CASE REPORT

Therapeutic rehabilitation for Klippel-Feil syndrome: a case report

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Abstract

Klippel-Feil syndrome (KFS) is a genetic condition presenting with a triad of symptoms: short neck, limited range of motion in the neck, and a low posterior hair line. Herein, we report the case of an eight-year-old girl who presented with complaints of pain, numbness, and a triad of KFS at the Paediatric Physical Therapy and Neuro-Rehabilitation Centre, Lahore. The aim of this report is to show that a rare syndrome can be coped very effectively through efficient goal setting. Physiotherapy intervention protocol was designed and given to the patient for four weeks and prognosis was established.

Keywords: Klippel-Feil syndrome, Physiotherapy Intervention, Genetic Condition.

DOI: https://doi.org/10.47391/JPMA.11563

Introduction

Klippel-Feil Syndrome (KFS) is a known rare genetic condition that involves abnormal development of the cervical region that causes the fusion of two or more cervical vertebrae in early foetal development. The specific symptoms present in this syndrome are a triad of short neck, limited range of motion (ROM) in the neck, and a low posterior hair line. Pain and neurological manifestations such as limb numbness or weakness are also present.[1] The acuteness of KFS can be varying, and the management of this syndrome initially focuses on symptomatic treatment and prevention of complications. Physical therapy and surgical interventions work side by side to improve the patient's quality of life. Sometimes surgical corrections are required to manage complications like scoliosis and spinal cord compression.[2] The occurrence of this syndrome is approximately 1 in 42,400 live births. This condition is more common in females with female to male ratio of 4:1. Risk factors for this condition are unknown.[3] Physiotherapy plays a crucial role in managing this condition and enhancing the patient's quality of life.[2]

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Submission complete: 05-01-2024 **First Revision received:**26-04-2024

Acceptance: 02-11-2024 **Last Revision received:** 01-11-2024

Case Report

An eight-year-old school going girl came to the Paediatric Physical Therapy and Neuro Rehabilitation Centre, Lahore, with her parents on August 18, 2023, with the complaints of pain in cervical region, limited neck ranges, and slight intermittent paraesthesia in the right upper limb. Birth history was full-term via spontaneous vaginal delivery followed by an immediate birth cry. There were no complications throughout the pregnancy. The child achieved her milestones at the right age. She was independent in all activities of daily living. On observation, the child was short necked with tilt towards the right side, and a chest wall deformity called pectus excavatum. On palpation, there was bilateral tenderness in trapezius and levator scapule muscle and tightness of right sternocleidomastoid. On examination, the patient was well oriented to time, place, and surroundings. Pain intensity was 5 out of 10 on Visual Analogue Scale. Restricted cervical ranges were measured through goniometer that are mentioned in Table.

Magnetic Resonance Imaging findings showed vertebral segmentation defect in the form of hemi vertebra extended from the second cervical vertebra to the seventh cervical vertebra with resultant scoliotic deformity in the spine, having concavity towards the left side. Consent was taken from the parents prior to the start of treatment. A treatment protocol was designed according to the International Classification of Functioning and Disability model.[4] It consisted of management of pain and tenderness by giving circular soft tissue massage, muscle conditioning exercises to improve strength and mobility, including active range of motion exercise of cervical region and upper extremities, scapular retraction, and stretching of sternocleidomastoid.[5] Manual traction of the neck was given using traction belt for 10 seconds hold and 10

Table: Range of Motion of Cervical Region

Neck ranges	Pre-Treatment		Post-Treatment	
	Right	Left	Right	Left
Rotations	15	22	19	24
Side rotations	8	10	10	13
Flexion	38		40	
Extension	42		42	

seconds for two minutes, it was followed by positioning of the neck in correct alignment using soft neck collar.[6] The patient was given guidance of kinaesthetic awareness regarding correct posture. Breathing techniques to maintain lung capacity, including active cycle of deep breathing technique, segmental breathing, and diaphragmatic breathing along with chest expansion exercises were introduced. Both the parents and the child were counselled regarding physical appearance and social stigma. Furthermore, prevention of secondary complications was also considered. Home exercise plan was given and feedback was taken daily through videos and logbook to ensure compliance. Re-evaluation was done after one month of therapy. Significant improvement was seen in pain reduction, score on VAS remained 0. Improvement was seen in neck mobility and stiffness. Kinaesthetic awareness helped to maintain correct posture. Intermittent paraesthesia resolved almost completely within weeks after cervical traction. Daily physiotherapy sessions helped the patient not only to develop the habit of regular exercise and well-being but also boost the confidence level of the child.

Discussion

Klippel-Feil syndrome (KFS) is a rare congenital disorder characterised by the fusion of at least two cervical vertebrae, leading to various spinal and extra-spinal anomalies. Although traditionally thought to manifest in adulthood, recent studies indicate that cervical spine symptoms can present much earlier in life, affecting nearly half of paediatric KFS patients. This study presents the case of an eight-year-old child presenting with KFS.[7]

KFS was first reported by Maurice Klippel and André Feil in 1912. Classical triad was noticed in approximately 50% of KFS cases. Skeletal system malformation, genitourinary system abnormalities, hearing problems, inborn diseases of the heart, and brainstem anomalies may go along with the clinical trio of KPS. In the present case the patient has congenital pectus excavatum. The family did not agree to include the child's photograph in the case report. Consent to publishing the case report was provided.

In 2014 a study was conducted by Agarwal et al. in which they advised neck collar, cervical and shoulder work out, and manual cervical traction.[8] A recent study draws attention towards patients having a possibility of neurological symptoms that can be managed with activity alterations. The results of this case confirms that, a patient can be well treated conservatively unless severe neurological symptoms or neck instability exists.[9, 10]

In a 2019 case study by Mukherjee et al., the authors explored the treatment of a patient with Klippel-Feil Syndrome (KFS) complicated by Benign Hypermobile Joint Disease (BHJS). They emphasised the use of conservative physiotherapy interventions, including manual cervical traction, targeted exercises, and activity modifications. These interventions led to significant improvement in pain relief, increased cervical mobility, and resolution of neurological symptoms, closely aligning with the present study's findings. Both studies underscore the efficacy of conservative physiotherapy management in improving patient outcomes without the need for surgical intervention unless severe symptoms or instability arises.

Conclusion

By means of appropriate physiotherapy protocol, this rare condition can be coped with very effectively and can be a blessing for those children who are going through cosmetic and social stigma in the community.

Consent: Guardian's consent was obtained for publishing this case.

Disclaimer: This article has not been presented or published before.

Conflict of Interest: None.

Source of Funding: None.

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AUTHORS' CONTRIBUTIONS:
WR: Concept, design and writing.
SS: Critical revision, concept and final approval.
SL: Data collection and assembly.
SS: Editing, formatting, trial registration and submission.
MJ, MA: Data entering and analysis.