CASE REPORT

Klippel feil syndrome: A case report

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ABSTRACT
Klippel-Feil Syndrome is an uncommon disorder characterized by the congenital fusion of any 2 of the 7 cervical vertebrae. The most common signs are short neck, low hairline at the back of the head, and restricted mobility of upper spine. We present a case of patient presented to us with breathlessness due to restrictive type of defects due to abnormality of spine and association with tuberculosis.

Keywords: Klippel-Feil Syndrome, Kyphoscoliosis, synkinesia, Cervical Vertebrae, Congenital Malformation

INTRODUCTION
Klippel-Feil Syndrome (KFS) is a rare congenital malformation characterized with the triad¹ ² of short neck, low hair line and restricted neck movements due to insufficiency of segmentation of two or more cervical vertebrae.

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History: This case was presented to us with pulmonary tuberculosis and patient was coincidentally had klippel-Feil syndrome. Patient Delivered normally in some Govt. Hospital 16 years back. Patient then developed normally and patient’s developmental milestones were normal. Patient started walking at the age of 1 year and patient was able to do his routine work normally. His physical and mental status are normal. He had studied till 7th standard and left study due to social reason. Patient developed exertional breathlessness and cough for 1 month and presented to us. He was admitted in our hospital for the same. Examination: On general and systemic examination patient had short webbed neck, low posterior hair line, restricted neck movements & associated kyphoscoliosis, hypoplasia of right ear, mirror movements (synkinesis or mirror movements, refer to involuntary movements in a group of muscles or limb of one side of the body as a response to intentionally made move of the opposite side of the body) and microtia of left ear (Figure 1). Investigation: Addon to the previous examination findings we evaluated further more by doing X-ray lateral view and MRI of cervical spine which showed occipitalisation of C1 and fusion of c2 and c3 along with, Basilar invagination at C2 vertebral body with compression over cervico medullary junction and resultant angulation & thinning of cord (Figure 2&3). On USG Abdomen Lt Renal fossawas not seen and Rt kidney was normal. From above findings plus added clinical examination findings like presence of abnormally low hair line, presence of kyphoscoliosis, restricted lateral movements of neck, were noted and patient diagnosed as klippelfeil syndrome.

Figure 1: Patient Photograph

Figure 2: X-ray cervical spine (lateral view)

Figure 3: MRI Cervical Spine

Management: Although there is no cure of this disease itself, the comorbidities can be treated to make the life of the patient better. Generalised weakness were cited to tuberculosis and we started anti tuberculosis treatment under supervised DOTS Program along with supplemental nutritional food and physiotherapy to make the maximum movement of neck possible and the breathing exercises to improve lung function deranged due to kyphoscoliosis [Fig-3], other symptoms of tuberculosis were treated accordingly. Patient then advised for regular follow up for the further management.

DISCUSSION
KFS was first described by Klippel and Feil in 1912. It is seen at a rate of approximately 1 in 42,000.KFS arises from insufficiency of the normal segmentation and cleavage of the cervical vertebrae in the early weeks of fetal development. It is a congenital malformation characterized with the triad
of short neck, low hair line and restricted neck movements.\textsuperscript{1,2} Cases in which the classical triad is observed constitute approximately 50% of KFS cases. Skeletal system abnormalities, urinary system abnormalities, synkinesis, hearing loss, congenital heart disease and brainstem abnormalities may accompany the clinical triad of Klippel-Feil syndrome.\textsuperscript{3} Klippel-Feil syndrome has been organized into three basic types. In type I, all of the cervical and upper thoracic vertebrae are fused together into one block. In type II, one or two pairs of cervical vertebrae are fused together. In type III, there is lower thoracic or lumbar fusion as well as cervical fusion.\textsuperscript{4,5} While type 1 and 3 show an autosomal recessive trait, type 2 is rather autosomal dominant. Although Type 2 is observed more associated with skeletal abnormalities, skeletal system anomalies show more severe outcome. Our case was consistent with type 2 KFS. Studies have suggested that the prevalence of KFS may have a predilection for sex type, with 60% to 70% of KFS cases occurring in females.\textsuperscript{6,7,8} The exact cause of Klippel-Feil syndrome remains unknown; but researchers have linked mutations of GDF6 and GDF3 genes to the syndrome. Other Symptoms associated with Klippel-Feil Syndrome are Kidney, rib and heart malformations, Respiratory problems, Neurological deficits, Syndactyly (webbed fingers) and hypoplastic thumb (abnormality of the thumb), A condition called synkinesia or mirror movement—where movement in one hand involuntarily mimics the deliberate movement of the other hand, Sprengels deformity, where the scapulae (shoulder blades) are underdeveloped and sit high on the back causing weakness of the shoulders, Cleft palate, Hearing issues. Several authors report the association of partial or complete conductive hearing impairment, underdeveloped low-set ears and facial asymmetry in patients with type II KFS.\textsuperscript{9,10,11} Children with Klippel-Feil syndrome may have other health issues too, such as kidney, heart or lung problems. This is because the child’s organs may have experienced disrupted development in utero. Therefore, an ultrasound—using high-frequency sound waves to create an image like those taken during pregnancy of the developing baby—also may be conducted on your child’s organs to detect any anomalies. Additional tests that may be needed include cardiac evaluation and hearing tests. There is no cure for Klippel-Feil syndrome. Treatment is ordered when certain issues—such as spinal curvatures, muscle weaknesses or heart problems—occur and need to be treated, accordingly with the consultation of respective physicians and surgeons.

\textbf{REFERENCES}