Klippel Feil Syndrome Type III with Associated Rare Congenital Anomalies: A Rare Case Report

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Abstract

Klippel Feil Syndrome (KFS) is a congenital anomaly having fusion of two or more cervical vertebral bodies characterized by presence of classic triad of low posterior hairline, short neck and restricted range of motion. We have reported a rare case of Type III Klippel Feil Syndrome having classical clinical triad with sprengel deformity and atlantooccipital assimilation. The baby was born at Sanjay Gandhi Memorial Hospital, Mangolpuri, New Delhi, INDIA. The detailed discussion is done in case report. Prognosis is based on radiological classification by Samartzi’s et al. which classifies KFS under 3 types with type II as commonest variety. Associated anomalies includes scoliosis or kyphosis, renal disease, sprengel deformity, loss of hearing, synkinesis or mirror movements, congenital heart defects, craniofacial malformations & skeletal abnormalities of ear, nose, mouth and larynx. Proper management requires multidisciplinary approach including neurologist, orthopedic surgeon, pediatrician, nurse practioner, physical therapist, and neurosurgeon.

Keywords: Klippel Feil syndrome, sprengel deformity, scoliosis, atlanto - occipital assimilation, congenital anomalies
Introduction

Klippel Feil Syndrome (KFS) is defined as congenital anomaly with fusion of two or more cervical vertebral bodies characterized by presence of classic triad which includes low posterior hairline, short neck and restricted range of motion(1,2). Typical classic triad however is found only in about 50% of cases(3,4). It was initially reported by Maurice Klippel and Andre Feil in 1912(3). Incidence as reported in different studies is 1:40,000 - 1:42000 per live birth with slight female predominance(1,2,5,6).

It results due to faulty segmentation along the embryo’s developing axis around 3-8 weeks of gestation(1). Pathogenesis of KFS involves various genetic mutations in genes like GDF 6, GDF 3, MEOX 1 and RIPPLY 2 (4). Associated anomalies includes scoliosis or kyphosis (60%), renal disease (35%), sprengel deformity (30%), loss of hearing (30%), synkinesis or mirror movements (20%), facial asymmetry with flattening of neck (20%), congenital heart defects (4-14%), craniofacial malformations & skeletal abnormalities of ear, nose, mouth and larynx(5,7). Most commonly associated congenital heart defect is Ventricular Septal Defect (VSD) (7).

Prognosis is based on radiological classification by Samartzi’s et al. which classifies KFS under 3 types; Type I - Single congenitally fused cervical segment; Type II – Multiple, non contiguous congenitally fused cervical segments; Type III - Multiple, contiguous congenitally fused cervical segments with thoracic and lumbar spine anomalies (1,2,4,6).

Work up includes detailed skeletal evaluation with the help of radiographs of cervical region including lateral & anteroposterior views, CT scan for evaluation of bony abnormalities, MRI brain and spine for detailed insight regarding spinal cord, disc space, nerve rootlets, ligaments, soft tissue and other associated anomalies (3,6).

Management of patient with KFS involves multidisciplinary approach including neurologist, orthopedic surgeon, pediatrician, nurse practioner, physical therapist, and neurosurgeon(3). Management is mostly conservative & symptomatic. Patient with 1 or 2 fusions below C3 are monitored and treated conservatively. They can play contact sports like hockey, rugby. Patient with fusions above C3, especially to the occiput are mostly symptomatic and should avoid contact sports. Surgical decision is based on both spinal deformities as well as instability(3,4). Surgical correction, if required, should be done at a young age of 3-8 years(2).

Case Report

We report a case of newborn male baby who was born full term to a G2 P1 L1 mother with birth weight of 1.815 kg (low birth weight) by LSCS in view of Meconium stained liquor with Fetal distress at Sanjay Gandhi Memorial Hospital, Mangolpuri, New Delhi, INDIA. Baby was born with gross congenital anomalies in the form of short neck, coarse facial features, elevated right shoulder and had respiratory distress immediately after birth. The antenatal period was uneventful as per mother. Only iron, folic acid and calcium supplementation were taken during antenatal period.

Baby was shifted to NICU where he was kept on CPAP support and oxygen by hood as per the requirement. Treatment was given in the form of IV fluids, IV antibiotics and other supportive care. On
examination, baby had multiple congenital anomalies including microcephaly (HC- 30cms), short neck, low posterior hairline, restricted neck mobility, right side elevated scapula (sprengel deformity), scoliosis, low set ears and right foot congenital talipes equinovarus (CTEV). He was irritable probably because of associated radiculopathy of cervical vertebrae. Respiratory distress was present in the form of tachypnea with mild Intercostal and subcostal retractions. Neonatal reflexes were sluggish and cardiovascular examination revealed presence of pansystolic murmur grade IV which was well appreciated on day 3 of life in 3rd & 4th ICS probably due to congenital heart disease (Figure 1-3).

**Figure 1:** Short Neck with Low set Ears

![Short Neck with Low set Ears](image1)

**Figure 2:** Sprengel Deformity Right Scapular Bone

![Sprengel Deformity Right Scapular Bone](image2)
Skeletal evaluation in the form of cervical x-rays and infantogram was done which showed contiguous fusion of cervical vertebrae with scoliosis of dorsal spine. MRI spine was also done which showed fusion of cervical and upper dorsal vertebrae upto D3 vertebrae with atlanto-occipital assimilation. The posterior elements of vertebral columns (cervical & dorsal) formed a bone block. Cervico-medullary junction showed dorsal flattening with prominent posterior subarachnoid space. Scoliosis was present in upper dorsal spine with convexity towards right side associated with prominent posterior & right lateral epidural space (Figure 4- 8).

**Figure 3:** Right Foot CTEV

**Figure 4:** X- Ray Cervical Spine – showing contiguous cervical fusion with cardiomegaly
**Figure 5:** Infantogram showing scoliosis

![Infantogram showing scoliosis](image)

**Figure 6:** X-Ray LS Spine- confirming Scoliosis

![X-Ray LS Spine- confirming Scoliosis](image)
**Figure 7:** Sagittal and axial T2W images show fusion of cervical and upper dorsal vertebrae upto D3 vertebrae with atlanto-occipital assimilation. There is dorsal flattening of cervico-medullary junction with prominent posterior subarachnoid space.
MRI brain revealed restricted diffusion in rostrum and splenium of corpus callosum and right parasylvian region on DW images. Subdural haemorrhage was seen along bilateral tentorial leaflets, posterior falx and left posterior parietal convexity shown as blooming on GRE images. These findings were suggestive of hypoxic ischemic encephalopathy.

Baby was gradually weaned off from CPAP support and put on oxygen support by hood. Thereafter, parents took LAMA on day 17 of life of the baby. Telephonic conversation was initially being done but gradually follow up was lost as parents were not willing for any further investigation or treatment. Parents were also advised for genetic evaluation for future pregnancy as well as to see the inheritance for the baby whether it was autosomal recessive or sporadic case.

Regarding the previous pregnancy, the ante natal period was uneventful. Previous sibling was born normal without any congenital anomaly and at present, is alive and healthy.

**Discussion**

KFS is a congenital disorder of cervical vertebrae that may show a familial or sporadic pattern(1). Cervical vertebral segmentation defects referred to as Klippel Feil Anomaly involving fusion of two or more cervical spine segments which occurs due to failure of normal segmentation & fusion processes of mesodermal somites occurring during 3rd-8th week of embryonic life(7).

Incidence reported in multiple studies ranges from 1:40,000- 1:42,000 live births with slight female preponderance(6).

Pathogenesis of KFS involves various dominant & recessive genetic mutations including GDF 6, GDF3, MEOX 1 and RIPPLY 2. GDF6 and GDF3 are responsible for transcription, regulation and signaling pathways involved in somite development during embryogenesis(2)(3). MEOX 1 lead to formation of MOX 1 protein which regulates the process of separation of vertebrae from one another during early gestation period. KFS associated with GDF6 and GDF3 inherited as autosomal dominant while that with MEOX 1 gene is inherited as autosomal recessive. Few KFS presents as sporadic case(3).

Classical clinical triad includes low posterior hairline, short neck and restriction of movements. It is seen only in approximately 50% of cases with most common clinical sign as restriction of movements of neck which is found in 50 – 76 % of cases(1). This classic triad was present in our case.

The clinical presentation in KFS could be diverse, ranging from asymptomatic to serious signs such as sensory abnormality, asymmetric reflexes, altered cerebellar functions, long tract signs and pain(6). Associated anomalies as enumerated earlier like congenital scoliosis or kyphosis (60%), renal disease (35%), sprengeal deformity (30%), synkinesis or mirror movements (20%), torticollis, hearing loss (30%), congenital heart defects (4-14%) could be seen(7). Atlanto occipital assimilation aka occipitalization of atlas is found to be reported in 30% of KFS case with overall incidence of 0.08 – 3% in general population(8). In our case, scoliosis, sprengeal deformity, atlanto occipital assimilation and congenital heart defect were present.
Samartzi’s et al. proposed a classification based on radiographic findings. Of these 3 types, type II is the most common\(^\text{(1)}\). Our case belongs to type III as per MRI spine report. Type I fusion is mostly associated with axial neck symptoms while myelopathic & radicular symptoms are associated with type II & type III\(^\text{(6)}\). In our case, baby was irritable probably because of associated painful radiculopathy.

In studies conducted by Samartzi’s et al. in 2006 and 2011, it was reported that most commonly fused cervical segments are C2-C3 (71 %) followed by C5-C6 (67.7 %), C6-C7 (67.5 %) and C3-C4 (29\%).\(^\text{(6)}\) In our case, fusion from atlanto-occipital joint till D3 was found.

The main steps in proper management of KFS include a regular radiographic surveillance for hypermobile cervical vertebrae, early identification of high-risk skeletal abnormalities and physical therapy. Surgical treatment is indicated to relieve cranio-cervical instability and spinal cord constriction, and to correct scoliosis. Emphasis should be made on lifestyle modifications as per fusion pattern. Majority of children with KFS carry a good prognosis provided the diagnosis is made early and measures are taken to prevent cervical spine damage\(^\text{(9)}\).

**Conclusion**

KFS is a rare heterogenous condition with varying clinical manifestations. As per literature, it has been classified into 3 types with Type II as the most common variety. Among the associated anomalies, sprengel deformity is a rare anomaly mostly associated with Type II. Atlanto occipital assimilation is also a rare associated congenital anomaly. We have reported a rare case of Type III Klippel Feil Syndrome having classical clinical triad associated with sprengel deformity and atlantooccipital assimilation. All cases should be thoroughly investigated with the help of various appropriate screening tests like radiographs, CT scan, MRI brain and spine to have detailed evaluation of associated anomalies. This is important for close follow up and decision on appropriate therapy. Parental counseling should be done in all cases. This case report and other similar reports demand for extensive alertness in identifying and diagnosis of such cases which would help in further management. Newer treatment modalities to be researched to reduce the morbidity and mortality associated.

**References**


