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TIED AT THE NECK, STRAINED AT THE CORD, AND BURDENED BY THE KIDNEYS: A MULTISYSTEM CASE OF KLIPPEL-FEIL SYNDROME

General Medicine

Dr Shah Devansh Dipak — 3 Year MD General Medicine Resident (R3), Dhiraj Hospital Sumandeep Vidyapeeth
Dr Mahajan Nikhil Ramdas — 3 Year MD General Medicine Resident (R3), Dhiraj Hospital Sumandeep Vidyapeeth
Dr Pattigilli Siva Krishna — 3 Year MD General Medicine Resident (R3), Dhiraj Hospital Sumandeep Vidyapeeth
Dr Amal Bhattacharya* — Professor MD General Medicine, Dhiraj Hospital Sumandeep Vidyapeeth
*Corresponding Author

ABSTRACT

Klippel–Feil syndrome (KFS) is a rare congenital condition characterized by failure of segmentation of the cervical vertebrae, commonly presenting with the classical triad of a short neck, low posterior hairline, and restricted cervical range of motion, though all three features are present in fewer than half of affected individuals. The syndrome is frequently associated with multisystem involvement, including scoliosis, Sprengel deformity, renal anomalies, cardiac defects, and neurological manifestations. We report a rare case of a 34-year-old male who presented with progressive symmetrical limb weakness, spasticity, and dysarthria over several years, culminating in a bedridden state. Neuroimaging revealed partial fusion of the C2–C3 vertebrae consistent with Klippel–Feil syndrome, along with narrowing of the cervical canal. Additional investigations demonstrated bilateral small kidneys with hydronephrosis, highlighting renal involvement. Notably, there was no evidence of congenital heart disease, significant sensory deficits, or acute compressive myelopathy. The patient also had a history of epilepsy and old bilateral capsular–ganglionic infarcts, which likely contributed to the progressive neurological deficits. This case emphasizes the importance of comprehensive evaluation for associated systemic abnormalities in patients with Klippel–Feilsyndrome. Early diagnosis and multidisciplinary management are essential not only to prevent neurological deterioration but also to address cosmetic, functional, and psychosocial concerns, thereby improving overall quality of life.

KEYWORDS

Klippel–Feil syndrome, congenital cervical fusion, multisystem involvement, neurological deficits

INTRODUCTION

Klippel-Feil syndrome was first described by Maurice Klippel and Andre Feil in 1912 in patient with congenital fusion of cervical vertebrae [1]. In KF syndrome, classically there is a triad of short neck, a low posterior hairline & a limited range of neck movements especially of lateral bending. In fewer than 50% of cases have all the three elements. Klippel-Feil syndrome occurs in one of every 42,000 births, and 60% of cases are female [2]. KF syndrome is group of deformities that result due to failure of segmentation of cervical spine. This syndrome is associated with Sprengel deformity, high scapula, scoliosis, urinary tract anomalies, congenital heart disease & hearing lost in 30% of cases. Usually in Sprengel deformity there is loss of abduction & forward flexion after 90 deg. In 30 % of cases with Sprengel deformity, the scapula is bound to cervical spine by fibrous tissue, cartilage or an omovertebral bone which restrict abduction of shoulder after 90 degrees [3].

CASE DESCRIPTION

Patient Profile

A 34-year-old male, newspaper vendor, resident of Vadodara, presented with progressive limb weakness and speech difficulty.

History of Present Illness

The patient was reportedly well until approximately **5 years prior**, when he sustained a **road traffic accident (RTA)** resulting in **left upper limb injury**. He received treatment at S.S.G. Hospital with adequate recovery and resumed routine activities.

A few months later, he developed **difficulty standing from a sitting position**, followed by **progressive difficulty walking**. Over the next **2–3 years**, the weakness gradually worsened, involving both lower limbs symmetrically and later affecting upper limbs. He developed increasing **spasticity/stiffness** of both upper and lower limbs. He also noted **slurring of speech** for the past **4 years**, gradually progressive.

For the past **1 year**, he has been **bedridden**, unable to ambulate, and requiring assistance for all activities of daily living, including bathing and feeding.

There is **no history of sensory loss**, bowel/bladder incontinence, headache, visual complaints, cranial nerve deficits, trauma apart from RTA episode, or toxin exposure.

He additionally presented with **fever for 1 day**, high grade, associated with chills and rigors. No cough, vomiting, or diarrhea.

Past Medical History

Epilepsy since 2021
Seizure episodes initially **8–10 years ago**
Treated for **2–3 years**, later stopped on medical advice
No recent episodes

COVID-19 infection – remote
No history of: Tuberculosis, diabetes, hypertension, thyroid disorders
Surgical history: Left elbow/olecranon fracture treated with ORIF
No history of blood transfusion

Medications

Sodium Valproate 500 mg – 1-1-1
Folic Acid 5 mg – 1-0-0
Atorvastatin 40 mg – 0-0-1
Famotidine 20 mg – 1-0-1
Domperidone 10 mg – 1-0-0

Family History

Father: Hypertension × 2–4 years
No family history of similar neurological illness

Social History

Occupation: Newspaper seller
Resides in Vadodara
No addictions

Examination:

HMF : Conscious and oriented to Time, Place and Person
Attention: Normal
Memory (immediate, recent, remote) : intact

CNS Examination:

B/L Pupil Reactive to Light

EOM- Normal
7th CN – Left sided OMN palsy
Dysarthria present

Investigations
MRI Brain (2022), epilepsy protocol (P+C):
Old **bilateral capsular–ganglionic infarcts**
MRI Cervical Spine (2022):
Normal
MRI Lumbosacral Spine (2022):
Diffuse L5–S1 disc bulge
Facet joint arthritis
Ligamentum flavum hypertrophy indenting the thecal sac
CT- KUB (2025)
B/L small size kidney
B/L hydronephrosis
2D Echo (2025)
Normal
MRI LS Spine (2025)
Narrowing of cervical canal
MRI WSS (2025)
Partial fusion of C2-C3 noted

DISCUSSION

This patient presented with **chronic progressive symmetrical limb weakness** with **spasticity** and **bulbar involvement (slurred speech)** over a course of several years, eventually leading to a bedridden state. The timeline suggests onset months after an RTA, though no major brain/spinal lesion was documented at that time.

MRI brain demonstrated **old bilateral capsular–ganglionic infarcts**, possibly explaining the pyramidal weakness and speech involvement.

However, the slow progression raises consideration for:
Post-stroke spastic sequelae with delayed deterioration
Hereditary spastic paraplegia
Leukodystrophy
Motor neuron disease (less likely due to long course + early post-stroke onset)
Chronic compressive myelopathy (lumbar level findings insufficient to explain quadriparesis/bulbar signs)
Lumbar imaging shows changes unlikely to account for upper limb weakness or bulbar dysfunction.
Recent fever is nonspecific, requiring evaluation for infectious causes.

CONCLUSION :

Patients with Klippel Feil Syndrome should be assessed for associated systemic abnormalities beside cervical fusion, and such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to the patient.

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