Caudal regression syndrome (CRS)  
Published on 17.10.2019  
DOI: 10.35100/eurorad/case.16515  
ISSN: 1563-4086  
Section: Musculoskeletal system  
Area of Interest: Musculoskeletal system  
Imaging Technique: MR  
Case Type: Clinical Cases  
Authors: AIKATERINI SOLOMOY1, VASILEIOS PATRIARCHEAS2, MINAS KOSTIS3, ANTONIOS PIKOULAS4, ASPASIA RIGOPOULOU5  
Patient: 2 years, male  
Clinical History:  
A 2-year-old male patient with abasia and deformities of lower extremities. He was the second child of a diabetic mother whose obstetric history showed absence of any formal prenatal examinations. Family history did not indicate any congenital disorders. Clinical examination revealed hypoplastic lower limbs with muscular paralysis and atrophy.  
Imaging Findings:  
A full spine MRI was performed on a 1.5 Tesla MRI scanner, consisting of T1 and T2 sequences in sagittal and axial plane. Paramagnetic contrast agent was administered intravenously. The MRI showed minor abnormalities of the spinal architecture with biconvex formation of the vertebral bodies C4 to C6 (Fig. 1) and L4 to L5. In addition, there was marked hypoplasia of the sacral vertebrae and agenesis of the coccyx (Fig. 2). The spinal cord was shorter than normal and terminated at the level of the T12. Moreover, the medullary cone was thickened and wedge-shaped (Fig. 3). Syringomyelia was recognised at the T4-T11 level and hydromyelia from the medullary cone up to the filum terminale. The cauda equina was thickened and fixed (Figs 3,4).  
Discussion:  
Caudal regression syndrome pathogenesis is thought to derive from a combination of underlying genetic predisposition and environmental factors. Genetic defects are thought to contribute in the pathogenesis of CRS, include mutations of CYP26A1, HOXD13 & HLXB9 genes [1]. Maternal diabetes mellitus is a well-established environmental risk factor and insulin-dependent diabetic women with poor glycemic control are 300 times more predisposed to have a child with CRS [2,3].  
In our case, there was a positive history of uncontrolled maternal diabetes mellitus, indicating its contribution to CRS pathogenesis. However, genetic testing was not performed due to patient’s rejection. The clinical presentation of CRS mainly depends on the level and the extent of the spinal cord dysplasia.  
Renshaw [4], introduced the classification of the syndrome into five types, based on the extent of the skeletal anomalies:  
Type I: Total or partial unilateral sacral agenesis.
Type II: Partial sacral agenesis with a bilaterally symmetrical defect and a stable articulation between the ilia and the first sacral vertebra, which can be normal or hypoplastic.

Type III: Variable lumbar and total sacral agenesis with the ilia articulating with the sides of the lowest vertebra present.

Type IV: Variable lumbar and total sacral agenesis, with the caudal endplate of the lowest vertebra resting above either fused ilia or an iliac amphiarthrosis.

Type V: Syringomyelia.

Severe cases of CRS can be diagnosed by observing the shorter than normal crown-rump length in antenatal sonography during the first trimester. MRI is the gold standard imaging technique for diagnosis of CRS and its deformities. According to imaging findings CRS patients are categorised into two groups:

Group 1: patients with a blunt conus medullaris which terminates above the normal level (L1); sacral deformities are more extensive in this group.

Group 2: patients with an elongated conus medullaris which terminated below the normal level, linked to a thickened filum terminale or intraspinal lipoma; the neurologic symptoms are more serious [5].

Treatment of patients with CRS is individualised, depending on the degree of the lower limb deformities and the presence of motor paralysis. When hydrocephalus and Chiari malformations are absent, attempts can be made to reconstruct the lower extremities to improve ambulation. Other syndromes and abnormalities associated with CRS are the VACTERL syndrome [6,7], as well as Currarino syndrome [8].

Learning Points:

- CRS is rare congenital abnormality arising from an insult before the fourth week of gestation and exhibits an extended range of malformation.

- MRI investigation is the gold standard the evaluation of CRS and its deformities.

- Treatment is decided per the clinical symptoms and the degree of the combine defects.

Written informed patient consent for publication has been obtained.

Differential Diagnosis List: Caudal regression syndrome (CRS), Sirenomelia, Meningocele, Lipomyelomeningocele

Final Diagnosis: Caudal regression syndrome (CRS)

References:


**Description:** 2-year-old male patient with caudal regression syndrome. Sagittal T2 WI depicted biconvex formation of C4 to C6 vertebral bodies (arrows)  
**Origin:** © Department of Radiology, General University Hospital of Patras, Greece, 2018
Description: 2-year-old male patient with caudal regression syndrome. Sagittal T1 WI shows low-signal filum terminale (arrows), hypoplasia of the sacral vertebrae and coccygeal agenesis (arrow)

Origin: © Department of Radiology, General University Hospital of Patras, Greece, 2018
Description: 2-year-old male patient with caudal regression syndrome. Sagittal T2 WI reveals wedge-shaped medullary cone, which terminates at the level of T12 (arrow) and syringomyelia at the thoracic spine (thin arrow) Origin: © Department of Radiology, General University Hospital of Patras, Greece, 2018
Description: 2-year-old male patient with caudal regression syndrome. Axial T2 WI shows syringomyelia at the level of T10 (arrow) Origin: © Department of Radiology, General University Hospital of Patras, Greece, 2018