliber. Multiple vertebral fusions were observed, including partial C2–C3 fusion (Figures 1 and 2), complete C7–T1 fusion (Figure 3), and partial T4–T5 fusion. Fusion of the spinous processes was also identified at C2–C3 and T1–T2. Importantly, the scan confirmed Sprengel's deformity of the right scapula, Rigault grade II (Figure 4). No omovertebral bone was present.

Additional investigations were performed to exclude systemic associations. Both renal ultrasound and echocardiography were normal.

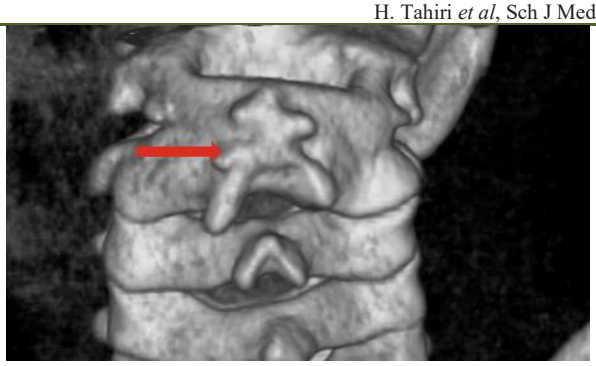

Figure 1: 3D reconstructed CT scan demonstrating partial fusion at C2–C3

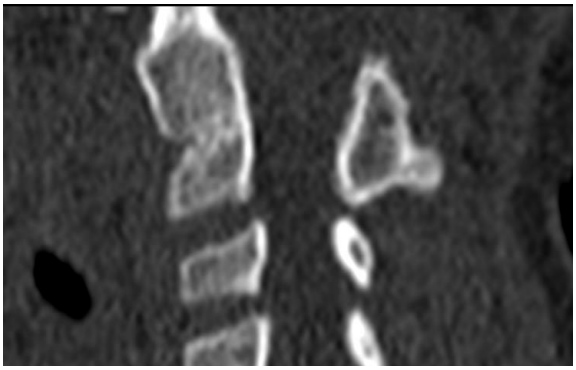

Figure 2: sagittal CT scan with bone window shows a partial fusion at C2-C3

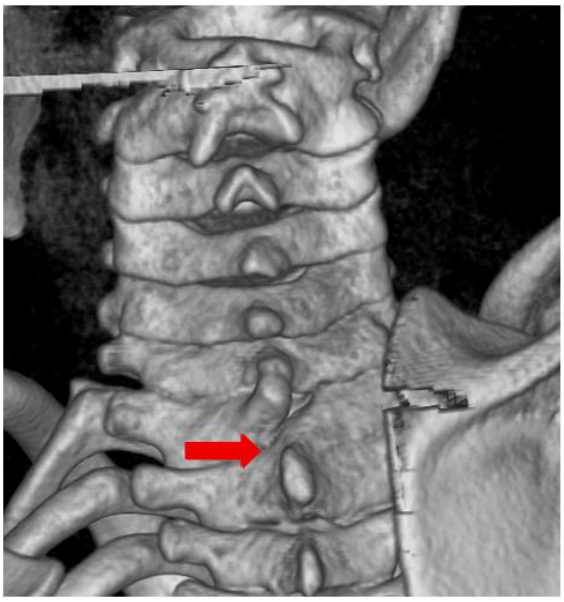

Figure 3: 3D reconstructed CT scan showing complete vertebral fusion at C7–T1

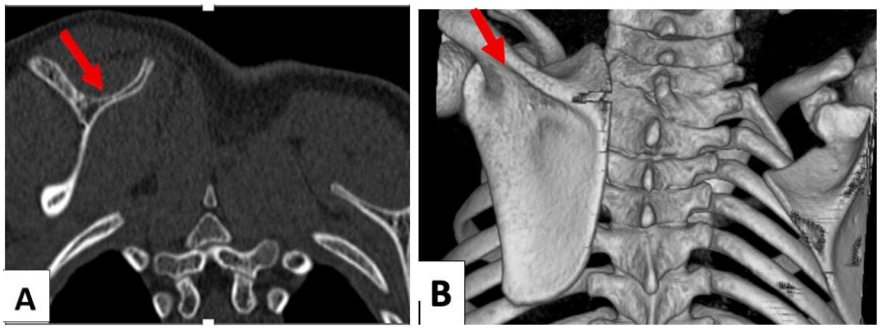

Figure 4: coronal CT scan and 3D reconstructed CT demonstrating right Sprengel's deformity, classified as Rigault grade II

DISCUSSION

Klippel–Feil syndrome (KFS) arises from a failure of cervical somite segmentation during embryonic development, typically between the third and eighth weeks of gestation [1]. The clinical spectrum is highly heterogeneous, ranging from mild cervical stiffness to severe deformity with neurological compromise [11]. The classical triad—short neck, low posterior hairline, and limited cervical motion—is present in fewer than half of patients [2]. Classification systems, such as the one proposed by Samartzis et al., stratify patients into type I (single fused segment), type II (multiple noncontiguous fusions), and type III (multiple contiguous fusions) [1]. Our patient, with noncontiguous fusions at C2–C3 and C7–T1, corresponds to type II.

Sprengel's deformity, first described by Eulenberg in the 19th century, represents the most frequent skeletal anomaly associated with KFS. It results from incomplete caudal migration of the scapula during fetal development, leading to congenital elevation of the scapula. Its prevalence among KFS patients varies significantly: 6.36% in large pediatric cohorts [9] but as high as 42% in smaller series [7]. The deformity can cause both cosmetic and functional impairment, the latter due to restriction of shoulder abduction. The severity is commonly assessed clinically with the Cavendish classification [4] and radiologically with the Rigault classification [5]. In our case, the deformity corresponded to Rigault grade II, with moderate elevation of the scapula but preserved shoulder mobility.

Imaging. Cross-sectional imaging plays a crucial role in the evaluation of KFS and associated anomalies. CT remains the gold standard for assessing the extent of osseous fusion and the degree of scapular elevation [6,7]. Three-dimensional CT reconstructions offer superior visualization of complex skeletal malformations, allowing for accurate detection of anomalies such as the omovertebral bone, a fibro-osseous bridge between the scapula and the cervical spine, which is present in 25–50% of Sprengel cases [7]. MRI complements CT by evaluating neural structures and identifying associated intraspinal anomalies, such as Chiari malformation, syringomyelia, and tethered cord, reported in up to 30% of patients [3]. In our patient, CT was sufficient given the absence of neurological symptoms, but MRI remains essential in cases with clinical suspicion of cord involvement.

Associated anomalies. KFS is often part of a broader syndrome involving multiple systems. Reported associations include scoliosis, present in up to 97% of cases [9], renal anomalies (e.g., renal agenesis, ectopia, or horseshoe kidney), congenital heart disease, and limb malformations such as radioulnar synostosis [6,10]. Because of this broad spectrum, systematic screening with renal ultrasound, echocardiography, and audiological evaluation is recommended. In our case, no visceral malformations were detected, highlighting the heterogeneity of phenotypic expression in KFS.

Management. The therapeutic approach depends on symptomatology, severity of deformity, and presence of complications. Most patients without neurological deficits are managed conservatively, with physiotherapy and regular clinical monitoring. Surgical treatment is generally reserved for Sprengel's deformity in cases of significant cosmetic concern or functional limitation. Procedures such as those described by Woodward and Green aim to reposition the scapula caudally and release restrictive attachments, improving both cosmesis and range of motion [2]. The best outcomes are achieved when surgery is performed before the age of 8, due to greater tissue plasticity and potential for remodeling. In our patient, conservative management was chosen because of the absence of neurological deficits and relatively preserved function.

Comparison with the literature. Our case aligns with previously published reports emphasizing the frequent association between KFS and Sprengel's deformity [1,2,7,9]. However, the absence of neurological manifestations distinguishes our case from series such as that of Moses et al., who documented a higher incidence of myelopathy in patients with multilevel fusions [11]. This underlines the importance of long-term follow-up, as neurological complications may arise later due to abnormal spinal biomechanics and adjacent segment hypermobility.

CONCLUSION

We present a 6-year-old boy with the rare association of Klippel–Feil syndrome and Sprengel's deformity. CT scan allowed precise delineation of vertebral fusions and scapular elevation. Although asymptomatic neurologically, the patient requires close follow-up to monitor for potential complications. This case reinforces the importance of early recognition, systematic screening for associated anomalies, and coordinated multidisciplinary care.

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