Case Report

Klippel feil syndrome rare presentation: Case report

Yogesh Ashok Edge*, J C Awalekar, C D Awalekar, Harshad Adhav

Department of General Medicine, Bharati Vidyapeeth University Medical College and Hospital Sangli, Maharashtra, INDIA.
Email: yogeshedge9592@gmail.com

Abstract

Klippel feil syndrome is a bone disorder characterised by abnormal joining (fusion) of two or more spinal bone in the neck (cervical vertebrae).which is present from birth. Three major feature result from this abnormality A short neck, A limited range of motion in the neck, and Low hairline the back of head. In this case report we have presented the clinical and radiological finding of the patient with klippel feil syndrome.

Key Words:

***Address for Correspondence:**
Dr. Yogesh Ashok Edge, Department of General Medicine, Bharati Vidyapeeth University Medical College and Hospital Sangli, Maharashtra, INDIA.
Email: yogeshedge9592@gmail.com

Received Date: 14/06/2017 Revised Date: 11/07/2017 Accepted Date: 20/08/2017
DOI: https://doi.org/10.26611/1004912

Access this article online	
Quick Response Code:	Website: www.medpulse.in
	Accessed Date: 23 September 2017

INTRODUCTION

Klippel –feil syndrome is a rare disease initially reported in 1912 by maurice klippel and andré feil from france (Klippel and Feil, 1912) characterized by the congenital fusion of any 2 of the 7 cervical vertebrae. The syndrome occurs in a heterogeneous group of patients unified only by the presence of a congenital defect in the formation or segmentation of the cervical spine and is believed to result from faulty segmentation along the embryo's developing axis during weeks 3-8 of gestation. Mutations in the gdf6 and gdf3 genes can cause klippel-feil syndrome. These genes provide instructions for making proteins that belong to the bone morphogenetic protein family, which is involved in regulating the growth and maturation (differentiation) of bone and cartilage (Genetic Home Page).. In people with klippel-feil syndrome, the fused vertebrae can cause a limited range of movement of the neck and back as well as pain in these areas. It manifests as a short neck with reduced mobility and a low posterior hairline (Yuksel *et al.,* 2006), occurring only in 40-50% of patients. Decreased range of motion is the most frequent clinical finding. Patients with upper cervical spine involvement tend to present at as earlier age than those whose involvement is lower in the cervical spine. In addition, a wide spectrum of associated anomalies may be present. This heterogeneity has complicated elucidation of the diagnosis and management of the syndrome. The actual prevalence of klippel–feil syndrome is unknown due to the fact that there was no study done to determine the true prevalence (Angeli *et al*., 2010). Although the actual occurrence for the kfs syndrome is unknown, it is estimated to occur 1 in 40,000 to 42,000 new-borns worldwide (Yuksel *et al.,* 2006). In addition, females seem to be affected slightly more often than males (Floemer *et al.,* 2008).

CASE REPORT

A 18 yr male presented in opd with difficulty in movement of neck, giddiness,, headache, vomiting. h/ o fall on bed. h/o deaf anddumb since childhood. h/o head injury. Family history of consacigious marriage 2 degree. Father has hypertension. Mother has diabetes mellitus, hypertension, hypothyroidism. On examination pt conscious oriented vital are stable, short neck with low hairline pattern of hair. All cranial nerve are normal expect pt had left rectus palsy. Respiratory syetem are normal cardiovascular system soft pan systolic murmur is heard. per abdomen system are normal.

Chest x ray: Lung parenchyma normal. with fussed c1-c2 vertebrae. cardiomegaly.

How to cite this article: Yogesh Ashok Edge, J C Awalekar, C D Awalekar, Harshad Adhav. Klippel feil syndrome rare presentation: Case report. *MedPulse – International Medical Journal.* September 2017; 4(9): 959-961. http://www.medpulse.in

CT brain plain: There is fusion of anterior arch of c1 vertebra with inferior end/last segment of clivus and fusion of posterior arch with occiput. Resultant widened foramen magnum noted and protruding dense noted.
Impression: No obvious traumatic brain parenchymal abnormality detected at present scan. Occipitalization of atlas /Atlanto occipital synostomosis/vertebra basal assimilation.
MRI brain plain: Normal MRI Barin plain study.
MRI CV Junction: Occipitalalisation of c1 vertebra noted. fused upper cervical vertebrae with short neck. Basilar invagination. E/O bifid upper cervical cord. No evidence of cervical cord and brain stem compression.

Figure 1:

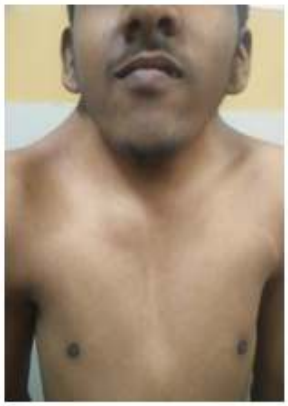

Figure 2:

Figure 3:

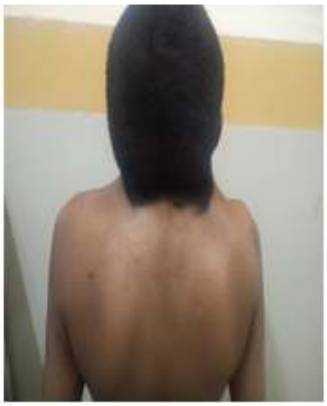

Figure 4:

Figure 5:

Figure 6:

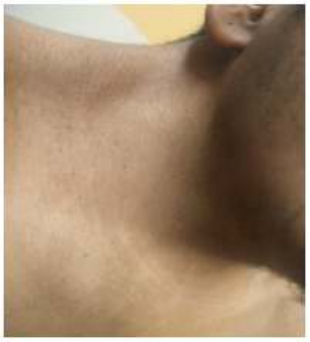

Figure 7:

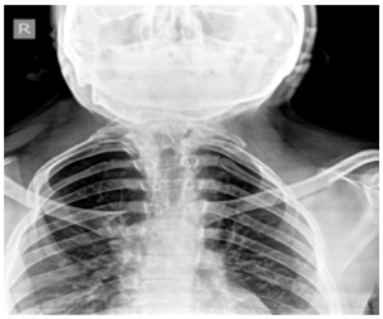

Figure 8:

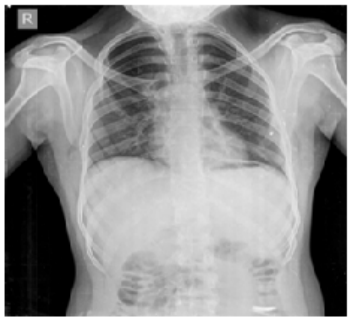

Figure 9:

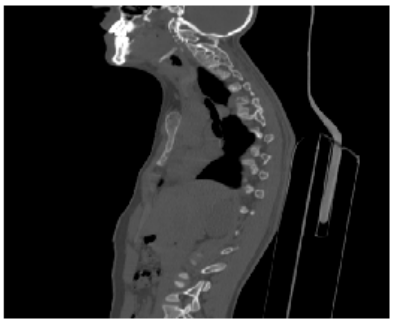

Figure 10:

Table 1

Class of klippel feil syndrome	Inheritance	Verterabal fusion and associated anomalies	Overlap with Klippel and Fell's original classification
KF1 (214300)	Autosomal recessive	Rostral fusion at C1 and severe associated anomalies ((Short neck, cardiac defects, and craniofacial anomalies)	Type I, II and III
KF 2 (118100)	Autosomal dominant	C2-3 fusion and possible craniofacial anomalies	Type I, II and III
KF 3 (613702)	Reduced penetrance	Singular isolated fusion, most rostral at C3	Type II
KF S4	X-linked inheritance	Vertebral fusion and ocular anomalies	Commonly referred to as wildervank syndrome

DISCUSSION

Feil (1919) defined three morphological sub-types of this anomaly (table 1) (Clarke *et al*., 1998) Different classifications have been proposed and 4classes (KF1, 2,3and4) was identified according to position of cervical vertebra fusion, status of familial trait and its characteristics. Syndrome can present late in childhood when the anomaly is in lower cervical spine in contrast to the children presenting early or soon after birth who are having upper cervical spine anomalies (Naikmasur *et al*., 2011). Klippel-Feil syndrome is usually diagnosed in the patients during childhood. However; Clinical presentation varies because different associated syndromes and anomalies may occur in these patients lately (NAIKMASUR *et al.*2011). Thus; the challenge of the specialist is to recognize the associated anomalies that can occur with Klippel Feil syndrome and to perform the appropriate workup for diagnosis.

REFERENCES

1. Angeli E and Wagner J et al., (2010). General format title. Retrieved from http://owl.english.purdue.edu/owl/resource/560/01/.
2. Clarke Catalan G et al., (1998). Heterogeneity in Klippel-Feil syndrome: a new classification. Pediatric Radiology (28) 967-974.
3. Floemer F and Magerkurth O et al., (2008). Klippel-Feil syndrome and Sprengel deformity combined with an intraspinal course of the left subclavian artery and a bovine aortic arch variant. American Journal of Neuroradiology (AJNR) 29(2) 306-7.
4. Klippel M and Feil A (1912). Un cas d'absence des vertebres cervicales. Avec cage thoracique remontant jusqu'a la base du crane (cage thoracique cervicale). Nouv Iconog Salpetriere (25) 223–250.
5. Naikmasur VG and Sattur AP et al., (2011). Type III Klippel-Feilsyndrome: case report and review of associated craniofacial anomalies. Odontology 99 197-202.
6. Ulmer JL and Elster AD et al., (No date). Klippel-Feil syndrome: CT and MR of acquired and congenital abnormalities of cervical spine and cord. Journal of Computer Assisted Tomography 17(2) 215-24.
7. Yuksel M and Karabiber H et al., (2006). Diagnostic importance of 3D CT images in Klippel-Feil Syndrome with multiple skeletal anomalies: a case report. Korean Journal of Radiology 6(4) 278-81.

Source of Support: None Declared
Conflict of Interest: None Declared