Multi-Disciplinary Journal
ISSN No- 2581-9879 (Online), 0076-2571 (Print)
www.mahratta.org, editor@mahratta.org

# Klippel Feil syndrome : A Rare Case Report

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**Abstract**

In Klippel-Feil syndrome, classically there is a triad of short neck, a low posterior hairline and a reduced cervical range of motion significantly in lateral flexion. In less than 50% of the patients diagnosed with Klippel-Feil Syndrome have all the three components.

**Case Report**: In the present case we came to conclusion that patient has Scoliosis, Sprengel deformity and there were no complaints suggesting of any renal disease, congenital heart disease and neurological impairment. The present case has classical triad low posterior hairline, short neck and reduced cervical range of motion.

**Conclusion**: A rare case of Klippel-Feil Syndrome is being presented with the aim that such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to her and to her parents.

**Keywords**: Congenital, fusion, Klippel-Feil syndrome.

**Introduction:** Klippel-Feil syndrome was first discussed by Maurice Klippel and Andre Feil in 1912 in patient with congenital fusion of cervical vertebrae [1]. In Klippel-Feil syndrome, classically there is a triad of short neck, a low posterior hairline and reduced cervical range of motion significantly in lateral flexion. In less than 50% of cases have all the three components. Klippel-Feil syndrome occurs in one of every 42,000 births, and about 60% of cases are female [2]. Klippel-Fiel syndrome is group of deformities that result due to failure of segmentation of cervical spine. This syndrome is associated with Sprengel deformity, raised scapula, scoliosis, urinary tract anomalies, congenital heart disease and loss of hearing in 30% of cases. it is commonly seen that in Sprengel deformity there is reduced shoulder abduction and flexion range of motion. In 30 % of cases with Sprengel deformity, the scapula is attached cervical spine by fibrous tissue, cartilage or an omovertebral bone which reduced the shoulder abduction and flexion range of motion[3]

Classification :

Type 1 = A Massive fusion of cervical spine

Type 2 =Fusion of one or two cervical vertebrae

Type 3 =Type 1 or 2 Klippel-Feil syndrome with thoracic and lumbar spine ananomlies.

Causes of Klippel-Feil Syndrome

- In most individuals with Klippel-Feil syndrome the cause is unknown.

- Mutation in the GDF6 and GDF3 gene or MEOX-1 gene can cause KlipF syndrome.
- Fetal alcohol syndrome.
- Goldenhar syndrome.

**Case Report:**

A girl child, 10 year old attended physiotherapy outpatient department with painless congenital neck deformity i.e. Torticollis of right side. There was no antenatal history of pyrexia, increased blood pressure, significant medical and any other event during pregnancy. Natal: Mother delivered premature baby girl of 1.5kg at 32 weeks of gestation by normal delivery. Post natal History: after three days of birth it was observed that there was yellowish discoloration of eyes so she was kept in NICU under observation and was discharged after 3-4 days. After few days her parents observed that her wrist was in completely extended position and child was unable to flex her wrist, so they consulted to her paediatrician where X-ray and investigation done were and the baby was diagnosed as Klippel-Feil syndrome. She was having normal overall Gross motor and mental development. She started neck holding, sitting, crawling & standing as per the development schedule. She started walking in ninth month. She goes to normal school with satisfactory progress (only has problems in writing and bimanual activities) as per her class teacher. She is shy,intelligent and understand all exercises herself. She is dependent in all her Activities of daily living such as grooming and eating etc. On examination, she was having normal built, short height; head tilted to right side, low hair line, short neck, and the distance between from tip of ear pinna to Trapezius upper border is grossly reduced as compared to left side. Mild cervical scoliosis present on right side neck. Range of motion of cervical spine: She can perform complete cervical flexion, partly extension, rotation and bilateral lateral flexion showed reduced range of motion. There is no tenderness in cervical spine. Right scapula is elevated. Bilateral shoulder had limitation of range of movement especially abduction and flexion after 90 degree. Cardio-respiratory system and urinary system examination revealed no abnormalities. Radiological examination of cervical spine revealed blocked vertebrae C1, C2, C3 and radiological examination of chest revealed high scapula on right side as compared with left shoulder. The girl was also examined by a plastic surgeon and was recommended for conservative management to begin with. As our case was not having any neurological or any other systemic involvement she was advised regular physiotherapy treatment of neck and shoulder exercises 5 times a week, Corrective cervical collar was prescribed.Cock up splint with thumb in opposition was prescribed to avoid wrist drop. Milwaukee brace for scoliosis correction and exercises for postural correction. Also the technique of manual traction of cervical spine was explained & demonstrated to her father. Prognosis was explained to parents and the patient in detail.

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**Postural evaluation**

**Anterior View:**

Ear lobes not at level (R<L)

Shoulders not at level (right shoulder elevated)

Elbow not at level (carrying angle R < L)

Both wrist extended and ulnar deviation seen.

ASIS not at level (right ASIS higher than left)

**Posterior View:**

Cervical spine laterally flexed on right side

Bilateral winging of scapula

Elbows not at level

Thoraco lumbar Scoliosis

Right PSIS raised

**Discussion**

Klippel-Feil syndrome is abnormalities in Cervical vertebra segmentation i.e. they involve fusion of 2 segments or the entire cervical spine. Klippel-Feil syndrome is caused due to failure of the normal segmentation and mesodermal somites fusion, during the third and seventh week of gestation [4, 5]. Various abnormality are present in Klippel-Feil syndrome. A torticollis seen along with the disease overlaps the shortness of neck. In torticollis, head is bent to one side while chin points to opposite side. Torticollis is either seen since birth (congenital) or afterbirth (acquired) [6]. In congenital Torticollis usually there is a history of difficult/prolonged labour followed by sternocledomastoid tumour. The acquired Torticollis can be caused due to various injury of cervical spine, inflammatory- due to inflamed/enlarged cervical lymph nodes, spasmodic i.e spasm of sternocledomastoid muscle along with posterior cervical muscle, compensatory i.e. secondary from scoliosis or due to a defect in vision, tuberculosis of cervical spine and lastly it may be caused due to burn contracture [7]. In cases of congenital muscular Torticollis, the contracture of sternocledomastoid muscle is painless and

newborn child tilts head toward and rotate his chin away because of the contracted sternocleidomastoid muscle. Usually the lesion is identified in 2-3 month after birth [3]. Stretching of affected muscle, lateral rotation and side bending is affective in 90% cases provided stretching is initiated within one year of age and if not surgical intervention is desirable to prevent facial deformity [3]. Klippel-Feil syndrome is often having other congenital lesions like congenital scoliosis or kyphosis, renal disease, synkinesis , Sprengal deformity and torticollis, loss of hearing, facial asymmetry and flattening of neck, congenital heart diseases, brainstem lesions, congenital cervical stenosis. Adrenal aplasia, ptosis, facial nerve palsy, syndactylia, diffuse hypoplasia of upper limb may also be seen. Intervertebral disc degeneration has also been reported in almost all cases [8-9]. Beside orthopaedic clinical evaluations, patients with Klippel-Feil syndrome should be assessed by anteroposterior and lateral cervical flexion/extension and thoracolumbar radiographies, abdominal ultrasonography, and were subjected to systemic examination to detect any urological, cardiological, oto-rhino-laryngeal, neurological and psychiatric finding. Computed tomography and magnetic resonance imaging can be done where necessary [10]. Mechanical symptoms because of degenerative diseases respond to traction, cervical collar and analgesics. Surgical correction of sprengel deformity is usually opted to improve cosmesis. Minimally involved cases of Klippel-Fiel syndrome have good prognosis and live normal life with no significant restrictions or symptoms [11]. In patients having hyper mobility in vertebra and not having fusion; prophylactic fusion should not be performed unless patient is having any neurological problems because of disc compression that may appear later. Severely involved patients have good prognosis if cardiopulmonary, genitourinary and auditory problems if any are treated early [12]. In anomalies of occipito-cervical passage, high morbidity and mortality rates have been frightening, as cervical cord and brain stem are in proximity to each other[13]. In the present case we have found congenital Scoliosis, Sprengel deformity and there were no evidence of renal disease, congenital heart disease & neurological problems. The present case has classical triad low posterior hairline, short neck& limited cervical range of motion.

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

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