

Klippel-Feil Syndrome and Crossed Renal Ectopia: Case Report

Klippel-Feil Sendromu ve Çapraz Renal Ektopi

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ABSTRACT Klippel-Feil syndrome (KFS) is associated with various congenital systemic anomalies in musculoskeletal, cardiovascular and genitourinary systems. We report a case with KFS associated with crossed renal ectopia. The typical short neck, low hair-line and restricted neck motion of the patient reported, was unrecognized until adulthood. He was diagnosed as KFS incidentally after referral to the urology clinics with flank pain. On cervical direct X-ray examination, congenital fusion of the cervical vertebrae was seen and urological imaging techniques demonstrated crossed renal ectopia. Congenital genitourinary anomalies including crossed renal ectopia should be kept in mind and assessed thoroughly in patients with KFS.

Key Words: Klippel-Feil syndrome; urogenital abnormalities

ÖZET Klippel-Feil sendromu (KFS) kas-iskelet, kardiyovasküler ve genitouriner sistem gibi birçok sistemi ilgilendiren konjenital anomalilerle birliktelik gösterir. Burada çapraz renal ektopi ile birlikte görülen KFS olgusu sunulmuştur. Olgumuz, erişkin çağa kadar farkedilmemiş olup, bu hastalar için tipik olan kısa boyun, düşük saç çizgisi ve boyun hareketlerindeki kısıtlılık özelliklerine sahiptir. Hasta, üroloji polikliniğine yan ağrısı yakınması ile başvurmuş ve rastlantısal olarak KFS tanısı almıştır. Servikal vertebraların direkt radyografik incelemesinde konjenital füzyon ve ürolojik görüntüleme tekniklerinde de çapraz renal ektopi saptanmıştır. KFS ile birlikte görülebilen, çapraz renal ektopi ve diğer konjenital urogenital anomaliler bu hastalarda akılda tutularak dikkatle incelenmelidir.

Anahtar Kelimeler: Klippel-Feil sendromu; ürogenital anomaliler

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Klippel-Feil Syndrome (KFS) is defined as congenital fusion of two or more cervical vertebrae as a result of failure in normal segmentation of cervical mesodermal somites during embryonic development. In 1912 Maurice Klippel and Andre Feil independently described this syndrome in patients with short and webbed neck, a low hair line and restricted neck motion.¹ First classification of this syndrome by Feil was based on the extent of fusion. Later, Clarke et al suggested another classification depending on the inheritance patterns.²

Many other associated abnormalities have been described such as scoliosis, kyphosis, Sprengel's deformity, hemivertebra, craniocervical junction abnormalities, hearing impairment, heart malformations, synkinesis and

kidney abnormalities.³ The most common renal anomalies reported in KFS are unilateral renal agenesis, crossed renal ectopia and duplication of the renal collecting system.⁴ In medical literature, the prevalence of genitourinary defects in patients with KFS ranges between 2% and 64%. Here, a patient with KFS associated with crossed renal ectopia is presented.

CASE REPORT

A 58-year-old male patient was referred to urology outpatient clinics with the complaint of right flank pain for a couple of days. Past medical history revealed that he had been experiencing neck pain and limitation of neck motion for many years but no diagnosis was established up to date. On physical examination, he had torticollis, a low hair line and facial asymmetry (Figure 1, 2). The range of motion of neck was limited. Abdominal examination did not reveal any palpable mass. There was a mild tenderness of lower costovertebral junction on the right side. Direct X-ray radiography of the cervical region revealed block vertebra involving all vertebra from C2 to C7 (Figure 3). Based on the radiologic findings of the cervical vertebra, diagnosis of KFS was made. Abdominopelvic ultrasonography revealed a normal right kidney but no renal tissue was present on the left renal fossa. Intravenous urography showed a crossed ectopic left kidney (Figure 4). A renal scan with technetium-99m dieti-



FIGURE 2: Low hair line of the patient.



FIGURE 3: Direct X-ray radiography of the cervical region revealed block vertebra.



FIGURE 1: Patient had torticollis and facial asymmetry.

lentrilamine-penta-acetic acid (DTPA) confirmed absence of left renal tissue and crossed ectopic left kidney. Besides renal abnormality of the patient,



FIGURE 4: Intravenous urography showing crossed ectopic left kidney.

other commonly associated visceral abnormalities were also searched. Echocardiographic investigation for an associated congenital heart malformation and audiometric tests for hearing impairment were both negative. Written informed consent was obtained from our patient for publication of this case report and any accompanying images.

DISCUSSION

The prevalence of KFS is unknown as majority of the patients are asymptomatic. In a study, all cervical spine X-rays from a hospital in Copenhagen were reviewed and an incidence was found to be 0.2 cases per 1000 patients.⁵ Most of the KFS cases are sporadic, however, different fusion patterns, associated skeletal and visceral anomalies with different modes of inheritance were described in family members with KFS.²

Patients with KFS present at different ages with varying clinical presentations. This syndrome may also be detected as an incidental finding. In symptomatic patients, most common symptoms are axial, including neck pain and restriction of head and neck movements. The classical triad of short neck with a low hair line and restriction in neck movements has been reported in approximately half of the patients.⁶ Other musculoskeletal abnor-

malities associated with KFS include torticollis, congenital scoliosis, Sprengel's deformity, spina bifida and hypoplastic rib.⁷⁻⁹

In association with the spinal anomalies, malformations of other viscera are the greatest threat to these patients. Congenital anomalies of the cardiovascular system mainly atrial and ventricular septal defects, dextrocardia, aortic stenosis and subclavian artery anomalies have been reported.^{10,11} Auscultation and echocardiographic examination should be done for patients with fused cervical spine, before cardiac complications occur. Hearing loss has been reported in approximately 36% of these patients but ear deformities with or without hearing impairment can also be seen.¹²

In the case presented here, the patient underwent echo-cardiographic and audiometric examination in order to exclude an associated cardiac and hearing anomaly respectively. For detecting presence of a genitourinary malformation, abdominopelvic ultrasonography and intravenous pyelography were performed which revealed crossed renal ectopia. In medical literature, the prevalence of genitourinary defects in patients with KFS ranges between 2% and 64%. In the study by Moore et al, it has been reported that, out of thirty-nine patients with KFS, twenty-five (64%) had significant genitourinary tract anomalies including unilateral renal agenesis, crossed renal ectopia, renal pelvic and ureteral duplication, renal dysgenesis, hypospadias, cryptorchidism and vaginal agenesis.⁴ Unilateral renal agenesis is the most commonly associated renal anomaly followed by crossed renal ectopia with or without fusion.¹³

Crossed renal ectopia is a congenital malformation in which both kidneys lie on the same side of the spine. The ectopic kidney is generally fused to the lower pole of the normal kidney,¹⁴ however, there may or may not be a fusion between the two renal units. It is a rare anomaly of urinary tract, occurring only in one out of 7.000 cases autopsied.¹⁵ Most often the anomaly is right sided. The ectopic kidney is smaller than normal and is malrotated.¹⁶

During fourth-fifth weeks of fetal life, each mesodermal somites form a pair of segmental sclerotome-

me which eventually develop into the vertebrae. The cervical spine develops approximately between the seventh and fourteenth somites. The pronephros, which eventually differentiates to adult kidney, lies between the seventh and eleventh somites. This spatial relationship explains the high incidence of associated congenital anomalies of genitourinary tract in KFS. Any insult causing faulty development of cervical spine may lead to some derangement in the genitourinary tract.¹⁷ Although

most of the patients with crossed fused renal ectopia are asymptomatic, they present with increased susceptibility to develop complications such as urinary infections, urolithiasis and abdominal mass.¹⁸

The high incidence of renal anomalies in KFS suggests that the assessment of genitourinary system by renal ultrasound should be considered in every patient. An intravenous pyelogram should be done only if abnormalities are found on the ultrasonographic examination.

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