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Case Report

Pediatrics for better child health

A Rare Case of Klippel Feil Syndrome Associated with Mirror Movements

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Abstract: Klippel Feil Syndrome (KFS) is a multisystem disorder with the typical clinical trial of short neck, restriction of head and neck movements and low posterior hairline and associated with anomalies of the urogenital, musculoskeletal, neurological and cardiovascular systems. Here is a case report of a young boy with history of abnormal head and upper limbs movements along with repeated bowing of head since the age of 6 months. He also had frequent falls with repeated minor trauma to the face. Antenatal and peripartum period was uneventful and child had no neonatal complications with normal development. On examination the child had abnormal head position with bowing, short neck and low hair line. Musculoskeletal system examination revealed decreased range of passive movements at cervical spine extension and lateral flexion with no focal neurological deficits. Further examination revealed Mirror movements (synkinesis) in the upper limbs. Mirror movements are the involuntary movements of one body part that mirror intentional movements on the opposite side. Other Systemic examinations were normal. CT Cervical spine showed fusion of the upper three cervical vertebrae and the child was diagnosed as a case of klippel feil syndrome type 2 presenting with mirror movements. This rare case of klippel feil syndrome is being presented with the aim that such cases should be detected and treated at an early stage to reduce the cosmetic & social stigma to the patient and parents. This case report also gives a brief overview of associated features of KFS along with its management.

Keywords: Klippel Feil Syndrome, mirror movements, cervical fusion, spine, magnetic resonance imaging, congenital

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1. INTRODUCTION

Klippel Feil syndrome (KFS) being an uncommon disease, estimated to occur 1 in 42000 newborns worldwide. It is usually sporadic but seldom may be inherited. Initially Maurice Klippel and Andre Feil from France reported this rarity independently.[1,2] It is mainly due to the normal cervical mesodermal somites segmentation failure during third to eighth week of embryonic development resulting in synostosis of cervical vertebrae. The exact etiology is not known however, certain genes such as MEOX1, GDF6, GDF3 (growth differentiation factor), and RIPPLY2 responsible for transcription regulation and somite development signaling pathway are the causes of pathogenesis.[3,4] KFS is a fusion of at least two cervical vertebrae congenitally and is characterized by presence of triad of short neck, restricted neck movements, and low posterior hair line.[5,6] Classification of KFS to three types was introduced in 1919 by Maurice Klippel and Andre Feil in to 3 types based on the extent and location of fusion of vertebrae as well as associated vertebral abnormalities:[7]

- Type I: many cervical and upper thoracic vertebral fusion
- Type II: two or three vertebral fusion with associated hemivertebrae, fusion of occipito-atlantal or other cervical spine abnormalities
- Type III: fusion of cervical with lower thoracic or lumbar vertebral segmental fusion.

The recent classification by Samartzis et al 2006 suggested three types [8]

- Type I: patients present as single level fusion
- Type II: patients depict a picture of numerous, noncontiguous fused segments
- Type III: patients reveal multiple, contiguous fused segments

Complete or focal hypoplasia of upper limbs may also be seen. Patients with KFS usually present in early childhood, but may also present in the 2nd decade of life.[9] Clinical presentations may be varied due to the different associated anomalies. The challenge to the clinician is to recognize the associated anomalies and perform appropriate diagnostic workup and symptomatic treatment.

2. CASE REPORT

2.1. Medical history

A 13-year-old adolescent boy presented with history of abnormal movements of head and upper limbs since the age of 6 months. The movements noticed were repeated bowing of head. Since the age of 2 years' mother had also noticed that involuntary movements of upper limb were mimicked by the movements on the other side. He had frequent falls with repeated minor trauma to the face. Antenatal and peripartum period was uneventful except gestational diabetes mellitus. Child had no neonatal complications. Developmentally normal child and average scholastic performance. There was no history of trauma, feeding difficulty, fever, stiffness of limbs, seizures, failure to gain weight, pain or weakness of limbs, bowel or bladder dysfunction, no gait abnormalities.

2.2. Observation\ Physical examination

On examination the boy was well built, with the height of 139 cm (normal for his age) and weight of 32 kg with BMI of 15.5 kg/m^2, abnormal head position with bowing, short neck 6% (neck length % of linear height measured by two-point discriminator with head in neutral position), low hairline (below C4 spine) (Figure -1), normocephaly, no scoliosis. His vital parameters were normal for age. Musculoskeletal system examination revealed decreased range of passive movements at cervical spine extension (30 degree) and lateral flexion (30 degree). No focal neurological deficits were noticed. Mirror movements (synkinesis) were noticed in the upper limbs. Systemic examinations were normal.

2.3. Mirror movements

Since the age of 6 months, the child had repeated bowing of head. Since the age of 2 years' mother also noticed that whenever one hand had movements, the other hand copied. His mirror movements [synkinesia] steadily improved as he grew older. Unfortunately, no neurologic evaluation was performed during the years when his mirror movements were most severe.

2.4. Test\Investigations

A routine blood examination showed mild anaemia (Hb-11g/dl) with normal cell counts and a normal erythrocyte sedimentation rate. Cervical spine radiograph (Figure-2) showed fusion of the upper three cervical vertebrae in line with KFS type 2. Ultrasonography of the abdomen revealed no renal anomalies (Figure-3). Echo were normal (Figure-4). CT Cervical spine showed fusion of the upper three cervical vertebrae (Figure-5). Other investigations done to rule out associated anomalies were normal. Since he had only mild symptoms, he was counselled about the KFS.

2.5. Diagnosis

A patient with a routine blood examination showing mild anemia (Hb-11g/dl) and normal cell counts was diagnosed with Klippel-Feil syndrome which was also support by Cervical spine radiographs and CT showed fusion of the upper three cervical vertebrae. Abdominal ultrasonography and echocardiography revealed no renal or cardiac anomalies.

2.6. Treatment and follow up

Since the patient is asymptomatic except for a movement disorder and also the symptoms were not hampering his daily activities, currently surgical interventions were kept on hold. The need for lifestyle interventions were educated and advised for regular follow up. Parents were informed about the foreseeing possible risks of neurological weakness and the need for early interventions in case of decompression and fusion of vertebrae to avert catastrophes.

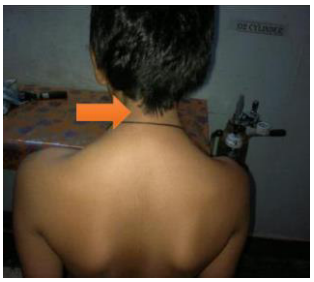

Fig 1 - Neck shortness with posterior low hairline

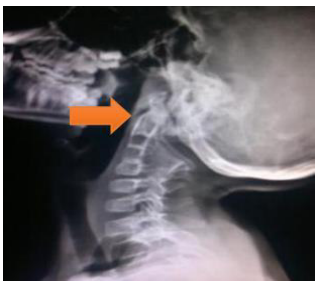

Fig 2: X ray Cervical spine showed fusion of upper three cervical vertebrae

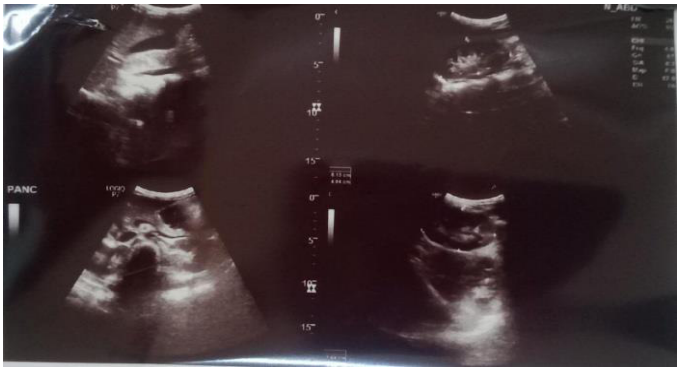

Fig 3: Ultrasound image of abdomen were normal

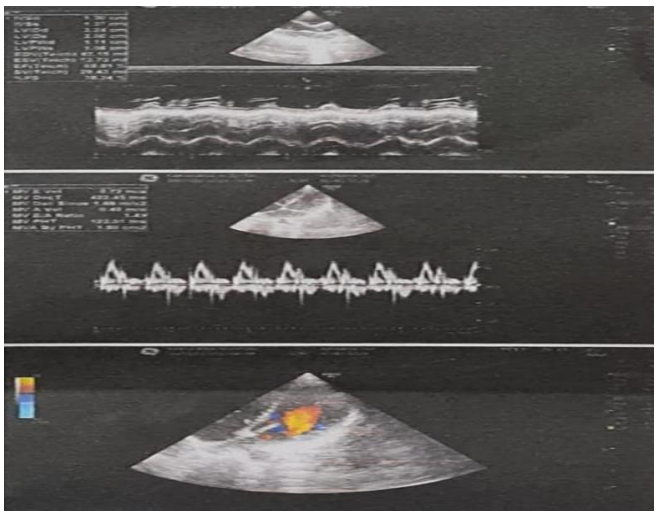

Fig 4: Echocardiogram were normal

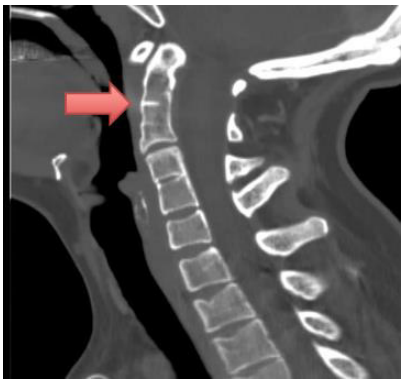

Fig 5: CT Cervical spine revealed fusion of upper three cervical vertebrae

Table 1: Anomalies seen in Klippel Feil syndrome

Anomalies	Incidence
Kyphosis or scoliosis	60%
Sprengel scapular deformity	30%
Renal anomalies	35%
Hard of hearing	30%
Nuchal Flattening	20%
Mirror movements	20%
Congenital cardiac anomalies	4.2%

3. DISCUSSION

Klippel–Feil syndrome is a complex, congenital condition with heterogeneity in both phenotypic expressions and etiological background. It is associated with deformities like Sprengel's deformity, facial asymmetry, hearing impairment, synkinesia, scoliosis, ocular abnormalities, cleft palate, congenital heart diseases, lung defects, and genitourinary problems such as abnormal kidneys or reproductive organs. [10,11] The exact cause of KFS has yet to be determined, although some have suggested a genetic aetiology, while others attribute environmental factors such as alcohol consumption. The importance of recognizing KFS is mainly due to its strong association of anomaly with significant multisystem abnormalities.[12] The usual associations seen in KFS are listed in Table 1. Other rare associations seen include: brainstem malformations, congenital stenosis of cervical vertebrae, congenital aplasia of adrenal gland, ptosis, paralysis of lateral rectus muscle, seventh cranial nerve paralysis, fixed or webbed fingers.[13,14]Mirror movements (MM) is defined as the simultaneous contralateral, involuntary and identical movement that accompany voluntary movements. Definition for MM as involuntary, synkinetic mirror reversals of an intended movement of opposite side was coined by Cohen *et al* in 1991[15] and the term mirror movements was first described by Erlenmeyer in 1879.[16] Mirror movements are seen at the age of 4 years and mostly disappear before 10 years of age coincident with myelination of the corpus callosum. MM has been associated with several neurologic conditions like cerebral palsy, Parkinson's disease, certain symptomatic epilepsies cervico-medullary junction anomaly, cerebrovascular disease, high cervical spinal cord malformation and psychiatric conditions such as schizophrenia and obsessive–compulsive disorder.[17] Recent molecular studies have suggested that mutations of the MEOX1 gene may cause a recessive subtype of the syndrome.[18] Mutations in the GDF6 gene, expressed in vertebrae and within the intervertebral disc, GDF3 genes, and PAX1 have all been implicated in the pathogenesis of the disease.[19,20] More recently, a homozygous frame shift mutation in a protein-coding gene (RIPPLY2) has been found to be responsible for a new type of autosomal recessive KFS.[21] The identification of mutations in these genes may lead to new advancement of treatment and possible explanation of associated anomalies. Depending on the severity, Klippel-Feil syndrome is seen on ultrasound in the first trimester of pregnancy. Diagnostic radiographic investigations like x-ray can show fused vertebrae and other abnormalities of entire spine, CT scan provides better imaging when compared to x-ray and may demonstrate canal stenosis and in patients with neurologic deficits MRI is recommended.[22,23] It is clearly in demonstrating stenosis of canal and compression of cord. When this syndrome is identified in childhood, the exact age of onset of spine-related neurologic symptoms is between 10 to 11 years in KFS patients.[24] However, patients with milder forms of Klippel-Feil Syndrome not identified in childhood can present with neurologic symptoms in their 40 years of age.[25] For this reason, the patient was encouraged to continue routine follow-up to evaluate for future development of neurological deficit. Conservative medical management like cervical collars, braces and traction is helpful in people who don't need surgery. In case of cervical or craniocervical instability and spinal cord constriction, and to correct scoliosis surgery is recommended.[26,27] Rehabilitation in a form of Physiotherapy may also be useful. The prognosis of Klippel-Feil Syndrome is good with early treatment.[28] Activities that can injure the neck should be avoided.

4. CONCLUSION

Early detection and orthopedic and neurological intervention in a form of rehabilitation will be the mainstay approach in the confirmed cases of KFS. Familial counselling regarding KFS should be in view of detailing about the regular follow up and the potentiality of sustaining a neurological deficit following a minor brain and spine injury as a preventive approach of disease worsening in the near future. Health professionals treating the patients with KFS should screen the latter for all associated anomalies as an interspeciality team approach so that no anomaly is left unidentified.

5. ETHICAL STATEMENT

Verbal explanation was given to the parents and an informed written consent was obtained both for the writing up and taking the pictures. A written informed consent was obtained from the parents for publication of this case report.

6. CONFLICT OF INTEREST

Conflict of interest declared none.

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