International Journal of Radiology Research
www.radiologyjournal.in
E-ISSN: 2663-4562, P-ISSN: 2663-4554
Received: 09-02-2025; Accepted: 10-03-2025; Published: 25-03-2025
Volume7, Issue 1, 2025, Page No. 16-20

Caudal regression syndrome– Case series

Dr. Sagar H V[1], Dr. Shanth Kumar[2]
[1] Junior Resident, Department of Radio Diagnosis, KVGMCH, Karnataka, India
[2] Senior Resident, Department of Radio Diagnosis, KVGMCH, Karnataka, India

Abstract
Caudal Regression Syndrome (CRS), also known as caudal agenesis or sacral agenesis, is a rare congenital disorder characterized by abnormal development of the lower (caudal) portion of the spine. This condition typically involves the partial or complete absence of the sacrum and coccyx, and in severe cases, may also affect the lumbar spine. CRS is often associated with a range of additional anomalies, including spinal cord malformations, genitourinary abnormalities, gastrointestinal defects, and lower limb deformities. The severity of the syndrome can vary widely, from mild cases with minimal symptoms to severe cases that may result in significant physical disabilities.
The exact etiology of CRS remains unclear, but it is believed to result from a combination of genetic and environmental factors. Maternal diabetes mellitus has been identified as a significant risk factor, though cases have also been reported in non-diabetic mothers. Diagnosis is typically made through prenatal imaging, such as ultrasound or MRI, or shortly after birth based on physical examination and radiographic findings.

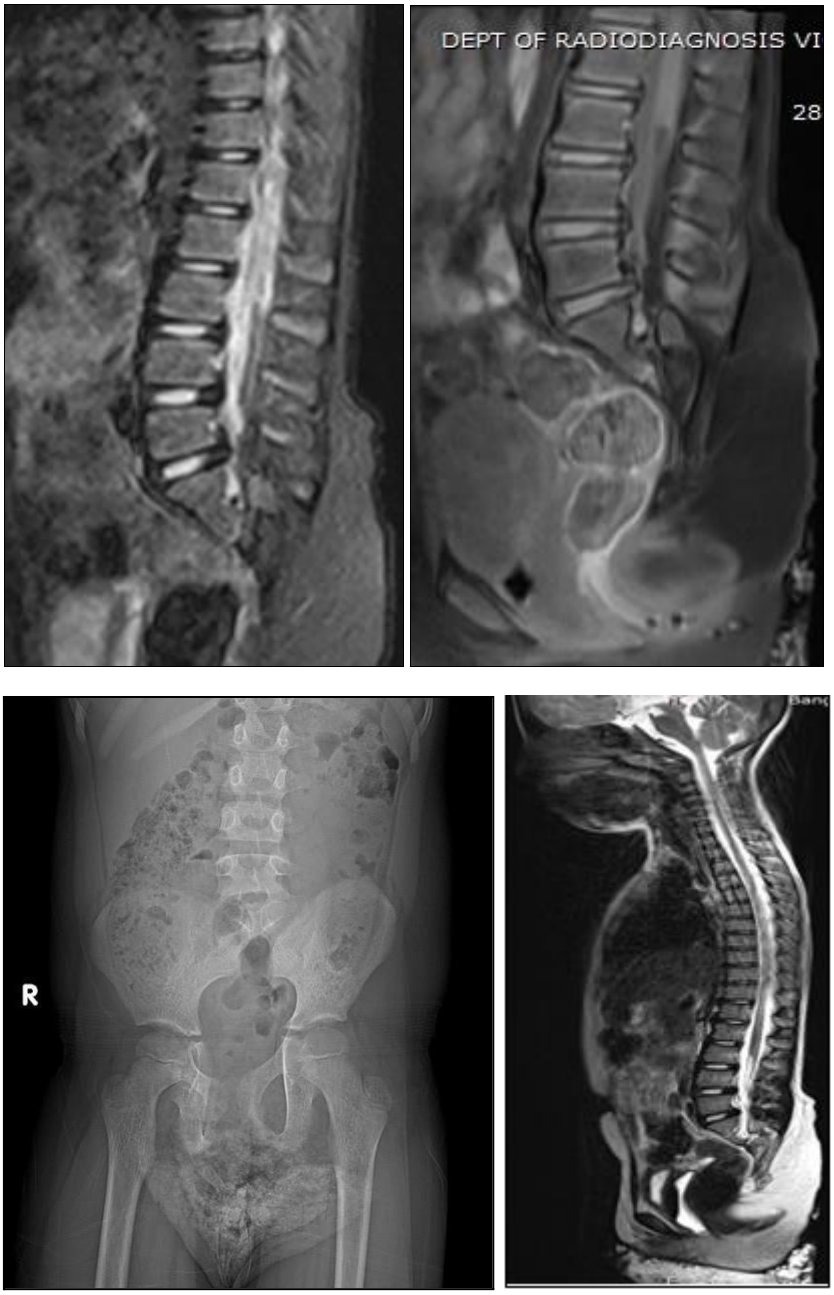
Keywords: Caudal Regression Syndrome (CRS), sacrum, lumbar vertebrae, Computed Tomography (CT), Magnetic Resonance Imaging (MRI), chronic kidney disease (CKD), vesico-ureteric reflux (VUR).

Introduction
Caudal Regression Syndrome (CRS) is a rare congenital malformation characterized by the underdevelopment or absence of the caudal (lower) portion of the body, which includes the sacrum, lumbar vertebrae, and associated structures. The syndrome results from abnormal embryologic development during the early weeks of gestation, leading to defects that are most often identified through radiological imaging. CRS varies in severity, with manifestations ranging from minor sacral anomalies to complete sacral agenesis, where the sacrum is entirely absent.
Radiological imaging, particularly X-ray, CT, and MRI, plays a pivotal role in diagnosing CRS and assessing the extent of the malformations. On plain radiographs, findings may include sacral agenesis or hypoplasia, abnormal formation of the lumbar vertebrae, and fusion of vertebral segments. In more severe cases, a truncated or absent sacrum can be observed. Additionally, there may be lumbar spine abnormalities, such as vertebral body anomalies or fusion of the vertebral elements. Advanced imaging techniques like MRI provide more detailed visualization of soft tissues and can reveal associated abnormalities in the pelvic organs, such as renal agenesis, malrotation, or ectopia, which are commonly seen in CRS. MRI also allows for better assessment of any neural involvement or spinal cord abnormalities, which can range from tethered cord to incomplete development of the caudal spinal cord.
Patients with CRS may present with a variety of clinical signs, including motor and sensory deficits, bladder and bowel dysfunction, and lower limb abnormalities. Radiologic evaluation is crucial for determining the severity of the syndrome, guiding the clinical management, and planning for potential interventions, such as surgical correction of associated malformations or rehabilitative therapies. Early and accurate radiological assessment helps in providing prognostic information, especially regarding mobility and other functional outcomes for affected individuals.

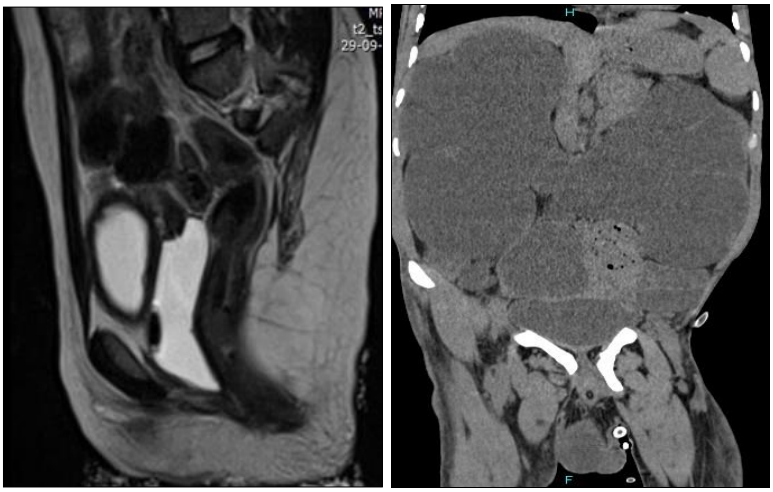
Materials and methods
Images from Conventional Radiography, Computed Tomography, and Magnetic Resonance Imaging were retrospectively gathered for three patients who presented with urinary incontinence at the Department of Radiodiagnosis, KVGMCH.
Demographic information, clinical history, and features were recorded. The imaging findings were analyzed in relation to the spectrum of Caudal Regression Syndrome.
One patient was identified as having Caudal Regression Syndrome Group I, while two others were classified under Group II. These patients exhibited additional neural, vertebral, and genito-urinary anomalies.

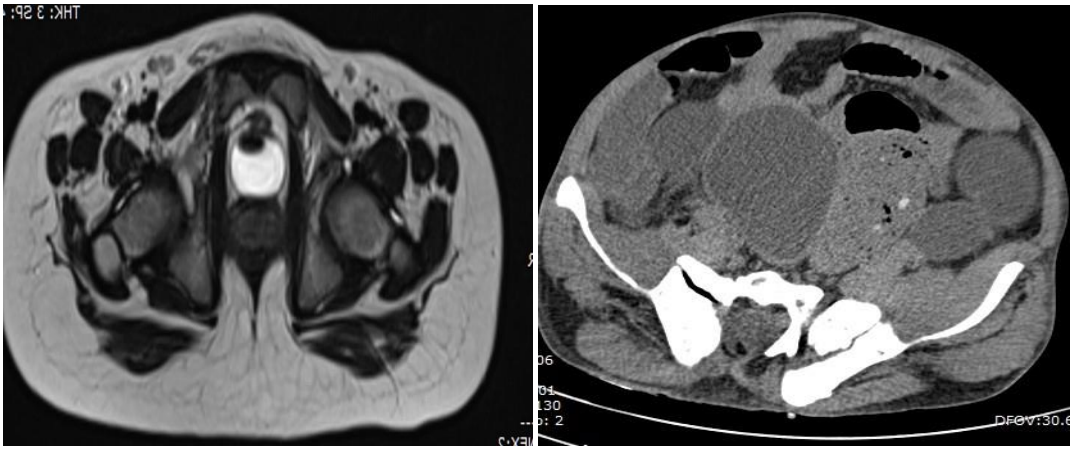
Case series
Case -1
Clinical data
A 7-year-old female patient presents with a history of frequent urination and urge incontinence, which has been ongoing since she was 2 years old. She was born prematurely and has a maternal history of diabetes mellitus.

Imaging features
There is a medial orientation of the bilateral iliac bones.
The spinal cord ends at the level of the L2 vertebra, accompanied by a terminal lipoma.
There is a collection in the vaginal canal, likely due to vaginal dysgenesis or an imperforate hymen.



The sacrum and coccyx are not visualized, with the L1 vertebra articulating with both iliac bones. MR findings-
There is focal outpouching observed on the anterior wall of the urinary bladder, suggestive of diverticula. The uterus appears normal for the patient's age.
A T2 hyperintense and T1 hypointense collection is seen within the vagina, likely due to vaginal outlet obstruction.



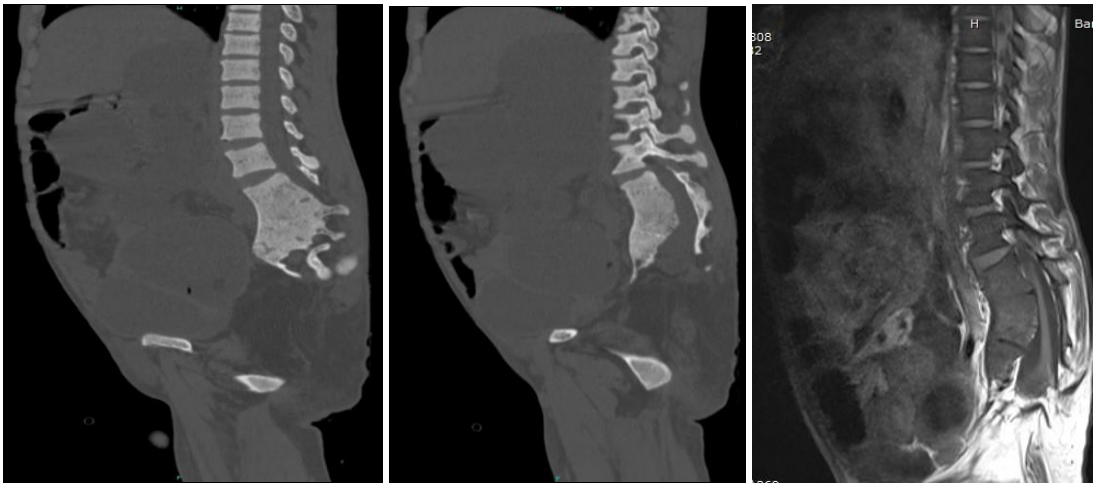


Diagnosis- Caudal Regression syndrome Group I with terminal lipoma.

Case: 2

History - A 40-year-old male, who has been paraplegic since childhood, presents with recurrent urinary tract infections and urinary incontinence. He also has a known history of chronic kidney disease (CKD).

Imaging features



CT findings

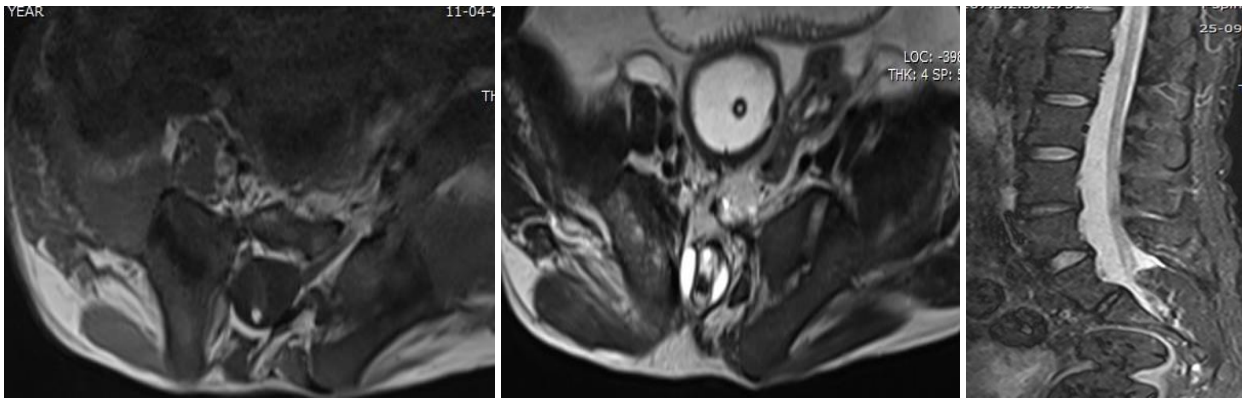
The L3, L4, and L5 vertebrae are fused, with a portion of the sacrum also visible.

The sacrum appears hypoplastic, and the S4 and S5 segments of the sacrum are not visualized. Additionally, the spinous processes of the L5 and S1 vertebrae are absent.

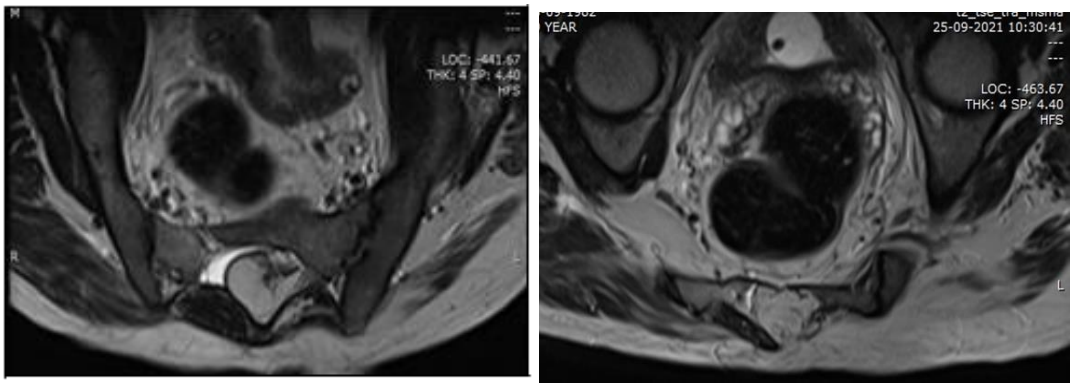
Bilateral significant hydro-uretero-nephrosis observed, likely resulting from vesico-ureteric reflux secondary to neurogenic bladder.

The bladder wall appears abnormally thickened, suggesting a possible neurogenic bladder. An outpouching is noted in the posterosuperior bladder wall, with the Foley catheter's bulb inside, indicating the presence of a diverticulum.

MR findings



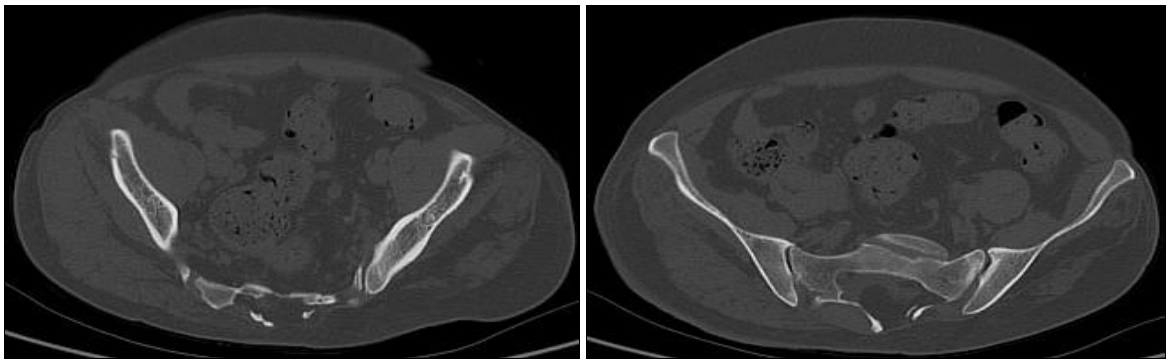
The spinal cord terminates at the lower margin of the S1 vertebra, indicating a low-lying and tethered spinal cord.
The neural placode-fat interface is positioned outside the spinal canal, with an associated enlargement of the subarachnoid space. This suggests a possible lipomyelomeningocele.
A lesion within the spinal cord is observed to be hyperintense on T1/T2 imaging and hypointense on STIR sequences, which is consistent with a lipoma.



Diagnosis - Caudal regression syndrome - Group II with lipomyelomeningocele with Neurogenic bladder and tethered cord.

Case 3
Clinical data
A 39-year-old male patient with a history of right flank pain and urinary incontinence for the past two years, along with recurrent urinary tract infections and low back pain, underwent MRI of the lumbar spine with whole spine screening and a CT scan of the lumbar spine.

Imaging findings
CT findings



The S4 and S5 segments of the sacrum and coccyx are not visualized.
There are defects observed at the junction of the right lamina and the ala of the S1 segment. The posterior elements of the S1, S2, and S3 sacral segments show splaying.
A lumbosacral transitional vertebra (Castellvi II a) is present at the L5 vertebra. Mild lateral rotation of the L5 vertebra is noted.

MRI findings
The S4 and S5 segments of the sacrum and coccyx are not visible.
There are defects observed at the junction of the right lamina and ala of the S1 segment.
The lower margin of the spinal cord is located at the level of the lower margin of the L5 vertebra, indicating a low-lying and tethered spinal cord.
Syringohydromyelia is present from the superior endplate of L3 to the inferior endplate of L5.
A well-defined lesion extending from the inferior endplate of L5 to the inferior margin of the S3 segment is identified in the posterior aspect of the spinal canal. The lesion is hyperintense on T1/T2 sequences and hypointense on STIR sequences, with fat suppression, suggesting it is most likely a lipoma.
Mild right sided hydronehrosis with proximal hydroureter? secondary to VUR

Diagnosis
Caudal regression syndrome - Group II with tethered cord and filum terminals lipoma with Neurogenic bladder

Conclusion
This case series provides a comprehensive analysis of the radiological features of Caudal Regression Syndrome (CRS), a rare congenital disorder characterized by abnormalities in the caudal portion of the body. The imaging findings observed in this series reveal a spectrum of abnormalities, including sacral agenesis, partial or complete absence of the lower lumbar vertebrae, and malformations

of the pelvic bones. In more severe cases, there were associated defects in the lower limbs, gastrointestinal, and genitourinary systems, further complicating the clinical presentation.

The radiological features are critical in establishing an early diagnosis of CRS, as they help distinguish it from other congenital anomalies. X-rays, CT scans, and MRIs provided detailed information regarding the degree of sacral agenesis, spinal curvature, and the involvement of surrounding structures, which are essential for accurate prognosis and management planning.

This series underscores the importance of a multidisciplinary approach, combining clinical evaluation and imaging studies, to assess the extent of malformations in affected individuals. Early recognition of CRS through radiological imaging allows for prompt intervention, which can improve the quality of life and

functional outcomes for patients. Furthermore, this case series contributes valuable insights into the diverse manifestations of CRS, aiding in better-informed clinical decision-making.

Reference

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):2478-2486.
2. Kylat RI, Bader M. Caudal Regression Syndrome. Children (Basel),2020:7(11):211.
3. Dayasiri K, Thadchanamoorthy V, Thudugala K, Ranaweera A, Parthipan N. Clinical and Radiological Characterization of an Infant with Caudal Regression Syndrome Type III. Case Rep Neurol Med, 2020.
4. Thottungal AD, Charles AK, Dickinson JE, Bower C. Caudal dysgenesis and sirenomelia-single centre experience suggests common pathogenic basis. Am J Med Genet A,2010:152A(10):2578-87.
5. Sharma P, Kumar S, Jaiswal A. Clinico-radiologic Findings in Group II Caudal Regression Syndrome. J Clin Imaging Sci,2013:3:26.
6. Cho PS, Bauer SB, Pennison M, Rosoklija I, Bellows AL, Logvinenko T, *et al*. Sacral agenesis and neurogenic bladder: Long-term outcomes of bladder and kidney function. J Pediatr Urol,2016:12(3):158.e1-7.
7. Sen KK, Patel M. Caudal Regression Syndrome. Med J Armed Forces India,2007:63(2):178-9.
8. Krishnan V, Jaganathan S, Jayappa S, Glasier C, Choudhary A, Ramakrishnaiah R, *et al*. Clinical and radiological evaluation of caudal regression syndrome. Pediatr Radiol,2024:54(9):1451-1461.