KLIPPEL-FELL SYNDROME: A CASE REPORT AND REVIEW OF THE LITERATURE

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SUMMARY: Klippel-Feil Syndrome is clinically well recognized and commonly associated with many congenital malformations. Skeletal anomalies represent a common feature of this syndrome. This case report describes a rarely seen Type I Klippel-Feil Syndrome with a cervical block vertebrae.

Key Word: Klippel-Feil Syndrome

INTRODUCTION

Klippel-Feil Syndrome (KFS) was originally described in 1912 by Klippel and Feil in a patient with a shot neck, low hairline and severly restricted neck movement. At autopsy this lack of neck mobility was found to be due to the total fusion of the cervical spine. More recently Harris stated that the KFS refers to patients with any degree of cervical fusion, only a minority of whom will have all three original clinical findings described in 1912 (9).

The classic triad of low posterior hairline, short neck and limitation of neck motion is present in only 50% of patients with KFS. Restriction of neck motion is the most common finding (14). Numerous other abnormalities recorded in association with this syndrome have been collected by Hensinger *et al.* (10): renal anomalies, Sprengel's deformity (congenital upward displacement of the scapula), deafness, facial asymmetry, webbing of the neck, congenital heart disease and synkinesis or mirror movements.

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Following their initial description of the syndrome, Klippel and Feil proposed a classification based on the site and extent of the cervical fusion. Type I applied to patients with extensive cervical and upper thoracic spinal fusion. These patients have the classical triad and often have other anomalies elsewhere. This type is rare and sporadic occurrence. Type II referred to patients with one or two interspace fusions, often associated with hemivertebrae and occipitioatlantal fusion. This class is probably 50 times more common than Type I. Type III classified individuals with both cervical and lower thoracic or lumbar fusion. Type II is considered the most usual and asymptomatic form of this anomaly, and the C_{2-3} and C_{5-6} interspaces are most often fused (23).

In this report, we describe a KFS patient who has a block vertebrae between C_2 and C_7. We classify this patient as Type I which is rarely seen.

CASE REPORT

A 40 year-old married woman admitted to Physical Medicine and Rehabilitation Department of Hacettepe

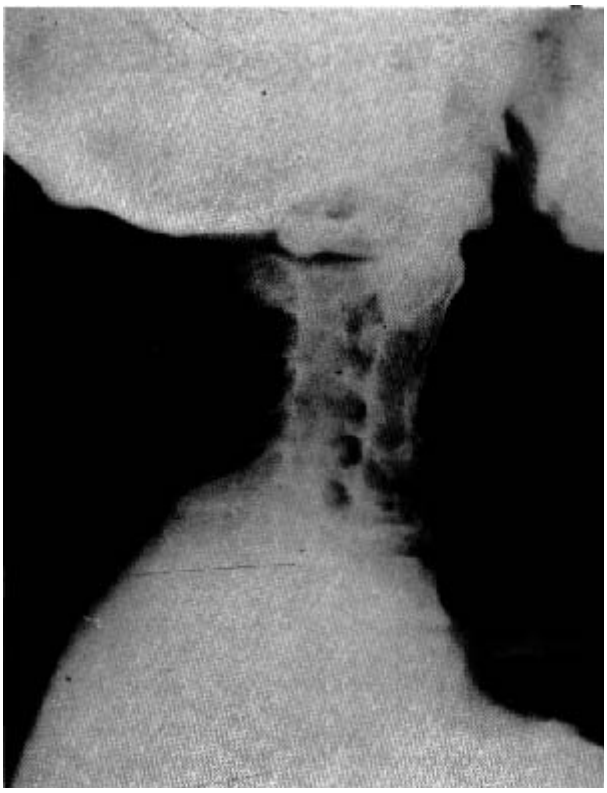

Figure 1: Lateral view of the cervical spine demonstrating the fusion between C_2 and C_7.

University Hospital in May 1988 with the complaints of severe neck pain and restricted neck movements. She has carried the diagnosis of KFS since early childhood. The patient who has a negative family history, complains about neck pain for thirty years. In her past medical history, she has been referred to many hospitals and given nonseroid anti-inflammatory medication and physical therapy.

When she first admitted to our department, on examination she had severly limited neck movements, dorsal kyphosis, short neck and low hairline. Her neurological examination was normal. Compression tests were negative. She had no musculoskeletal abnormalities except the restricted left elbow extension because of a previous trauma sequel. On her cervical radiological examination, a block vetebrae between C_2 and C_7 was seen. In order to find out probable associated anomalies, she was referred to otolaryngology, urology, gynecology and internal medicine departments. All her further physical examination and specific tests were normal (Figure 1).

After ten sessions of physical therapy, her pain diminished and she returned to her job as a secretary.

DICUSSION

The patient described here is similar to the one which was reported by Klippel and Feil in 1912. Except the classical triad, no associated anomaly could be found.

Visceral congenital anomalies associated with this syndrome may be the greatest threat to these patients. Cardiovascular anomalies have been recognized in patients with KFS. An incidence of 42% to 14% has been reported by several authors. Various lesions occur, but ventricular septal defects are the most common (19). Associated genitourinary anomalies have been identified in 64% of patients by Moore (20). Congenital absence of the uterus and upper vagina, as well as other abnormalities involving the genital tract have also been recognized (1). Kords reported a KFS associated with Rokitansky-Küstner Syndrome (vaginal aplasia, rudimentary uterus) renal aplasia and focomelia (15). The high incidence of genitourinary anomalies in patients with KFS has been defined several times. Unilateral renal agenesis is the most common finding, but rarely the crossed fusion of renal pelvis may occur and is usually associated with fusion of the kidneys (8). Jalladeau first reported the association of deafness with KFS. Since then, several reports of other otolaryngological anomalies have appeared (12). It is reported that the underdevelopment of the bony and membranous labyrinth, cochlea and auditory nerve causes deafness (18). A rare case which always destroys the hearing of the patient is described by Daniidilis: Stapes gusher. The stapes gusher which is the most dramatic complication of stapedectomy arises from an abnormal communication between the subarachnoid and perilymphatic spaces. This congenital abnormality of the labyrinth, may be associated with other anomalies such as the KFS (5).

A variety of ocular disorders have been recognized in association with KFS. Wildervanck first described the cervico-oculo-aoustic dysplasia usually seen in females with the disease (23). Cleft palate, mandibular molformations and micrognathia have also been described in patients with KFS. It is considered that fusion of the cervical vertebrae causes the mandible to remain compressed against

the chest and consequently forces the tongue to remain between the palatal shelves, at a time palatal closure would normally be taking place. This would tend to lead to the formation of a palatal cleft but with no interference in the normal development of the lip and alveolus (4). The basis of many facial congenital abnormalities begins with maldevelopment of neural crest tissue that give rise to much of the skeletol and connective tissue primordial of the face. This embryonic dysmorphogenesis can be induced by infectious diseases, drugs and chemical agents (2).

Skeletal anomalies represent a common feature of the syndrome and include skullasymmetry, platybasia, basillar invagination and brachycephaly. A 60% incidence of significant scoliosis was reported. Sprengel's deformity has been identified in 25% to 35% of the patients. Radial and ulnar hypoplasia, finger and toe deformities, and the presence of an omovertebral bone are rarely encountered anomalies (3).

In patients with KFS, dissociate movements of the two hands were identified by several authors. An impaired pyramidal tract decussation has been proposed as a possible underlying mechanism by some investigators (22).

A case of multiple meningiomas confined solely to the cervical spinal cord association with multiple bony abnormalities of the cervical spine is presented by Holliday *et al.* It is suggested that multiple meningiomas unassociated with other central or peripheral tumors may present a distrinct clinical entity and may occur in an as yet uncharacterized familial pattern (11).

Although most cases with KFS are sporadic, there can be also rather rare familial cases. Especially there are a number of familial cases of KFS associated with Sprengel's deformity, omovertebral bone, supernumerary digits and patent ductus arteriosus (23,26).

Some of these familial occurrence are atributed to single factor inheritance such as the familial $C_{2\text{-}3}$ fusion which may be an autosomal dominant trait at times. The evidence for a reccesive form of cervical vertebrae fusion is less convincing and to data some exogenous factors have been proposed as causative agents (16,23).

Lowry described a brother and sister with malformations and handicaps consistent with both the KFS and the Fetal Alcohol Syndrome. The mother was known to be a chronic alcoholic throughout both pregnancies. He suggested that these anomalies are not purely fortuitous but rather maternal alcoholism may cause errors in cervical vertebrae segmentation (16).

Patients with KFS are often at high risk for neurological injury. The cervicomedullary junction and cervical spinal cord are especially vulnerable. Three potentially unstable cervical fusion patterns are described: fusion of C_2 and C_3 with occipitalization of the atlas, a long fusion with an abnormal occipitocervical junction and a single open space between two fused segments. Commonly, the risks for neurological damage are due to abnormalities other than the fusion pattern of the cervical vertebrae. These include abnormalities at the occipitoatlantal junction, spinal canal stenosis and scoliosis. With the inhertent tendency in individuals with cervical fusion for the development of spondylosis and hypermobility of the adjacent segments, a congenitally stenotic canal provides a narrow margin of safety for any additional encroachment on the canal (24).

The importance of the KFS lies in the secondardy effects produced on the nervous system. There can be symptoms like weakness, spasticity, staggering, dysmetria, nystagmus, sensory loss and lower cranial nerve involvement due to progressive cord and brain stem compression (7, 25).

It is known that the deformities of KFS increases the probability of spinal cord injury with trauma. The marked deformities of the cervicothoracic spine result in mechanical distortion altering the compensatory properties of the spine to react to decelerating and rotatory forces (27).

Especially when there is multiple block vertebrae, as in our patient, the normal segments may become hypermobile and subjected to significantly increased stress. Potentially fatal subluxations may then occur at these levels (6). Sudden neurologic compromise or death following minor trauma have been reported in the KFS (17,21). The mobile articulations are under greater mechanical demands and are less capable of protecting the patients against traumatic injuries. Therefore we believe that these patients, like described here, should be guided to minimize activity that can be potentially harmful. If there is no need for surgical correction of associated anomalies, patients with KFS can be treated semptomatically with NSAI medications and physical therapy.

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