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CASE REPORT

# Caudal regression syndrome: A case report of a rare congenital anomaly

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**Abstract**

Caudal regression syndrome (CRS) is a spectrum of disorders of caudal vertebral agenesis or dysgenesis, involving the caudal spinal column, that is associated with other congenital anomalies. The majority of caudal regression cases are sporadic but maternal pre-gestational diabetes is the main risk factor. Imaging is an integral part of the management of CRS. Antenatal diagnosis on obstetric ultrasound and evaluation with fetal magnetic resonance imaging (MRI) is ideal. Early postnatal diagnosis and/or detailed evaluation with MRI is essential for early management to improve outcomes.

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## Introduction

Caudal regression syndrome (CRS) is a rarely occurring genetic disorder defined as total or partial agenesis of the lower vertebrae, including the sacral and thoracolumbar spine. This syndrome is a part of spinal dysraphism, which is a spinal disorder that occurs due to disruption in the development of the spinal cord [1].

Although the etiological factors remain unknown, evidence suggests that genetic predisposition, maternal diabetes, and ischemic or infectious etiologies before the 4th week of gestation may be associated with CRS [2].

CRS has a broad spectrum of systemic anomalies that must be investigated thoroughly at the earliest age. Often reported as sacral agenesis, this rare condition is associated with complex abnormalities of multiple organs such as genitourinary tract, anorectal, and neural systems [1]. We present clinical and radiologic findings of a CRS case in our center.

## Case Report

A 7-year-old woman with no particular maternal history except for a poorly monitored pregnancy presented walking disability and sphincter incontinence since birth. The patient presented a sacral fossa without cutaneous abnormalities.

Magnetic resonance imaging (MRI) and Computed Tomography scan (CT-scan) of the lumbosacral spine revealed: vertebral agenesis of the spinal column below S2 (Fig. 1) with truncated shape of spinal cord at the L1 vertebral level with non-conical termination (Fig. 2a and b).

Associated findings include an aggregation of cauda equina roots, which are attached to an intracanal fibro lipoma (Fig. 2c). There is also bilateral ureterohydronephrosis (Fig. 3) and neurological bladder.

These findings are characteristic of a CRS.

The patient remains in follow-up with urology, traumatology, neurology, and general pediatrics, in order to keep a close watch on all the risk factors and possible complications.

## Discussion

CRS is a rare disorder characterized by abnormal development of the caudal spine of the developing fetus.

Two possible causes are suspected: disruption of the primary neurulation process or derailment of the process of degeneration and differentiation of an initially normally developed neural tube [3].

CRS has been associated with poorly controlled maternal diabetes [7,8]. Our patient had no history of maternal diabetes.

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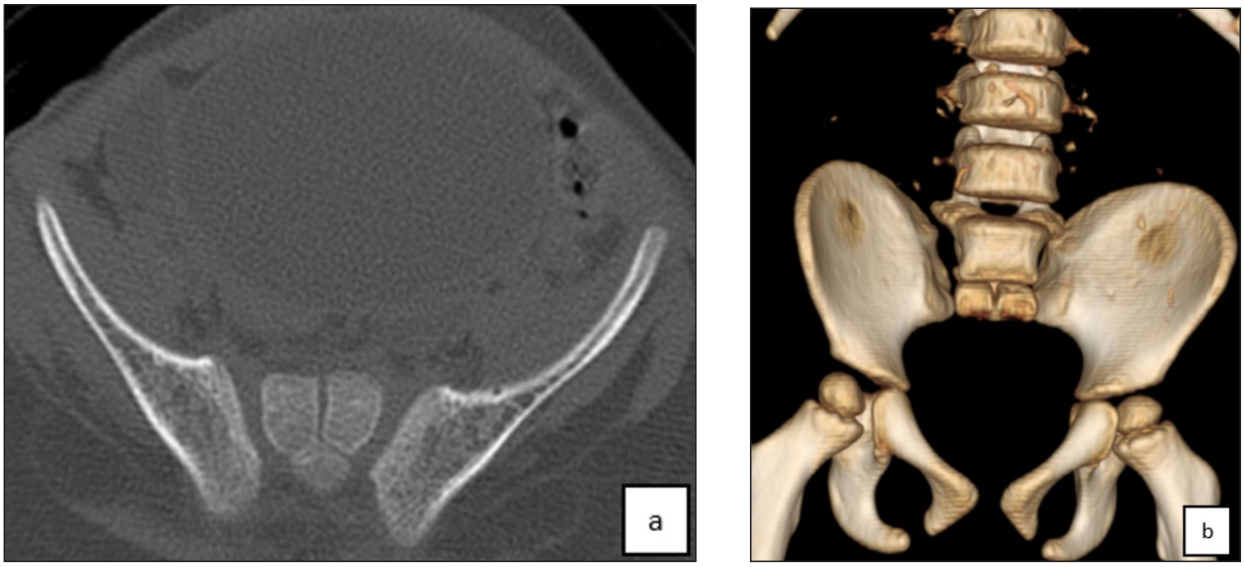



**Figure 1.** (a, b): The spine X-ray and CT scan show hypoplastic S2 vertebrae, agenesis of vertebrae below S2 level, and apposition of both iliac bones.

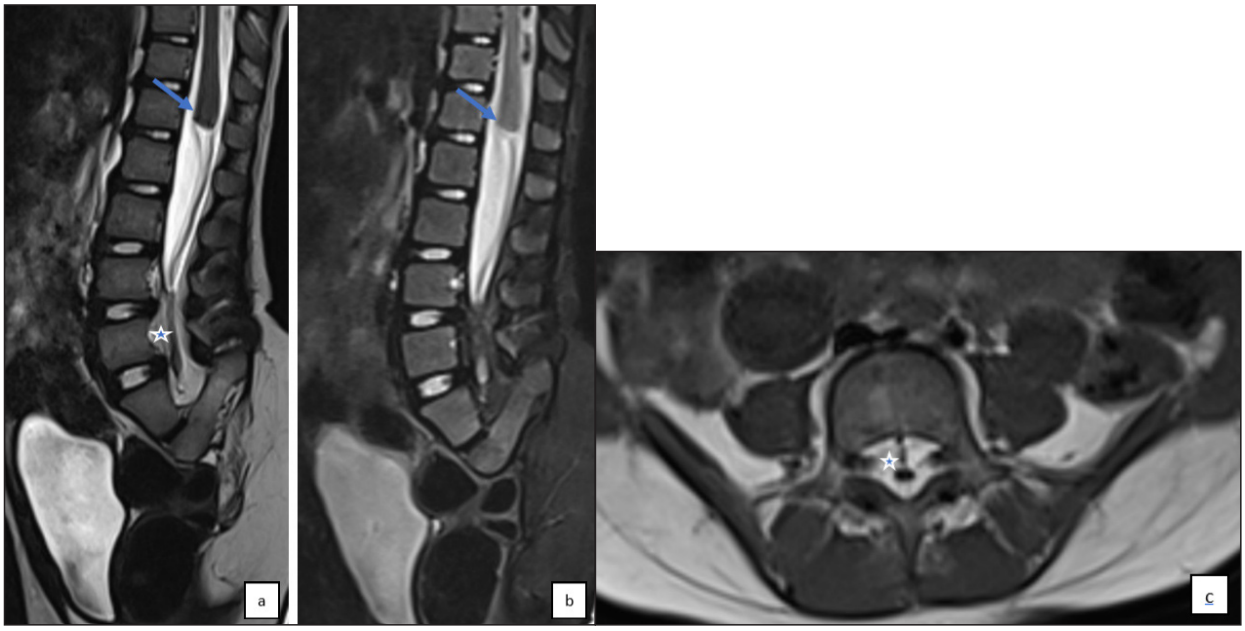



**Figure 2.** (a–c): Sagittal T2, STIR, and axial T2; MRI sequences show a characteristic wedge shape of spinal cord ends (arrow) at L1 vertebral body level. At the L5 vertebral body level the filum terminal is stretched and filled by a fibrolipoma (asterisk).

The most characteristic feature of the spinal cord ends in CRS is the wedge-shaped cord terminus (cigar-shaped conus medullaris) [4]. When this characteristic of cord terminus is seen, imaging of neurologic anomalies in CRS includes segmental deficit corresponding to the level of vertebral agenesis, neurogenic bladder, bowel incontinence or obstipation, dimples, and absent or deep tendon or skin reflexes. Additionally, the lumbosacral sensation is much better preserved than the motor functions. Urinary and bladder symptoms are unusual [5]. The level of vertebral agenesis correlates with the motor level but not the sensory level [1].

CRS may be associated with several additional anomalies, both spinal and extraspinal. A number of genitourinary (vesicoureteral reflux, hydronephrosis, fused kidneys, renal agenesis, ectopic ureters, transposition of external genitalia due to Müllerian duct agenesis) and gastrointestinal anomalies

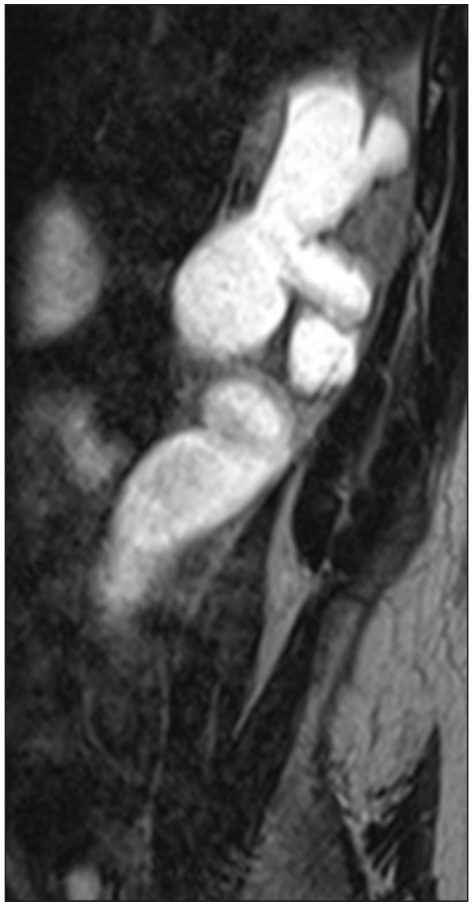


**Figure 3.** Sagittal T2 MRI sequence shows severely dilated ureters and pyelocaliceal tracts.

(anorectal atresia, inguinal hernia, abdominal wall defects, malrotation of gut, imperforate anus) have been described in association with CRS [3,6].

Early recognition of CRS is crucial for improving patient outcomes and quality of life through timely diagnosis. Diagnosis should be done in prenatal ultrasound and MRI [8,10].

Antenatal sonographic findings of CRS are variable. There may be a complete absence of the sacral vertebrae; we can also find a tapered and low-lying conus that ends caudal to the first lumbar segment, and the elongated conus is often tethered by a thickened filum terminale, terminal myelocystocele, or even transitional lipoma [11].

Subsequent Ultrasound (US) can also demonstrate relative hypoplastic musculature of the fetal legs. US also showed an atypical position of legs and feet and abnormally positioned iliac wings close to the midline or fusion of the iliac bones can also be noted [12,13].

Particularly in the setting of oligohydramnios and maternal obesity, MRI is valuable and can be used to assess genitourinary, gastrointestinal, and musculoskeletal anomalies that might be associated with CRS. It can be helpful to assess the degree of vertebral body dysgenesis and to characterize the location and shape of the conus medullaris [11].

The multisystem nature of CRS calls for a multimodal approach to the evaluation and management of this pathology [8]. The appropriate early management with surgical correction as necessary can significantly decrease the risk of complications and thus improve the prognosis and survival in CRS [9,10].

## Conclusion

Caudal regression syndrome is a rare congenital condition characterized by malformations of the vertebral or sacrococcygeal bones, which can lead to a range of complex abnormalities involving multiple systems. These include the neural, orthopedic, gastrointestinal, and genitourinary systems. The severity of CRS can vary greatly, often depending on the extent of the vertebral agenesis or malformation. Due to the wide variety of possible clinical presentations, a multidisciplinary approach to diagnosis and treatment is essential. A structured diagnostic algorithm is important to identify the specific type of CRS in each patient, enabling clinicians and radiologists to manage the condition more effectively, avoid unnecessary diagnostic tests, and reduce radiation exposure. This approach ultimately improves patient outcomes and quality of life by facilitating targeted interventions and comprehensive care.

## Conflict of interest

None declared.

## References

1. Purbasari U, Nazar H, Miraj F, Aprilia D, Widiani W, Suprihatin M, et al. Caudal regression syndrome from radiology and clinical perspective: a case series and a proposed new integrated diagnostic algorithm. Radiol Case Rep 2023; 18(7):2478–86.
2. Bouchahda H, El Mhabrech H, Ben Hamouda H, Ghanmi S, Bouchahda R, Soua H. Prenatal diagnosis of caudal regression syndrome and omphalocele in a fetus of a diabetic mother. Pan Afr Med J 2017; 27:1–7.
3. Mehmet B, Akif A, Yunus A, Temel T, Deniz K, Mehmet K, Can Y, Hakan E, Aysegul B, Erol T. Caudal regression syndrome (Sacral Agenesis) with

associated anomalies. J Turkish Spinal Surg 2013; 3(April):191–19.
4. Sharma P, Kumar S, Jaiswal A. Clinico-radiologic findings in group II caudal regression syndrome. J Clin Imaging Sci 2013; 3:26.
5. Sen KK, Patel M. Caudal regression syndrome. Med J Armed Forces India 2007 Apr; 63(2):178–9; doi:10.1016/S0377-1237(07)80071-2
6. Kylat RI, Bader M. Caudal regression syndrome. Children 2020; 7(11):211. doi: 10.3390/children7110211
7. Chen CP, Chen CY, Lin CY, Shaw SW, Wang W, Tzen CY. Prenatal diagnosis of concomitant alobar holoprosencephaly and caudal regression syndrome associated with maternal diabetes. Prenat Diagn 2005; 25(3):264–6.
8. Boulas MM. Recognition of caudal regression syndrome. Adv Neonatal Care 2009; 9(2):61–9.
9. Altaf F, Hakel W, Sivaraman A, Noordeen H. Orthopaedic management of caudal regression syndrome. Eur J Orthop Surg Traumatol 2008; 18(7):531–5.
10. Mudekereza PS, Balemba GM, Murhega RB, Murhula GB, Nteranya DS, Lekuya HM. Atypical caudal regression syndrome with lumbar agenesis, hypoplastic sacrum without sacroiliac joints in the eastern Democratic Republic of Congo: a case report. Childs Nerv Syst 2023 Sep; 39(9):2551–6.
11. Mahmoud MH, Asghar T, Elkammash TH, Housseini AM, Gad AA. Fetal magnetic resonance imaging in association with antenatal ultrasound in the diagnosis of caudal dysgenesis: report of two cases. Cureus 2023 Feb 26; 15(2):e35485.
12. Negrete LM, Chung M, Carr SR, Tung GA, Tung GA. In utero diagnosis of caudal regression syndrome. Radiol Case Rep 2015; 10(1):1049.
13. Heuser CC, Hulinsky RS, and Jackson GM. Caudal Regression Syndrome. Obstetric Imaging: Fetal Diagnosis and Care 2018. 291–294.e1. doi:10.1016/b978-0-323-44548-1.00059-0