

High abrupt cord termination: a hallmark of caudal regression syndrome

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DESCRIPTION

A 10-month-old female infant, born to a diabetic mother, presented with dribbling of urine since birth. Physical examination was unremarkable. Plain radiograph of lower spine showed partial sacral agenesis. MRI of the spine (figure 1) revealed sacrococcygeal hypoplasia (dysplastic S₄ vertebra and absent S₅ and coccyx). Conus medullaris was blunt ending, club-shaped and terminated at L₁ vertebral level. The caudal nerve roots were arranged in anterior and posterior bundles. No associated syrinx was seen. Imaging findings were characteristic of caudal regression syndrome type 1.

Caudal regression syndrome designates a constellation of anomalies resulting from insult in the early stages of gestation. Although uncommon in the general population, there is a 200-fold increase in the incidence in infants of diabetic mothers, where 1 in 350 newborns is affected.¹ One per cent of infants born to diabetic mothers will have a form of this syndrome, and 16 % of infants with the syndrome have diabetic mothers.² The spectrum of clinical presentation and caudal structural defects varies from being asymptomatic to extensive. Motor involvement is common and deficits correspond to the level of vertebral agenesis. In type 1, there is abnormally high abrupt termination of the conus, while in type 2, the cord is low-lying and tethered. Club-shaped or wedge-shaped appearance of the conus with double-bundle arrangement of the caudal nerve roots is typical of type 1.^{3 4} Type 1 is associated with major sacral dysgenesis and urinary and bladder dysfunction.

Large sacral motor deficit occurs, while sensations are relatively preserved. Severe neurological disturbances and deterioration are common in type 2.⁵ Associated cardiopulmonary, genitourinary and gastrointestinal malformations must be looked for and managed accordingly. The prognosis depends on the severity of the defect and presence of associated abnormalities. Surviving infants usually have normal mental function but require extensive urological and orthopaedic assistance.⁶ Prenatal sonographic diagnosis is possible but findings are variable depending on the severity of defect. Small crown-rump length in the first trimester and short spine with missing lumbosacral vertebrae later in pregnancy can suggest the diagnosis.^{6 7}

Learning points

- ▶ Abnormally high club-shaped abrupt termination of the cord with sacral deformities is a hallmark of caudal regression syndrome type 1.
- ▶ In type 2, the spinal cord is low-lying and tethered.
- ▶ Bladder and anorectal problems, neurological deficits and clinical/radiographical evidence of sacral dysgenesis should raise the suspicion of caudal regression syndrome, especially in infants of diabetic mothers.

Contributors All authors have contributed equally to the design and construction of the case report.

Competing interests None.

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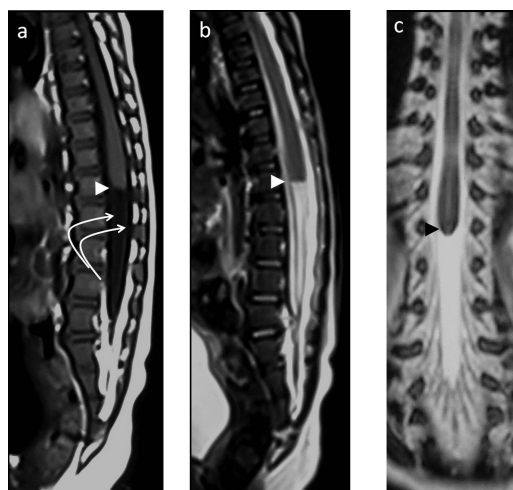


Figure 1 Sagittal T1-weighted (A), T2-weighted (B) and coronal T2-weighted (C) MRI showing abrupt club-shaped termination of the conus medullaris (arrowheads) with double-bundle arrangement of nerve roots (curved arrows). There is a partial sacral agenesis.



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