

Case Report

Klippel-Feil Syndrome: A Case Report

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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.

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