

Case Report

Klippel-Feil Syndrome: A Case Report

Dr. Abdullah Al Saleh

Department of Family Medicine and Primary Care, National Guard Comprehensive Specialized Clinic, King Abdulaziz Medical City, Riyadh, Saudi Arabia

ABSTRACT

Klippel-Feil Syndrome (KFS) is defined as a congenital fusion of two or more cervical vertebrae. The most common signs are short neck, low hairline at the back of the head, and restricted mobility of neck. I report on a one year-old Saudi female who presented for a routine one-year vaccination and was later diagnosed with a case of KFS.

Keywords: Klippel-Feil syndrome, Posterior hairline, Cervical vertebra, Short neck.

INTRODUCTION

KFS is a rare disease, initially reported in 1912 by Maurice Klippel and Andre Feil from France (Klippel and Feil, 1912) and characterized by the congenital fusion of two or more cervical vertebrae. The incidence is 1:40000 in 1:42000 births. [1] Females are affected slightly more often than males. [2] KFS, or synostosis of the cervical spine, occurs as a result of failure in normal segmentation of cervical mesodermal somites during embryonic development. This failure occurs at two to eight weeks of gestation yet its cause is unknown. [3] The most common signs are short neck, low posterior hairline, and restricted mobility of upper spine. Associated abnormalities may include scoliosis or kyphosis, Sprengel deformity, hemivertebrae, basilar impression, spina bifida, anomalies of the kidneys and the ribs, cleft palate, respiratory problems, deafness or hearing impairment, and heart malformations. [3]

Most patients who have this syndrome are first seen due to restricted motion of the neck, torticollis, webbing of the neck, or Sprengel deformity. [4]

KFS is listed in the Online Mendelian Inheritance in Man database as being of sporadic autosomal dominant inheritance with reduced penetrance and variable expression. Almost all cases of this syndrome occur sporadically; nevertheless, close evaluation of the immediate family is recommended.

CASE REPORT

A one-year-old Saudi female was seen in the pediatric clinic in National Guard Comprehensive Specialized Clinic, Riyadh, Saudi Arabia. Her visit to the clinic was for a routine one-year vaccination. No past medical or family history of significance. On physical examination, she was noticed to have a short neck with low hair line, torticollis, and Sprengel deformity of the left shoulder (Figure 1). The growth parameters were appropriate for her age. The cardiovascular system and abdominal examinations were normal as well. Further investigations were done for the child. A radiography showed a C2-C3 fusion of the cervical spine (Figure 2), with the thoracolumbar spine being normal, and renal ultrasound revealed ectopic right kidney. Hence, a clinical diagnosis of KFS

was made. Other relevant investigations such as echocardiogram and a hearing test were normal.



Figure 1 : Short neck with low hair line, torticollis, and Sprengel deformity of left shoulder



Figure 2 : fusion of cervical vertebrae

DISCUSSION

KFS is a congenital disorder with fusion of cervical vertebra, which may be familial or sporadic. Classical clinical triad of KFS is lower posterior hair line, short neck, and restriction of head and neck movements. This classical triad is seen in about 40-50% of patients, while the most common finding is restriction of movements. [4-7]

Feil classified the syndrome into 3 categories: Type 1 includes cases with fusion of cervical vertebrae; Type 2 includes cases with fusion of cervical and thoracic vertebrae, along with an associated cervical hemi vertebrae anomaly and abnormal fusion of the atlantoaxial joint with the occiput. Type 3 KFS is characterized by the fusion of cervical, thoracic, or lumbar vertebrae with associated rib anomalies. [8] Updated classification (by Clarke et al.) grouped patterns of inheritance, associated anomalies, and the axial level of the most anterior fusion (Table 1).

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

Classification of Klippel-Feil syndrome	Inheritance	Vertebral fusion and associated anomalies	Overlap with Klippel and Feil's original classification
KF1	Autosomal recessive	Rostral fusion at C1 and severe associated anomalies (short neck, cardiac defects, and craniofacial anomalies)	Types I, II, and III
KF2	Autosomal dominant	C2-3 fusion and possible craniofacial anomalies	Types I, II, and III
KF3	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

Congenital heart defects may sometimes accompany KFS at 4%-14%. The most common cardiac anomalies associated with KFS are ventricular septal defect and aortic arch anomalies.

As the cervical vertebrae and genitourinary system differentiate during the same embryological period, urinary system abnormalities may be encountered in 30-35% of KFS cases, the most common being unilateral renal agenesis. [9] Therefore, urinary ultrasonography and intravenous pyelography, if necessary, should be performed on KFS patients to eliminate

renal anomalies; this includes agenesis, double collecting system, horseshoe shaped kidneys, renal ectopia, or hydronephrosis.

Treatment for KFS is symptomatic and may include surgery to relieve cervical or craniocervical instability and constriction of the spinal cord, and to correct scoliosis. Physical therapy may also be useful. [10]

CONCLUSION

KFS is a very rare congenital disorder, and is often accompanied by sporadic clinical diagnosis. A thorough clinical examination and investigation are

needed to detect associated deformities associated. Treatment of this syndrome is geared towards specific symptoms that present in each individual. Parental counseling should be done in all cases.

REFERENCES

1. Thomsen MN, Schneider U, Weber M et al. Scoliosis and congenital anomalies associated with Klippel-Feil syndrome types I-III. *Spine*. 1997;22:396-401.
2. Floemer F, Magerkurth O, Jauckus Cet al. Klippel-Feil syndrome and Sprengel deformity combined with an intraspinal course of the left subclavian artery and a bovine aortic arch variant. *Am. J Neuroradiol*.2008;29(2):306-7.
3. Fernandes T& Costa C. Klippel-Feil syndrome with other associated anomalies in a medieval Portuguese skeleton (13th-15th centuries). *J Anat*. 2007;211:681-5.
4. Heninger RN, Lang JE, Macewen GD. Klippel-Feil Syndrome. A constellation of associated anomalies. *J Bone Joint*. 1974;56-A:1246-53.
5. Guille JT & Sherk HH. Congenital osseous anomalies of the upper and lower cervical spine in children. *J Bone Joint Surg*. 2002;84:277-88.
6. Tracy MR, Dormans JP, Kusumi K. Klippel-Feil syndrome: clinical features and current understanding of etiology. *Clin Orthop Relat Res*. 2004;424:183-90.
7. Van Kerckhoven MF & Fabry G. The Klippel-Feil syndrome: a constellation of deformities. *Acta Orthop Belg*. 1989;55:107-18.
8. Samartzis DD, Herman J, Lubicky JP et al. Classification of congenitally fused cervical patterns in Klippel-Feil patients: epidemiology and role in the development of cervical spine-related symptoms. *Spine*. 2006;31(21):E798-804.
9. Moore WB, Matthews TJ, Rabinowitz R. Genitourinary anomalies associated with Klippel-Feil syndrome. *J Bone Joint Surg*. 1975;57(3):355-57.
10. Samartzis DD, Herman J, Lubicky JP et al. Classification of congenitally fused cervical patterns in Klippel-Feil patients: epidemiology and role in the development of cervical spine-related symptoms. *Spine*. 2006;31:E798-804.

How to cite this article: Al Saleh A. Klippel-Feil syndrome: a case report. *Int J Health Sci Res*. 2017; 7(4):474-476.
