

## Case Report

# Klippel-Feil syndrome: a case report

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### ABSTRACT

Klippel-Feil Syndrome (KFS) is defined as congenital fusion of two or more cervical vertebrae. The most common signs are short neck, low hairline at the back of head and restricted mobility of neck. We report a case of a neonate who presented with complaint of respiratory difficulty and later diagnosed as case of Klippel-Feil syndrome.

**Keywords:** Cervical vertebra, Congenital, Klippel-Feil syndrome, Posterior hairline

### INTRODUCTION

Klippel-Feil Syndrome (KFS) is defined as congenital fusion of two or more cervical vertebrae, results from faulty segmentation along the embryo's developing axis during 3-8 weeks of gestation with incidence of 1:40000 to 1:42000 births.<sup>1</sup>

KFS, or synostosis of the cervical spine, occurs as a result of failure in normal segmentation of cervical mesodermal somites during embryonic development. This failure occurs at second-eight weeks of gestation and its cause is unknown.<sup>2</sup> The most common signs are short neck, low posterior hairline and restricted mobility of upper spine. Associated abnormalities may include scoliosis or kyphosis, Sprengel's deformity, hemivertebrae, platybasia, basilar impression, spina bifida, anomalies of the kidneys and the ribs, cleft palate, respiratory problems, deafness or hearing impairment, and heart malformations.<sup>2</sup>

Most patients who have this syndrome are first seen with restricted motion of the neck, torticollis, webbing of the neck, or Sprengel deformity.<sup>3</sup>

### CASE REPORT

We report a case of just born new-born female gestational age  $34 \pm 2$  weeks Low Birth Weight (LBW) 1.625 kg with AGA (appropriate for gestational age) born to 25 year old mother. The patient presented to department of paediatrics, S.S. medical college, Rewa (M.P.) with the symptoms of respiratory distress immediately after birth. Mother was booked case with no antenatal complications. She had four antenatal visits with history of full immunisation with tetanus toxoid with normal vaginal delivery.

On careful examination of the new-born we observed short neck, low posterior hairline and restricted neck mobility (Figure 1 & 2). Head circumference was 30 cm, length 40 cm, chest circumference 25 cm and new Ballard score 26. The ratio of upper segment and lower segment was 1.35:1. On respiratory system examination mild intercostal and substernal recessions were seen. On auscultation bilateral crepitations were found. On CNS examination neonatal reflexes were sluggish. On cardiovascular examination systolic murmur was appreciated after 48 hours in third and fourth intercostal spaces. On Per abdomen examination bilateral kidney

was palpable. Septic screening was normal. X-ray neck anteroposterior view revealed fusion of cervical vertebra. X-ray thoracolumbar region revealed scoliosis and hemivertebra. On ultrasonography abdomen bilateral dilated mild calyces were seen. Patient died on 6<sup>th</sup> day.

CPAP was administered to the new-born.

No family history was reported and he was third birth order.



**Figure 1: Showing short neck and low posterior hairline.**



**Figure 2: X-ray of short neck and low posterior hairline.**

## DISCUSSION

KFS is a congenital disorder with fusion of cervical vertebra which may be familial or sporadic.

Classical clinical triad of Klippel-Feil syndrome is lower posterior hair line, short neck and restriction of head & neck movements. This classical triad is seen in about 40-50% and the most common finding is restriction of movements with a ratio of 50-76%.<sup>3-6</sup> In present case the classical triad was observed. Klippel and Feil proposed a classification:

Type I: Extensive cervical and upper thoracic spinal fusion.

Type II: One or two interspace fusions, often associated with hemivertebrae and occipitioatlantal fusion.

Type III: Both cervical and lower thoracic or lumbar fusion.

Type II is commonest; C2-3 and C5-6 inter-spaces are most often fused.<sup>7</sup>

Lateral and anteroposterior radiographs of cervical spine in flexion and extension should be done to assess the location and severity of the hypermobility. Magnetic Resonance Imaging (MRI) of the cervical spine in flexion and extension may be performed to further delineate other associated anomalies. As in present case scoliosis and fusion of cervical vertebra were observed. Also hemivertebra in thoracolumbar region was seen.

Treatment for KFS is symptomatic and may include surgery to relieve cervical or craniocervical instability and constriction of the spinal cord, and to correct scoliosis. Physical therapy may also be useful.<sup>8</sup>

## CONCLUSION

All the cases of KFS should be thoroughly investigated to formulate the further line of management. The disorder should be diagnosed timely, to provide close follow-up and appropriate therapy and counselling. The parental counselling should be done in all such cases.

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