

Case report

Klippel-Feil syndrome – the risk of cervical spinal cord injury: A case report

Subramanian Vaidyanathan*¹, Peter L Hughes², Bakul M Soni¹,
Gurpreet Singh¹ and Pradipkumar Sett¹

Address: ¹Regional Spinal Injuries Centre, District General Hospital, Southport, PR8 6PN, UK and ²Department of Radiology, District General Hospital, Southport, PR8 6PN, UK

E-mail: Subramanian Vaidyanathan* - vaidyanathansiu@hotmail.com; Peter L Hughes - Peter.Hughes@mail.soh-tr.nwest.nhs.uk;
Bakul M Soni - Bakul.Soni@mal.soh-tr.nwest.nhs.uk; Gurpreet Singh - gurpreet.singh@mail.soh-tr.nwest.nhs.uk;
Pradipkumar Sett - Pradip.Sett@mail.soh-tr.nwest.nhs.uk

*Corresponding author

Published: 11 April 2002

Received: 15 February 2002

BMC Family Practice 2002, **3**:6

Accepted: 11 April 2002

This article is available from: <http://www.biomedcentral.com/1471-2296/3/6>

© 2002 Vaidyanathan et al; licensee BioMed Central Ltd. Verbatim copying and redistribution of this article are permitted in any medium for any purpose, provided this notice is preserved along with the article's original URL.

Abstract

Background: Klippel-Feil syndrome is defined as congenital fusion of two or more cervical vertebrae and is believed to result from faulty segmentation along the embryo's developing axis during weeks 3–8 of gestation. Persons with Klippel-Feil syndrome and cervical stenosis may be at increased risk for spinal cord injury after minor trauma as a result of hypermobility of the various cervical segments. Persons with Klippel-Feil Syndrome often have congenital anomalies of the urinary tract as well.

Case presentation: A 51-year male developed incomplete tetraplegia in 1997 when he slipped and fell backwards hitting his head on the floor. X-rays of cervical spine showed fusion at two levels: C2 and C3 vertebrae, and C4 and C5 vertebrae. Intravenous urography (IVU) revealed no kidneys in the renal fossa on both sides, but the presence of crossed, fused renal ectopia in the left ilio-lumbar region. This patient had a similar cervical spinal cord injury about 15 years ago, when he developed transient numbness and paresis of the lower limbs following a fall.

Discussion and Conclusion: 1) Persons with Klippel-Feil syndrome should be made aware of the increased risk of sustaining transient neurologic deterioration after minor trauma if there is associated radiographic evidence of spinal stenosis.

2) Patients with Klippel-Feil syndrome often have congenital anomalies of the urinary tract. Our patient had crossed, fused, ectopia of kidney.

3) When patients with Klippel-Feil syndrome sustain tetraplegia they have increased chances of developing urinary tract calculi. Treatment of kidney stones may pose a challenge because of associated renal anomalies.

4) Health professionals caring for cervical spinal cord injury patients with Klippel-Feil syndrome and renal anomalies should place emphasis on prevention of kidney stones. A large fluid intake is recommended for these patients, as a high intake of fluids is still the most powerful and certainly the most economical means of prevention of nephrolithiasis.

Background

Klippel-Feil syndrome is defined as congenital fusion of two or more cervical vertebrae and is believed to result from faulty segmentation along the embryo's developing axis during weeks 3–8 of gestation. The classic clinical triad of short neck, low hair line, and restricted neck motion is present in less than 50% of patients with this syndrome. Three types of Klippel-Feil syndrome have been described.

Type 1: Cervical spine fusion in which elements of many vertebrae are incorporated into a single block.

Type 2: Cervical spine fusion in which there is failure of complete segmentation at only one or two cervical levels and may include an occipito-atlantal fusion.

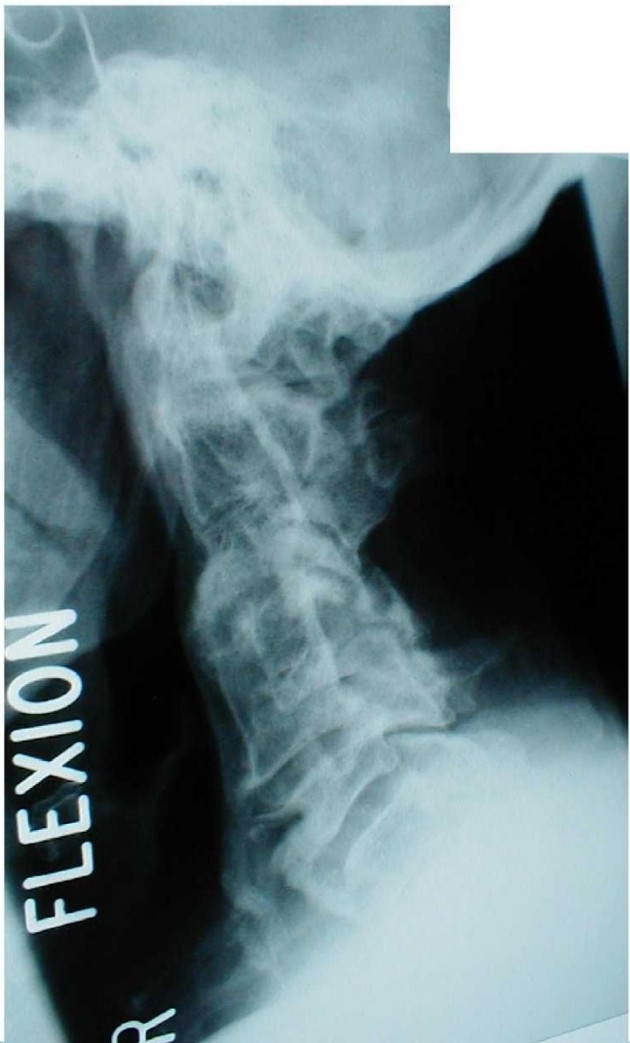


Figure 1
Lateral view of cervical spine shows congenital fusion of bodies of C2 with C3, and vertebral bodies of C4 with C5.

Type 3: Type 1 or type 2 fusion with co-existing segmentation errors in the lower dorsal or lumbar spine. [1]

Case presentation

A 51-year male developed an incomplete tetraplegia (ASIA scale D) in 1997 when he slipped and fell backwards on his head. X-rays of the cervical spine showed fusion at two levels: the C2 and C3 vertebrae, and C4 and C5 vertebrae. MRI of the cervical spine showed congenital fusion of the bodies of C2 and 3, and vertebral bodies of C4 and C5. There was a large disc protrusion at C3/4 with cord compression. Microdiscectomy of the intervertebral disc between the third and fourth cervical vertebra and a Cloward fusion was done elsewhere. This patient had a similar cervical spinal cord injury about 15 years ago. Following that fall, he developed transient numbness and paresis of the lower limbs. A recent X-ray of the cervical spine (Figure 1) and MRI of the cervical spinal cord (Figure 2) showed congenital fusion at two levels, thus confirming a diagnosis of Klippel-Feil syndrome – Type 2.

A routine intravenous urography (IVU) revealed no kidneys in the renal fossa on both sides, but the presence of crossed, fused renal ectopia in the left ilio-lumbar region. (Figures 3 and 4). Ultrasound examination confirmed the IVU findings. Two separate renal sinus fat echoes could be identified sonographically in the ectopic fused kidney. (Figure 5). MAG 3 renogram showed functioning renal tissue in the left ilio-lumbar region. (Figure 6). This patient has been managing his bladder by reflex voiding, and has been doing well.

Discussion

Patients with Klippel-Feil syndrome and cervical stenosis may be at increased risk of sustaining a transient neurologic deficit after minor trauma. This is probably related to the fused segments and the resultant altered mechanical force transfer that makes the adjacent non-fused segments excessively mobile. [2]. When multiple block vertebrae are present, the normal segments may become hypermobile and be subjected to significantly increased stress. Potentially crippling or fatal subluxations may occur at these levels. Strax and Baran [1] reported two patients with Klippel-Feil syndrome who developed tetraplegia after minor trauma. A 13-year old girl was rendered tetraplegic by falling out of bed trying to shut off her alarm clock. A second case involved a 17-year old male with Klippel-Feil syndrome who became tetraplegic when he sustained a so-called whiplash injury in an automobile accident. Elster [3] reported 35-year old male with Klippel-Feil syndrome who developed tetraplegia after only a minor trauma. Our patient developed cervical spinal cord injury twice, which highlights the fact that persons with Klippel-Feil syndrome may be at high risk for developing a transient neurologic deficit following minor trauma.



Figure 2
MRI of cervical spine shows continuity of the vertebral body of C2 and C3. There is fusion of the vertebral bodies of C4 and C5. Marked disc narrowing and spondylotic changes at C3-4 (level of Cloward procedure).

As persons with Klippel-Feil syndrome may be at increased risk of sustaining a neurologic deficit in the setting of spinal stenosis after minor trauma, they should be provided appropriate guidance to alter their behavior if they experience an episode of neurologic compromise. Knowledge and realization of potential for spinal cord injury following a previous episode of neurologic compromise will enable persons with Klippel-Feil syndrome to make an informed choice of their occupation and leisure activities. Doctor-patient communication is vital to improve overall patient care. [4]. Patients with Klippel-Feil syndrome should also be informed of the medical implications of associated anomalies in other body systems, especially the kidneys.

There is high incidence of congenital anomalies of the genito-urinary tract in patients with Klippel-Feil syndrome. Twenty-five of 39 patients with Klippel-Feil syndrome (64 per cent) had significant genitourinary-tract



Figure 3
20-minute film of intravenous urography shows crossed, fused renal ectopia in the left ilio-lumbar region.

anomalies as demonstrated by intravenous urogram and physical examination. The incidence of these anomalies in three types of the syndrome was essentially the same, unilateral renal agenesis being the most common. [5]. Our patient had crossed, fused, renal ectopia.

Spinal cord injury patients are at high risk for developing renal stones. [6]. It is estimated that within 10 years after injury, 7% of persons with spinal cord injury will develop their first kidney stone. [7]. Stones in an ectopic kidney pose a challenge for percutaneous nephrolithotripsy and for extra corporeal shock wave lithotripsy. Health professionals, caring for cervical spinal cord injury patients with Klippel-Feil syndrome and renal anomalies, should place emphasis on prevention of kidney stones. A large fluid intake is recommended for these patients. [8-10]. A high intake of fluids is still the most powerful and certainly the most economical means of prevention of nephrolithiasis. [11]. Although renal stones are an infrequent cause of renal failure, infectious-related urolithiasis, associated with anatomic and functional urinary tract anomalies, and spinal cord injury, present a great risk for renal failure. [12]. Therefore, prevention of urinary infection and avoidance of indwelling urinary catheters are extremely important in the patients with Klippel-Feil syndrome and tetraplegia.



Figure 4
Coned view of kidney shows anteriorly rotated large extra-renal pelvis, which is occupying the centre and a further renal pelvis with normal configuration facing laterally.

Conclusion

- Persons with Klippel-Feil syndrome and cervical stenosis should be made aware of the potential for sustaining a neurologic deficit after minor trauma.
- Patients with Klippel-Feil syndrome often have congenital anomalies of the urinary tract. Our patient had crossed, fused, ectopia of kidney.
- When patients with Klippel-Feil syndrome sustain tetraplegia they have increased chances of developing urinary tract calculi. Treatment of kidney stones may pose a challenge because of associated renal anomaly.



Figure 5
Ultrasound examination of kidney shows two distinct renal sinus echo patterns thus confirming the presence of a crossed, fused renal ectopia.

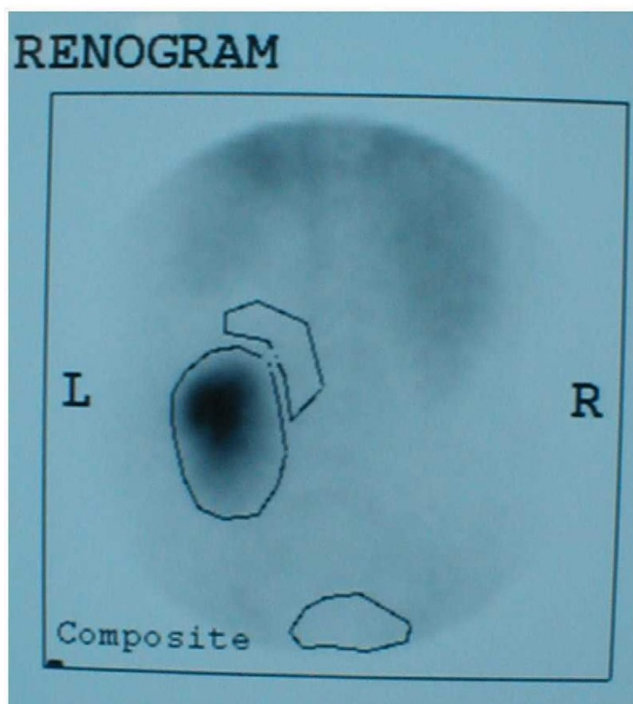


Figure 6
MAG 3 renogram (images obtained in prone position) shows no kidney in the right side. There is functioning renal tissue in the left ilio-lumbar region.

- Health professionals, caring for cervical spinal cord injury patients with Klippel-Feil syndrome and renal anomalies, should place emphasis on prevention of kidney stones. A large fluid intake is recommended for these patients, as a high intake of fluids is still the most powerful and certainly the most economical means of prevention of nephrolithiasis.

Competing interests

None declared

Author contributions

SV generated the idea and wrote the manuscript. PH performed ultrasound scans of kidneys. BMS is the consultant in charge of the patient. All authors contributed to the final version of the manuscript.

Acknowledgement

The authors thank Dr Alexander Vaccaro, Department of Orthopaedics, Thomas Jefferson University Hospital and The Rothman Institute, Philadelphia, PA 19107, U.S.A. Dr Vaccaro kindly reviewed the manuscript and provided valuable advice and comments, which helped the authors to revise the manuscript accordingly. The authors are grateful to the patient, who provided written informed consent for publication of his case in BioMed Central journals. The financial support provided by AstraZeneca (Ms Charlotte Lawledge), Pharmacia (Ms Joanne Thomas), and Shire Pharmaceuticals (Mr Patrick Tierney) is acknowledged with thanks. This enabled the Regional Spinal Injuries Centre, Southport, United Kingdom to become an institutional member of BioMed Central.

References

1. Strax TE, Baran E: **Traumatic quadriplegia associated with Klippel-Feil syndrome: discussion and case reports.** *Arch Phys Med Rehabil* 1975, **56**:363-365
2. Karasick D, Schweitzer ME, Vaccaro AR: **The traumatized cervical spine in Klippel-Feil syndrome: imaging features.** *AJR Am J Roentgenol* 1998, **170**:85-88
3. Elster AD: **Quadriplegia after minor trauma in the Klippel-Feil syndrome: A case report and review of the literature.** *J Bone Joint Surg Am* 1984, **66**:1473-1474
4. Vaidyanathan S, Glass CA, Soni BM, Bingley J, Singh G, Watt JWH, Sett P: **Doctor-Patient Communication: Do people with spinal cord injury wish to receive written information about their medical condition from the physicians after an outpatient visit or after a readmission in the spinal unit?** *Spinal Cord* 2001, **39**:650-653
5. Moore WB, Matthews TJ, Rabinowitz R: **Genitourinary anomalies associated with Klippel-Feil syndrome.** *J Bone Joint Surg Am* 1975, **57**:355-357
6. Vaidyanathan S, Soni BM, Biering-Sorensen F, Bagi P, Wallberg AH, Vidal J, Borau A, Singh G, Sett P, Krishnan KR: **Recurrent bilateral renal calculi in a tetraplegic patient.** *Spinal Cord* 1998, **36**:454-462
7. Chen Y, DeVivo MJ, Roseman JM: **Current trend and risk factors for kidney stones in persons with spinal cord injury: a longitudinal study.** *Spinal Cord* 2000, **38**:346-353
8. Vaidyanathan S, Parsons KF, Krishnan KR, Soni BM, Singh G, Sett P: **What is the optimum fluid intake in male patients with spinal cord injury and neuropathic bladder?** *Spinal Cord* 1999, **37**:594-595
9. Pak CY: **Medical prevention of renal stone disease** *Nephron* 1999, **81**:60-65
10. Pearle MS: **Prevention of nephrolithiasis.** *Curr Opin Nephrol Hypertens* 2001, **10**:203-209
11. Borghi L, Meschi T, Schianchi T, Briganti A, Guerra A, Allegri F, Novarini A: **Urine volume: stone risk factor and preventive measure.** *Nephron* 1999, **81**:31-37
12. Gambaro G, Favaro S, D'Angelo A: **Risk for renal failure in nephrolithiasis.** *Am J Kidney Dis* 2001, **37**:233-243

Pre-publication history

The pre-publication history for this paper can be accessed here:

<http://www.biomedcentral.com/1471-2296/3/6/prepub>

Publish with **BioMed Central** and every scientist can read your work free of charge

"BioMedcentral will be the most significant development for disseminating the results of biomedical research in our lifetime."

Paul Nurse, Director-General, Imperial Cancer Research Fund

Publish with **BMC** and your research papers will be:

- available free of charge to the entire biomedical community
- peer reviewed and published immediately upon acceptance
- cited in PubMed and archived on PubMed Central
- yours - you keep the copyright

Submit your manuscript here:

<http://www.biomedcentral.com/manuscript/>



editorial@biomedcentral.com