Klippel–Feil Syndrome with Benign Hypermobile Joint Disease in a Young Indian Female: A Rare Case Report

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Abstract
Klippel–Feil syndrome (KFS) is a congenital segmentation anomaly of cervical vertebra leading to vertebral fusion, often accompanied by other systemic anatomical and functional anomalies. Benign hypermobile joint syndrome (BHJS) is a frequently overlooked cause of chronic musculoskeletal pain. Early identification and management of both conditions are essential to prevent long-term damage and suffering due to chronic pain and impaired activities of daily living (ADLs). The authors present this rare case report of KFS with BHJS in a young Indian female.

Keywords: Cervical, Congenital, Fusion, Hypermobility, Klippel–Feil.


Introduction
Klippel–Feil syndrome (KFS) is a rare congenital anomaly characterized by the fusion of at least two cervical vertebra. This syndrome was first defined by Maurice Klippel and Andre Feil in 1912.1 The classical triad of KFS consists of short neck, low posterior hairline, and limitation of range of motion of cervical spine,2 with less than 50% cases having all three features. It is often associated with a wide array of musculoskeletal and visceral anomalies such as congenital heart defects, deafness, genitourinary abnormalities, cleft palate, scoliosis, Sprengel's deformity, facial asymmetry, etc. Klippel–Feil syndrome occurs in 1 in every 42,000 live births, with 60% female predilection.³

Benign hypermobile joint syndrome is characterized by joint hypermobility and is a commonly underdiagnosed cause of chronic musculoskeletal pain. It is seen in up to 3% of the general population.⁴ The term “benign” is used to differentiate it from more severe syndromes also presenting with hypermobility such as Ehlers–Danlos syndrome, Marfan syndrome, etc.

Early diagnosis and management of both the syndromes are required for patient education and adaptation of precautionary steps at an early age.

Case Description
A 16-year-old Indian female student presented in our outpatient department with multiple joint pain involving bilateral elbow joints, knee joints, and wrists since 4 months and neck pain for 3 months. The pain was gradual in onset and progressive in nature. There was no radiation of neck pain or any neuropathic symptoms accompanying it.

There was no history of recent trauma, any preceding infection, or early morning stiffness. The patient has a history of surgery for congenital inguinal hernia at 4 years of age.

Clinical Findings
On clinical examination, it was found that the patient had polyarthritis, hypermobile joints, facial asymmetry, left laterocollis (Fig. 1), decreased range of motion of the cervical spine, low posterior neck hairline (Fig. 2), low set ears (Fig. 3), and dextroscoliosis of lumbar spine.

Features of synovitis, synkinesia, abnormal eye movements, limb length discrepancy, muscle weakness, and sensory abnormalities were absent.

Severity of both neck and joint pain as measured on the numerical rating scale (NRS) were 8/10.

Fig. 1: Facial asymmetry and left laterocollis

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The patient was advised X-rays of the cervical spine and whole spine. Hematological investigations for complete blood count, thyroid profile, serum uric acid, urea, creatinine, calcium, vitamin D (25(OH)D) level, and alkaline phosphatase were also advised. An ultrasonography of the whole abdomen, echocardiography, pulmonary function test, and pure tone audiometry were carried out to rule out other systemic anatomical anomalies.

The patient was diagnosed as a case of benign hypermobile joint syndrome (BHJS) according to the revised Brighton criteria (1998), fulfilling two major criteria: (a) Beighton score of 4/9 (Fig. 4) and (b) polyarthralgia of more than 2 months, and two minor criteria: (a) arm span and height ratio > 1.03 (Fig. 5) and (b) history of congenital inguinal hernia.

X-rays showed definite fusion of cervical vertebra C6–C7, possible fusion of C5–C6 (Figs 6 and 7), dextroscoliosis of
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Thoracolumbar spine with apex at L2 and Cobb’s angle of 25° (Fig. 8). Magnetic resonance imaging (MRI) and computed tomography (CT) scan of cervical spine were carried out to rule out other anomalies, which revealed a bulky anterior arch of atlas with an aplastic posterior arch (Figs 9D and E) in addition to congenitally fused vertebra C5–C7 (Figs 9A to C). Based on the radiological findings, the patient was diagnosed as a case of KFS.

Hematological investigations were within normal limits except for a deficiency of serum vitamin D (10.52 ng/mL) and an elevation of thyroid stimulating hormone (TSH) (6.9 μIU/mL). Ultrasonography of the whole abdomen, echocardiography, pulmonary function test, and pure tone audiometry showed no abnormalities.

**TREATMENT**

The patient was treated pharmacologically with analgesics and muscle relaxants. Vitamin D and levothyroxine supplementation were done.
The patient was counseled about her condition and educated about proprioceptive training, posture care, joint protection, and energy conservation techniques. Static neck exercise, range of motion exercises for cervical spine, stretching of left sternocleidomastoid, strengthening of right sternocleidomastoid, and isometric strengthening of key muscles were advised. Supervised physical therapy was prescribed for correction of scoliosis. Six monthly follow-up with X-ray of whole spine has been planned for monitoring of progression of scoliosis and bracing, if required.

The patient experienced symptomatic improvement with the above management. The severity of joint pain decreased significantly as measured by the NRS score of 2/10. There has been minimal improvement in the range of motion of cervical spine but the decrease in neck pain severity as measured by NRS score of 3/10 has led to marked improvement in the activities of daily living (ADLs).

**Discussion**

Klippel–Feil syndrome is a congenital disorder characterized by cervical vertebral fusion abnormalities due to failure of normal segmentation of mesodermal somites, an event occurring between the third and seventh week of the embryonic life. It is more often than not accompanied by diverse anatomical and sensory abnormalities involving primarily the musculoskeletal, cardiovascular, auditory, and neurological systems.

Guille et al. distinguished cervical fusion defects in KFS into three types: type I single congenitally fused cervical segment; type II multiple, noncontiguous, congenitally fused segments; and type III multiple contiguous, congenitally fused segments. A fourth type has been suggested to be associated with sacral agenesis. Type I fusions are most commonly associated with axial neck symptoms, while types II and III are associated with myelopathic and radicular symptoms. In this case, our patient can be classified as type III.

The MRI is the imaging modality of choice for spinal cord anomalies. It is useful for early diagnosis of KFS and associated spinal cord anomalies such as Chiari malformation, syringomyelia, myelomalacia, etc. The CT myelography may be advised in patients having contraindications to MRI.

Most patients with KFS can be treated symptomatically, surgical management being the option for those with refractory radiculopathy or myelopathy.

Benign hypermobile joint syndrome is a commonly overlooked cause of chronic musculoskeletal pain in the general population, as its diagnosis requires a high level of clinical suspicion and specific maneuvers to be performed during clinical examination. Diagnosis is carried out by Beighton scoring and the revised Brighton criteria (1998). Early diagnosis and initiation of therapy are useful to prevent damage to the affected ligaments and joints.

Lifestyle modification is the most effective and important intervention. Patient education regarding good body mechanics, joint protection, and energy conservation techniques is essential. Though regular exercise is important, overtraining and excessive joint movement may be harmful leading to joint, cartilaginous, and meniscal injury. Bracing and neuromuscular taping are cheap but useful interventions for injury prevention and gait improvement.

Strengthening regimens for periarticular musculature are essential for stabilizing the joints. Customization of physical therapy according to each patient is necessary as training regimens need modifications according to variations in body habitus and occupation. Nutritional supplementation, especially with vitamin D for those with reduced bone mineral density, has been proposed as an important treatment modality.

**Conclusion**

The coexistence of these two syndromes in a single patient was found to be extremely rare. As discussed, the patient was managed conservatively with pharmacological and nonpharmacological approaches, and the results were quite satisfactory.
REFERENCES