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Case Report

Complex presentation of Klippel-Feil syndrome with scoliosis and Sprengel's deformity: a rare case

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ABSTRACT

Klippel-Feil syndrome is a rare congenital anomaly. It is characterized by fusion of two or more cervical vertebrae. In this case report, we have presented a rare case of a nine years old male child suffering from Klippel-Feil syndrome associated with congenital scoliosis and sprengel deformity with no other abnormality in accordance with heart, kidneys, and nervous system. As per the literature review very few cases have been reported on this syndrome thus, this case report is a significant contribution to existing literature.

Keywords: Scoliosis, Vertebrae, Fusion, Congenital, Spine, Screening

INTRODUCTION

Klippel-Feil syndrome is a complex condition occurring due to improper segmentation in the early fetal development phase.[1] This syndrome is typically marked by a triad of lower hairline at the back of head, fused cervical vertebrae, and a short neck. This classical triad is seen in less than 50% affected patients. It affects 1 in 40,000 to 42,000 newborns globally.[2] Maurice Klippel and Andre Feil were the first to report this condition in 1912.[3] Most of the KFS cases get undiagnosed due to asymptomatic presentation thus, making it difficult to estimate its incidence and prevalence rate.[4]

KFS may involve other parts of the body, including lungs, kidneys, heart, eye, ears, muscles, nerves, spinal cord, and bones. It affects 30% to 50% of patients with scoliosis. 30% of patients with sensorineural hearing loss and 30% with kidney diseases. In 30% of cases this syndrome is associated with Sprengel deformity (Abnormal shoulder blade development). In this deformity there is abnormal elevation of the shoulder associated with defective descent, position and anatomy of scapula.[5]

CASE REPORT

A nine years old male child presented to the paediatric outpatient department with abnormal development of shoulder girdle and restricted neck movements. There was no deviation of the neck. The antenatal history was unremarkable. There was no history of fever, diabetes, hypothyroidism, hypertension, and any drug intake. The child was born at term via normal vaginal delivery at home. The child cried immediately after birth. The vaccination schedule was complete up to now. There was no family history of spinal deformities.

The child had normal development of milestones. The child is satisfactory in studying as per the school records. The child has a normal IQ and completes his daily activities by himself. On physical examination the child was normally built, short height, short neck, no deviation of neck, low hairline at the back of head, elevated left shoulder, and cervicothoracic scoliosis with convexity towards right side. There was no soft tissue swelling and no tenderness in the spine.

The child had limited movement of the left shoulder during abduction above 90°C. The range of motion of the right shoulder is normal. The neck had limitations in performing extension and lateral bending on both sides. The vitals were within normal range. Before this the patient had never visited any doctor for these concerns.

Management and outcome

The child was advised to have a radiological examination of the neck, which showed fusion of cervical vertebrae at C2-C3 and C5-C6 levels on lateral X-ray of neck. The vertebral bodies appeared joint with loss of intervertebral disc space. The left scapula was raised. The patient had no cardio respiratory or urinary system abnormality.

The child had no neurological symptoms. He was advised to perform exercises to increase the mobility of neck and shoulder as conservative management. The child was prescribed a cervical collar. For the management of scoliosis regular follow-up was advised to monitor the curve progression along with stretching and strengthening exercises to increase mobility and relieve any spasm. The parents were explained about this syndrome and its prognosis in detail.

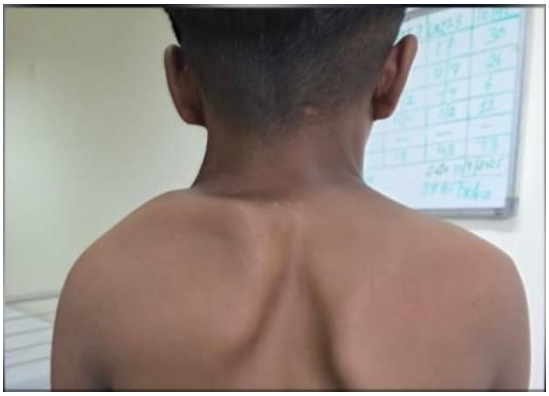

Figure 1: Klippel-Feil syndrome with sprengel deformity.

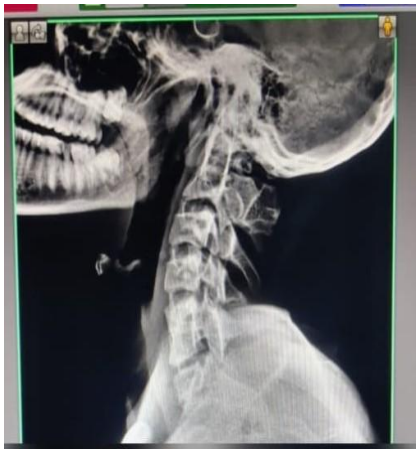

Figure 2: X-ray showing fusion of cervical spine.

DISCUSSION

Klippel-Feil syndrome is a multifactorial disease. Andre Feil in 1912 categorized this syndrome into three types on the basis of involvement of the spine as type 1 (cervical and upper thoracic spine fusion), type 2 (incomplete segmentation at one or two cervical levels and association with atlanto-occipital anomaly), type 3 (fusion of cervical levels with thoracic or lumbar vertebrae).[6] The cause of Klippel-Feil syndrome is hypothesized by several studies stating that neural tube defects, vascular disruption, fetal distress, or genetic factors may be responsible for it. The heritable mutation occurring in GDF6, GDF3, and MEOX1 genes can be a cause.[7,8] In our case it was sporadic.

Various cases of Klippel-Feil syndrome are accompanied by torticollis. Torticollis is a condition in which the head is bent to one side and the chin points away to the other side. In comparison to the case study by Agarwal AK et al where the child suffered from torticollis on the left side our patient did not have any neck deviation.[9] This condition can be either acquired or congenital. The causes for acquired type can be trauma, spasm of sternocleidomastoid muscle, inflammation, enlargement of lymph nodes, tuberculosis, and scoliosis. While congenitally it can occur due to exhaustive labour, pelvic disproportion, and contracture of muscles.[10]

As per the recent research, it is discovered that most of the sporadic cases go undiagnosed and incidental radiological examination reveals this syndrome. This condition can predispose the patients to spinal stenosis, neurological defects, audiovisual impairment, and visceral inclusion. There are 4.4% to 14% cases affected by cardiac anomalies including ventricle septal defect, coarctation of aorta, hypoplasia of aortic arch, aneurysm, and abnormal pulmonary vessels.[11] In a case study by Shoaib et al the child had Klippel-Feil syndrome with dandy-walker spectrum and occipital cephalocele unlike our case report, where the patient was devoid of these anomalies.[12]

During pregnancy the prenatal ultrasound may report cervical spine deformities in affected neonates. Besides, that it might detect associated visceral anomalies, if present. For children, X-rays (anteroposterior, lateral, and odontoid) can detect the fusion of cervical spine and other segments. To check for scoliosis thoracic and lumbar spine imaging is also required. For patients with neurological defects, magnetic resonance imaging is helpful, and for the assessment of spinal canal stenosis computed tomography is useful.[13]

Most cases undergo non-operative management which include modification of activities, avoidance of contact sports, stretching exercises, analgesics, cervical collar, monitoring and follow-up. Depending on the severity of symptoms, age group, associated anomalies, and risk-benefit ratio for the operative candidate surgical decision is taken. In this case report, our patient suffered from

Scoliosis and Sprengel deformity with no other evident abnormality and visceral impairment. We offered conservative management to the patient and provided all the details to the parents as well.

CONCLUSION

Patients with Klippel-Feil syndrome and its associated disorders should undergo early screening and management to facilitate conservative treatment options whenever possible. Education about the condition is essential to empower patients and reduce societal stigma. Awareness can significantly improve quality of life and outcomes for affected individuals.

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