

# Klippel Feil Syndrome: A Rare Case Report

Ashok Kumar Agarwal<sup>1</sup>, Mohit Goel<sup>1</sup>, Jeetendra Bajpai<sup>1</sup>,  
Sourav Shukla<sup>1</sup>, Nikhil Sachdeva<sup>1</sup>

## What to Learn from this Article?

Presentation and diagnosis of Klippel Feil syndrome

### Abstract

**Introduction:** In Klippel Feil syndrome, classically there is a triad of short neck, a low posterior hairline and a limited range of neck movements especially of lateral bending. In fewer than 50% of cases have all the three elements.

**Case Report:** In the present case we have found congenital Scoliosis, Sprengel deformity and there were no evidence of renal disease, congenital heart disease and neurological impairment. The present case has classical triad low posterior hairline, short neck and limited cervical range of motion.

**Conclusion:** A rare case of Klippel Feil Syndrome is being presented with the aim that such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to her and to her parents.

**Keywords:** congenital, fusion, Klippel-Feil syndrome, cervical, vertebrae.

### Introduction

Klippel-Feil syndrome was first described by Maurice Klippel and Andre Feil in 1912 in patient with congenital fusion of cervical vertebrae [1]. In KF syndrome, classically there is a triad of short neck, a low posterior hairline & a limited range of neck movements especially of lateral bending. In fewer than 50% of cases have all the three elements. Klippel-Feil syndrome occurs in one of every 42,000 births, and 60% of cases are female[2].

KF syndrome is group of deformities that result due to failure

of segmentation of cervical spine. This syndrome is associated with Sprengel deformity, high scapula, scoliosis, urinary tract anomalies, congenital heart disease & hearing lost in 30% of cases. Usually in Sprengel deformity there is loss of abduction & forward flexion after 90 deg. In 30 % of cases with Sprengel deformity, the scapula is bound to cervical spine by fibrous tissue, cartilage or an omovertebral bone which restrict abduction of shoulder after 90 degrees (3).

### Author's Photo Gallery



Dr. Ashok Kumar Agarwal



Dr. Mohit Goel



Dr. Jeetendra Bajpai



Dr. Sourav Shukla



Dr. Nikhil Sachdeva

#### Access this article online

Quick Response Code:



Website:  
www.jocr.co.in

DOI:  
10.13107/jocr.2250-0685.197

<sup>1</sup>Department of Orthopaedics, Vivekanand Polyclinic and Institute Of Medical Sciences, Lucknow, Uttarpradesh. Pin- 226007. India

#### Address of Correspondence

Dr. Jeetendra Bajpai, M.S orthopaedics, Senior Resident Department of Orthopaedics, Vivekanand Polyclinic and Institute of Medical Sciences, Lucknow, Uttarpradesh. Pin-226007. India. Email: dr.jbajpai@gmail.com

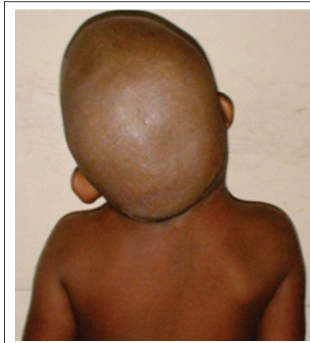
Copyright © 2014 by Journal of Orthopaedic Case Reports

Journal of Orthopaedic Case Reports | pISSN 2250-0685 | eISSN 2321-3817 | Available on www.jocr.co.in | doi:10.13107/jocr.2250-0685.197

This is an Open Access article distributed under the terms of the Creative Commons Attribution Non-Commercial License (<http://creativecommons.org/licenses/by-nc/3.0>) which permits unrestricted non-commercial use, distribution, and reproduction in any medium, provided the original work is properly cited.



**Figure 1:** Flexion of neck without facial asymmetry.



**Figure 2:** Mild cervicodorsal scoliosis present on left side neck.

## Case Report

A girl child, 4 year old attended outpatient department of orthopaedic surgery of VPIMS, Lucknow in July 2013 with painless deformity since birth i.e. torticollis of left side. There was no antenatal history of fever, hypertension, drug intake and any other significant event during pregnancy. Natal History: Full term normal delivery at home and child cried soon after birth. Post natal History. No history of high fever, trauma & child was having complete course of vaccination.

She was having normal over all physical & mental development & started yearly neck holding, sitting, crawling & standing. She started walking in ninth month earlier than her elder brother. She goes to school with satisfactory progress as per her class teacher. She is shy, intelligent and understand all exercises herself. She is independent in all her daily activities. Till the time of presenting to us she was not shown to any doctor.

On examination, she was having good built, short in height, head tilted to left side, low hair line, short neck, the distance between from tip of ear pinna to Trapezius upper border is grossly short as compare to healthy side. Flexion of neck without facial asymmetry (Fig 1).

Mild cervicodorsal scoliosis present on left side neck.(Fig 2). Range of movement of cervical spine. She can perform full flexion, partly extension, rotation & lateral bending showed Limitation of movement. There is no tenderness in cervical spine. Shoulder scapula is elevated on left side.

Right shoulder showed normal range of movement whereas left shoulder had limitation of range of movement especially abduction after 90 degree. Cardio respiratory system, urinary system –examination revealed no abnormality.

Radiological examination of cervical spine revealed blocked vertebrae C1,C2,C3 along with spinal bifida C6 & C7(fig 3) and radiological examination of chest revealed high scapula on left side as compared with normal right shoulder joints(Fig4).

The girl was also examined by a Senior Plastic Surgeon & was advised for conservative management to begin with. As our case was not having any neurological or any other systemic involvement she was advised exercises of neck & left shoulder 5 to 6 times a day, Corrective cervical collar was prescribed and the technique of manual traction of cervical spine was explained & demonstrated to her father. Prognosis was explained in detail.

## Discussion

Cervical vertebral segmentation anomalies are referred to as the Klippel-Feil anomaly whether they involve fusion of 2 segments or the entire cervical spine Klippel-Feil syndrome appears to be failure of the normal segmentation and fusion processes of mesodermal somites, which occurs between the third and seventh week of embryonic life[4,5].

Various abnormality are present in Klippel-Feil syndrome. A torticollis accompanying the disease may mask the shortness of neck. In torticollis, head is bent to one side while chin points to other side. Broadly torticollis is either since birth (congenital) or after birth(acquired) [6]. In congenital usually there is a history of difficult labour followed by sternomastoid tumor. The acquired lesion is due to various injury of cervical spine, inflammatory-due to enlarged/inflamed cervical lymph node, spasmodic i.e spasm of sternomastoid muscle along with posterior cervical muscle, compensatory from scoliosis or due to a defect in sight. Tuberculosis of cervical spine and lastly it may be due to burn contracture[7].

In cases of congenital muscular torticollis, the contracture of sternomastoid muscles is painless and newborn child tilts head toward & rotate his chin away from contracted sternocleidomastoid muscle. Usually the lesion is identified in 2-3 month after birth[3]. Stretching of affected muscle, lateral rotation & side bending is affective in 90% cases provided stretching is initiated within one year of age and if not surgical intervention is desirable to prevent facial deformity[3].

Klippel feil syndrome is often having other congenital lesions like congenital scoliosis or kyphosis(60%), renal disease(35%), synkinesis or mirror movements(20%), Sprengel deformity(30%) and torticollis, loss of hearing(30%), facial asymmetry and flattening of neck(20%), congenital heart diseases(4% to 14%), brainstem lesions, congenital cervical stenosis. Adrenal aplasia, ptosis, facial nerve palsy, syndactylia, diffuse hypoplasia of upper limb may also be seen. Disc degeneration has also been reported in almost all cases[8-9].

Beside orthopaedics clinical evaluations, patients with Klippel-Feil syndrome should be assessed by anteroposterior and lateral cervical flexion/extension and thoracolumbar radiographies, abdominal ultrasonography, and were subjected to systemic examination to detect any urological, cardiological, otorhinolaryngological, neurological and psychiatric finding. Computed tomography and magnetic resonance imaging where necessary[10].

Mechanical symptoms caused by degenerative diseases respond to traction, cervical collar and analgesics. Surgical correction of Sprengel deformity is done to improve cosmesis. Minimally involved cases have good prognosis and live normal



**Figure 3:** Radiological examination of cervical spine revealed blocked vertebrae C1,C2,C3 along with spinal bifida C6 & C7.



**Figure 4:** Radiological examination of chest revealed high scapula on left side as compare with normal right shoulder joints.

life with no significant restrictions or symptoms [11]. Patients with hypermobility on vertebra not having fusion, prophylactic fusion shouldn't be performed unless patient have neurological problems because of disc problem that may appear later. Severely involved patients have good prognosis if cardiopulmonary, genitourinary and auditory problems are treated early [12]. In anomalies of occipito-cervical passage, high morbidity and mortality rates have been frightening, as cervical cord and brain stem are very close to each other [13].

In the present case we have found congenital Scoliosis, Sprengel deformity and there were no evidence of renal disease, congenital heart disease & neurological impairment. The present case has classical triad low posterior hairline, short neck & limited cervical range of motion.

## Conclusion

Patients with Klippel Feil Syndrome should be assessed for associated systemic abnormalities beside cervical fusion, and such cases should be identified and treated at an early stage to minimize cosmetic & social stigma to the patient.

## Clinical Message

Though klippel-feil syndrome is rare syndrome encountered less commonly and classical triad are present in almost 50% cases, one should closely investigate for other anomalies associated with it for better, early management and rehabilitation.

## References

1. Jones KL. Smith's recognizable pattern of human malformation. 5th ed. Philadelphia: WB Saunders Company;1997.
2. Stephen R Pledger. Cervical Spine Disease. Ortho Secrets, 3rd Ed,2003.
3. Paul W Esposito and Brain E Brighman. Miscellaneous Congenital Disorders. Ortho Secrets, 3rd Ed,2003.
4. Vaidyanathan S, Hughes PL, Soni BM, Singh G, Sett P. Klippel-Feil syndrome – the risk of cervical spinal cord injury: A case report. BMC Fam Pract. 2002;3:6.
5. Boraz RA, Irwin DH, Van Blarcom C, The dental rehabilitation of a patient of klippel-feil syndrome and sprengel's deformity. Spec Care Dentist 1986; 6(1):22-4.
6. Mahirogullari M, Ozkan H, Yildirim N, Cilli F, Gudemmez E. Klippel-Feil syndrome and associated congenital abnormalities: evaluation of 23 cases. Acta Orthop Traumatol Turc. 2006;40(3):234-9.
7. S. Das. A manual of Clinical Surgery, 10th edition 2013, kolkotta.
8. Van Kerckhoven MF, Fabry G. The Klippel-Feil syndrome: A constellation of deformities. Acta Orthop Belg. 1989;55:107-18.
9. Sudhakar AS, Nguyen VT AND Chang JB. Klippel-Feil syndrome and supra-aortic arch anomaly: A case report. Int J Angiol. 2008; 17(3): 118.
10. Nagib MG, Maxwell RE, Chou SN. Klippel-Feil syndrome in children: clinical features and management. Child Nerv Syst. 1985; 1(5):255-63.
11. Warner WC jr. Pediatric cervical spine. In: Canale ST, editor. Campbell's operative orthopaedics. Vol 2. 12th ed. St. Louis: Mosby; 2013. P. 1677-83.
12. Pizzutillo PD, Woods M, Nicholson L, MacEwen GD. Risk factors in Klippel-Feil syndrome. Spine. 1994;19:2110-6.
13. Disorder of the neck. In: Herring JA, editor. Tachdjian's pediatric orthopaedics. Vol 1. 3rd ed. Philadelphia: W. B. Saunders; 2002. P. 173-87.

Conflict of Interest: Nil  
Source of Support: None

## How to Cite this Article

Agarwal AK, Goel M, Bajpai J, Shukla S, Sachdeva N. Klippel Feil Syndrome: A Rare Case Report. Journal of Orthopaedic Case Reports 2014 July-Sep;4(3): 53-55