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Klippel-Feil syndrome with auxiliary anterior cervical meningomyelocele and thoracic syringomyelia: A case report

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Abstract

Study Design: Case report.

Objectives: Since this is the first ever case of a male patient with Klippel-Feil syndrome with anterior cervical meningomyelocele and syringomyelia. All four previously reported cases were female patients. This makes this case unique.

Summary of Background Data: Klippel-Feil syndrome with auxiliary anterior cervical meningomyelocele is a rare entity. To best of our knowledge so far only four cases are reported.

Methods: A 22-year-old male patient was presented to neurology outpatient department with two years history of left- hand paresthesia and progressive weakness. The diagnostic evaluation showed Klippel-Feil syndrome with auxiliary anterior cervical meningomyelocele and thoracic syringomyelia.

Results: Patient was sent to neurosurgery department for intervention. After discussing the possible risks & complications of intervention he opted for conservative therapy and declined the surgery.

Conclusions: The paucity of data is the key reason for any recommended protocol for management of such patients but the available literature recommends neurosurgical intervention in symptomatic patients.

Key Words: paresthesia, muscle weakness, Klippel-Feil syndrome, meningomyelocele, syringomyelia, MRI Spine, CTA neck, congenital anomalies, Spinal cord, anterior horn cell disease

Level of Evidence: 5

Introduction

Klippel-Feil syndrome (KFS) is a rare congenital anomalous fusion of one or more cervical vertebrae. It is characterized by a triad of short neck, restricted neck movements & low hairline, seen only around 50% of patients with KFS. [1] Many extra vertebral manifestations including cardiopulmonary, urogenital & gastrointestinal system are reported in association with KFS. Associated neurological anomalies include hydrocephalus, syringomyelia, hearing loss, thin corpus callosum, atlanto-occipital fusion, split cervical spinal cord, meningomyelocele, and pyramidal tract decussation failure.[1-2] KFS with auxiliary anterior cervical meningomyelocele is extremely rare and so far only four cases are reported. We discuss the 5th case which presented to us with progressive left-hand paresthesia and wasting.

Case Report

A 22-year-old right-handed man presented to the outpatient clinic with two years history of progressive clumsiness, weakness, and numbness of the left hand. No history of neck pain or radiculopathy and the right hand was asymptomatic. There is no history of urinary urgency or increased frequency. There is no significant past medical history of note. On examination, there was significant wasting of intrinsic muscles of the left hand (Figure 1). The weakness of C8 to T1 myotomes muscles (finger flexion and abduction) was more marked (MRC grade 1/5) compared to C6 and C7 myotomes (wrist extension, elbow extension, wrist flexion and finger extension) whose strength was MRC grade 3/5. The tricep and brachioradialis reflexes were absent. Pain & temperature sensation was reduced in the C6-T1 dermatomal distribution in the affected hand. Although asymptomatic, depressed right tricep and brachioradialis reflexes and a mild functional weakness of right hand was also noted. The deep tendon reflexes were exaggerated in lower limbs. There was no restriction of the neck flexion and extension and the neck hairline was normal. No obvious spine deformity noted. The rest of his neurological & general physical examinations were normal. The plain X-ray cervical spine showed fused vertebral bodies from C6 to T1. An MRI scan of whole spinal cord demonstrated a large bony defect in anterior parts of C5 to T1, through which meninges and cervicodorsal cord were protruding forming a sac measuring 3.2×5×6.1 cm. It extended into posterior mediastinum lying

predominantly on the left side (Figure 2A-C). The dural sac widened at this level & having two hemi-cords with a dilated central canal. Moreover, there was a syrinx extending from T9 to T12 (Figure 3AB). The CT scan & 3D CT reconstruction of the cervical spine revealed the fusion of last three contiguous cervical & first thoracic vertebrae along with anterior defect at the same level. Additionally, there was mild focal scoliosis with concavity towards the left side (Figure 4A-D). The CT scan head and CT angiogram (CTA) neck were normal. His nerve conduction studies and EMG were consistent with focal anterior horn cell disorder affecting C6-T1 on the left side. Interestingly similar but less severe findings were also noted on the right side as well. The results of lumbosacral spine X-ray, echocardiography, and abdominal, pelvic & scrotal ultrasound were normal. Based on these imaging features we made the diagnosis of Klippel-Feil syndrome with anterior cervical meningomyelocele. The neurosurgical consultation was taken. The patient declined surgical option due to the possible risks associated with the procedure. He opted for a conservative approach and has been referred to physiotherapy services. He will be followed in the neurosurgery outpatient clinic.

Discussion

Klippel-Feil syndrome (KFS) is a rare perplexing congenital condition of inappropriate fusion of one or more cervical vertebrae. Various associated spinal & extraspinal manifestations are reported with KFS which include neurological, skeletal, cardiopulmonary, urogenital and gastrointestinal system.[1] Meningomyelocele (MCC) is the most common malformation in the spectrum of neural-tube defects. The MCC can occur at any level of the spine but most frequently affects the lower lumbar and/or sacral spine. Consequently, anterior cervical MMC associated with KFS is an extremely uncommon condition.[2] Going through English literature we found only four reported cases of anterior cervical MMC with KFS. All the four patients were women. The clinical histories and examinations findings and imaging features of all four reported cases are summarised in table 1. Among these four, only one patient undergone through surgical intervention and only one had associated syringomyelia.[2,5]

The classic triad of KFS is neck webbing, low hairline & limited neck mobility, which is found around 50% patient. The usual presenting complaint is reduced cervical spine mobility. The

facial asymmetry & hearing loss are two other frequent presenting features.[1] Various multi-systemic abnormalities are associated with KFS. The neurological findings include developmental anomalies, nonspecific headache & neck pain, hydrocephalus, myelopathy, radiculopathy, synkinesia (pyramidal tract decussation failure results in mirrored movements[6]), and split-spinal cord. Scoliosis and Sprengel deformity (high scapula) are the most common musculoskeletal anomalies associated with KFS. Other associated abnormalities include renal agenesis, absent vagina, uterus and ovaries, undescended testes, ventricular septal defect, aortic arch aneurysm, aberrant vertebral arteries, have also been reported.[1-5] Diagnostic workup includes plain X-ray cervical spine & chest, MRI cervical spine & brain, and CTA neck. The latter is done for any possible aberrant artery. Additionally echocardiography, abdominal & pelvic (including scrotal in men) ultrasound and imaging of thoracic & lumbar spine should also be done for any possible associated anomalies. The treatment of KFS depends upon the severity of symptoms. Symptomatic patients need neurosurgical interventions[2].

In conclusion, patients with KFS with or without associated anterior MMC can present to neurological clinics and proper diagnostic workup and then referral to spine surgeon is crucial.

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Figure Legends

Figure 1: The left hand of the patient showing extensive wasting of intrinsic muscles including the marked atrophy thenar and hypothenar muscles.

Figure 2: Sagittal T1 (A) & T2 (B) and coronal T2 (C) weighted MRI of cervical spine showing anterior meningocele protruding through vertebral body defect.

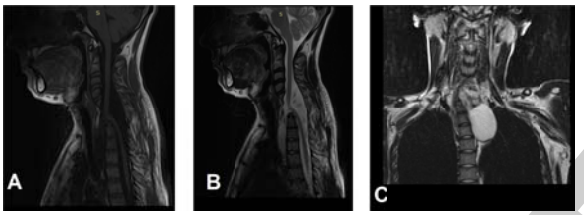

Figure 3: Sagittal T1 (A) & T2 (B)) weighted MRI of a thoracolumbar spine showing syrinx extending from the lower border of D9 to the lower border D12.

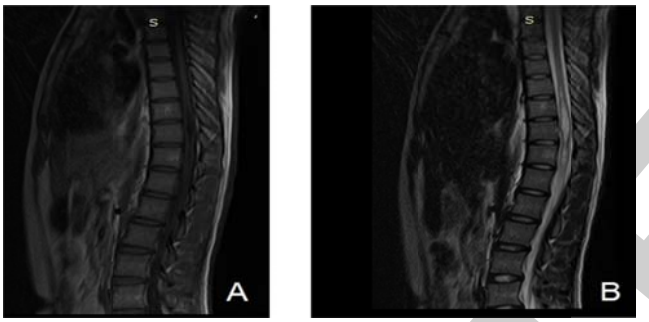

Figure 4: Sagittal (A) and coronal (B) reconstruction of the CT scan and anterolateral (C) and anterior (D) views of the 3D CT reconstruction showing fusions of C5-T1 vertebrae and anterior bony defect at the same level. Note the mild focal scoliosis.

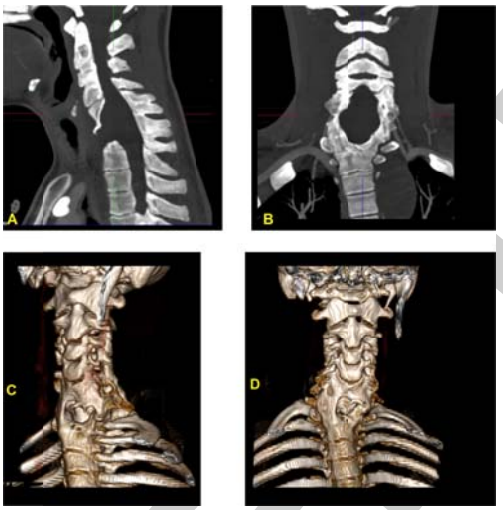

Table 1: Brief summary of all four published case of anterior Meningomyelocele (MCC) with Klippel-Feil syndrome (KFS) [2-5]

Author	Case description	Imaging findings
Balachandran G[3]	A 36-year-old woman with a bilateral symmetrical weakness of intrinsic muscles of hand.	MRI cervical spine showed fused cervical vertebrae & neural arches and anterior MMC at C6 and C7 level.
Jiang et al[4]	A 23-year-old woman with rapidly progressive weakness in both hands. There was mild restriction of neck flexion & extension and decreased hands grip strength.	MRI cervical spine revealed anterior MMC at the C6 and C7 level. Additional findings like prolapsed & kinked spinal cord extending into enlarged thecal sac, hydrocephalus, thinning trunk & genu of corpus callosum, expansion of 4th ventricle and maldevelopment of cerebellar tonsils were also noted.
Brokinkel et al[2]	A 26-year-old woman who was diagnosed as a case of anterior MMC 7 years back, presented with progressive weakness and hypaesthesia in both hands. Bilateral hearing loss, reduced neck mobility, symmetrical hypaesthesia of C6 and motor weakness of C7 & C8. Lower limbs hyperreflexia, posterior column impairment & subtle signs of myelopathy.	MRI cervico-thoracic showed incomplete fusion of anterior parts of C2 to T1 with anterior MMC and Stretching of nerve roots between cord & the neuroformina. MRI brain revealed left frontal arachanoid cyst.
Gallagher et al[5]	A 45-year-old woman who was referred from emergency department where she presented after mechanical fall and her plain X-ray of cervical showed fusion of C5-C7 vertebrae. There was subjective weakness of upper limbs.	MRI cervical spine revealed fusion of C5- C7 vertebrae with anterior MMC at the level of C6-C7. Additionally there was syringomyelia extending from the disc space of C4/5 to the disc space of C7/T1.