Radiology Case Reports 19 (2024) 6603–6608

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Case Report

Caudal regression syndrome with incidental brain tumor in a woman: A case report ☆

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ARTICLE INFO

Article history:
Received 10 August 2024
Revised 9 September 2024
Accepted 15 September 2024

Keywords:
Caudal regression syndrome
Low lying tethered cord
Brain tumor

ABSTRACT

Caudal regression syndrome is a rare inherited neural tube disorder. It may be associated with urological, gastrointestinal, and other spine malformations. It usually presents with variable neurological deficits of varying severity. Maternal diabetes mellitus has been postulated to be a significant risk factor for this syndrome. Most of the cases are diagnosed in antenatal or early childhood with very few cases been reported in adulthood. We have tried to illustrate a rare case of caudal regression syndrome in a 31-year female presenting with low back pain with incidental finding of brain tumor.

Introduction

Caudal regression syndrome (CRS) encompasses a group of rare congenital malformations primarily affecting the lower spinal region, including the spinal cord and vertebrae along with other systems predominantly gastrointestinal and genitourinary. The estimated incidence of CRS ranges from 1 to 2 per 100,000 live births, with an equal distribution between genders [1,2]. Various risk factors have been postulated as the etiology of CRS, including folate deficiency, maternal obesity, and exposure to teratogenic medications during pregnancy. The most significant risk factor has been found to be gestational diabetes which markedly increases the risks estimated to be 200 to 400 times higher compared to the general population. Hyperglycemia and teratogenic agents probably lead to abnormal retrogressive differentiation of the developing notochord and spine as well as disturbance of the caudal mesoderm leading to failure in development of lumbosacral spine [3]. Majority of cases are diagnosed in infancy or early childhood with rare cases being diagnosed in adulthood [2].

Clinical presentation

A 31-year female presented with low back pain radiating to left lower limb for 2-3 months. Numbness, tingling and sharp shooting pain were present. Bowel and bladder functions were

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2024.09.073

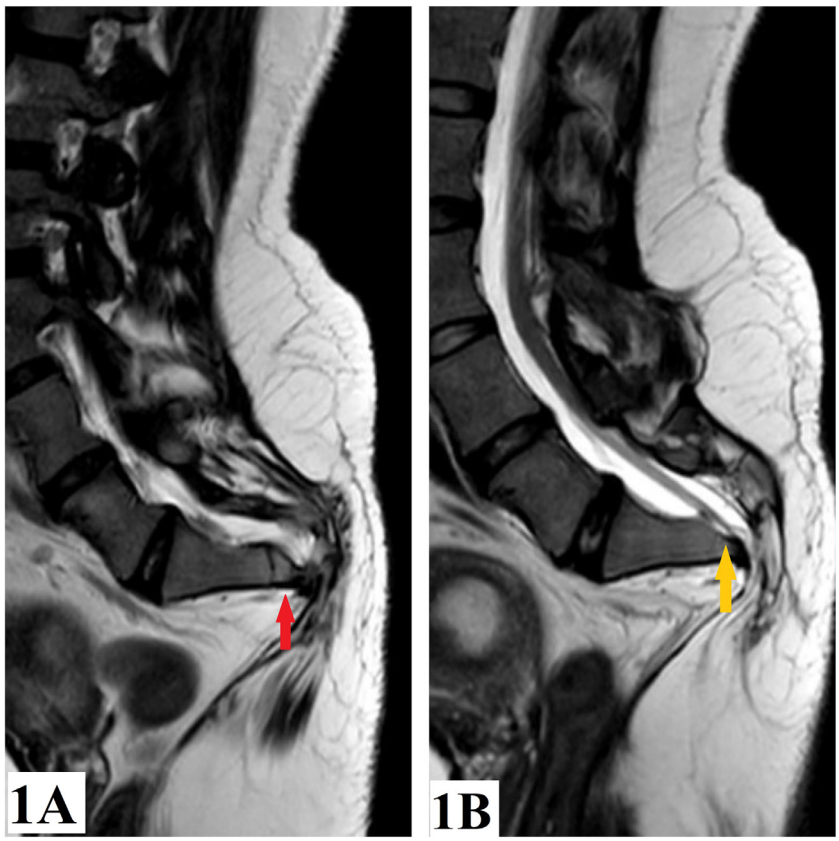

Fig. 1 – T2W Sagittal images (A and B) show visualization of S1 and a small part of S2 with nonvisualization of rests of the sacrococcygeal vertebrae (red yellow) and low-lying tethered cord (yellow arrow).

normal. The patient had no history of hypertension or diabetes mellitus. The patient followed a nonvegetarian diet.

On examination, higher mental function assessment showed no significant abnormality. Motor assessment showed 4/5 power of left lower limb and 5/5 power of rests of the limbs. Left lower limb showed brisk reflexes. Sensory assessment showed decreased vibration and temperature sensations in lateral aspect of left lower limb. Examination of other systems showed no remarkable findings.

On the basis of clinical history and examination, provisional diagnosis of left L4-L5 radiculopathy was made.

Patient was advised for MRI lumbosacral spine with screening of whole spine. A brain parenchymal lesion was seen in the screening images of cervical spine and contrast enhancement MRI (CEMRI) brain was planned for better evaluation of the brain lesion.

MRI of spine revealed following findings:

1. Deformity in sacral vertebrae with nonvisualization of most of the S2 vertebrae and rests of the lower sacrococcygeal vertebrae (Fig. 1A).
2. Low-lying tethered cord (Fig. 1B).
3. Spinal dysraphism with fat-neural placode interface within the spinal canal (lipomyelocele) (Figs. 2A-C).
4. Diastematomyelia with the hemicords lying within the single thecal sac (Type II) (Figs. 2D).
5. Hydrosyringomyelia formation in lower cord (Figs. 3A and B).

CEMRI brain revealed ill-marginated lesion in right frontoparietal lobe with significant perilesional edema. The displays hypo to isointense signal in T1W images, intermediate to hyperintense signal in T2W and FLAIR images. The lesion showed areas of diffusion restriction and blooming within (Figs. 4A-F). On postcontrast images, the lesion showed heterogeneous enhancement. Multivoxel MRS showed raised choline peak with significantly reduced Creatinine and NAA peak in the enhancing portion of the lesion (Figs. 5A-D).

No other major gastrointestinal or genitourinary malformations associated with this syndrome was seen in our case.

On the basis of imaging findings, diagnosis of caudal regression syndrome (Pang group 2) with other associated anomalies and incidental high grade glioma in right frontoparietal lobe was made.

The patient was referred to neurosurgery and spine surgery department for the management of brain tumor and spinal deformities. The patient opted to go to government hospital for further management. The patient was, therefore, lost to follow-up.

Discussion

Caudal regression syndrome is a rare congenital anomaly that results from an abnormal development of the caudal aspect of the spinal cord and vertebral column. Most of the cases are

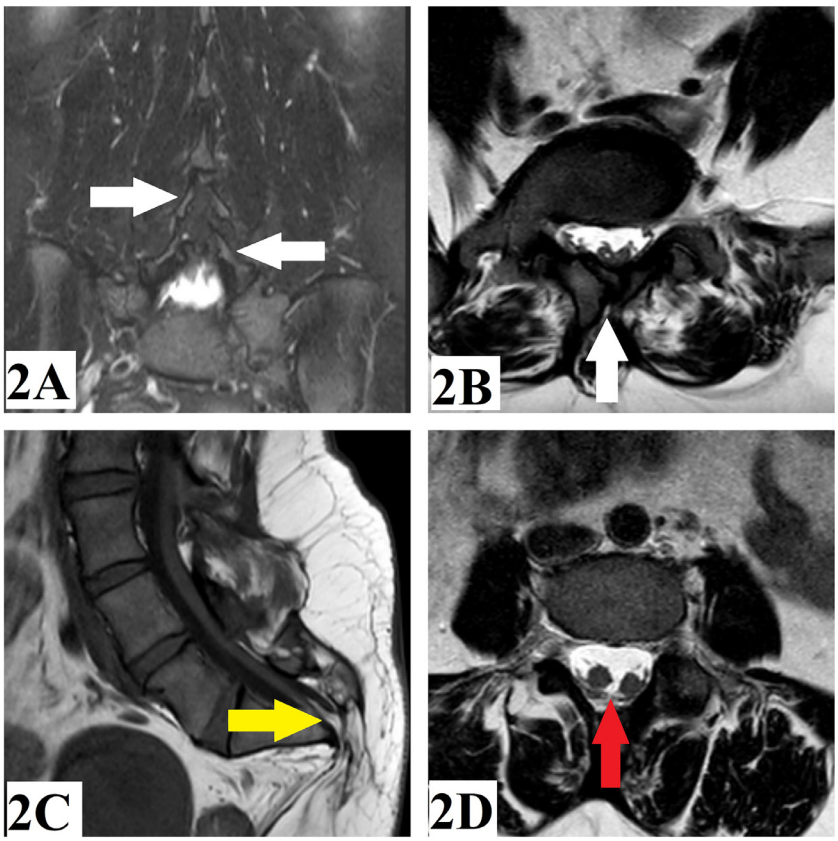

Fig. 2 – STIR Coronal (A) and T2W Axial (B) show defects in posterior elements of lower lumbar vertebrae (white arrows). T1W Sagittal image (C) shows protrusion of fat into the spinal canal with fat-neural placode interface within the spinal canal (yellow arrow). T2W Axial image (D) shows 2 hemicords lying within the single thecal sac (Type II) (red arrow).

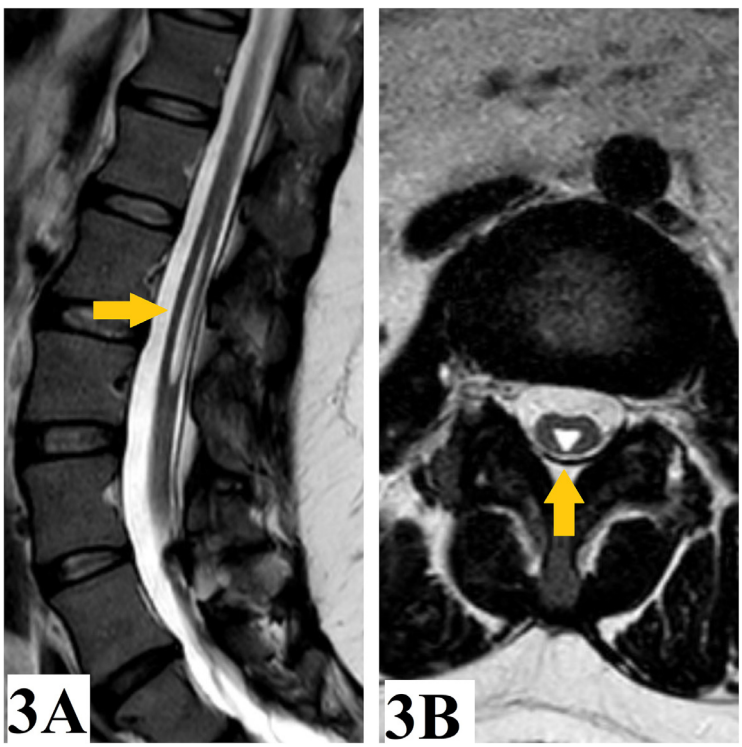

Fig. 3 – T2W Sagittal and Axial images showing dilatation of central canal. (Yellow arrows).

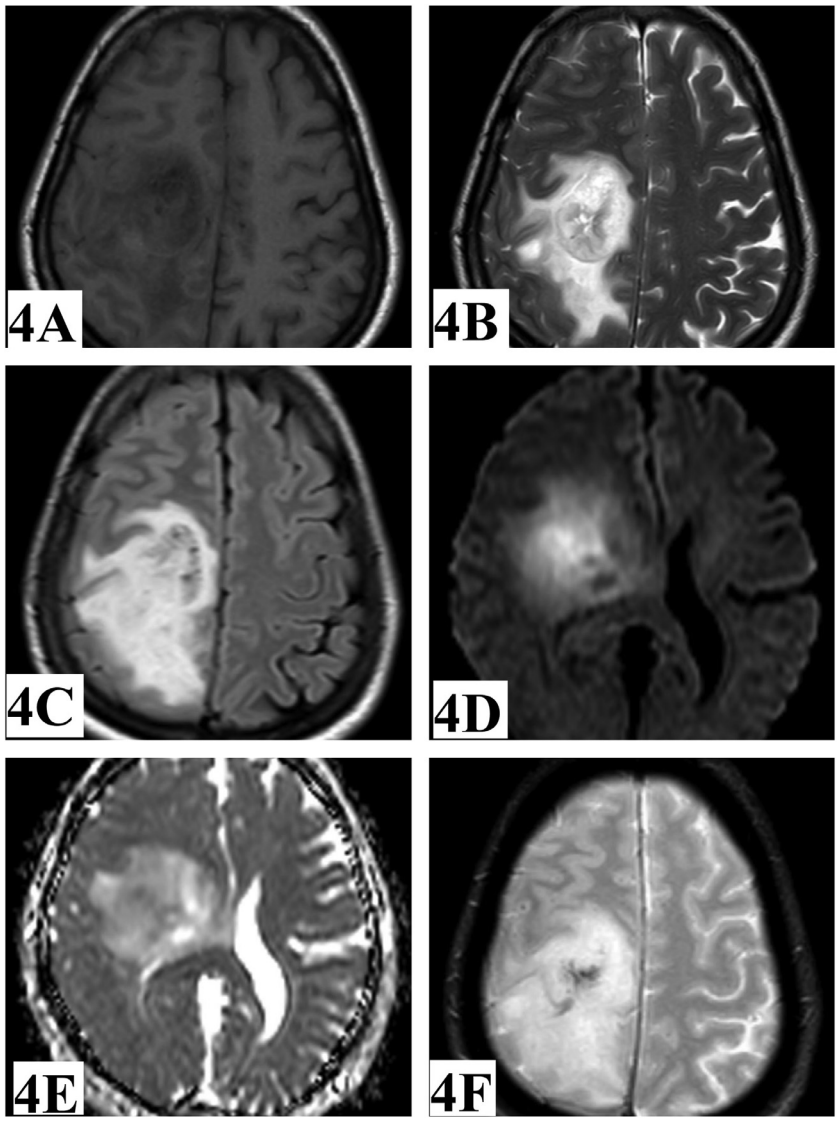

Fig. 4 – T1W, T2W and FLAIR (A-C) Axial images show ill-marginated lesion in right fronto-parietal lobes with significant perilesional edema. Areas of diffusion restriction are seen in DWI and corresponding ADC images (D and E) with blooming in GRE images (F).

sporadic and an association with maternal diabetes has been made. Adequate preconception control of diabetes and during the first few weeks of pregnancy has shown to reduce the incidence of this syndrome [4]. Second or trimester ultrasound can be used to make prenatal diagnosis of CRS in severe form. Ultrasound can also be helpful in diagnosing other associated anomalies. Antenatal ultrasound can be used to suspect or make the diagnosis and fetal MRI can be helpful in confirming the diagnosis and evaluation of other associated anomalies. However, it is difficult in milder forms.

Based on position of conus, CRS can be divided into 2 groups [5]:

a) Group 1: Conus ends cephalic to the lower border of L1 vertebrae and is blunt-ending. The vertebral deficit is large ending at or above S1 vertebrae.

b) Group 2: Conus ends caudal to the lower border of L1 vertebrae and is tethered (low-lying tethered cord). Vertebral deficit is less with identifiable portions of S1 or lower vertebrae.

Patients in group 1 have major sacral deformities, whereas neurologic disturbances are more severe in group 2.

Patients usually present in infancy or early childhood with diverse range of neurological symptoms arising from intricate spinal and neural abnormalities. These symptoms encompass neurogenic bladder dysfunction (urinary incontinence or retention), bowel disturbances (constipation or fecal incontinence), lower extremity weakness and sensory deficits in specific dermatomal distributions. This syndrome in adults often manifests as lumbosacral pain exacerbated by activities and bladder dysfunction [6].

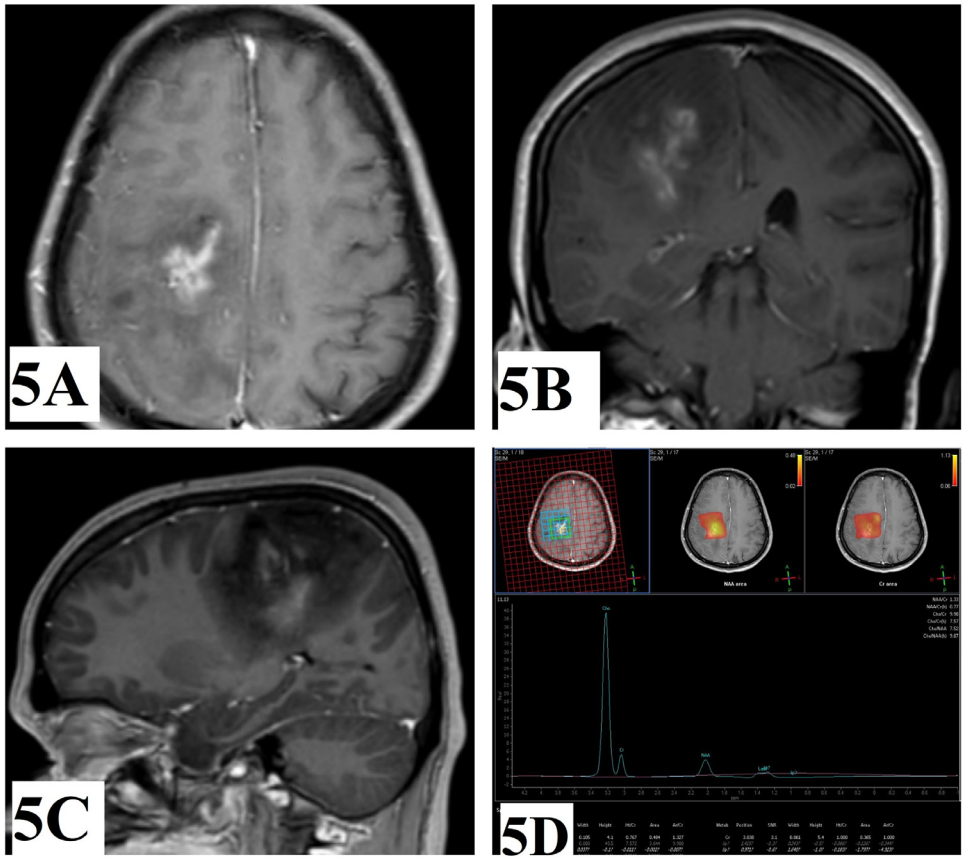

Fig. 5 – T1 Postcontrast images Axial, Coronal and Sagittal (A-C) show heterogeneous enhancement with raised Choline peak and significantly reduced creatinine and NAA peak in MRS (D).

Other clinical manifestations that may be observed are short intergluteal folds, flat buttocks, narrow hips, lower limb atrophy, hip dysplasia, and congenital talipes equinovarus (CTEV). Genitourinary abnormalities include horseshoe kidney, cystic dysplasia, neurogenic bladder, and Mullerian duct abnormalities, while gastrointestinal abnormalities may include duodenal atresia, imperforate anus, and intestinal malrotation [7].

Conclusion

Caudal regression syndrome is a rare congenital malformation predominantly involving lower vertebrae along with other constellations of malformation and not uncommonly involving the gastrointestinal and genitourinary syndrome. Most of the cases are diagnosed in early infancy or childhood and rarely in adulthood.

Our case illustrates an incidental finding of brain tumor which highlights the importance of thoroughly examining the surrounding structures to avoid missing major findings during imaging focused on a specific area of interest. In our case, while the spine was the primary focus, a partially visualized brain lesion was detected through meticulous evaluation. This prompted further investigation with contrast-enhanced MRI of the brain, ultimately leading to the radiological diagnosis of a high-grade glioma.

Patient consent

I, the author of the article "Beyond expectations: caudal regression syndrome with incidental brain tumor in an adult female: A case report" approve that the patient gives the consent for information to be published as a Case Report.

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