

Klippel-Feil Syndrome with Situs Inversus - A Rare Association

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ABSTRACT

Klippel-Feil Syndrome (KFS) is a congenital anomaly characterized by a defect in the formation or segmentation of the cervical vertebrae. The clinical triad consists of short neck, low posterior hairline and limited neck movement. Multiple congenital anomalies have been associated with this disease. This is a case of KFS in a young girl along with situs inversus, which is an extremely rare association. Various systemic associations occurring in this multi-system disorder are also discussed.

Key words: *Klippel-Feil syndrome. Situs inversus. Sprengel's deformity.*

INTRODUCTION

Klippel and Feil in 1912, first described the disorder in a 46-year-old French man who had a short immobile neck with massive fusion of cervical and upper thoracic vertebrae,¹ which came to be known as the Klippel-Feil syndrome. Later on, other anomalies were described in association with it thus making it a multi-system congenital disorder. It is characterized by fusion of cervical vertebrae as a result of defect in their formation or segmentation. The resulting triad consists of short neck, low posterior hairline, and limited neck movement, but less than 50% of patients demonstrate all the three clinical features.² The importance of recognizing Klippel-Feil syndrome lies in the fact that there is strong association of this anomaly with other significant conditions like scoliosis and/or kyphosis (60%), Sprengel's deformity (30%), torticollis, urinary system abnormalities (35%), loss of hearing (30%), facial asymmetry and flattening of neck (20%), synkinesis or mirror movements (20%), congenital heart diseases (4.2-14%), brain stem anomalies, congenital cervical stenosis, adrenal aplasia, ptosis, lateral rectus muscle paralysis, facial nerve paralysis, syndactily, diffuse or focal hypoplasia in upper extremities and agenesis of lungs and gallbladder may also be seen.^{2,3}

We present here a 5-year-old girl, who presented with reduced mobility of neck, Sprengel's deformity and torticollis. On diagnostic work-up, she was found to have situs inversus, an extremely rare association.

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CASE REPORT

A 5-year-old girl presented to the outpatient department with complaints of reduced mobility of the neck since early infancy. There were no other associated complaints. She was 3rd of five siblings from consanguineous parents. All were alive and healthy. On examination, she was a cooperative child. There was marked torticollis with the head tilted to right side. Her neck was short with prominent webbing. She was unable to turn her head to either side. There was Sprengel's deformity of the left scapula, which was placed higher and was more prominent (Figure 1). Examination of the musculoskeletal system, spine and motor system revealed no abnormality. Height was 102 cm (25th centile) and weight was 13kg (< 3rd centile). Upper and lower segment of the body both measured 51cm and the arm span was 103 cm. Her apex beat was palpable in right 4th intercostal space in mid clavicular line. Examination of abdomen revealed that liver was palpable on right side. X-ray cervical spine revealed congenital block vertebrae involving C2-C3 and C5-C6 with a characteristic "wasp waist" appearance at the line of fusion.

X-ray of the spine showed multiple wedge, butterfly and hemi-vertebrae in the upper thoracic region. Mild scoliosis of thoracic spine was also evident with concavity to the right side. Chest X-ray revealed that cardiac apex was on right side with stomach bubble visible on the left side. There was abnormal spacing and orientation of ribs secondary to vertebral anomalies (Figure 2). Left scapula was small, equilateral in shape and located superiorly, consistent with Sprengel's deformity. No omovertebral body was visible.

Electrocardiography findings were consistent with dextrocardia. Ultrasonography of the abdomen revealed situs inversus. No genitourinary abnormality was detected. Complete blood count, renal and liver functions were within normal limits. CT scan of the chest and abdomen showed situs inversus but no other structural abnormality was detected.



Figure 1: Short webbed neck, torticollis and Sprengel's deformity.

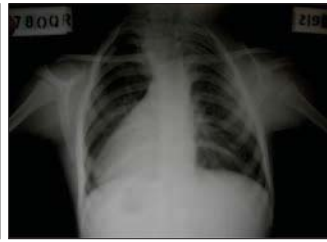


Figure 2: Dextrocardia, left sided stomach bubble and scoliosis.

DISCUSSION

It has been hypothesized that Klippel-Feil syndrome results from the interruption of the early embryonic blood supply in the subclavian arteries, the vertebral arteries and/or their branches. The term Subclavian Artery Supply Disruption Sequence (SASDS) was suggested for this group of birth defects.⁴ Most cases are sporadic, like the present case.

The syndrome has been classified into various types. Gunderson classified into three types.⁵ Type-I (autosomal recessive) consists of massive fusion of many cervical and upper thoracic vertebrae into bony blocks; type-II (autosomal dominant) has fusion at only one or two interspaces (although hemivertebrae, occipitoatlantal fusion, and other anomalies might be associated); and type-III (autosomal recessive) comprises both cervical fusion and lower thoracic or lumbar fusion. Raas-Rothschild suggested the existence of a fourth type of Klippel-Feil anomaly associated with sacral agenesis.⁶ Clarke⁷ have reclassified Klippel-Feil syndrome into four classes: KF1 (autosomal recessive) with C1 fusion; KF2 (autosomal dominant) with fusion of C2 and C3; KF3 (autosomal recessive) with marked fusion of C3; KF49 (X-linked) with cervical fusion and eye anomalies. The present case with multiple fusions of cervical vertebrae and various anomalies including hemi and wedge vertebrae of the upper thoracic spine falls in the type-II of the Gunderson classification.

There is strong association of this condition with other anomalies. In a study of 23 cases, scoliosis and fusion of the cervical vertebrae (between 2-5 vertebrae), low hairline and short neck were found in all the cases. Lumbar fusion was detected in one patient. Other findings included renal agenesis in one patient, different types of hearing loss in 9 patients, cardiac pathologies in 5 patients, epilepsy in one patient, and mirror movements in 2 patients.⁸

Renal anomalies are common in individuals with Klippel-Feil syndrome, and they can be quite serious. Minor renal anomalies include a double collecting system, renal ectopia, and bilateral tubular ectasia and major renal anomalies include hydronephrosis, absence of kidney, and horseshoe kidney.⁹

Deafness is a well-known feature of KFS and may be of sensorineural, conductive, or mixed type. Extensive audiological evaluation of this patient revealed no sensorineural or conductive deafness in our patient.

Cardiovascular anomalies, mainly septal defects, were found in 7 patients in Hensinger's series, with 4 of those individuals requiring corrective surgery.⁹ Situs inversus is a very rare association only once reported before in literature in association with compressive myelopathy from India.¹⁰ In this case, situs inversus was the only systemic abnormality.

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