



# Caudal Regression Syndrome: A Rare Cause of Chronic Constipation

Global Pediatric Health  
Volume 9: 1–4  
© The Author(s) 2022  
Article reuse guidelines:  
sagepub.com/journals-permissions  
DOI: 10.1177/2333794X221118162  
journals.sagepub.com/home/gph



Ibrahima Dokal Diallo, MD<sup>1</sup> , Amine Cherraqi, MD<sup>1</sup> ,  
Wend-Yam Mohamed Traore, MD<sup>1</sup>, Siham El Haddad, PhD<sup>1</sup>,  
Latifa Chat, PhD<sup>1</sup>, and Nazik Allali, PhD<sup>1</sup>

## Abstract

A malformative syndrome of the lower spine pole, caudal regression syndrome is a rare condition (1-5 cases/100 000 births). It usually occurs in children of diabetic mothers and includes a wide range of disorders. This case illustrates a subtype of this syndrome (type IV, group I) in a non-diabetic mother.

## Keywords

regression, caudal, type IV, group I

Received July 2, 2022. Accepted for publication July 19, 2022.

## Introduction

Following the description of a malformative syndrome of the caudal part of the spine in 1852 by Geoffroy Saint-Hilaire and Hohl,<sup>1</sup> the term caudal regression syndrome was first used by Duhamel in 1964. It refers to an agenesic malformative disorder of the distal part of the spine and associates many other anomalies that may be orthopedic, neurological, genitourinary, cardiovascular, or gastrointestinal. While its etiopathogeny is still poorly understood, research has established a clear relationship between maternal diabetes and caudal regression syndrome.<sup>2</sup> The diagnosis of this malformation is easy after birth thanks to the clinical confrontation and standard radiography while ultrasound and MRI allow the search for other associated visceral anomalies.

## Observation

We received a 5-month-old infant from a non-consanguineous marriage and second of a sibling group for a chronic constipation exploration. The interrogation revealed a poorly followed pregnancy and a dystocic delivery which was responsible for a fracture of the left femur and a brachial paralysis; there was no history of malformation among the siblings, and the mother never had diabetes before. While the clinical examination showed a deformation Manuscript (without Author Details) Click here to view linked References of the caudal part of the spine (Figure 1), there was also a caudal

skin dimple and a medial dorsal lumbar bony protrusion. No other striking signs stood out during the rest of the somatic examination.

Standard radiographs of the pelvis, lower limbs (A, B) and dorsolumbosacral spine in profile (C) (Figure 2), showed, in addition to agenesis of the last lumbar vertebrae and sacro-coccyx, a consolidated fracture of the left femur, a shrunken pelvis, shortened femurs, and abduction of the thighs with knee flexion. The diagnosis of caudal regression syndrome was then retained. Abdominal ultrasound and spinal cord MRI were also performed in order to search for other potentially associated lesions (Figure 3).

## Discussion

The caudal regression syndrome is a rare malformative syndrome (1-5 cases/100 000 births) of poorly known etiopathogeny, usually occurring in children of diabetic mothers.<sup>2</sup> It includes all agenesis or hypoplasia of the caudal pole of the fetus or the newborn and is often manifested by the absence or hypoplasia of the last lumbar vertebrae and the sacrococcyx. This multifactorial condition is thought

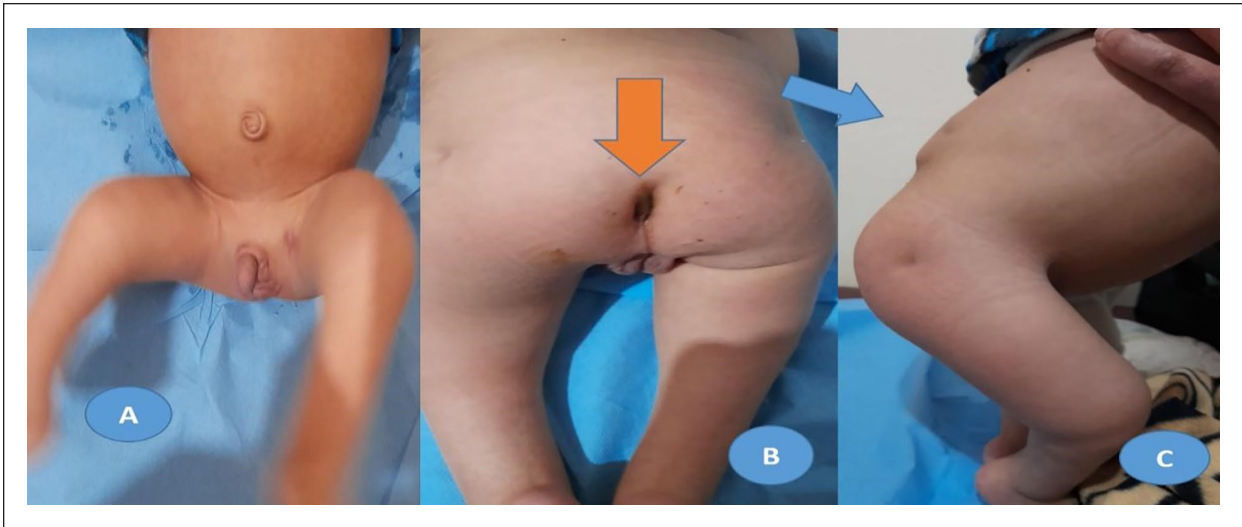
<sup>1</sup>Mohammed V University, Rabat, Morocco

### Corresponding Author:

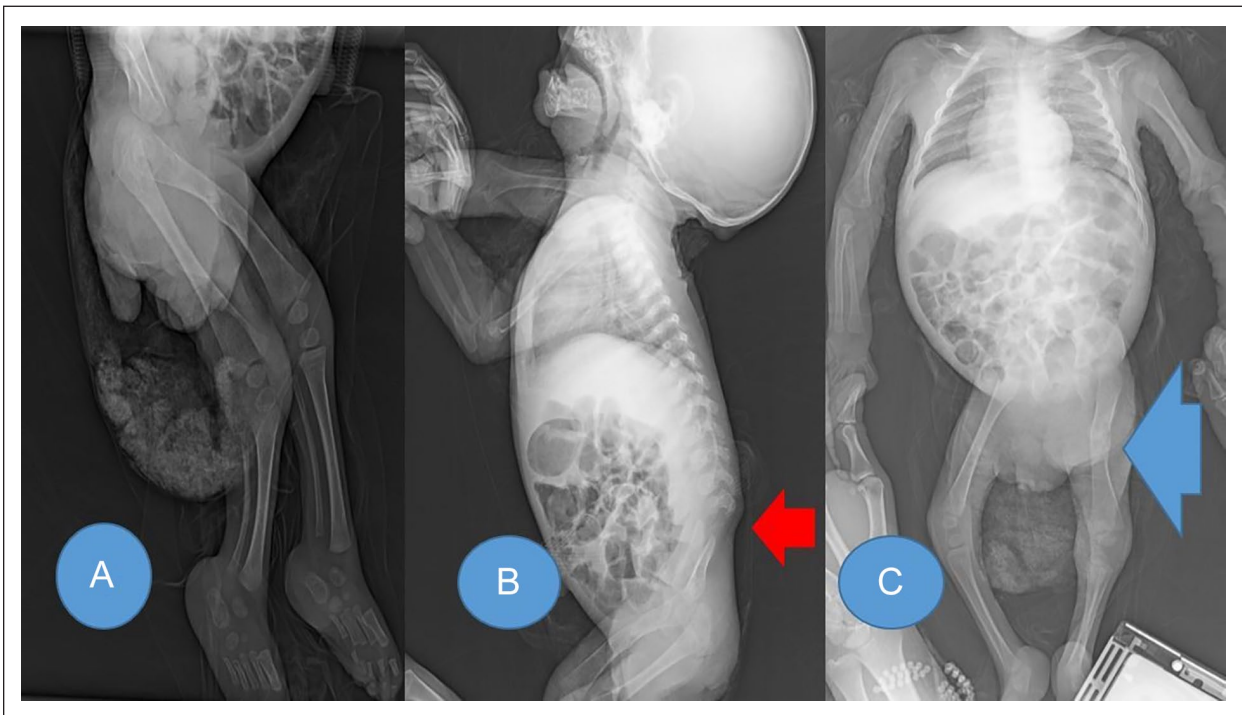
Ibrahima Dokal Diallo, Imaging Department, Children Hospital, Mohammed V University, avenue Maghreb el araabi, 5 Bis at entrance B Residence, Apt 16.4eme arrondissement rabat, Rabat 10500, Morocco.

Email: dialloibrahimadokal@gmail.com





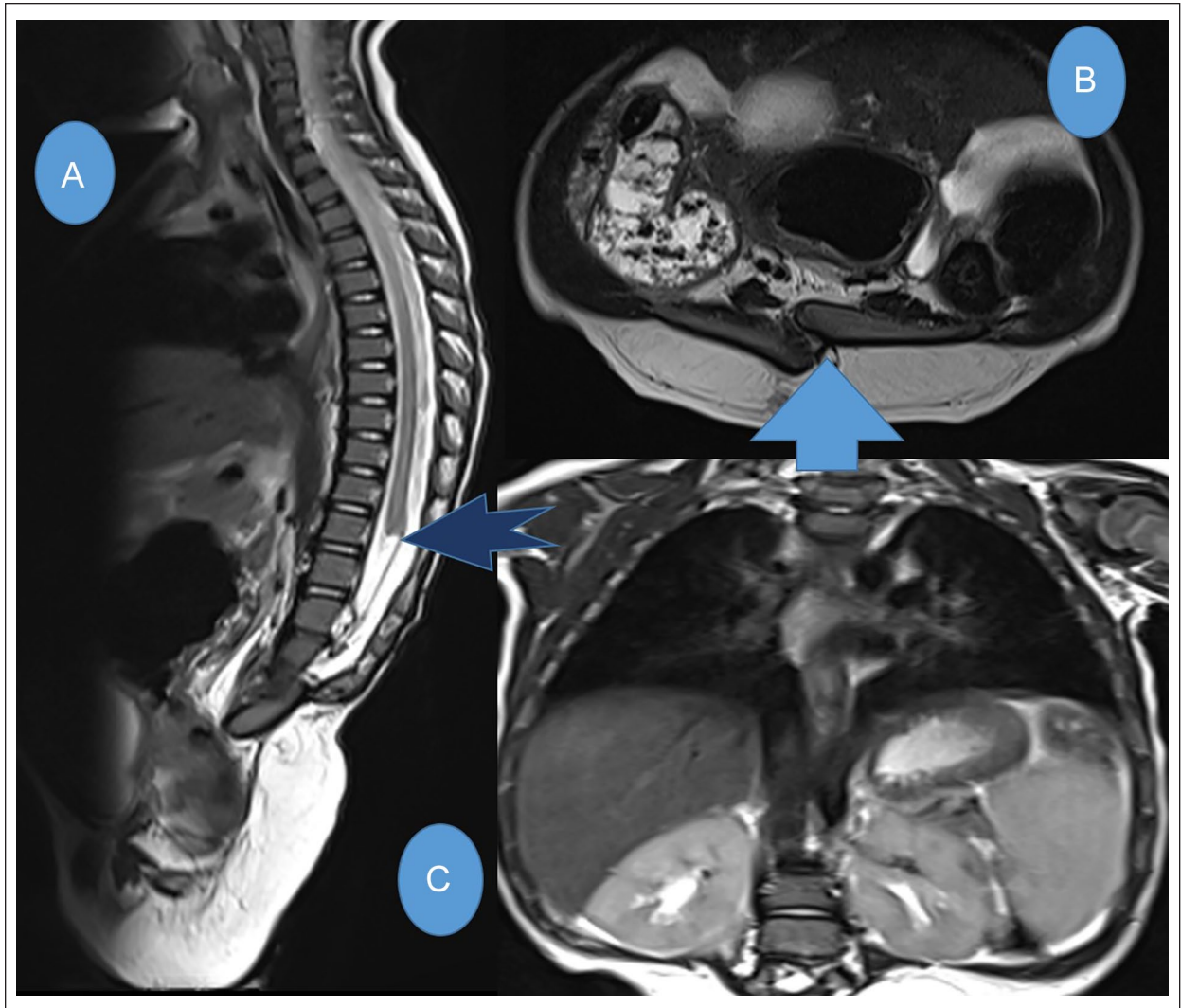
**Figure 1.** (A) Hip abduction and irreducible knee flexion attitude. (B) Caudal skin dimple. (C) Medial dorsal lumbar bone protrusion.



**Figure 2.** Standard radiographs of profile (A, B) and front (C): absence of visualization of the last lumbar vertebrae and sacrococcyx, consolidated fracture of the left femur.

to be due to abnormal mesodermal development before the fourth week of gestation and is often associated with maternal hyperglycemia, infectious, ischemic, toxic, or genetic conditions.<sup>3</sup> A range of disorders can be observed in this condition, including abnormalities<sup>2,4,5</sup>:

- orthopedic such as femoral shortening, clubfoot, kypho-scoliosis;
- genitourinary anomalies such as agenesis, dysplasia (horseshoe kidney, ureteral duplication) or ectopy;



**Figure 3.** Sagittal (A), coronal (B), and axial (C) sections STIR and T2: agenesia of the last three (3) lumbar vertebrae (L3, L4, and L5) and of the sacro coccyx with overlapping of the two (2) iliac wings; Squaring of the medullary cord in front of D11 extending at the posterior level by a tapered cord stopping at L2; no visceral abnormalities.

- gastrointestinal issues such as esophageal and duodenal atresia or anorectal malformations which, associated with sacro-coccygeal agenesia and a pre-sacral mass, define the curarino syndrome;
- neural tube defects such as attached cord, diastematomyelia, lipomyelomeningocele, and congenital narrow spinal tract;
- cardiovascular conditions including pulmonary hypertension, atrial and ventricular septal defects, and patent ductus arteriosus.

Clinically, children with this syndrome present a variety of symptoms dominated by genitourinary

disorders such as incontinence, gastrointestinal disorders represented by chronic constipation and incontinence, as well as neurological disorders affecting the lower limbs.<sup>6</sup> The diagnosis of this condition is first made antenatally by obstetrical ultrasound which notes a shortening of the craniocaudal length and femoral length at the end of the first trimester or the absence of visualization of the sacrum in the second and third trimesters. Postnatally, the diagnosis can be easily made thanks to standard radiology confirming a strong clinical suspicion of a “frog-like” or “Buddha-like” appearance of the lower limbs, which corresponds to an abduction deformity of the hips with irreducible flexion of the knees.<sup>2</sup>

The role of ultrasound and magnetic resonance imaging in this syndrome is mainly to study the marrow and to determine other associated visceral disorders which may in some cases be life-threatening. Thus 5 forms of increasing severity have been described by Renshaw<sup>7</sup>: type 1 (unilateral total or partial sacral agenesis); type 2 (variable total lumbar and sacral agenesis and ilia articulating with the sides of the lowest vertebra); type 3 (variable lumbar and total sacral agenesis); type 4 (partial fusion of the soft tissues of the 2 lower limbs); and type 5 (sirenomelia or sirenomelia syndrome corresponding to complete fusion of the lower limbs forming a single femur and tibia with additional anomalies).

Another classification that is based on the position of the cone had been proposed by Pang<sup>8</sup> divides this syndrome into 2 groups: group I corresponds to a cone ending above the inferior border of the L1 vertebrae at the risk of a significant sacral deficit, whereas group 2 corresponds to a cone ending caudally to the inferior border of the L1 vertebrae. In the latter group, the medullary cone is prolonged by a thick terminal filum (65%).

In the present case, ultrasound and MRI allowed to eliminate associated visceral anomalies, and there was an incomplete agenesis of the lumbar vertebrae and total of the sacro coccyx with a medullary cone ending at the height of D11. This allowed us to retain the diagnosis of caudal regression syndrome type IV group I in accordance with the respective classifications of Renshaw and Pang. In terms of chronic constipation, the diagnosis was obvious in this patient due to the presence of caudal dysmorphic syndrome—which is not always the case, especially for minor injuries (type I).

This implies the inclusion of a spinal exploration in the workup of chronic constipation of non-obvious cause in children. Furthermore, given the plurality of forms, it follows that the management of this condition will depend on the subtype in question. That will be based largely on supportive and symptomatic measures, during which we may have to resort to surgery depending on the systems affected.

## Conclusion

The caudal regression syndrome is a rare complex malformation associated with chronic constipation. Its discovery implies a complete malformative assessment with magnetic resonance imaging as the gold standard. It can occur in children of non-diabetic mothers, hence the interest of a regular follow-up of pregnancies.

## Author Contributions

All authors contributed equally to this work.

## Declaration of Conflicting Interests

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

## Funding

The author(s) received no financial support for the research, authorship, and/or publication of this article.

## Ethics Approval

Our institution does not require ethical approval for reporting individual cases or case series.

## Informed Consent

Written informed consent was obtained from a legally authorized representative(s) for anonymized patient information to be published in this article.

## ORCID iDs

Ibrahima Dokal Diallo  <https://orcid.org/0000-0002-2754-8274>

Amine Cherraqi  <https://orcid.org/0000-0003-1838-4631>

## References

1. Duncan MA, Cantú-Salinas AC, Villarreal-Rodríguez DL, Muñoz Landeros CE, Villarreal Velázquez HJ. Caudal regression syndrome: a case report. *Med Univ.* 2014;16:74-77.
2. Tilfene C, Laamrani FZ, Dafiri R. Caudal regression syndrome: about a case. *Pan Afr Med J.* 2018;30:219.
3. Kang S, Park H, Hong J. Clinical and radiologic characteristics of caudal regression syndrome in a 3-year-old boy: lessons from overlooked plain radiographs. *Pediatr Gastroenterol Hepatol Nutr.* 2021;24:238-243.
4. Bicakci I, Turgut ST, Turgut B, Icagasioglu A, Egilmez Z, Yumusakhuylyu Y. A case of caudal regression syndrome: walking or sitting? *Pan Afr Med J.* 2014;18:92.
5. Mehdi SM, Baig U, Zia MH, Iftikhar N. Caudal regression syndrome - a rare congenital disorder: a case report. *J Pak Med Assoc.* 2021;71:2847-2849.
6. Boruah DK, Dhingani DD, Achar S, et al. Magnetic resonance imaging analysis of caudal regression syndrome and concomitant anomalies in pediatric patients. *J Clin Imaging Sci.* 2016;6:36.
7. Sen KK, Patel M. Caudal regression syndrome. *Med J Armed Forces India.* 2007;63(2):178-179.
8. Kanagasabai K, Bhat V, Pramod GK, Patil SJ, Kiranmayi S. Severe caudal regression syndrome with overlapping features of VACTERL complex: antenatal detection and follow up. *BJR Case Rep.* 2016;3(2):20150356.