KLIPPEL FEIL SYNDROME: A RARE CASE REPORT

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ABSTRACT

INTRODUCTION: Klippel-Feil syndrome is rare congenital, musculoskeletal condition that is characterized by the fusion of at least two vertebrae of the neck. The incidence of which is 0.71% as reviewed by Brown et. al in 1400 skeletons. In fewer than 50% of cases, the three classical diagnostic findings of Klippel-Feil syndrome includes a low posterior hairline, a triad of short neck and a limited range of neck movements especially of lateral bending are present. **CASE REPORT:** In this particular case, we found congenital fusion of C6 and C7 vertebrate, levoscoliosis, basilar invagination and fusion of atlantooccipital joint on right side with no evidence of renal disease, congenital heart disease and neurological impairment except low IQ. This case has classical triad short neck, low posterior hairline and limited cervical range of motion. **CONCLUSION:** A rare and typical case of Klippel-Feil Syndrome is very important to present for better identification and prompt treatment.
Keywords: Fusion, congenital Klippel-Feil syndrome, cervical, vertebrae.

Introduction

In 1921, a patient was diagnosed Klippel-Feil syndrome (KFS) by Maurice Klippel and Andre Feil due congenital fusion of vertebrae.[1] The classical clinical triad of, limitation of head and neck, short neck and low posterior hairline is present in a KFS that is a complex syndrome of osseous and visceral anomalies.[2] This rare disease occurs in 1 of every 42,000 births globally and interestingly, 60% of cases are in females.[4] The patients with KFS exhibit a smaller lower third of the face and facial asymmetry with no dental implications.[1-3] There are several defects reported from KFS including congenital heart defects in which most common is the ventricular septal defect, ear defects causing deafness either conductive or neural. Mental deficiency including lower IQ and different neurological disorders, rib defects; the Sprengel sequence (elevated scapula), cleft plate and scoliosis are also commonly observed in diagnosed cases of Klippel-Feil syndrome (KFS).[3]

With the reduced penetrance and variable expression, KFS is enrolled in the Online Mendelian Inheritance in Man Database as being of sporadic autosomal dominant inheritance.[5] Poland syndrome, spondyloepiphysial dysplasia, spondylocostal dysostoses and short-rib polydactyly syndromes are main differential diagnosis for Klippel-Feil syndrome.[3] It is strongly recommended to evaluate close members of immediate family of diagnosed KFS because in almost all cases this syndrome occur sporadically.[4] Various anomalies and fetal alcohol syndrome is somehow, related with KFS but evidence is not strong due to fewer cases and lower prevalence.[6-7] There are speculations that fetal alcohol syndrome causes Klippel-Feil syndrome (KFS).[7]

In the patient, the bony malformation at age of 16 didn t entrap or damage brain and spinal cord. The rapid growth of female body as adolescence and adult will not obviously symptomatic due to malfunction

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in lower vertebral region.[8] In this case report, we will present radiological and clinical findings of a 14 years old girl who got diagnosed with KFS.

Case Report

A 14 years old, Pakistani girl from Pashton tribe was brought to orthopedic outdoor of our hospital in Islamabad on 15-05-2019 by her mother with history of fall and presented with neck stiffness. In antenatal history patients mother revealed it was a normal vaginal delivery, patient was healthy in her neonatal period. Achieved mile stones slightly delayed as compared to her siblings and had poor understanding of things. No proper workup was done. Now patient presented with complain of neck pain and restriction of neck movement. On physical examination observations are made of short neck, low posterior hair line, head tilted on left sided and restricted neck movements. No tenderness of cervical spine is noted however mild elevation of left scapula is observed. Patient was referred to our department for CT cervical spine.

CT cervical spine revealed malalignment of the cervical spine with levoscoliosis centered on C6-C7. There is fusion of posterior elements of C6 and C7 is noted with reduced anteroposterior diameter of the vertebral bodies giving wasp-waist appearance (Fig.1 and Fig. 2).

Invagination of odontoid into the base of skull by taking chamberlain line as reference is observed (Fig.3 and 4). Increased atlantodens distance is noted (Fig. 5a and 5b).

Further MRI spine is advised to assess the cord and craniocervical junction. MRI showed craniocervial junction tightening (Fig. 6). Patient was managed conservatively by the orthopedic specialist with cervical collar correction and manual traction technique of the cervical spine was explained to the parents, along with neck and shoulder exercises were also advised by the physiotherapist. Prognosis was explained in detail.

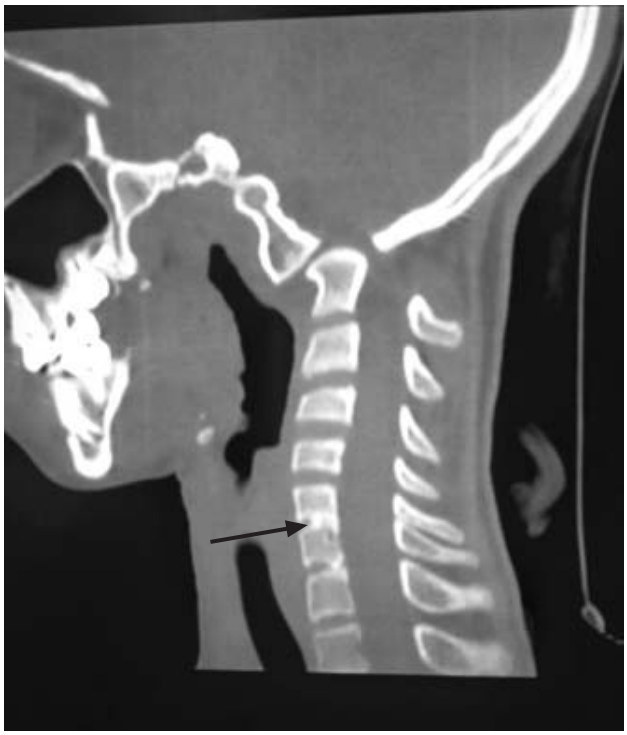

Figure 1: CT Sagittal reformat- Wasp- Waist Sign (Reduced AP diameter of the vertebral bodies).

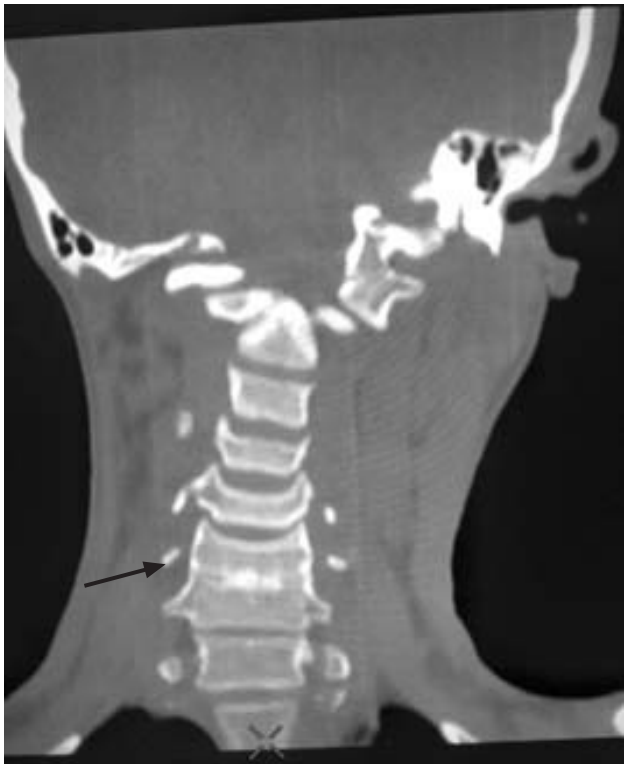

Figure 2: CT coronal reformat Fusion of C6 and C7 vertebral bodies

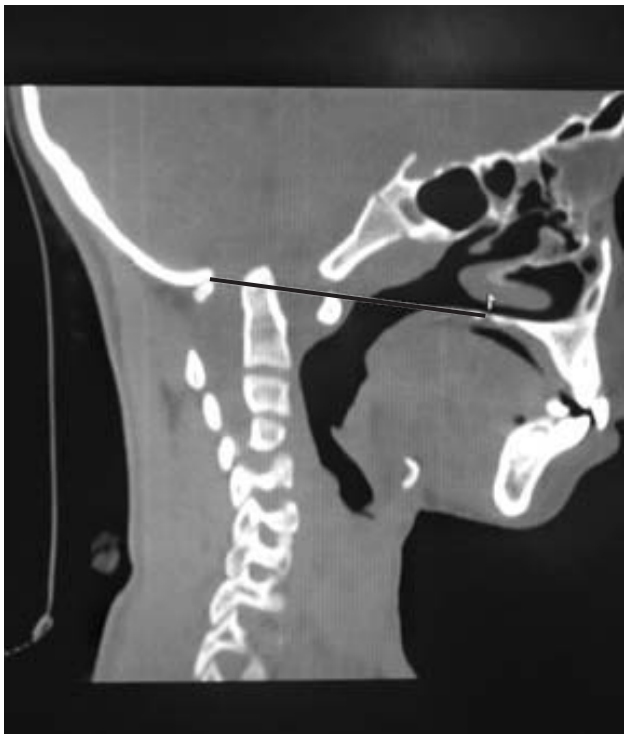

Figure 3: CT Sagittal reformat Odontoid process projecting above the Chamberlian line.

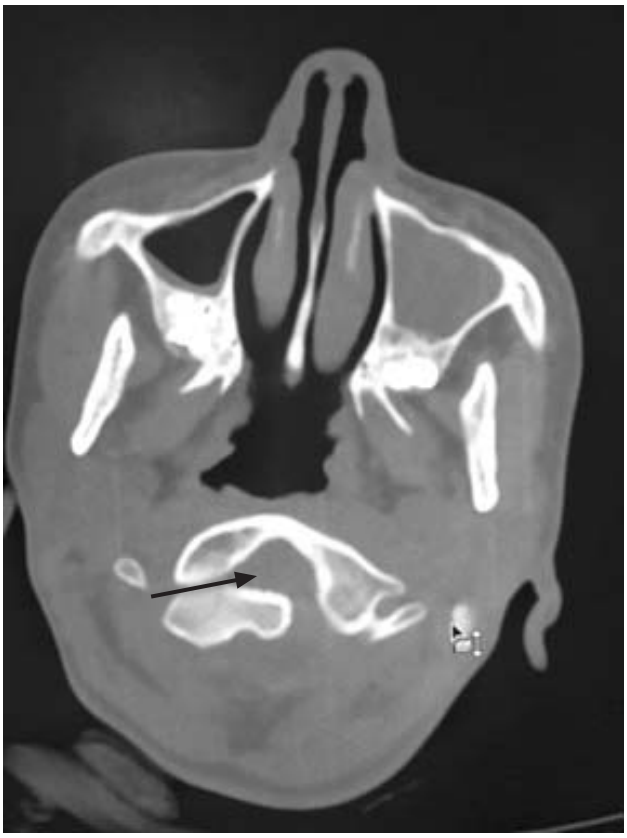

Figure 5a: CT axial section Bone window Increased atlantodens distance.

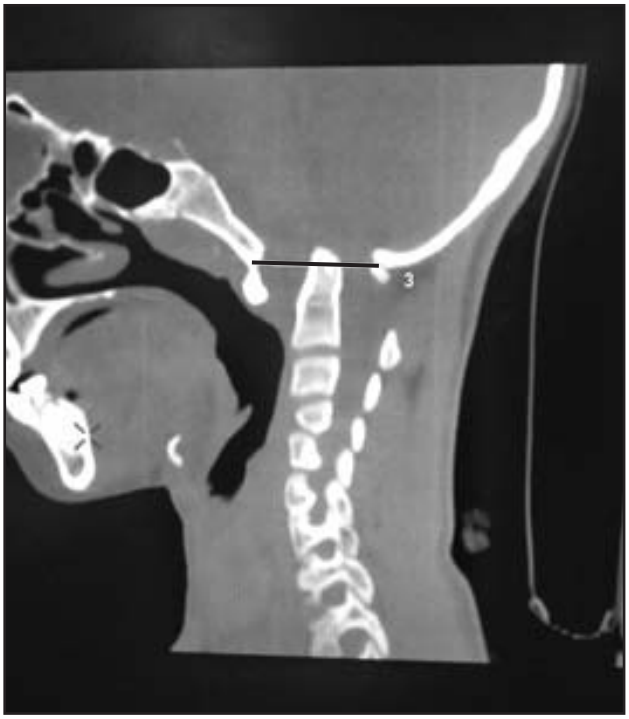

Figure 4: CT Sagittal reformat Odontoid process projecting into the foramen magnum.

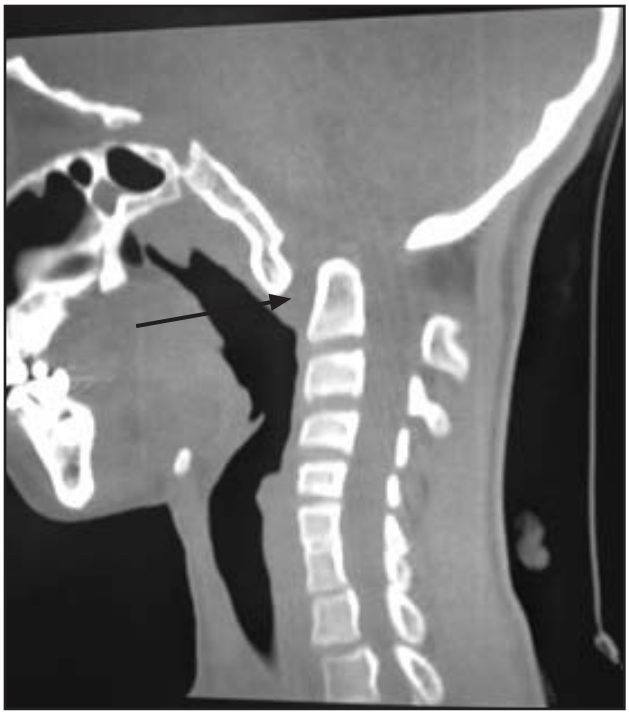

Figure 5b: CT Sagittal reformat Bone window Increased atlantodens distance.

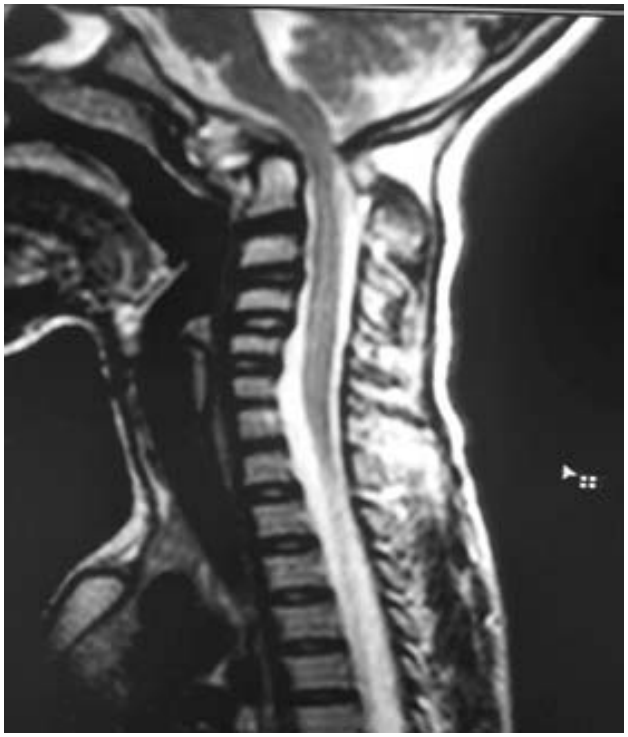

Figure 6: MRI Sagittal section Craniocervical tightening is seen.

Discussion

KFS is a spectrum of various abnormal features predominantly congenital fusion abnormality of cervical vertebra. It may follow familial or sporadic course. This condition was first reported by Drs. Klippel and Feil in 1912, when a patient with short neck, low posterior hair line and restricted neck movement was presented.9 These characteristics are considered to be the classic triad in this patient population. Samartzis proposed in 2016 all three characteristics may not all be present in up to 50% of cases.[10] Multiple extra-spinal manifestations may coexist with this condition. The classic triad of Klippel-Feil Syndrome include presence of lower posterior hair line, short neck and restriction of head movements. There is fusion of all or few of the cervical vertebral bodies of the cervical vertebrae may be fused (1, 2, 4, 6).[11] The number of the cervical vertebrae may be diminished, the spinous processes may be fused, and often there is an irregular formation of the lateral masses and bodies. The arches of the vertebrae may fail to unite posteriorly, resulting in a spina bifida occulta. Often there are associated malformations of the ribs, such as cervical ribs, crowding of the ribs, fusion of the ribs, as well as congenital anomalies in other parts of the body. 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 atlantooccipital 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.[12] MRI of cervical spine both in flexion and extension is modality of choice to assess the craniocervical junction tightening and associated abnormalities of the cord. As in our case craniocervical tightening is demonstrated by MRI. (Fig. 6) Management of the KPS is usually conservative however surgery may be considered in few cases for craniocervical instability and relieve the cord constriction if present. Physiotherapy along with shoulder and neck exercises also play a prominent role.

Conclusion

Klippel-Feil syndrome (KFS) is one of the rare heterogeneous spectrum with varying clinical and physical manifestations. Different factors according to patient s presentations should be considered for choosing particular management for KFS case. Various imaging modalities with initial evaluation starting from plain radiographs, followed by CT scan cervical spine and MRI play important role for assessing the extent and severity of bony manifestations and cord abnormalities. In this particular case cervical spine CT and MRI cervical spine was performed which aid in patient s further management. Last but not the least role of physiotherapy and proper exercises cannot be ignored in cases with conservative management plan.

Conflict of Interest: Declared none by authors.

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