

Case Report

Case presentation: A 45-year male developed complete tetraplegia in 2012 when he had an attack of seizure and fell backwards hitting his head on the floor. On assessment the level of C4 complete quadriplegia. Previous history includes a surgery for cystic lesion in the posterior fossa of the skull and known case of Noonans disease with cryptogenic cirrhosis of the liver requiring liver transplant.



Your diagnosis?

Diagnosis

KLIPPEL-FEIL SYNDROME WITH ACUTE TETRAPLEGIA

Definition: 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.

Triad

1. Short neck; Webb neck [Pterygium Colli]
2. Low posterior hairline
3. Restriction of neck ROM [3 or more vertebral fusion]

Other findings:

4. Torticollis
5. May have excessive movement in the unfused segment to compensate. Flexion-extension better preserved than lateral bend or rotation
6. Pain is due to instability or degeneration or stenosis
7. Neurological 1. Radicular: from osteophytes at the mobile segment 2. Cord symptoms
8. Desmoid cystic swelling of the cerebellum

Types: Henisinger: 3 patterns

Type I: C2-3 fusion with occipitalisation of Atlas.

Causes C1-C2 instability with age can cause spinal cord problem

Type II: Long fusion with instability Occipito-cervical spine.

Type III: Single open interphase between two fused segments Spinal movements concentrated at the single open articulation.

Association

Genito-urinary system 35% Nervous, Cardiovascular, hearing impairment 20% Congenital Scoliosis [Hemivertebrae] 60% Sprengel's shoulder 30% Congenital Heart disease 14% [[Septal defects; dextro-cardiac]

Reported case

A 45-year male developed an complete tetraplegia at C4 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.

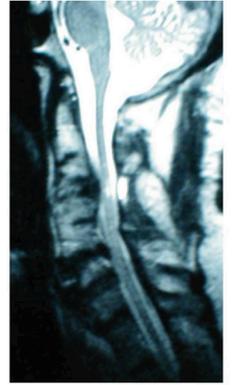
X ray

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

Henisinger type III



2. **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. There was significant narrowing of at C3 with measured canal dimension 50% less than normal due to stenosis and disc secondary to extensive spondylosis at this mobile segment.



3. 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.

Ultrasound examination confirmed the IVU findings.



Due to considerable medical comorbidities non-operative treated with collar and tender loving care. He died after 6 weeks with aspiration pneumonia.

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. 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.

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. However, this was not the issue in the present case as fall as accidental during the attack of a seizure and could not be avoided.

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-

per cent) had significant genitourinary-tract 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. Our patient had crossed, fused, renal ectopia. [2]. Spinal cord injury patients are at high risk for developing renal stones. A large fluid intake is recommended for these patients. A high intake of fluids is still the most powerful and certainly the most economical means of prevention of nephrolithiasis.

Strax suggested that patients with the Klippel-Feil syndrome might be at risk for sudden quadriplegia after minor Trauma. Other reports showed that the cervical quadriplegia not only followed trivial trauma but also due to the Symptoms and signs of myelopathy are secondary to spondylosis and stenosis due to stress concentration.

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.

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. BMC Family Practice 2002, 3; 1
3. African Health Sciences 302 Vol 10 No 3 September 2010: 302