



Physiotherapy Intervention in Klippel-Feil Syndrome: A Rare Case Report

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Abstract

Klippel-Feil syndrome is a complex condition caused by congenital fusion of the vertebrae due to failure in the division or normal segmentation of the cervical spine vertebrae in early fetal development. It is characterized by a triad of short neck, low hairline, and decreased neck mobility. The incidence of Klippel-Feil syndrome is approximately 1 in 40,000 to 42,000 newborns worldwide and found predominantly in females. Here a case of a 6-year-old girl child is presented having medial scapular pain with the classical triad of Klippel Feil syndrome. Physiotherapy intervention of 3 weeks was given and the prognosis was established.

Keywords: Klippel-Feil syndrome; Congenital fusion; Cervical spine

Introduction

Klippel-Feil syndrome is a complex condition caused by congenital fusion of the vertebrae due to failure in the division or normal segmentation of the cervical spine vertebrae in the early fetal development [1]. It is characterized by a triad of short neck, low hairline, and decreased neck mobility [2]. Klippel-Feil syndrome is present since birth and may be due to mutations in *GDF3*, *GDF6*, and/or *MEOX1* genes [1,3].

The incidence of Klippel-Feil syndrome is approximately 1 in 40,000 to 42,000 newborns worldwide and found predominantly in females [1,4]. Vertebral fusion becomes visible only after spinal ossification reaches completion in a young child [4].

This syndrome can be associated with scoliosis, Sprengel's shoulder, torticollis, hypoplasia of upper limb, genitourinary, cardiovascular abnormalities. More than 50% of the cases are associated with congenital scoliosis, 30% of them with deafness, 20% to 30% with Sprengel's deformity, 12% to 15% with cervical ribs and 4% to 29% can show cardiovascular abnormalities [2,5].

Case Presentation

A 6-year old girl came with a complaint of right scapular pain on December 24th, 2021 from the past 10 to 15 days. There was a gradual onset of pain and it was aggravated by doing overhead activities and relieved by rest. No other signs of heaviness, tingling or numbness were present. No history of recent injury or trauma was reported by the girl. According to mother-child was full term and C-section was done followed by an immediate birth cry. The mother didn't have any postnatal complications. A vaccination course was completed. There was no history of delayed milestones. Child started walking at age of 10 months. The child was having satisfactory physical and mental performance in the school as per parents and teachers. She was independent altogether activities of daily living. On observation, the child was having good build, short stature with low hairline, short neck, and decreased neck mobility. Facial expressions were symmetrical.

Right Cervico dorsal scoliosis along with kyphosis was present. Shoulder elevated on the right side, B/L wrist Radial deviation was there in the wrist along with hypoplastic thumb (one extra finger in the left thumb).

Mirror movements were present and gait was normal. Higher mental functions were normal and the child responded well to the commands.

Pain intensity on VAS score was 6. Tenderness was present in the medial border of the right scapula and bilateral tightness of sternocleidomastoid was present. Neck range of motion was restricted and is mentioned in Table 1.

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Table 1: Neck range of motion was restricted.

Neck (Range of motion)	Right	Left
Rotation	25 degree	15 degree
Side flexion	10 degree	11 degree
Flexion	35 degree	
Extension	40 degree	

**Figure 1:** Posterior view of the child.

ROM of the bilateral shoulder joint was full. Strength in the bilateral upper limb was within the normal limit. All the reflexes were normal and no neurological deficit was present. The child was not having any other cardiorespiratory or genitourinary problems.

(Figure 1) shows the posterior view of the child and (Figure 2) shows the radiograph of both AP and lateral view of cervical spine.

Goals of physiotherapy

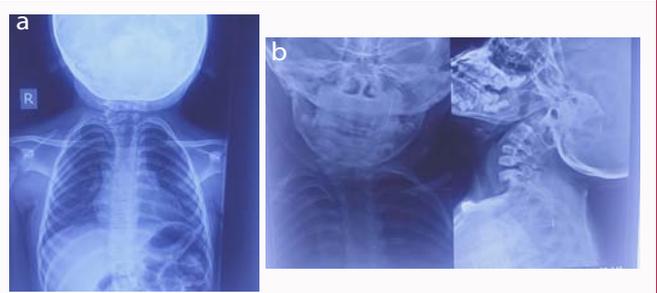
- To reduce pain and stiffness
- To improve mobility and strength
- Maintenance of correct posture
- Maintenance of respiratory functions
- Prevention of secondary Complication
- To reduce psychosocial stress

To reduce pain and stiffness

- Hot pack was given
- Palmer kneading was given kneading on the medial border of the scapula

To improve mobility and strength

- Active ROM exercise for B/L UL
- Active ROM neck exercises
- Stretching of B/L sternocleidomastoid
- Strengthening of B/L Ulnar deviators
- Scapular retractor and protractors
- Wall push-ups
- Clock exercises

**Figure 2:** Radiograph of both AP and lateral view.

Maintenance of correct posture

- Deep neck flexors strengthening exercise.
- Prone on elbow
- Strengthening exercise of lateral muscle chain for improving convexity
- Stretching exercise of lateral muscle chain for improving concavity
- All four positions then raise the right arm and leg while the spine stays aligned. Repeat the same exercise but change arm and leg.
- Hanging exercise

Maintenance of respiratory functions

- Diaphragmatic breathing exercise
- Deep breathing exercise
- Thoracic expansion exercise

Prevention of secondary complication

- Ergonomic advice
- Core strengthening exercises
- Swimming exercise

To reduce psychosocial stress

Counseling of both the parents and child was done as the child was aware of her short stature and the parents were concerned about the cosmetic appearance and social stigma.

Results and Discussion

Besides physiotherapy exercises, home exercise programs were also taught to the patient. Self-correction exercises and active exercises for both upper limb and neck were explained to the patient. After 3 weeks of physiotherapy intervention, the patient was reassessed. The pain was reduced to a VAS score of 0. Exercises help to improve mobility and reduce the stiffness in the neck. Postural correction also helps to create self-awareness. Every day physiotherapy sessions helped the girl not only to develop the habit of regular exercise and well being but also boost the confidence level of the child.

Klippel-Feil syndrome remains asymptomatic many times and intervention is only given only when symptoms are present. Due to altered biomechanics, hypomobility at one level will be compensated by the hypermobility at the other level of the spine which can lead to secondary complications and functional impairment. Early intervention with physiotherapy will help to maintain the strength and mobility of the joint and prevent secondary degenerative changes.

In 1985 a study was done where 11 children with Klippel-Feil

syndrome were classified into 3 categories, Group 1 patients with an unstable fusion pattern, Group 2 patients with craniocervical abnormalities Group 3 patients with cervical spinal canal stenosis. Various Surgical approaches and a thorough evaluation for early identification of neurological symptoms have been suggested [5].

Agarwal et al. [6] 2014 in their case study of Klippel-Feil syndrome, prescribed cervical collar, neck and shoulder exercise, and manual cervical traction as the child was not having any neurological problems [6].

Menger et al. [7] have emphasized that the patients having a high risk of neurological deficit can be managed with activity modification. According to his report, a patient can be well managed conservatively unless neurological deficit or cervical instability is present [7].

There have been lesser studies on physiotherapy management of Klippel-Feil syndrome. This study will provide an insight that a rare syndrome can be managed efficiently and can be a boon to those who are facing cosmetic and social stigma in society.

Conclusion

Early identification and physiotherapy intervention will improve mobility, prevent secondary complications and thus help to develop

the confidence of the patient. In absence of neurological deficit and spinal instability, the patients of Klippel-Feil syndrome can be well managed by conservative treatment.

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