

PHOTO QUIZ

What is your diagnosis?

A 14-year-old woman referred to our department for MR imaging to rule out a conus medularis lesion. She had history of incontinence and recurrent urinary tract infection since childhood.



Fig 1. Sagittal section on lumbosacral MRI, T1W FSE (Fast Spin Echo).

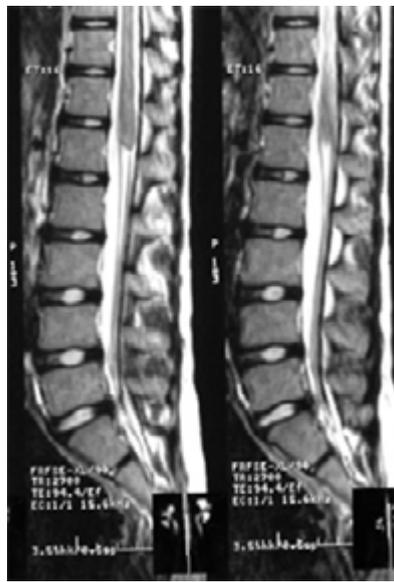


Fig 2. Sagittal section, T2W FSE (Fast Spin Echo).

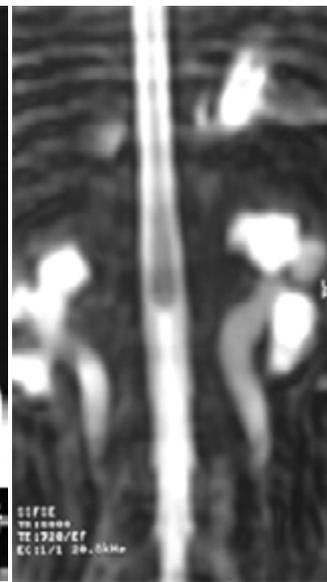


Fig 3. Coronal section, heavily T2W FSE (Fast Spin Echo).

What is your diagnosis?

Diagnosis: Caudal Regression Syndrome

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Caudal regression syndrome consists of the absence of caudal vertebrae, anal atresia, malformed external genitalia and renal abnormalities. Its prevalence is 1 in 7,500 live births, which 15–20% are infants of diabetic mothers and 1% of offsprings from diabetic mothers are affected.¹ Although most cases are sporadic, dominantly inherited forms as a result of a defect in the HLBX9 homeobox gene (chromosome 7) has been recently described. HLBX9 also expresses in pancreatic cells, which can describe the association between diabetic hyperglycemia and caudal regression syndrome.²

Common associations are VACTERL syndrome (10%), omphalocele, bladder exstrophy, imperforate anus, spinal anomalies (10%), and Currarino triad syndromic complexes. Patients with this syndrome should be investigated for neurological, urological and orthopedic complications.³

This syndrome includes a wide range of skeletal malformations, which range from agenesis of the coccyx to the absence of the thoracic spine below the T8 vertebral body. The spinal cord terminus is abnormal in 95% of the symptomatic patients. Commonly associated spinal cord abnormalities are tethered cord and thickened filum (with (/out) dermoids or lipomas).¹

Based on MRI findings, patients are divided into two groups: Group 1: vertebral body dysgenesis/hypogenesis, distal spinal cord hypoplasia ("wedge-shaped" cord termination) and severe sacra-

losseous anomalies. Group 2: Vertebral body dysgenesis/hypogenesis, tapered low-lying distal cord elongation with tethering, and other less severe sacral anomalies. Major differential diagnoses are tethered spinal cord, closed spinal dysraphism and occult intrasacral meningocele.³

Recommended imaging modalities are ultrasound for infant screening and MR imaging to confirm ultrasound findings and treatment planning.³

Figures Finding

Fig1. Sagittal T1W image shows wedge-shaped conus termination. Did you notice sacral hypogenesis? (You can only see S1 and S2).

Fig2. On T2W image, conus seems mildly expanded and wedge-shaped. Fluid between cauda equine roots should not be interpreted as abnormal signal in distal conus.

Fig3. Clubbing of distal cord. Bilateral hydronephrosis probably is due to neurogenic bladder.

References

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2. Merrelo E, De Marco P, Mascelli S, Raso A, Calevo MG, Torre M, et al. HLBX9 homeobox gene and caudal regression syndrome. *Birth Defects Res A Clin Mol Teratol* 2006 Mar;76(3):205-9.
3. Ross J, Brant-Zawadzki M, Chen M, Moore K. *Diagnostic Imaging: spine*. New York: W.B. Saunders; 2004.