KLIPPEL FEIL SYNDROME WITH PULMONARY CYST

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Introduction

Klippel Feil Syndrome (KFS) is a rare malformation due to congenital fusion of two or more cervical vertebrae (synostosis) resulting in brevicollis [1]. A triad of brevicollis, limited neck movements and a low hairline results in this syndrome. It is associated with various systemic anomalies. However, lung parenchymal lesion has not been described with this syndrome. A case of Klippel Feil syndrome with associated unilateral pulmonary cyst is being reported for its rarity.

Case Report

6 year old female child, 2nd in birth order of 3 siblings of non-consanguineous parents, was born by term vaginal delivery

Fig. 1 : Photograph of child showing classical features of Klippel-Feil syndrome (short neck, low hair line, Sprengel deformity)

with uneventful perinatal period. She achieved all motor and mental milestones normally. She was noticed to have deformity of neck, back and restricted neck movements since 1½ years of age. Presently she was admitted with an episode of generalized tonic clonic seizure. There was no focal neurodeficit or evidence of meningitis. Examination revealed weight - 13.5 Kg, height - 104 cms and normal vital parameters. She was having short webbed neck with restricted neck movements and low hairline. There was Sprengel deformity of left scapula with restricted movements of neck, scoliosis of dorsal spine with convexity to left. Systemic examination was within normal limit. X-ray cervico-dorsal spine showed occipitalization of Atlas with fusion of posterior elements of C3-4 vertebrae. There was marked cervico-dorsal scoliosis and kyphoscoliosis with convexity to left. Multiple cervico-dorsal hemi-vertebrae and fused vertebrae were seen. Left scapula was small, triangular and elevated. Multiple rib anomalies were present. X-ray chest revealed 2 cms translucent rounded shadow in RMZ. CECT neck and chest showed a cystic lesion in the apical segment of right lower lobe besides brevicollis. MRI of cervico dorsal spine showed fusion of bodies of C6-D1 vertebrae. The intervening disc spaces were rudimentary. Spinal cord was normal in size, contour, morphology and signal intensity. Dural ectasia was noted at C4-D2 level. The findings were consistent with KFS. CT scan cranium did not show any focal lesion. EEG showed generalized cerebral dysfunction. USG abdomen and echo study were within normal limit.

Discussion

KFS was first described by Klippel and Feil in 1912

Fig. 2 : CT scan of chest showing congenital pulmonary cyst in right lower lobe

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Fig. 3 : MRI scan of cervico-dorsal spine showing fusion of cervical spine

based on fusion of cervical vertebrae [1]. The incidence is about 1:42000 births with female preponderance of about 65% cases [2]. Short neck, low hairline, restriction of neck movements and fusion of at least 2 cervical segments are the cardinal features of this syndrome. It has autosomal mode of inheritance with variable expression, however, sporadic cases are also seen. The main underlying abnormality is failure of normal segmentation of cervical somites during 3rd and 8th weeks of gestation. The exact initiating factor for the same is not known. Three forms of syndromes have been identified based on type and extent of congenital malformation of vertebral column, features of which are shown in Table-1.

TABLE I

Types of Klippel - Feil Syndrome

Type	Features
I	Cervical and upper thoracic vertebrae are fused in one bony block.
II	Complete segmentation fails to occur at one or two cervical vertebrae.
III	Additional segmentation occurs in lower thoracic / lumbar vertebrae.

The anomaly may be a part of serious problem in early neural tube development such as anencephaly, cervical meningocele, syringomyelia or syringobulbia. CNS anomalies comprise paraplegia, hemiplegia, cranial nerve palsies, macrocephaly, hydrocephalus

and mental retardation. Other associated anomalies which occur with this syndrome are deafness (30%) [3], congenital heart defects, commonest being ventricular septal defect, rarely patent ductus arteriosus and coarctation of aorta [4]. The musculo-skeletal anomalies include thoracic scoliosis, spina bifida occulta, Sprengel deformity, abnormalities of ribs, webbing of neck, hypoplastic thumb, supernumery digits and unilateral hypoplasia of Pectoralis Major muscle. The latter is called as Poland anomaly if it is associated with syndactyly, hypoplasia of nipple and areola and facial asymmetry. Genitourinary anomalies including unilateral renal agenesis, horseshoe kidney, absent vagina and ovarian agenesis may be present. Dental abnormalities, cleft lip, cleft palate, abnormalities of gastro-intestinal tract, lung and skin may also be associated.

Differential diagnosis include bilateral Sprengel deformity, occipitalization of C1 vertebra, and acquired post inflammatory (Diskitis, Rheumatoid arthritis) or post-traumatic fusion. Treatment consists of correcting the associated anomalies wherever it is possible and appropriate genetic counselling.

In our case, the features of classical triad of KFS were present along with Sprengel deformity, scoliosis and seizures. An interesting hitherto not described finding was that of presence of unilateral pulmonary cyst in right middle zone. Perfusion-ventilation imbalance, decrease ventilatory turnover and expiratory flow from the alveolar space resulting from impaired respiratory mechanism leading to respiratory failure and pulmonary hypertension on account of kyphoscoliosis has been reported in literature [5]. In our case the presence of pulmonary cyst is likely to aggravate the process of respiratory failure and development of pulmonary hypertension.

References

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