
Klippel-Feil syndrome with basilar invagination and spinal cord compression**Dr. Ruthira Eshanth VN¹, Dr. Amish Aggarwal², Dr. M.S. Hemhnath³**^{1,2}Resident, Sree Balaji Medical college, 7, Work' S Road, Chrompet, Chennai, Tamilnadu, India³Assistant Professor, Sree Balaji Medical college, 7, Work' S Road, Chrompet, Chennai, Tamilnadu, India***Corresponding author**

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Abstract: Klippel-Feil syndrome (KFS) is defined as congenital fusion of two or more cervical vertebrae. Defective segmentation along the embryo's developing axis during 3-8 weeks of gestation results in KFS. It commonly manifests as short neck with reduced mobility and a low posterior hairline, found only in 40-50% of patients. We present a case of KFS with basilar invagination of C2 vertebra causing cervical myelopathy. We discuss here the clinical presentation, radiographic findings and various treatment options for cervical spine abnormalities in KFS.**Keywords:** Klippel-Feil syndrome, basilar invagination of C2, spinal cord compression, clinical triad

INTRODUCTION

Klippel-Feil syndrome (KFS) is a rare skeletal disorder characterized by the congenital abnormal fusion of two or more cervical vertebrae [1, 2, 3] and its incidence is 1 in 40,000 to 50,000 live births [3]. KFS is a condition presenting with diversity of skeletal/extra-skeletal manifestations. Studies have found that up to 50 % of young KFS patients encounter cervical-spine-related symptoms [1, 4, 5].

Individuals with KFS are at increased risk to neurologic injury due to altered spinal biomechanics, leading to greater incidence of hypermobility, spondylolisthesis, degenerative changes, and spinal canal stenosis [6,7]. It is important to recognize and diagnose KFS at the earliest to provide precautionary guidelines and proper non-surgical or surgical management.

CASE PRESENTATION

A 22-year-old male presented with complaint of bilateral upper and lower limb weakness and paresthesia's since last one year. Patient also complained of headache predominantly in frontal region since last one and a half year. There was no history of trauma. On physical examination, he was found to have short neck, low posterior hairline, and restricted range of neck motion. Neurological examination revealed reduced power in all four limbs.

Plain radiograph cervical spine was obtained in antero-posterior and lateral views. X ray lateral view showed abnormal high position of C2 vertebra

protruding into the skull base, fusion of anterior-posterior elements of cervical vertebrae at C4-C5 and C5-C6 levels (Figure 1A). Anteroposterior view demonstrates cervical scoliosis with convexity towards left side and degenerated cervical osteophytes (Figure 1B). Flexion and extension films of the cervical spine are necessary for evaluation of cervical instability, but in our case, we did not take flexion/extension films due to basilar invagination by high positioned C2 vertebrae compressing over the cord which may lead to further complications.

MRI cervical spine was done, Sagittal T1, T2, STIR; Coronal STIR; Axial T1, T2 sequences were obtained. MRI revealed Atlanto – occipital assimilation, abnormal high position of C2 vertebra protruding into the skull base. There was marked narrowing of foramen magnum (measuring 3.3 mm) with compression of cervico-medullary spinal cord. Fusion of anterior-posterior elements of cervical vertebrae at multiple levels was seen with omovertebral bony connection between the supromedial angle of the left scapula and left lateral posterior elements of lower cervical vertebrae. It also revealed cervical scoliosis with convexity towards left side. Degenerated cervical discs were seen (Figure 2 A & B).

CT could better image many of the features seen on plain radiograph and MRI. In addition, CT revealed multiple segmental abnormalities with fusion of the atlas and the base of occiput along with fusions at C4–C5, C5–C6, C7–D1, D2–D3 and D3–D4 vertebrae (Figure 3).



Fig- 1[A]: X ray lateral view show abnormal high position of C2 vertebra protruding into the skull base. Fusion of anterior-posterior elements of cervical vertebrae at C4-C5 and C5-C6 levels. [B]: X ray anteroposterior view shows cervical scoliosis with convexity towards left side.

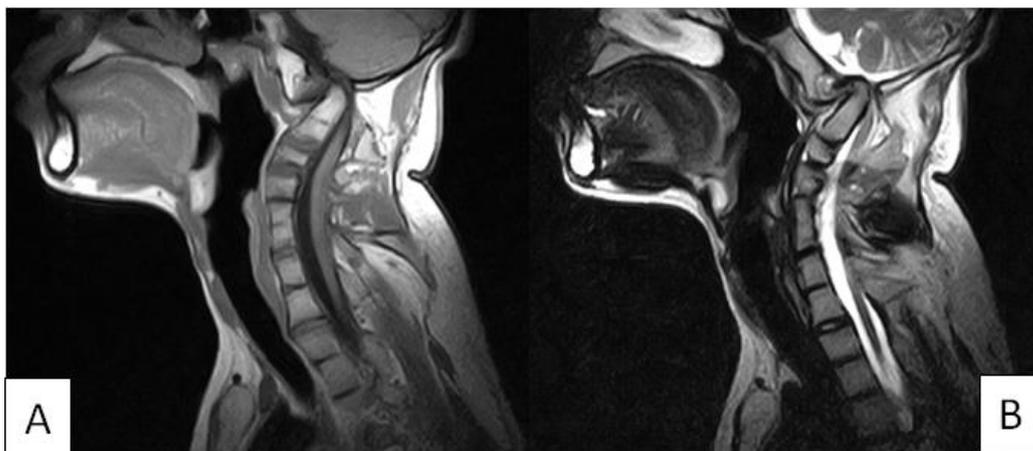


Fig. 2 A and B MRI of the cervical spine without contrast demonstrates abnormal high position of C2 vertebra protruding into the skull base with compression of cervico-medullary spinal cord



Fig-3: NCCT of the cervical spine reveals segmentation abnormalities and fusion of the atlas and the base of occiput along with fusions at C4-C5, C5-C6, C7- D1, D2-D3 and D3-D4 vertebrae.

DISCUSSION

Klippel-Feil syndrome (KFS) is a rare congenital skeletal disorder that results from abnormal fusion of two or more cervical vertebrae. Patients affected with KFS presents with classical triad consisting of short neck, low posterior hairline and restricted range of neck motion, found in approximately 40 - 50 % of patients [3]. Present case was a man who had mentioned triad.

Andre Feil classified the syndrome into three types [2]:

- Type I - Massive fusion of the cervical spine
- Type II - Fusion of one or two vertebrae
- Type III - Presence of thoracic and lumbar spine anomalies in association with type I or type II Klippel-Feil syndrome

Samartzis *et al.* in his subsequent articles proposed his own classification system [1], which stratified patients as follows:

- Type I - Single-level fusion
- Type II - Multiple, noncontiguous fused segments
- Type III - Multiple, contiguous fused segments

The most common fused cervical levels in KFS are C2–C3 (71.0 %) followed by C5–C6 (67.7 %), C6–C7 (67.6 %), and C3–C4 (29.0 %) [1]. Our patient had fusion at multiple cervical levels with an elevated left scapula and a bony prominence extending from the shoulder to the neck.

There are many symptoms and signs that point to KFS and it usually present as cosmetic problems or severe problem predisposing to neurologic damage. Thus, it may render as an emergency and is essential to diagnose it at the earliest to prevent serious neurologic sequelae.

Some patients with this syndrome had extra anomalies that can be accompanying to KFS. Torticollis and loss of symmetrical expression of the face has been reported in 10% and 11.5% of cases with KFS respectively. [7] However in our case, there is no torticollis.

Our patient that was a rare presentation in that age had atlanto-occipital assimilation, basilar invagination of C2, spinal cord compression, Sprengel deformity and scoliosis.

Sprengel deformity (SD) is seen in 7-42% of cases with KFS. Its most prominent feature is elevation of scapula. Patients with SD presents with cosmetic deformity and muscular dysfunction of the associated upper limb. Muscular dysfunction results from defects in the trapezius, rhomboids and levator scapulae. Cartilaginous or bony or fibrous tissue fuse the scapula and processes of cervical spines which leads to SD [8].

These patients are not benefited with conservative management and need surgical correction. [9].

MRI is the foremost imaging tool for evaluation of spinal cord abnormalities associated with KFS. MRI can reveal spinal abnormalities such as Chiari I malformation, syringomyelia, myelomalacia and diastematomyelia. Other associated radiographic findings include narrowing of the spinal cord, widening of the spinal canal, vertebral osteophytes or disc protrusion, ligamentum flavum hypertrophy, spinal canal stenosis, spondylolisthesis and spondylolysis. Nguyen and Tyrrel reported that 53.8 % of patients displayed a wasp-waist sign on MRI, a valuable sign to diagnose fusion of vertebrae [10]. CT myelography may be utilized in cases where MRI is contraindicated. CT scan is optimal imaging modality to visualize complex bony abnormalities, spinal instrumentation and can be used in surgical planning. Flexion and extension films of the cervical spine are necessary to evaluate cervical instability.

KFS patients develop hypermobility, degenerative changes, spinal stenosis, spondylolisthesis and neurologic injury resulting from altered spinal biomechanics because of improper vertebral fusion. Three particularly unstable fusion patterns are there which include the following: fusion of C2–C3 with occipitalization of the atlas, a long fusion with an abnormal occipitocervical junction and a single open space between two fused segments [3]. The patient presented in this case possesses an unstable combination of the fusion patterns, including an abnormal occipitocervical junction with fusion of C4–C5 and a long fusion in addition to other cervical and upper thoracic abnormalities.

Management is supportive and symptomatic in most of the cases. Treatment approach depends upon features and severity of the disease. Various conservative therapies are available, which include the use of cervical collars, braces, traction, physical therapy, non-steroidal anti-inflammatory drugs (NSAIDs) and various pain medications. However, in some patients, symptoms are progressive due to degenerative changes in the spine. The role of prophylactic surgery for spinal stabilization remains controversial and is not indicated in cases of hypermobility without neurological deficit. A study by Theiss *et al.* found that 22 % of patients with congenital scoliosis and KFS developed cervical or cervical-related symptoms, with only two of those patients requiring surgery [11]. The conservative treatment of radiculopathy should be aggressive, Surgery is reserved for patients with refractory radiculopathy and/or myelopathy.

Surgeons should look for individual patient characteristics that may predispose to future neurological injury, such as a type II or type III fusion.

Patients with congenital cervical fusion and underlying congenital cervical stenosis are at high risk of developing neuropathy, as noted by Theiss *et al.* who found a significantly greater incidence of upper extremity pain in these patients [11]. Early diagnosis of KFS is essential to minimize the risk of injury during routine daily activities or operative intervention. Surgical intervention is required in cases of myelopathy to improve neurologic function and prevent further neurologic compromise. Decompression of the nerve roots and spinal cord in cases of cervical spondylitic myelopathy can be achieved with multiple discectomies preferred over corpectomy or discectomy-corpectomy combined approaches for multilevel disease [11]. Cervical laminectomy is required in cases with extension of disease posteriorly.

The present case refused to undergo surgery, so he was managed medically and with physiotherapy.

CONCLUSIONS

Klippel-Feil syndrome is a rare condition with varying clinical manifestations. Our case as classical clinical triad with rare radiological presentation of Klippel-Feil syndrome with basilar invagination and spinal cord compression. Early diagnosis and understanding of the biomechanics of Klippel-Feil anomaly may facilitate prompt referral for appropriate management and avoidance of permanent disability.

Various imaging modalities, including MRI, CT, and X ray of the cervical spine are necessary for diagnosis and evaluation of extent of congenital deformity.

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