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Imaging findings in Klippel-Feil Syndrome with Unilateral Renal Agenesis and Ectopic Pelvic Kidney

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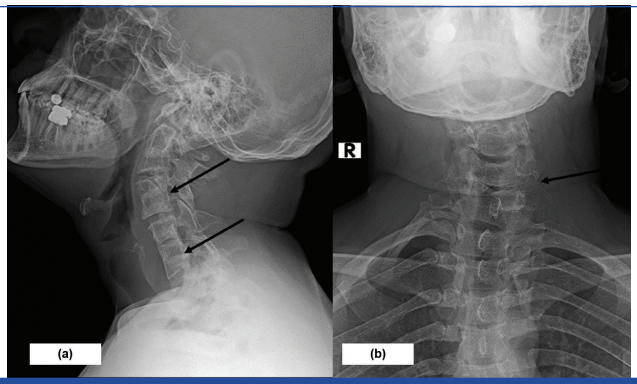
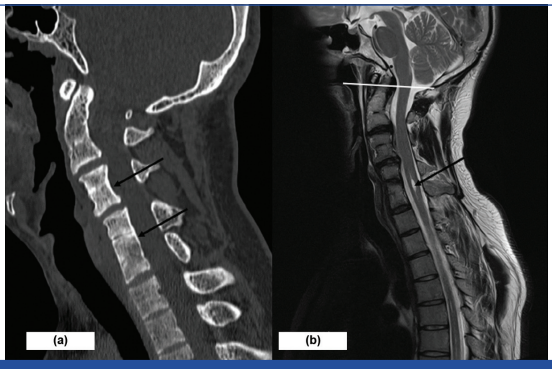
A 22-year-old male presented with a five-day history of insidious-onset neck pain, rated 5/10 on the numeric rating scale, aggravated by neck turning and relieved by rest. The pain was localised to the neck, non radiating, with no history of trauma, bowel or bladder incontinence, weakness, numbness, neurogenic claudication, or morning stiffness. The patient's past medical history was unremarkable, with no known co-morbidities. On examination, there was no gross deformity, scar, or sinus over the cervical spine. Palpation revealed no warmth, no local rise in temperature, or tenderness. Range of motion of the neck in side-to-side movement was painful. Neurological examination showed full power (5/5) in all cervical myotomes (C2-C7) and normal sensation. Provisional diagnosis was neck muscle spasm. Differential diagnoses included cervical spondylosis, congenital vertebral anomaly, inflammatory and neoplastic aetiology.

Plain radiographs of the cervical spine showed mild scoliosis with convexity to the left and fusion of the C3-C4 and C6-C7 vertebral bodies, suggestive of block vertebra [Table/Fig-1a,b]. Computed Tomography (CT) of the cervical spine confirmed the radiographic findings [Table/Fig-2a]. Cervical spine MRI revealed mild basilar invagination, with the tip of the odontoid process approximately 7 mm above Chamberlain's line [Table/Fig-2b]. A well-defined linear T2 hyperintensity was seen within the centre of the spinal cord at the C6-C7 vertebral level, suggestive of syrinx [Table/Fig-2b]. Dorsal-spine screening also showed a well-defined linear T2 hyperintensity within the centre of the spinal cord at D5-D10 vertebral levels, suggestive of syrinx [Table/Fig-3a]. Lumbar spine screening showed mild disc desiccation at L5-S1 and suggestion of pelvic kidney [Table/Fig-3b]. Further investigation with CT abdomen revealed that bilateral kidneys were not visualised in the renal fossae [Table/Fig-4a]. A single kidney, measuring approximately 9.7×5 cm, was noted in the pelvis, above the urinary bladder, with its convexity towards the right side [Table/Fig-4b]. This finding confirmed unilateral renal agenesis with an ectopic pelvic kidney. Based on the constellation of imaging findings, including multiple cervical vertebral fusions (C3-C4, C6-C7), cervical scoliosis, basilar invagination, along with the presence of syrinx, the possibility of Klippel-Feil Syndrome (KFS) was considered. The co-occurrence of unilateral renal agenesis with an ectopic pelvic kidney further supported this diagnosis, given the known association of KFS with genitourinary anomalies.

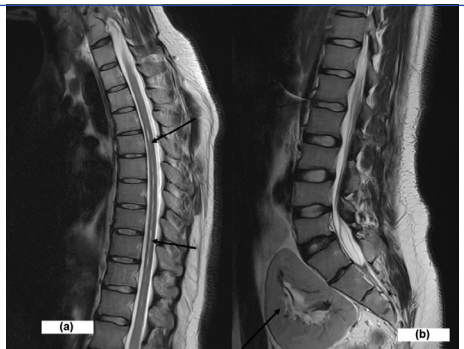
The KFS, first described in 1912 by Klippel and Feil, is a developmental anomaly resulting from failure of segmentation of the cervical somites during weeks 3-8 of gestation, leading to congenital fusion of the cervical vertebrae. The classic triad of a short neck, a low posterior hairline, and restricted neck movement is present in less than 50% of cases [1]. In the present case, the triad was not evident. Various classification systems have been proposed to categorise the diverse presentations of KFS, aiding in understanding its clinical significance. The condition is often associated with a wide spectrum of extra-skeletal abnormalities, with renal anomalies being among the most common [2,3]. These can range from agenesis, ectopia, and malrotation to duplication and hydronephrosis. Early



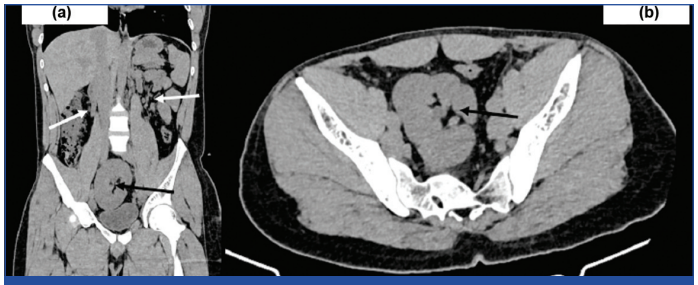
[Table/Fig-1a,b]: Plain radiograph of cervical spine lateral and anteroposterior view showing fusion of C3-C4 and C6-C7 vertebral bodies suggestive of block vertebra (black arrows) and scoliosis of cervical spine with convexity towards left.



[Table/Fig-2]: a) CT cervical spine sagittal sections shows fusion of C3-C4 and C6-C7 vertebral bodies suggestive of block vertebra; b) T2 weighted MRI images sagittal sections of cervical spine showing basilar invagination with tip of odontoid process 7 mm above chamberlain's line which connects posterior margin of hard palate with opisthion (white line) and also syrinx at C6-C7 level (black arrow).



[Table/Fig-3]: a) T2 weighted MRI images sagittal sections of dorsal spine shows evidence of syrinx from D5 to D10 levels (black arrow); b) T2 weighted MRI images sagittal sections of lumbar spine shows evidence of ectopic pelvic kidney in presacral region (black arrow).



[Table/Fig-4]: a) Coronal; and b) Axial plain CT abdomen section show ectopic pelvic kidney in presacral region (black arrow) with absent kidneys in bilateral renal fossa (white arrow).

detection of these associated anomalies is crucial for appropriate management and prevention of complications [3].

KFS is a complex disorder with varied clinical presentations, often due to associated systemic anomalies. The presence of cervical spine fusions, while the hallmark, necessitates thorough evaluation for other malformations. Renal anomalies are among the most frequent associated conditions, with agenesis representing a severe manifestation. The embryological basis for this association lies in the fact that vertebral column and renal development occur during similar periods of gestation and are influenced by common genetic pathways [2-4]. Unilateral renal agenesis, as seen in this patient, can often be asymptomatic if the contralateral kidney is healthy and functions adequately; however, it increases the risk of complications such as hypertension, proteinuria, and renal insufficiency later in life, especially if the solitary kidney is subjected to increased physiological demands or develops abnormalities [2]. The ectopic pelvic kidney in this case further emphasises the broad spectrum of renal malformations that can occur. The syrinx noted at multiple levels in the spinal cord is another significant neurological complication associated with KFS, often resulting from altered CSF dynamics due to underlying bony abnormalities such as basilar invagination and cervical fusions. This can lead to progressive neurological deficits if left unmanaged.

Similar to the present case, a report by Kelekçi S et al., described a 10-year-old girl with KFS and unilateral renal agenesis, emphasising the strong association between these developmental anomalies [4]. Another notable case by Eroz R et al., described a seven-year-old girl with KFS, bilateral Sprengel deformity, and congenital unilateral renal agenesis, with a unique association to an MEFV gene mutation, suggesting broader genetic and systemic implications that can accompany this syndrome [5]. This case underscores the critical need for a comprehensive diagnostic approach in patients suspected of having KFS. Imaging of the entire spine is essential to identify all vertebral anomalies and associated neurological complications such as syrinx [2]. Furthermore, routine renal ultrasonography or CT abdomen is highly recommended to detect any associated genitourinary malformations, even in asymptomatic patients, to facilitate early intervention and long-term follow-up [2]. Multidisciplinary management involving orthopaedic surgeons, neurologists, nephrologists, and geneticists is often required for optimal patient care [3-5].

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