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Case Report

KLIPPEL-FEIL SYNDROME, A RARE PRESENTATION – CASE REPORT

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ABSTRACT
Klippel-feil syndrome is a bone disorder characterized by the abnormal joining (fusion) of two or more spinal bones in the neck (cervical vertebrae), which is present from birth. Three major features result from this abnormality: a short neck, a limited range of motion in the neck, and a low hairline at the back of the head. Most affected people have one or two of these characteristic features. Less than half of all individuals with klippel-feil syndrome have all three classic features of this condition. Here we are reporting a rare case with associated anomalies. The aim of this paper was to review clinical heterogeneity; radiographic abnormalities in klippel-feil syndrome which simulate acute pathology. We insist in comprehensive evaluation and delineation of diagnostic and prognostic classes.

Keywords: *Klippel-Feil Syndrome, Associated Anomalies, Cervical Spine, Complex Heart Disease*

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 an 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).

CASES
A new born baby first issue of second degree consanguineous married couple delivered in our hospital through emergency LSCS in view of foetal distress (Non-reactive NST) was admitted to NICU for respiratory distress which developed soon after birth. On examination, baby had short neck with low hair line, micrognathia, heart sound predominantly over right side of chest. Suspecting congenital syndrome, baby was taken for further investigations. Imaging showed fused C2-C3 spine, dextrocardia with complex heart disease with single atrium, anomalous position of right kidney. Hence a clinical diagnosis of variant of type 1 Klippel-Feil syndrome was made. Baby was then managed by appropriate referral to pediatric surgical consultation for associated anomalies.

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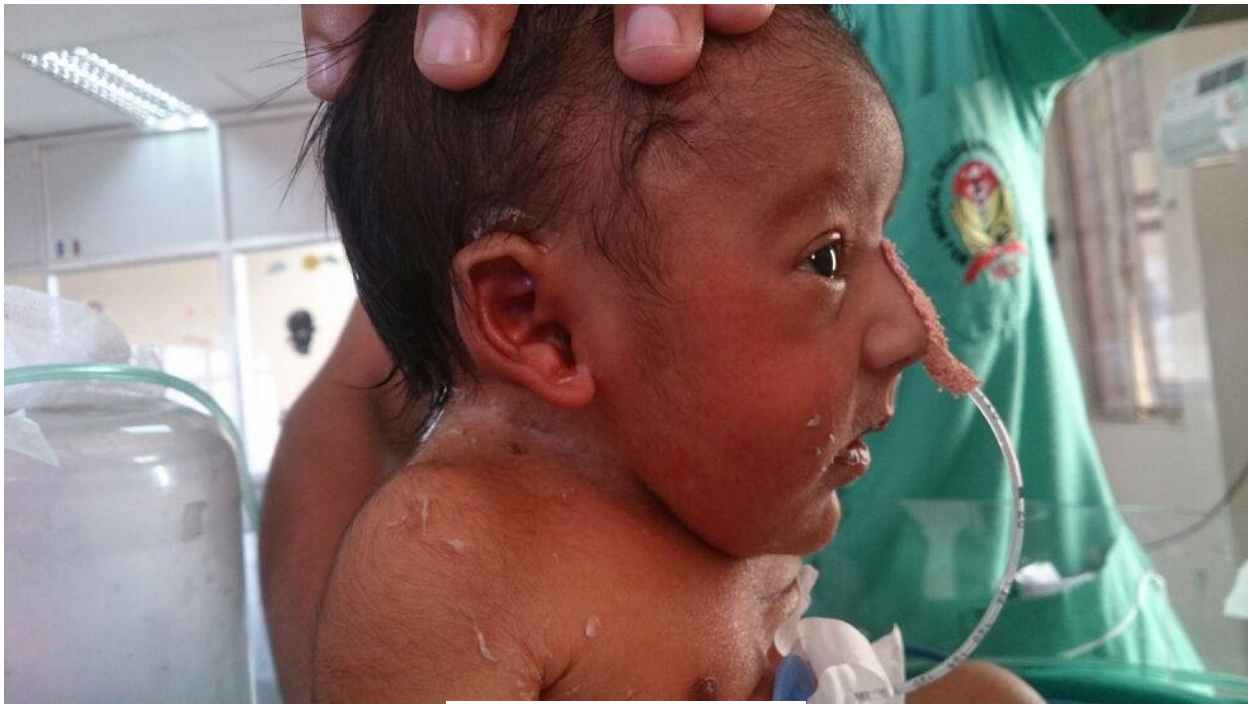

Figure 1: Short neck in baby

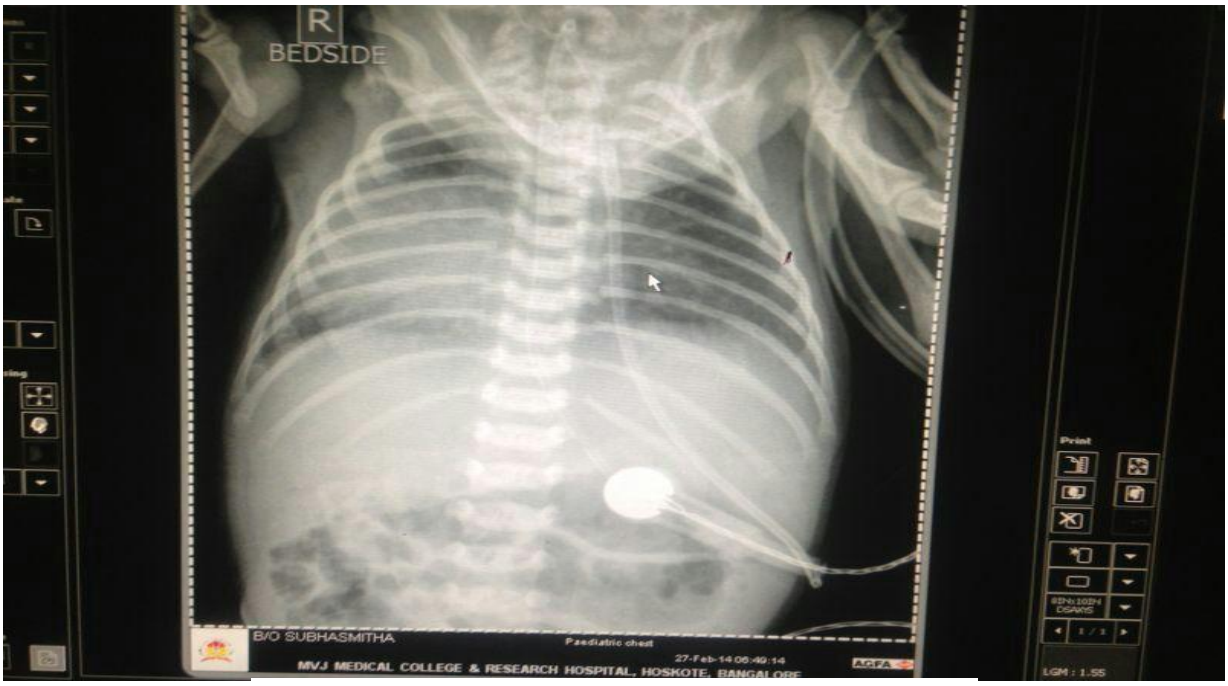

Figure 2: X-ray film showing dextrocardia & fused C2-C3

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).

Table 1: Classification of Klippel-Feil syndrome (Clarke *et al.*, 1998)

Class of Klippel-Feil syndrome	Inheritance	Vertebral fusion And associated anomalies	Overlap with Klippel and Feil's original classification
KF1 (214300)	autosomal recessive	Rostral fusion at C1 and severe associated anomalies((short neck, cardiac defects, and craniofacial anomalies)	Types I, II and III
KF2 (118100)	autosomal dominant	C2-3 fusion And possible craniofacial anomalies	Types I, II and III
KF3 (613702)	reduced penetrance	singular isolated fusion, most rostral at C3	Type II
KFS4	X-linked inheritance	vertebral fusion and ocular anomalies	Commonly referred to as Wildervanck syndrome

Associations

- Sprengel deformity of the shoulder
- Wildervanck syndrome
- Anomalies of the aortic arch and branching vessels, e.g. carotid, subclavian arteries
- Spinal Scoliosis
- Antervertebral disc herniation (Ulmer *et al*.,)
- Cervical spondylosis (Ulmer *et al*.,)
- Renal abnormalities, e.g. unilateral renal agenesis (Yuksel *et al.*, 2006)

Conclusion

We insist that clinical heterogeneity and radiographic abnormalities found in Klippel-Feil syndrome may simulate acute pathology and thus require comprehensive evaluation and delineation of diagnostic and prognostic classes. Lastly, the potential for other abnormalities should be considered and evaluated through appropriate referral.

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