Case 13171

Eurorad

Klippel-Feil Syndrome with associated meningocoele and Sprengel deformity

Published on 27.11.2015

DOI: 10.1594/EURORAD/CASE.13171
ISSN: 1563-4086
Section: Paediatric radiology
Area of Interest: Paediatric
Procedure: Diagnostic procedure
Imaging Technique: MR
Special Focus: Congenital Case Type: Clinical Cases
Authors: Bouachra S, Laamrani FZ, En-nouali H, Boumdin H, Amil T
Patient: 12 months, female

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

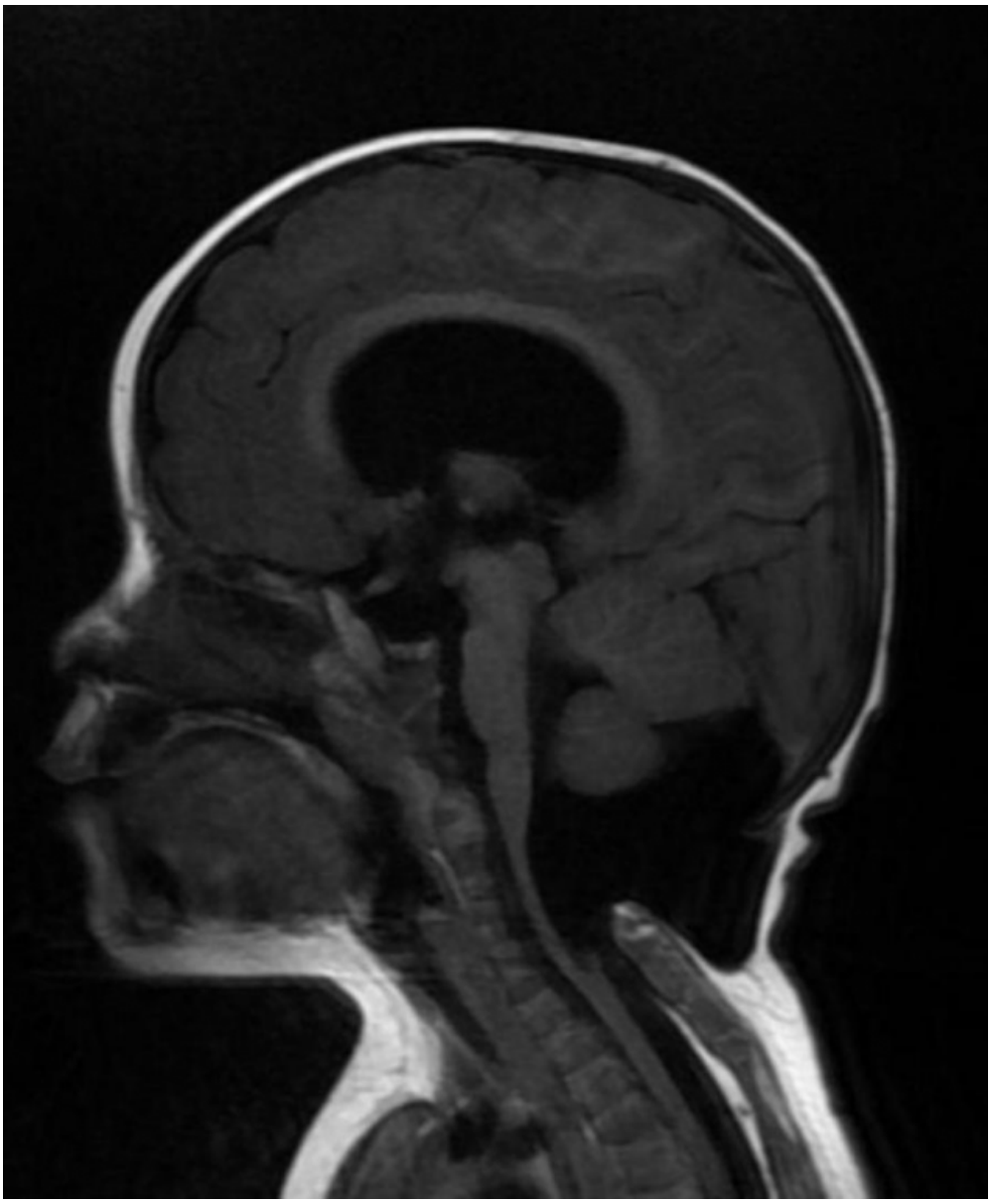
Final Diagnosis: Klippel-Feil syndrome associated to a Sprengel syndrome and neurological abnormalities

References:

Tracy MR, Dormans JP, Kusumi K (2004) Klippel-Feil Syndrome. Clin Orthop Relat Res 424:183–90 (PMID: 15241163)
O\'Donnel DP, Seupaul RA (2008) Klippel-Feil syndrome. The American Journal of Emergency Medicine 26(2): 252–252 (PMID: 18272131)
Samartzis D, Herman J, Lubicky JP, et al. (2007) Sprengel's deformity in Klippel-Feil syndrome. Spine 32:512–16 (PMID: 17700431)
Hensinger RN, Lang JE, MacEwen GD (1974) Klippel-Feil syndrome; a constellation of associated anomalies. J Bone Joint Surg Am 56(6):1246-53 (PMID: 4436358)
Yuksel M, Karabiber H, Yuksel KZ et-al. (2006) Diagnostic importance of 3D CT images in Klippel-Feil Syndrome with multiple skeletal anomalies: a case report. Korean J Radiol 6 (4): 278-81 (PMID: 16374086)
Karasick D, Schweitzer ME, Vaccaro AR ((199) The traumatized cervical spine in Klippel-Feil syndrome: imaging features. AJR Am J Roentgenol 170 (1): 85-8 (PMID: 9423605)
Vaidyanathan S, Hughes PL, Soni BM, Singh G, Sett P (2002) Klippel-Feil syndrome - the risk of cervical spinal cord injury: a case report. BMC Fam Pra 3-6 (PMID: 11985781)

Figure 1

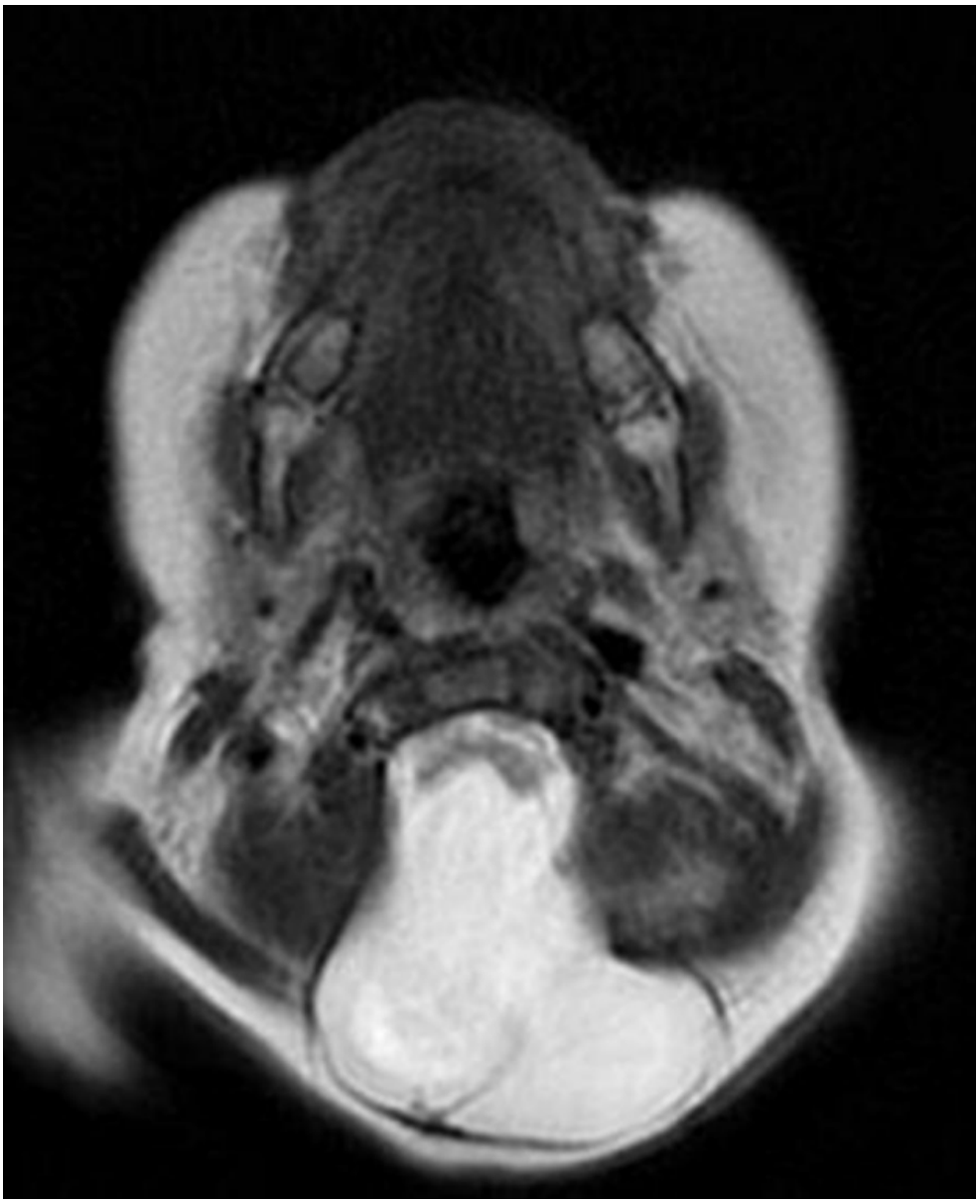
a



Description: Klippel-Feil syndrome associated with occipitocervical meningocoele **Origin:** Hopital militaire d\'instruction Mohamed V Rabat

Figure 2

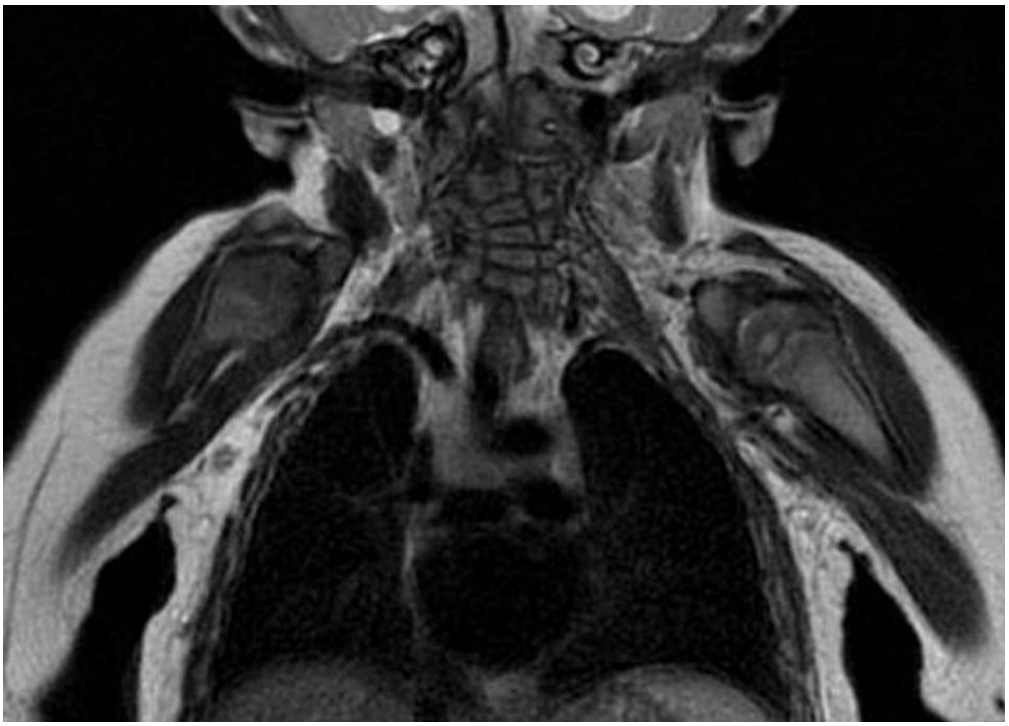
a



Description: Occipitocervical vermis hypoplasia with meningocoele **Origin:** Hopital militaire d\'instruction Mohamed V. Rabat

Figure 3

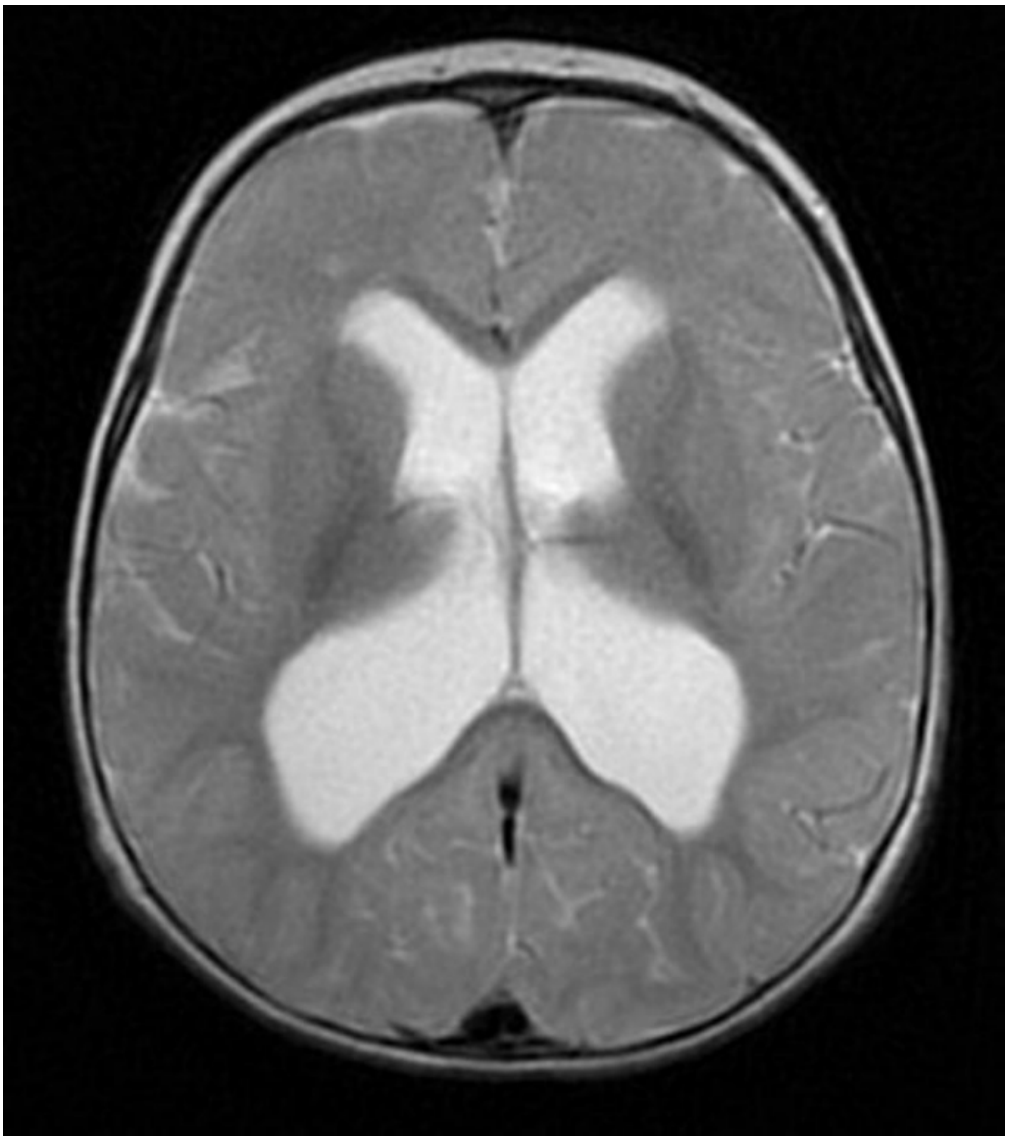
a



Description: Klippel-Feil syndrome with Sprengel syndrome **Origin:** Image origin: Hopital militaire d\'instruction Mohamed V. Rabat

Figure 4

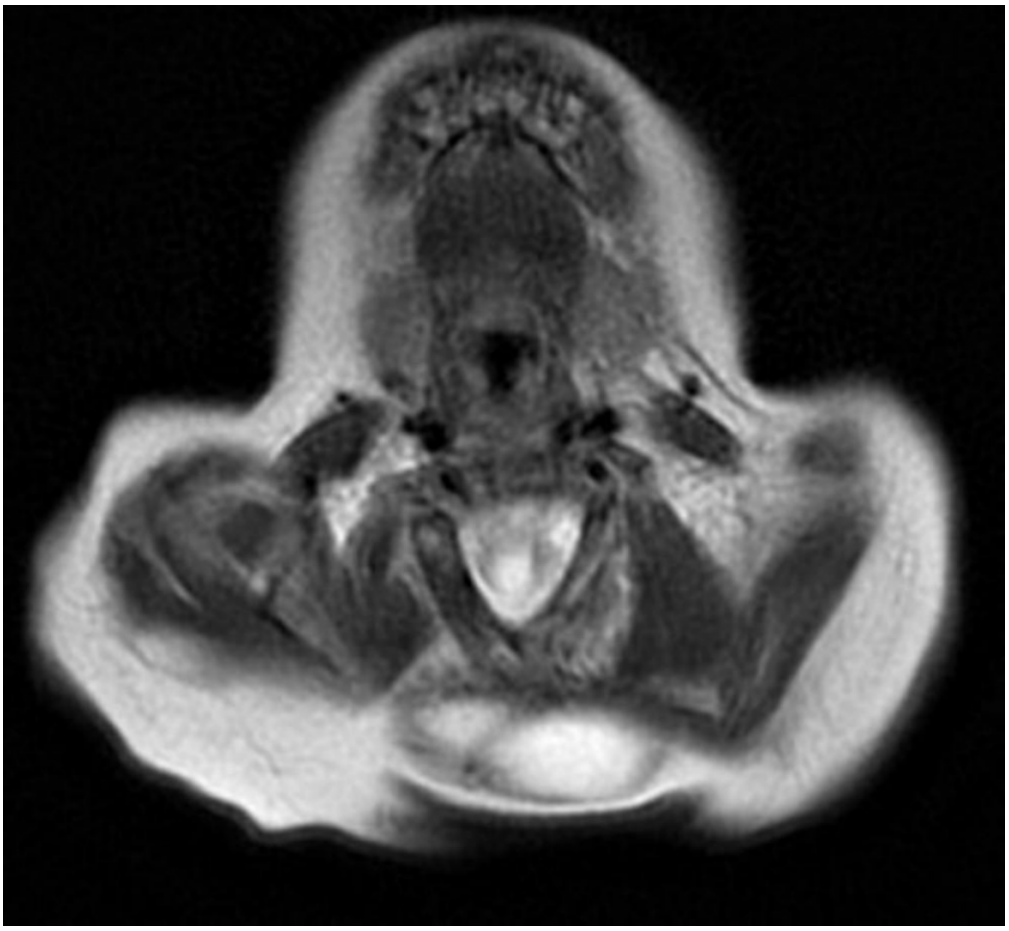
a



Description: Lateral ventricular dilatation, compatible with hydrocephalus **Origin:** Service de Radiologie . Hopital militaire d\'instruction Mohamed V

Figure 5

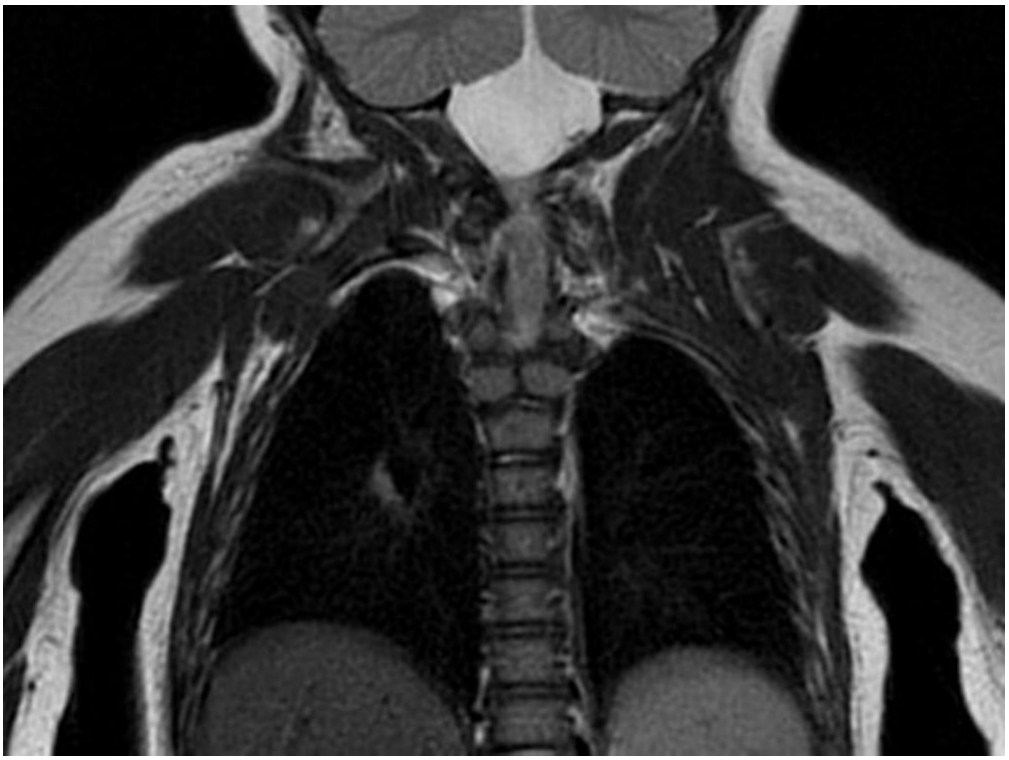
a



Description: Cervical spinal syringomyelia cavity **Origin:** Service de Radiologie . Hopital militaire d\'instruction Mohamed V

Figure 6

a



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