

RARE CASE OF KLIPPEL FEIL SYNDROME: A CASE REPORT

Sasirekha B¹, Pranavadhyani G², Serena Florence Francis³

1. Reader, Department of Oral Medicine and Radiology, JKKN Dental College and Hospital, Komarapalayam, Namakkal, TamilNadu.

2. Professor and Head of the Department, Department of Oral Medicine and Radiology, JKKN Dental College and Hospital, Komarapalayam, Namakkal, TamilNadu

3. MDS

ABSTRACT:

Klippel feil syndrome or congenital osseous torticollis is a congenital fusion of cervical vertebrae. It is a complex syndrome with a classical triad of short neck, low posterior hair line and limitation of head and neck movements. We report a case of a female child who consulted us for forwardly placed teeth. The occurrence of abnormal gait, kyphoscoliosis associated with hemivertebrae and fused ribs oriented the diagnosis and linked with the features of Klippel Feil syndrome.

Key words: Klippel Feil syndrome, Classification, Clinical triad, Radiological findings

INTRODUCTION:

Klippel Feil Syndrome is predominantly a skeletal anomaly characterised by failure of union of 2 or more segments of the cervical spine. May or may not involve the dorsal and/or lumbar spine.^[1] It is due to the failure in the normal segmentation/ division of the cervical vertebrae during the 3 – 8th wk of gestation. Clinical Triad includes the short neck, low hairline at the back of the head, and a limited range of motion in the neck^[2].

In the Online Mendelian Inheritance in Man database KFS is listed under autosomal dominant inheritance pattern with reduced penetrance and variable expression.^[3]

A case of Klippel Feil Syndrome associated with Bell palsy

and partial deafness in a 15 year old girl was presented in this article.

CASE DETAIL:

A 15 year old female patient came to our department with a complaint of forwardly placed upper front teeth. She has disproportionate skeletal growth since childhood and encountered with frequent fractures. The girl was alert, cooperative and responds to the questions. Examination revealed abnormal gait and kyphoscoliosis. Extraoral examination revealed a facial asymmetry and incompetent lips on the left side. Deviation of nose towards right side and partial impairment of hearing on left side was observed. Inability to furrow her forehead, close her eyes and inability to close her mouth while blowing on left side.

On intraoral examination, Class II molar relation with bimaxillary protrusion is observed. Depapillated & erythematous areas in the dorsal surface of the tongue was evident. Based on the findings, we provisionally diagnosed it as Bell's palsy associated with a syndrome. Otorhinolaryngology opinion pictures out partial deafness on left side.

OPG revealed no significant changes. Lateral cephalogram shows fused C1 – C2. Chest X-ray reveals fused ribs at multiple levels both medially & laterally. Scoliosis was also present. Pelvis X-Ray shows Hemivertebrae, unfused pubis and left side Pelvis appears translated superiorly. Spine X-ray points out multiple block vertebrae & hemivertebrae was noted at different levels. Orthodontic treatment was instituted to correct the malocclusion and symptomatic treatment for Bell's palsy was provided. Hearing aid was advised for partial deafness.

DISCUSSION:

The first case was reported long back in the history of Egyptian mummy from 500 BC. Klippel-Feil Syndrome (KFS), also known as synostosis of cervical spine, is sporadic, but seldom may be inherited. Maurice Klippel and Andre Feil independently described this syndrome in 1912. This was due to an alteration in the migration of mesodermal tissue at third and seventh week of embryonic life during when the cervical disc

formation & development of other organ system occurs.^[4,5] Etiology is unknown; mainly it is genetically determined. GDF6, GDF3, or MEOX1 gene produces a protein which is important in the formation of bones and joint, especially spine. MEOX1 gene produces protein MOX1 helps in separation of vertebrae from one another. Mutations in these genes lead to Klippel Feil syndrome.^[6]

Initially 3 types of KFS were described by Feil in 1912. Later it was modified by Tracy et al in 2004, based on the fusion of C2 – C3 and mode of inheritance.

[A] 3 groups depending on the level and extend of vertebral fusion (Feil, 1912)^[7]

I : Extensive fusion into bony blocks of cervical & upper thoracic vertebrae

II: Fusion of 2 or 3 vertebrae

III: Fusion in the cervical, lower thoracic and /or lumbar spine, together with rib fusions.

IV: Cervical fusion associated with sacral segmentation defects.^[8]

[B] Tracy et al, 2004:^[9]

KF1: Fusions at C1 with or without caudal fusions.

KF2: Fusions no higher than C2 -3. Autosomal dominant inheritance, 100% penetrance

KF3: Fusions no higher than C2-3. Autosomal recessive inheritance.

KF4: Cervical fusions, hearing loss & Dunane anomaly (Wildervanck syndrome). X-linked inheritance.

It occurs in 1 of 42,000 individuals, 57 – 70% patients are female.^[10] This remains asymptomatic with a Classic triad, 20% of them present with a facial asymmetry, torticollis and neck webbing. Axial symptoms of neck/headache, neck pain, and neck stiffness are the predominant symptoms in symptomatic KFS patients.

An associated symptom varies greatly from one another. Primary / secondary neurological deficits mainly facial nerve palsies, hemiplegia, quadriplegia & paraplegia, scoliosis, vertebral abnormalities and abnormalities in eyelids.^[11] Intelligence is normal in 90% of patient^[12] and 20 – 50% with Hearing deficits,^[13] Congenital heart defects, Rib defects, Sprengel anomaly (failure of descent & lateral migration of scapula), Aplasia of vagina & Uterus and renal abnormalities are reported.^[14]

Fibrous dysplasia of maxilla, Pseudoankylosis of TMJ and. Cleft palate or submucous cleft palate – 17%, Oligodontia of primary & permanent teeth, maxillary constriction and velopharyngeal insufficiency. Mouth breathing and facial asymmetries are frequently observed in patients with KFS.^[15]

Anteroposterior (AP) and lateral views of cervical spine shows Block vertebrae, hemivertebrae, atlanto-occipital fusion & stenosis of the cervical spinal canal and fused vertebra at the level of C2 - C3 segment.^[16] CXR may show cardiac abnormalities or fusion of ribs. CT scan can be very useful at the base of the skull, especially if abnormalities are unilateral. MRI – Identifies spinal cord abnormalities and assess the spinal canal. Flexion & extension Ultrasound and Intravenous pyelogram (IVP) are required for imaging of the urinary tract. Hearing assessment should be carried out in all children.

Our patient has presented with a facial asymmetry, short neck, neurological problems, partial deafness, fused at the level of C1-C2, hemivertebrae and unfused pubis is suggestive of KFS type III category.

Dental professionals should be aware of the characteristics of KFS when making an oral diagnosis and planning treatment. It is important to should check for the presence of a submucous cleft^[16,17] and congenitally missing teeth^[18] as the incidence of these findings are high in KFS patients.

Symptomatic procedures may include surgery to relieve cervical or craniocervical instability and constriction of the spinal cord, and to correct scoliosis. For mechanical symptoms like cervical collar analgesics, NSAIDS or careful traction can be used. To relieve neurologic

problems decompression may employed based on the site of the stenosis. Early identification of this condition helps to minimize the risk of injury in their daily activities or operative interventions. Special precautions are undertaken during sedation or anesthesia in the dental clinic as they should not be intubated.^[19]

CONCLUSION:

KFS is a rare sporadic and heterogeneous condition affecting the

spine with varying clinical manifestations. Various imaging modalities like antero posterior and lateral views, extension and flexion views of MRI, CT, USG and various neurological tests are performed. Considering all the clinical and radiographic features, treatment should be instituted. Dentist should follow a special precaution before performing the treatment procedures and patient should not be intubated for giving sedation or general anesthesia.

REFERENCES:

1. Jack Ramsey, Johnny Bliznak. Klippel-feil syndrome with renal agenesis and other anomalies. *American Journal of Roentgenology* 1971; 113:460-463.
2. Samartzis D, Kalluri P, Herman J, et al; "Clinical triad" findings in pediatric Klippel feil patients. *Scoliosis spinal Disord*. 2016 Jun 27; 11:15.
3. McKusick VA. Klippel-Feil Syndrome: Online Mendelian Inheritance in Man, John Hopkins University; 2004. Available from: URL: <http://www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM> (accessed 28 Sept. 2004).
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 sprenge's deformity. *Spec Care Dentist* 1986; 6(1):22-4
6. Mohamed JY, Faqeih E, Alsiddiky A, Alshammari MJ, Ibrahim NA, Alkuraya FS. Mutations in MEOX1, encoding mesenchyme homeobox 1, cause Klippel-Feil anomaly. *Am J Hum Genet*. 2013 Jan 10; 92(1):157-61.
7. Samartzis D, Kalluri P, Herman J, Lubicky JP, Shen FH. The extent of fusion within the congenital Klippel-Feil segment. *Spine (Phila Pa 1976)*. 2008 Jul 1; 33(15):1637-42.
8. Clarke RA, et al. Heterogeneity in Klippel Feil syndrome: a new classification. *PediatrRadiol*. 1998; 28:967-74.
9. Tracy MR, Dormans JP, Kusumi K. Klippel-Feil syndrome: clinical features and current understanding of etiology. *Clin Orthop Relat Res*. 2004 Jul;(424):183-90.
10. Wessell et al. Cervical instability in Klippel Feil Syndrome: a case report and review of the literature. *Chinese Neurosurgical Journal* (2015) 1:6
11. Soumeya Fedal, Haddam Ali el Mahdi, TadjineAicha, Ahmed Ali eyla. Klippel Feil syndrome: A case report and review of literature. *IOSR Journal of Dental and Medical Sciences* .Volume 15, Issue 12 Ver. III (December. 2016), PP 65-67.
12. Isidor B, David A. Two girls with Short Stature, Short neck, vertebral anomalies, Sprengel deformity and Intellectual disability. *Eur J Med Genet* 2014 Nov 18. pii:S1769-

- 7212(14)00204-3. Doi:10.1016/j.ejmg.2014.11.001.
13. McGaughran JM, Kuna P, Das V. Audiological abnormalities in the Klippel-Feil syndrome. *Arch Dis Child* 1998; 79(4):352–5.
 14. W.B. Moore, T J Mathews Ronald Rabinowitz. Genitourinary Anomalies Associated with Klippel-Feil Syndrome. *The journal of bone and joint surgery* 1975; Vol. 57-a(3): 355-357.
 15. Manuel O. Lagravère, María I. Barriga, Carla Valdizán, Augusto Saldarriaga, Juan F. Pardo, Martha Flores. The Klippel-Feil Syndrome: A Case Report. *J Can Dent Assoc* 2004; 70(10):685–8
 16. Dino Samartzis , Prakasam Kalluri, Jean Herman, John P. Lubicky and Francis H. Shen. Clinical triad findings in pediatric Klippel- Feil patients. *Scoliosis and Spinal Disorders* 2016; 11:15.
 17. Nagib MG, Maxwell RE, Chou SN. Klippel-Feil syndrome in children: clinical features and management. *Childs Nerv Syst* 1985; 1(5):255–63.
 18. Helmi C, Pruzansky S. Craniofacial and extracranial malformations in the Klippel-Feil syndrome. *Cleft Palate J* 1980; 17(1):65–88.
 19. Ozdiler E, Akcam MO, Sayin MO. Craniofacial characteristics of Klippel-Feil syndrome in an eight year old female. *J Clin Pediatr Dent* 2000; 24(3):249–54.

FIGURES:



Fig 1: shows kyphoscoliosis



Fig 2: Facial asymmetry was observed due to forwardly positioned maxillary teeth

Fig 3, 4 & 5: shows Inability to furrow her forehead on left side, Inability to close her eyes on left side & Incompetent lips on left side



Fig 6: Depapillated & erythematous areas in the dorsal surface of the tongue.



Fig 8: Lateral Cephalogram shows C1-2 appears fused



Fig 7: Ribs is present at a higher level on right side



Fig 8: Chest x-ray shows Ribs appear fused at multiple levels both medially & laterally



Fig 9: Pelvis x-ray shows Hemivertebrae, unfused pubis and left side Pelvis appears translated superiorly



Fig 10: Spine x-ray shows Multiple block vertebrae & hemivertebrae was noted at different levels

