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Late Presentation of Caudal Regression Syndrome Revealed by Bladder and Bowel Dysfunction in a 6-Year-Old Girl: Diagnostic Value of Spinal MRI

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Abstract **Case Report**

Caudal regression syndrome is a rare congenital malformation of the caudal pole, characterized by variable involvement of the lumbosacral spine, terminal spinal cord, and pelvic structures, often associated with urogenital and gastrointestinal anomalies [1,2]. We report the case of a 6-year-old girl evaluated for secondary enuresis associated with encopresis evolving over one year in an afebrile setting. Spinal MRI revealed caudal regression syndrome. This case highlights that a caudal malformation should be suspected in children presenting with urinary and anorectal disorders, even when the presentation is delayed, and emphasizes the major role of MRI in the assessment of associated spinal cord and osteorachidian abnormalities [3-5].

Keywords: Caudal regression syndrome; sacral agenesis; enuresis; encopresis; spinal MRI; child.

INTRODUCTION

Initially described by Duhamel, caudal regression syndrome is a rare spectrum of congenital malformations resulting from early developmental impairment of the embryonic caudal structures [1]. It encompasses anomalies of varying extent, ranging from partial agenesis of the coccyx or sacrum to more severe forms involving the lumbosacral spine, terminal spinal cord, and, to varying degrees, the urogenital, gastrointestinal, and musculoskeletal systems [6,7].

This condition is uncommon in the general population, but its frequency is significantly increased in children born to diabetic mothers, although not all cases occur in this context [2,3,6]. Clinical presentation is heterogeneous: some forms are evident at birth, whereas others are diagnosed later because of urinary, anorectal, or locomotor disorders [3,5,6].

MRI plays a central role in the positive diagnosis and lesion assessment. It allows precise evaluation of the level of conus medullaris termination, its morphology, abnormalities of the distal spinal canal, and associated vertebral malformations [4,8]. We report the case of a 6-year-old girl in whom caudal regression syndrome was suspected because of secondary enuresis associated with encopresis and subsequently confirmed by spinal MRI.

CASE REPORT

A 6-year-old girl was referred for evaluation of secondary enuresis associated with encopresis, evolving over one year in an afebrile setting and without deterioration in general condition. History-taking revealed no recurrent urinary tract infections and no significant past medical history. Prenatal and neonatal history was unremarkable, notably with no maternal diabetes and no neonatal distress. There was also no particular psychosocial or family context likely to explain the sphincter disorders, including the absence of any major psychoaffective event, significant social difficulties, or recent change in the child's environment.

Psychomotor development was normal for age, with no delay in milestone acquisition. Gait was autonomous and reportedly normal. Clinical examination was unremarkable, with no obvious neurological deficit, no static or locomotor abnormality, and no visible lumbosacral cutaneous anomaly, particularly no dermal sinus.

Spinal MRI was performed using sagittal T1-, T2-, and STIR-weighted sequences, together with axial T2-weighted sequences. At the cervical level, the examination showed a fluid-signal intramedullary lesion centered on the ependymal canal, extending from C4 to C7 and measuring 1.6 mm in diameter (Figure 1), consistent with mild canal dilatation. No abnormality of the craniocervical junction was identified. The cervical vertebral bodies and intervertebral discs were normal in appearance.

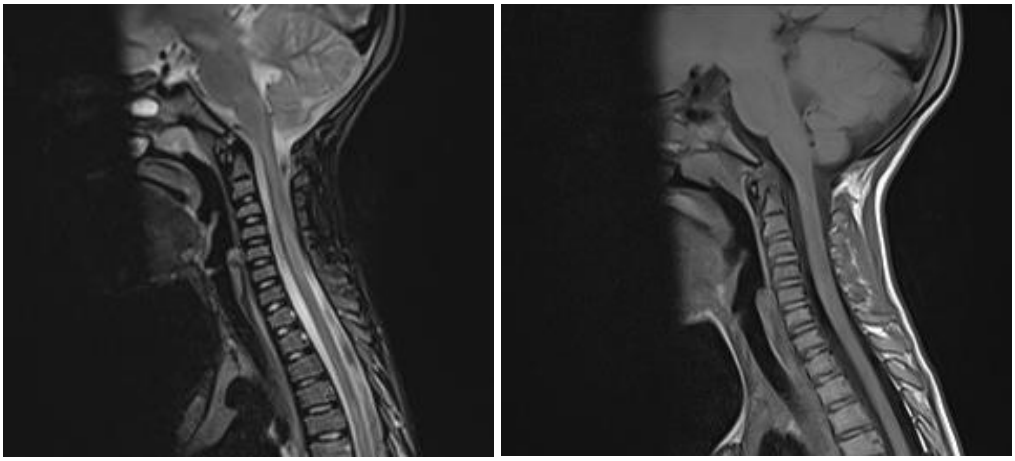

Figure 1: Sagittal T1- and T2-weighted cervical spinal MRI showing a fluid-signal intramedullary cavity centered on the ependymal canal, extending from C4 to C7 and measuring 1.6 mm in diameter

At the dorsolumbar level, the vertebral bodies and intervertebral discs were normal in height and signal intensity. There was regular dilatation of the ependymal canal involving the thoracic spinal cord and terminal conus medullaris, extending from D6 to D12. The terminal conus medullaris had a bulbous appearance and was abnormally high, terminating at the D12-L1 level (Figure 2).

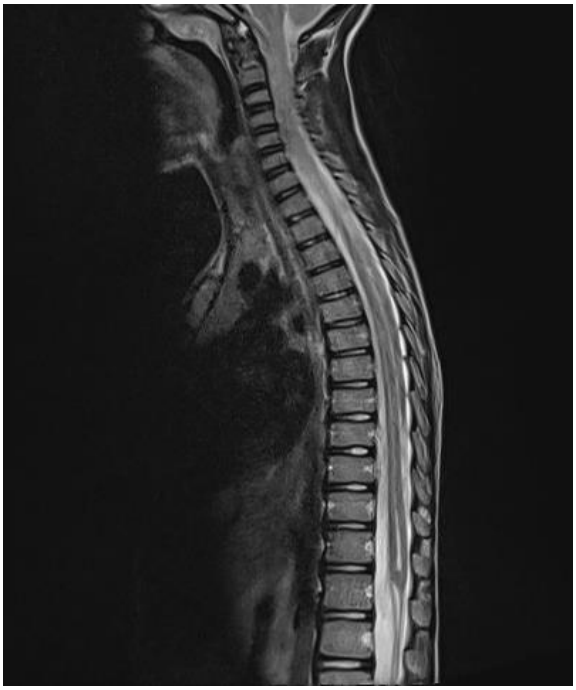

Figure 2: Sagittal T2-weighted cervicothoracic spinal MRI showing a high-riding, bulbous terminal conus medullaris ending at the D12-L1 level, with regular dilatation of the ependymal canal from D6 to D12

The examination also demonstrated agenesis of the sacrum and coccyx, with persistence of an S1-S2 vertebral block. No posterior vertebral arch defect was identified (Figure 3). Overall, the association of sacrococcygeal agenesis with a high-riding and dysmorphic terminal conus medullaris was consistent with caudal regression syndrome.

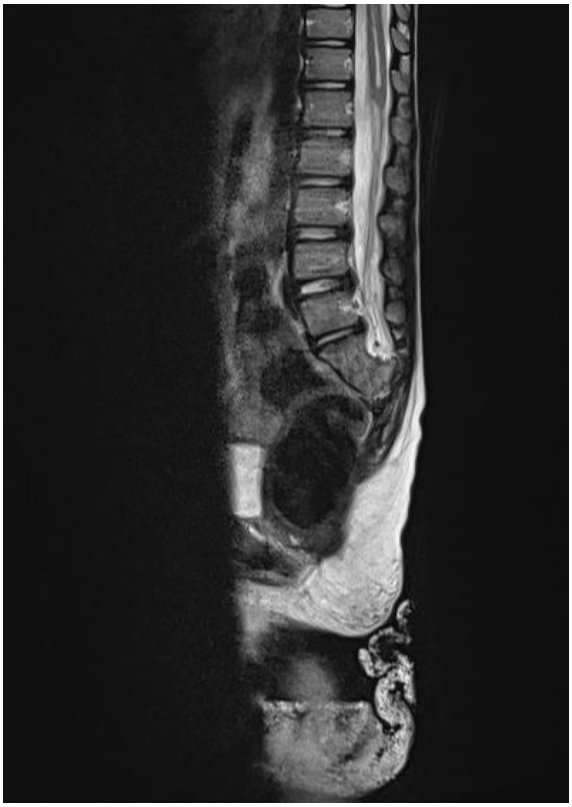

Figure 3. Sagittal T2-weighted spinal MRI centered on the lumbosacral region showing agenesis of the sacrum and coccyx with persistence of an S1-S2 vertebral block.

DISCUSSION

Caudal regression syndrome is a rare malformative entity related to defective development of the embryonic caudal pole, occurring early during organogenesis [6,7]. The most characteristic skeletal abnormality is partial or complete sacral agenesis, often associated with malformations of the terminal spinal cord and with urogenital and gastrointestinal involvement [6,7].

Clinical manifestations depend on the extent of the skeletal and neurological lesions. Bladder and bowel dysfunction are common and may represent the main presenting complaint, particularly in incomplete forms or in cases with few orthopedic manifestations [3,5]. In our patient, secondary enuresis and encopresis were the revealing symptoms, in the absence of any infectious context.

MRI is the reference imaging modality for evaluation of this condition [4,8]. It allows precise assessment of the level of conus medullaris termination, its morphology, the presence of associated ependymal canal abnormalities, and vertebral and sacral malformations [4]. Two major spinal cord patterns are classically described: on the one hand, a high and abrupt termination of the conus medullaris, often dysmorphic, and on the other hand, forms with a low-lying conus or tethered cord [7,8]. A high and truncated conus is considered a particularly suggestive imaging feature of the diagnosis [8].

In our case, the conus medullaris terminated at the D12-L1 level and had a bulbous appearance, associated with agenesis of the sacrum and coccyx. This constellation of findings fits the classic radiological profile of caudal regression syndrome. The cervical and thoracic-terminal ependymal canal dilatation may be regarded as an associated spinal cord abnormality, the extent of which is accurately delineated by MRI [4].

From an etiological perspective, maternal diabetes remains the best-established risk factor in the literature, although it is not constant [2,3,6]. In practice, the diagnosis should prompt a multidisciplinary work-up including at least neurological, urological, digestive, and orthopedic evaluation in order to detect neurogenic bladder, upper urinary tract involvement, anorectal dysfunction, and possible associated locomotor abnormalities [3,4,6].

CONCLUSION

Caudal regression syndrome should be suspected in children with unexplained urinary and anorectal disorders, even when presentation is delayed. Spinal MRI plays a decisive role in the diagnosis by demonstrating abnormalities of the terminal conus, the ependymal canal, and the caudal spine. Identification of this malformation allows prompt referral for appropriate multidisciplinary management.

CONSENT

Written informed consent was obtained from the patient's parents.

Conflict of Interest: The authors declare no conflicts of interest.

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