KLIPPEL-FEIL SYNDROME PRESENTING AS RECURRENT ABDOMINAL PAIN IN A TEENAGER: IMPORTANCE OF COMPUTED TOMOGRAPHY SCAN IN DIAGNOSTIC WORKUP

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Abstract

Klippel-Feil syndrome (KFS) is a segmentation and cleavage malformation of the cervical spine in the early weeks of foetal development. This is considered as a sporadic genetic abnormality, and is accompanied by multisystem disorders such as a short neck, cardiac disease, renal ectopia and other associated genitourinary syndromes. In this case report, we present the clinical and radiological findings of a 14-year old school boy who was referred to us from a private hospital for abdominopelvic ultrasound to ascertain his cause of recurrent abdominal pains since childhood. We also present the role of computed tomography in detecting multisystem abnormalities during a single hospital visit.

Keywords: Recurrent abdominal pains, renal ectopia, multiple cervicothoracic vertebral fusions, Sprengel's scapulae.

Introduction

In 1912, Maurice Klipplel and Andre Feil independently described a syndrome with the triad of short webbed-neck, limitation of neck motion and low posterior hairline.[1] It is described as a failure of separation of two or more segments of the cervical spinal vertebrae with or without similar fusions involving the dorsal or lumbar components.[1-4] It is a genetically determined disorder of segmentation that occurs between the 3rd and 8th weeks of foetal development,[3] with a frequency of 1 in 42,000 live births.[4] It is also established that females seem to be affected slightly more often than males.[5-7]

Klippel-Feil Syndrome is also known by other synonyms such as Cervical Fusion Syndrome or Congenital Brevicollis.[8] This syndrome was initially believed to be mostly sporadic,[9] but both the autosomal dominant and recessive forms have recently been reported.[10] This syndrome can present with a variety of other clinical syndromes, including renal ectopia and/or agenesis, Foetal Alcohol Syndrome, Goldenhar Syndrome and many other anomalies of the extremeties.[10-11] It is however known that these other abnormalities can occur independently.[12] We present the case of a Klippel-Feil syndrome presenting with recurrent abdominal pain from childhood who turned out to have bilateral suprapubic kidneys with other multiple organ abnormalities.

Case Report

AA is a 14-year old secondary school male student who presented to a diagnostic centre for abdominal ultrasound following a history of recurrent abdominal pain since childhood. He has also had repeated episodes of respiratory tract infections which were usually aggravated by exposure to cold environments. He is the fifth child of his parents. At the age of three, his parents noticed that he had an 'abnormally short neck', with restriction of movements including elevation of the shoulder when he wants to raise his hand up while playing with his siblings and friends. Apart from these concerns by the parents, he was said to be developing normally, with no mental, neural or otologic deficits noted.

Physical examination revealed a webbed-neck and a low hairline but no facial asymmetry or torticollis was observed. The palate was high-arched but with no cleft lip or palate defect seen. Abnormal disposition of the scapular was noted bilaterally. Bilateral thenar muscle wasting was noted (right > left), as well as a short and deformed thumb on the right hand. There were no abnormalities noted in the cardiovascular, respiratory,

central nervous systems, abdominal and genito-urinary systems on examination.

A chest radiograph confirmed the bilateral winged scapulae, with no thoracolumbar scoliosis seen. Abdominopelvic ultrasound showed a centrally located relatively big suprapubic mass that showed central echogenicity similar to those of renal sinuses. The usual renal angles were bilaterally empty. Computerised intravenous urography (CT IVU) showed bilateral centrally located suprapubic kidneys that were joined superiorly (horse shoe-shaped kidneys) with satisfactory contrast excretion (figs. 1a & 1b). No calculi or hydronephroses were seen. However, their bipolar lengths were comparatively shorter than those seen in normal individuals.

The cranio-cervical computed tomography (CT) showed reversal of the cervical curvature and scoliosis with concavity to the left (figs. 2a & 2b). The vertebrae were generally reduced in heights, with fusion of C1/2 and C7/T1/T2 bodies (fig.2b). There was relative stenosis of the antero-posterior diameter of the spinal canals at levels 2-6 vertebral bodies, measuring about 10.9mm, but the transverse diameters were normal. Serial 3 D volumetric images of the neck and thoracic inlet showed high shoulder blades (the typical winged-scapulae) as shown in figures 3a & 3b. A cervical rib was demonstrated on the left side of the seventh vertebral body, while there was fusion of the 1st and 2nd ribs posteriorly on the contralateral side (figs. 3b & 4). Sagittal views also confirmed the high palatine arch which was observed during the physical examination. Routine haematological examinations were normal.

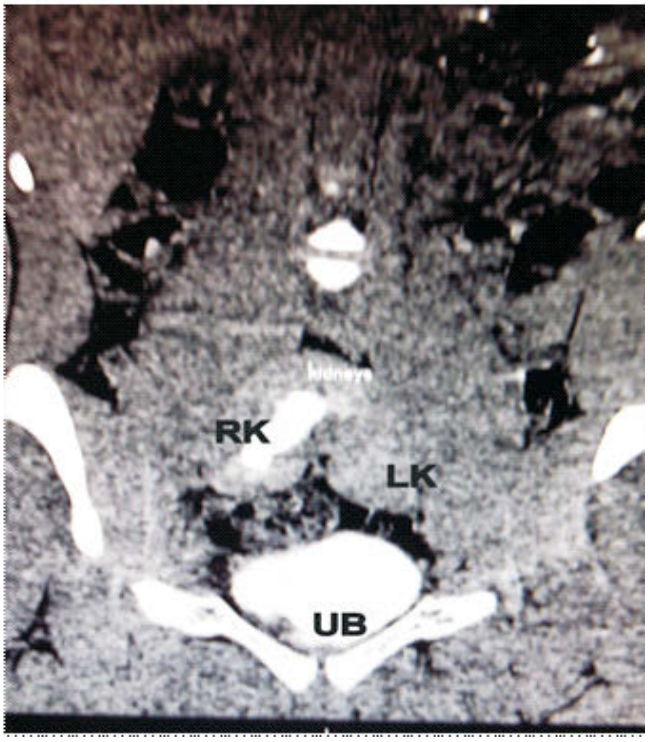

Fig. 1a shows bilateral suprapubic kidneys with fusion of the superior poles.
RK=Right kidney. LK=left kidney.
UB=Urinary bladder.

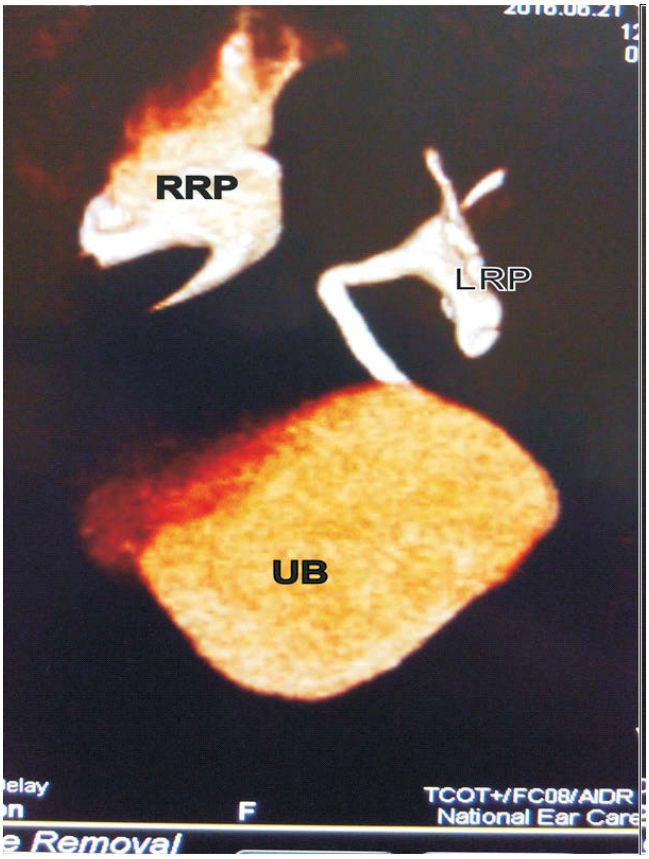

Fig. 1b shows suprapubic renal pelvis and ureters.
RRP=right renal pelvis. LRP=left renal pelvis.
UB=urinary bladder.

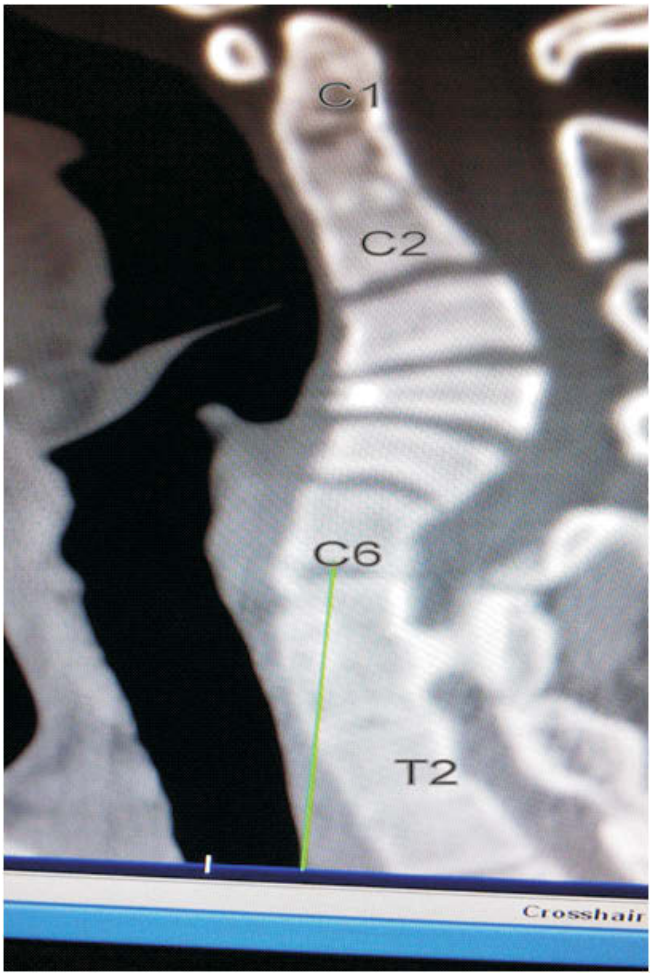

Fig. 2a shows the reversal of cervical curvature and the fused C1/2 and C7/T1/T2 vertebral bodies.

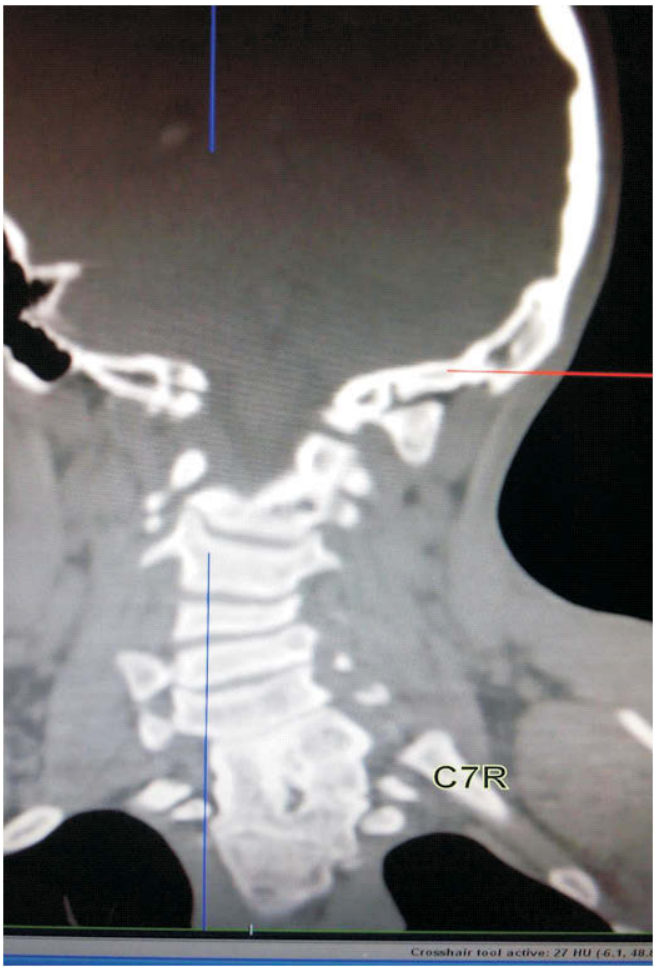

Fig. 2b shows scoliosis of the cervical vertebrae, with concavity to the left. There is also generalised reduction of vertebral heights. A left cervical rib is also seen (C7R)

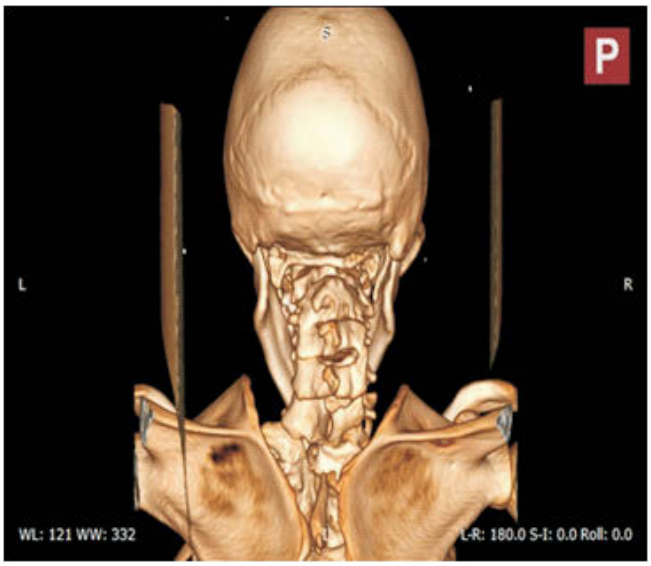

Fig. 3a shows elevated scapulae as seen from the back in a 3D image.

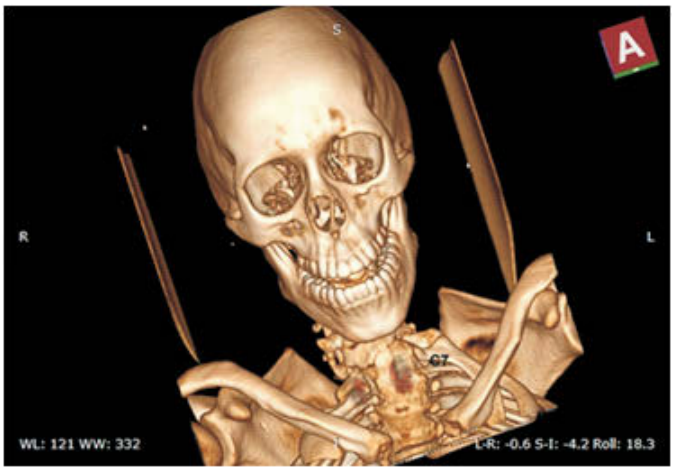

Fig. 3b shows high scapulae as seen in a 3D image anteriorly. There is also a left sided cervical rib demonstrated (C7).

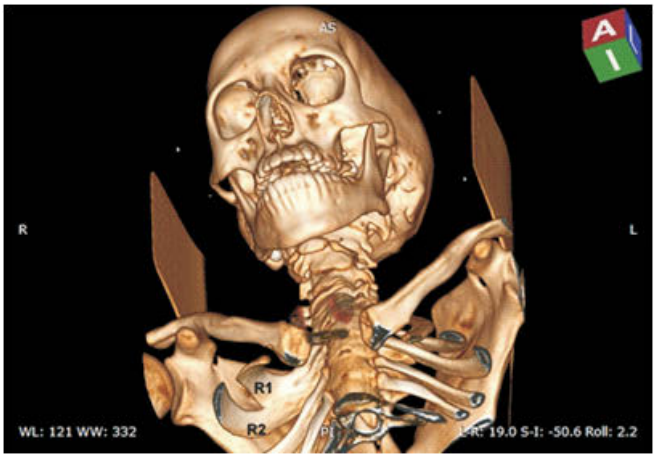

Fig.4 shows posteriorly fused 1st and 2nd right ribs in a 3D image

Discussion

The classical triad of short webbed-neck, restricted movement and low hairline are the clinical diagnostic features of a typical case of Klippel-Feil Syndrome.[3] Helsinger et al referred to this syndrome as a constellation of congenital anomalies.[13] The syndrome occurs in a heterogeneous group of patients, unified only by the presence of a congenital defect in the formation or segmentation of the cervical spine,[7] and is believed to result from faulty segmentations along the embryo's developing axis during the 3rd-8th week of gestation.[2-3] It is considered either as a sporadic disease[3, 14] or an inherited disease or both.[3] The syndrome was initially classified into three types by Clarke et al,[3] but the present classification has given us four types.[7] The radiological classification is called types I-III,[6] based on both clinical and radiological findings.[1,3,15] Recently, genetic mutations have been implicated.[16-20] Mutations in the GDF6, GDF3, MEOX1 or MYO18B genes can cause Klippel-Feil syndrome.[17, 19] This inheritance is either autosomal dominant or recessive.[18] Mutations in the GDF6 and GDF3 genes are autosomal dominant, and are classified as KFS types 1 and 3, while mutations in MEOX1 and MYO18B genes are autosomal recessive, and are called KFS types 2 and 4 respectively.[16, 19]

Even though the true aetiology of KFS is speculative and its phenotypic expression varies between individuals,[14] majority of young individuals with KFS are asymptomatic, with many presenting in childhood but a sizeable number of them are also identified in adulthood.[8-10, 13] The case of a 70-year old man has been reported in Nigeria.[20] The actual prevalence of Klippel-Feil syndrome is unknown due to the fact that many patients with the syndrome pass through life unnoticed, that is, they are symptoms free,[19-21] hence the prevalence of KFS may in fact be higher than has previously been reported.[21] Whatever the true prevalence is, females are affected slightly more often than males.[9]

The presentation of KFS is varied because of the clinical differences associated with the syndrome and the wide range of anomalies that can occur in patients with this syndrome.[21] Many associated congenital anomalies to KFS were described with little mention of the genitourinary system until Moore et al intensified the association of many renal diseases to this syndrome.[6] Their findings showed that 64% of KFS patients had significant genitourinary system anomalies demonstrated by physical examination and intravenous urography, with unilateral renal agenesis being the most common congenital defect.[6] These anomalies which were found by other authors include renal agenesis, malrotation, ureteral and pelvic duplication, renal ectopia, dysgenesis, hypospadias, cryptorchidism, vaginal atresia and bicornuate uterus.[6-9, 13] These findings are presently easily diagnosed by CT as a diagnostic tool.[8] Our patient had bilateral renal ectopia, which was the reason for the recurrent abdominal pain. He also had bilateral winged scapulae, multiple fused cervicothoracic vertebrae, cervical ribs and fused ribs. These features would probably have been missed without a modern modality of imaging like CT.

Conclusion

While KFS is a genetic and rare congenital condition with wide and varied symptoms associated with it, its prognosis is not grave if it is diagnosed early. Some of these symptoms can be cured or medically managed with or without surgical intervention. Though this condition is rare, a high index of suspicion is necessary in the event of the presence of abnormalities of the spine associated with other systems. Our case report underscores the importance of CT imaging in the diagnosis of KFS and should be used in suspected cases of the syndrome.

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