Klippel-Feil Syndrome with associated meningocoele and Sprengel deformity

Clinical History:

We present the case of a 12-month-old infant, whose parents consulted us due to the infant's short neck with decreased mobility of the cervical spine associated with psychomotor retardation.

Imaging Findings:

A cerebro-spinal MRI was performed to explore the shortness and fixity of the child’s neck. The MRI showed:

* Fusion of the anterior arc of the first 3 vertebrae with agenesis of their posterior arcs associated with a reduced overall depth of the cervical vertebral bodies, left lateral cervical spine curvature and cervical spinal syringomyelia cavity [Figures 1, 2, 4].

* A butterfly aspect of the third dorsal vertebra without abnormality of the thoracic curve associated with an elevation of the right scapula [Figures 3, 4].

* Basi-occipital meningocoele through a posterior spinal canal dehiscence enlarging the foramen magnum associated with cerebellar vermis hypoplasia and ventricular dilatation, compatible with hydrocephalus [Figures 1, 5, 6].

From these findings we conclude a Klippel-Feil syndrome associated with occipitocervical meningocoele, a vermis hypoplasia, hydrocephalus and Sprengel syndrome.

Discussion:

Klippel-Feil syndrome (KFS) is an unusual skeletal malformation, also known as synostosis of the cervical spine. It was reported for the first time in 1912 and its prevalence is 1 in 50 000 [1, 2]. It is due to a lack of normal cervical somite segmentation in the third and eighth weeks of gestation, but the exact aetiology is unknown [2].

Diagnosis is often based on the classical clinical triad generally present at birth: short neck, low hairline and stiffness of the cervical spine [1, 2]. However, 50% of patients do not have this triad, which leads to a late discovery through symptoms of neck pain, decreased range of motion of the neck, and radiculopathy and/or myelopathy [1, 2].

Some frequently associated abnormalities must be investigated: primarily neurological defects (brain and spinal cord), static spinal disorders, the ascent of one or two blades (Sprengel syndrome), as well as visceral malformations (genito-urinary, cardiac, respiratory and hearing) that can influence the functional and vital prognosis [3, 4].

Imaging is based primarily on standard radiographs of the cervical spine which shows the level and modality fusion...
of the cervical vertebrae [1, 2]. Spine CT is often helpful for 3D reconstructions for a specific balance sheet when surgical treatment is indicated.

Cerebro-spinal MRI gives a detailed study of spinal abnormalities, intraductal and any associated brain abnormalities [5, 6]. Further investigations should be carried out in search of associated visceral anomalies (Lung Rx, abdominal-pelvic ultrasound, echocardiography etc.) The prognosis of this syndrome is variable.

Treatment is mostly symptomatic, but a healthy lifestyle should be maintained in order to prevent neurological complications due to accidents and injuries. Surgical treatment is indicated in cases of progression of neurological signs [1, 2, 7]. Imaging plays an important role in the diagnosis, the search for associated anomalies, monitoring and research of neurological complications.

**Differential Diagnosis List:** Klippel-Feil syndrome associated to a Sprengel syndrome and neurological abnormalities, Juvenile rheumatoid arthritis, Discitis, Post-surgical fusion

**Final Diagnosis:** Klippel-Feil syndrome associated to a Sprengel syndrome and neurological abnormalities

**References:**


Description: Klippel-Feil syndrome associated with occipitocervical meningocoele Origin: Hopital militaire d'instruction Mohamed V Rabat
Description: Occipitocervical vermis hypoplasia with meningocoele
Origin: Hopital militaire d'instruction Mohamed V. Rabat
Figure 3

Description: Klippel-Feil syndrome with Sprengel syndrome

Origin: Image origin: Hopital militaire d'Instruction Mohamed V. Rabat
Description: Lateral ventricular dilatation, compatible with hydrocephalus
Origin: Service de Radiologie. Hopital militaire d'instruction Mohamed V
Description: Cervical spinal syringomyelia cavity 

Origin: Service de Radiologie. Hopital militaire d'instruction Mohamed V
Description: Butterfly vertebra with elevation of the right scapula compared to left scapula

Origin: Service de Radiologie. Hopital militaire d'Instruction Mohamed V