

Cervical Myelopathy in Klippel-Feil Syndrome

Shu-Yi Chen¹ and Pao-Yu Wang^{1,2}

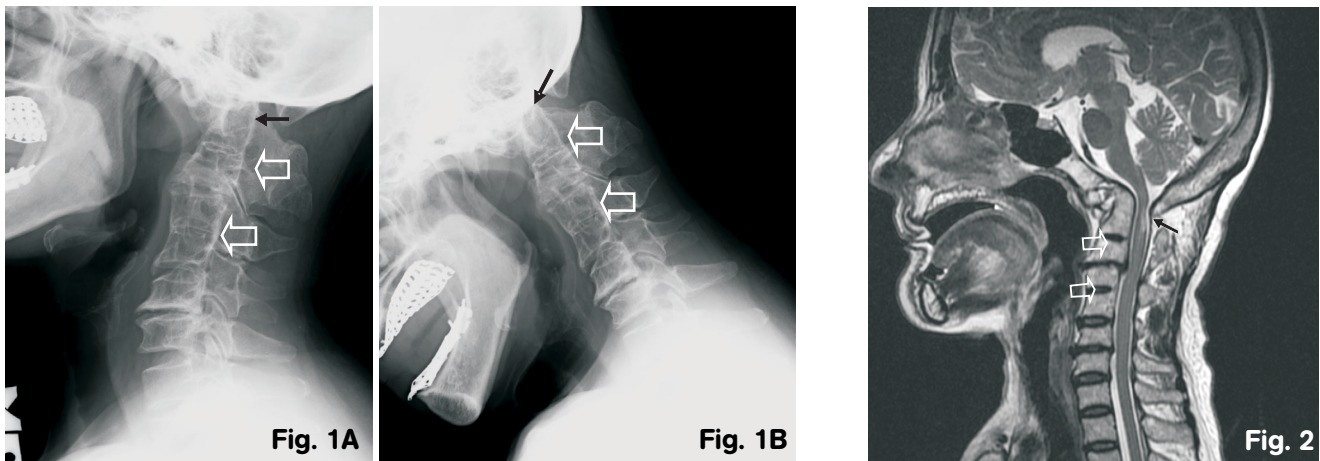


Figure 1. Cervical spinal X-rays. (A) lateral view extension. (B) lateral view flexion. Small dark arrow: Non-visualization of the C1. Limitation of neck range of movement was noted also. Large white arrow: fusion of C2-3, C4-5.

Figure 2. MRI of cervical spine, T2 sagittal view. Small dark arrow: non-visualization of the C1 vertebra with enlargement of the opisthon and basion and small size of the C2 vertebra, subluxation over the cranio-cervical junction with narrowing of the foramen magna and compression to the adjacent spinal cord. High signal intensity over C1-2 cord. Large white arrow: Fusion of the spinal process between C2 and C3, C4 and C5

A 65-year-old lady was admitted due to progressive weakness and hypethesia over limbs and trunk for one year. Tracing her history, she was relatively well before. She has had short neck since childhood and is 143 cm in height. There has been no major trauma, no surgery history, nor systemic disease. There is limited neck extension and rotation. Neurological examination revealed normal cranial nerve function. The muscle strength was grade 4 over limbs without muscle atrophy. Deep tendon

reflexes were increased bilaterally with positive bilateral Hoffman signs. The bilateral plantar responses were of flexor type. There were diminished pinprick and temperature sensation below C4 dermatome, but normal joint position and vibration. There was no cerebellar sign, no Lhermitte's sign, nor sphincter dysfunction.

Plain cervical spine X-ray showed absent C1 vertebra, fusion of C2-3, C4-5 (Fig. 1). Cervical magnetic resonance imaging (MRI) disclosed non-visualization of

From the ¹Section of Neurology, Department of Internal Medicine, Taichung Veterans General Hospital; ²School of Medicine National Yang-Ming University, Taiwan.

Received February 29, 2008.

Revised and Accepted March 14, 2008.

Reprint requests and correspondence to: Pao-Yu Wang, MD, Section of Neurology, Department of Internal Medicine, Taichung Veterans General Hospital, No. 160, Sec. 3, Taichung-Kang Road, Taichung, Taiwan.

E-mail: pywang@vghtc.gov.tw.

the C1 vertebra with enlargement of the opisthon and basion and small sized C2 vertebra with upward migration of the C2 odontoid process with basilar invagination (Fig. 2). Fusion of the spinal process between C2 and C3, C4 and C5 was noted. High signal intensity over C1-2 cord was revealed in T2 image. Cervical myelopathy at C1-2 was impressed. Electromyography and nerve conduction velocity of four limbs, and somatosensory evoked potential of median nerve were normal. No lumbar spine abnormality was noted in lumbar X-ray. Klippel-Feil syndrome type II with cervical myelopathy was impressed. Decompression surgery was suggested, but the patient refused.

DISCUSSION

Klippel-Feil Syndrome (KFS) is the congenital fusion of at least two of the seven vertebrae in the cervical spine, first described by Klippel and Feil in 1912. KFS is detected throughout life, often as an incidental finding. There may also be fusion or anomalies of vertebrae in the thoracic or lumbar spine. Short neck, reduced cervical range of motion, and a low posterior hairline are the most common features of the syndrome. Patients with upper cervical spine involvement tend to present at an earlier age than those whose involvement is lower in the cervical spine. Most patients present with a short neck and a decreased cervical range of movement, with a low hairline occurring in 40-50% of patients. Decreased range of motion is the most frequent clinical finding. Rotational loss usually is more pronounced than the loss of flexion and extension.

Estimated frequency of occurrence varies widely from 1 in 100 to 1 in 20,000 births^(1,2), about 65% of cases are females. Many people with this syndrome are never diagnosed due to minor symptoms or are asymptomatic throughout life. In persons with certain high-risk patterns of cervical fusion are at greater danger of sudden neurological injury or death after minor trauma.

In 1912, Feil subsequently classified the syndrome into 3 categories: Type I - a massive fusion of the cervi-

cal spine; Type II - the fusion of 1 or 2 vertebrae; Type III - the presence of thoracic and lumbar spine anomalies in association with type I or type II Klippel-Feil syndrome. In 2006, Samartzis and colleagues⁽³⁾ suggested a new classification system: type I patients were defined as having a single congenitally fused cervical segment; type II patients demonstrated multiple noncontiguous, congenitally fused segments; type III patients had multiple contiguous, congenitally fused cervical segments. Axial neck symptoms such as neck pain, stiffness and restriction of neck movement were highly associated with Type I patients, whereas predominant radicular and myelopathic symptoms occurred in Type II and Type III patients.

Klippel-Feil syndrome should be considered if patient has the characteristics of short neck, reduced cervical range-of-motion, and a low posterior hairline plus cervical myelopathy or radiculopathy. Plain cervical X-ray is the basis for diagnosis and computed tomography scanning with 3-dimensional reconstruction can be valuable in assessing anatomy. Magnetic resonance imaging is indicated in patients with neurological deficits. Besides, it may combine with other bony abnormality. So the chest, thoracic and lumbar spine X-rays are needed too.

The prognosis for most individuals with Klippel-Feil Syndrome is good if the disorder is treated early and appropriately. Activities that can injure the neck should be avoided.

References:

1. Gjorup PA, Gjorup L. Klippel-Feil's syndrome. *Dan Med Bull* 1964;11:50-3.
2. Brown MW, Templeton AW, Hodges FW III. The incidence of acquired and congenital fusions in the cervical spine. *Am J Roentgenol* 1963;94:1255-9.
3. 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;31:798-804.