

Klippel-Feil Syndrome and Thoracic Outlet Syndrome

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CLINICAL IMAGE

Klippel-Feil Syndrome (KFS) is a rare disease that was firstly described in 1912 by Maurice Klippel and Andre Feil. It is a bone disorder recognized by the abnormal fusion of two or more spinal bones, it seem in the cervical vertebrae. It has three major features which are short neck, limited age of motion in the cervical spine and low hairline at the beck [1].

We report the case of a 26-year-old male patient was admitted to cardiology clinic with complaint of left arm and chest pain. Mitral valve prolapse detected by transthoracic echocardiography. Patient was referred to physical therapy and rehabilitation clinic due to cervical scoliosis and the short neck. The diagnosis of KFS was established by the radiographic abnormalities with fusion defect at cervical spine, hemivertebrae and rib defects (Figure 1). Monophasic flow pattern showed by doppler examination during abduction of upper limbs, so the patient was diagnosed bilateral thoracic outlet syndrome additionally. The occurrence of thoracic outlet syndrome was thought to be the reason of cervical rib which can be seen at KFS (Figure 2).



Figure 1: Radiograph of the cervical spine (later view).

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Figure 2: ACT scan of the cervical vertebrae and 3D reconstructed image of the vertebrae and hemivertebrae (arrow) and bilateral cervical costa (asterisk).

Scoliosis (%60) is the most common associated anomaly in patients with KFS, followed by spina bifida, Sprengel's deformity, urinary tract problems, and congenital cardiovascular anomalies [2]. In conclusion KFS should be considered in the differential diagnosis of patients with cervical scoliosis and short neck. It should be taking for that upper extremity pain may be due to not only the cardiovascular diseases accompanying KFS, but also the disease like thoracic outlet syndrome.

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