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Pediatrics

KLIPPEL FEIL SYNDROME CASE REPORT ON TWO PEDIATRIC CASES

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ABSTRACT We report two children with Klippel Feil Syndrome with varying presentations. First case presented at 08 weeks with noisy breathing and respiratory distress. She had facial asymmetry, short neck, low set posterior hairline and restricted neck motion along with torticolis. Plain X-ray neck revealed occipitalization of C-1 vertebra,hemivertebra,S-shaped deformity of cervico-thoracic spine, Kyphoscoliosis, short neck and low hair line. Second case a 07 years old female child was brought with short stature, short neck, torticolis and an abnormal posture and Sprengel shoulder.

KEYWORDS : Fusion of vertebrae, short neck, respiratory distress, low hairline, Klippel-Feil Syndrome, KFS

INTRODUCTION

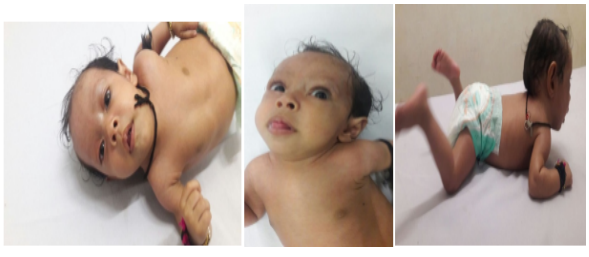
Klippel Feil Syndrome (KFS) was first described by Maurice Klippel and Andre Feil(1,2) in 1912 in a patient with congenital fusion of cervical vertebrae.KFS is a complex syndrome of osseous and visceral anomalies that include the classical clinical triad of short neck, limitation of head and neck movements and low posterior hairline(3). It is associated with several defects such as deafness conductive or neural, congenital heart defects most commonly ventricular septal defect, mental retardation, cleft palate, rib defects, Sprengel Sequence, and scoliosis. (1,2). KFS patients have a smaller face and facial asymmetry with no dental implications(4).KFS occurs in 1 out of every 42,000 to 60,000 births and 60 % of cases are in females(4).The gene locus of familial Klippel Feil syndrome is on the long arm of chromosome 8. Almost all cases of this syndrome occur sporadically. Although the prevalence of KFS is very low, it may be related to various anomalies and to fetal alcohol syndrome (6, 7). Children with KFS may become symptomatic during the rapid growth of adolescence. Bony malformations in KFS may entrap and damage the brain and spinal cord (1).The other name of this syndrome is congenital Brevicollis syndrome.(2).

CASE NOTE

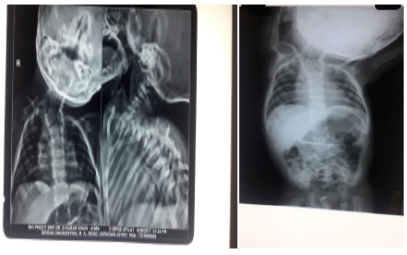
CASE-1.

This female 08 weeks old presented with recurrent noisy breathing and respiratory distress . The baby was born out of a non consanguineous marriage as a term vaginal delivery. The birth weight was 2300 grams. Mother was a 23 year old, non alcoholic Primigravida. Perinatal history was uneventful .Her detailed examination revealed a weight of 4kgs, head circumference of 38.6 cms, length of 53 cms. Her vital parameters revealed marked tachypnea with a respiratory rate of 68/min , She was also noticed to have restricted neck movements, a short neck with a low hairline, scoliosis, torticolis and facial asymmetry.

Figs.1 of case 1-short neck ,torticolis,scoliosis,low hair line



Figs 2 of case 1-Block vertebra at C-2,C-3level,poorly formedC-7 with rt sided hemivertebra anomaly, acute lordosis C-7TO D-1,spina bifida at upper thoracic D-1 to D-5 level, leftward tilted neck, Rt shoulder having Sprengel deformity

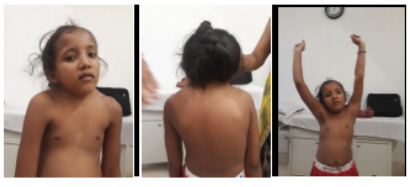


X-ray neck revealed block vertebra at C-2,C-3level,poorly formedC-7 with rt sided hemivertebra anomaly, acute lordosis C-7TO D-1,spina bifida at upper thoracic D-1 to D-5 level, leftward tilted neck, Rt shoulder having Sprengel deformity. Subsequent follow up revealed ventricular septal defect on 2D - echo. The ultrasonography of abdomen revealed non visualization of left kidney.

CASE -2.

06 yrs old female child, product of a non consanguineous marriage, a full term normal delivery presented with short stature. On examination she had facial asymmetry, torticolis, a short neck with webbing and limitation of head and neck movements along with a low posterior hairline. She also had Kyphoscoliosis and partial conductive hearing loss. Her height for age was less than 5 th centile, head circumference was 52 cms. Ultrasonography of abdomen and 2-D echo was normal. X-ray neck revealed occipitalization of C-1 vertebra, hemivertebra, S-shaped deformity of cervico-thoracic spine and Kyphoscoliosis.

Figs.2 of Case -2 - Short neck, Sprengel shoulder, Kyphoscoliosis, facial asymmetry, low hairline



DISCUSSION

Klippel Feil Anomaly is characterized by fused cervical spine or blocked vertebrae, restricted mobility of neck and cervical spine. In some patients there may be low hairline and short webbed neck also ,as is seen in the present case . Associated anomalies can be present as spina bifida, cleft palate, late dentition carries, oligo and hypodontia, respiratory distress, cardiac anomaly, short stature, Duane syndrome and Sprengel shoulder, and developmental abnormalities in sex organs, brain and spinal cord. (14,15,16). Prevalence of KFS is not known. Estimated incidence is 1 in 42000 to 60000 newborns worldwide. Females are more affected than males. The commonly fused cervical vertebrae are C2-C3(71%),C5-C6(67% and C3-C4(29.0%).There may be cervical stenosis in 25% at both fused and

non fused level. Side to side movement, flexion and extension and rotational movements are usually restricted. Torticolis and facial asymmetry occurs in 25-50% of cases. About 30 to 60% of patient with Klippel Feil syndrome may have genitourinary problem and unilateral renal agenesis is most common.

Exact cause remains obscure ,however various hypotheses put forward are vascular disruptions, global fetal insult, primary neural tube anomaly, genetic predisposition, facet joint segmentation failure ,maternal alcoholism due to fetal alcohol syndrome. Mutations in chromosome GDF-6 & GDF-3 have been described as causative factor .These genes regulate the growth and maturation of bone and cartilage. Mutations could be inherited in one of the following ways, autosomal dominant or autosomal recessive, or a rare autosomal dominant form(mapped on locus 8q (22.2) KFS with laryngeal malformation also called segmentation syndrome has been reported(6).Treatment in KFS remains supportive only.

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