Caudal Regression Syndrome and Associated Abnormalities in a Adult Patient with mild Neurological Deficit: A Case Report

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Abstract

Caudal Regression Syndrome (CRS) is a spectrum of congenital malformations that consist of anomalies of the urinary and genital systems, the rectum, the lower limbs, and the lumbo-sacral spine. Though the exact etiology leading to caudal regression syndrome is still unknown, it has been hypothesised that genetic influence as well as maternal pathologic factor like diabetes plays an important role. The severity of CRS depends on function of the residual spinal cord. Patients may present with mild to severe neurological involvement with or without visceral anomaly.

Here, we report a case of caudal regression syndrome with associated spinal and vertebral abnormalities in an 20 year old male who presented with complaints of lower limb twitching movement and associated left club foot.

Keywords: Caudal regression syndrome; sacral agenesis; diastematomyelia

Introduction

Caudal regression syndrome(CRS) is a rare congenital anomaly, with incidence between 0.1 and 0.25 out of every 10,000 pregnancies, and a male-female ratio of 2.7:1. [1] Geoffroy Saint-Hilaire and Hohl first reported the features of this condition in 1852. [2] CRS characteristically involves lumbosacral vertebra and corresponding segments of the spinal cord, which innervate structures in the pelvis and lower limbs. [3] Malformations such as renal and duplex ureters, ureter agenesis, vesicoureteric reflux, neuropathic bladder, hydronephrosis, anorectal atresia, and imperforated anus. [4] Caudal regression syndrome occurs in up to 1% of pregnancies of diabetic mothers and have up to 22% of cases associated with Type I or Type II maternal diabetes mellitus (DM). [9]

Case Report

An 20 years old male presented with the complaints of episodic involuntary twitching movements of lower limb since birth. Physical and neurological examination of the

patient was performed which showed he had associated left cub food. The patient also had positive family history maternal diabetes.

MRI being the investigation of choice was recommended by the neurologist MRI of brain was performed with was unremarkable (Image 1-4).

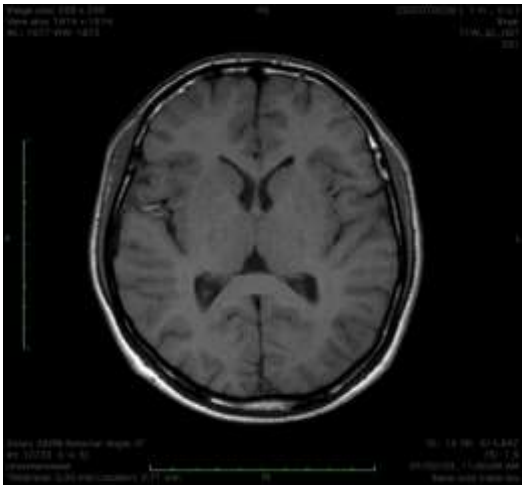
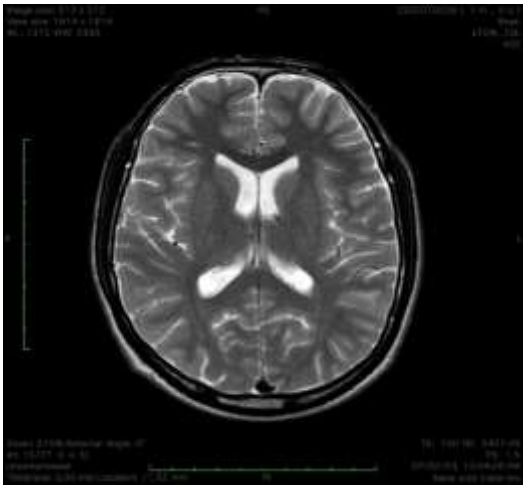


Image 1



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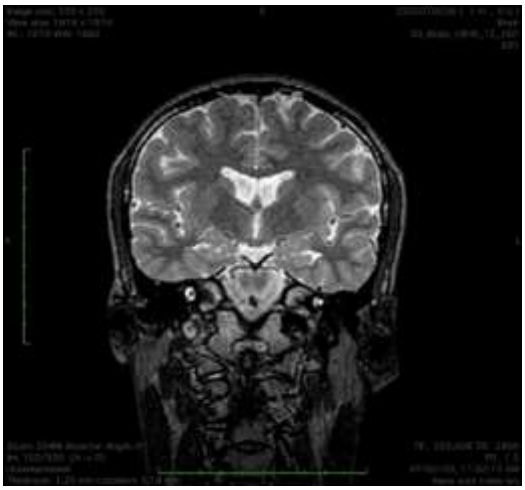

Image 3

Image 4

Images (1-4): Image 1: Axial T1W, Image 2: Axial T2W, Image 3: Thin Coronal T2W and Image 4: Sagittal FLAIR show no gross abnormality within the brain parenchyma.

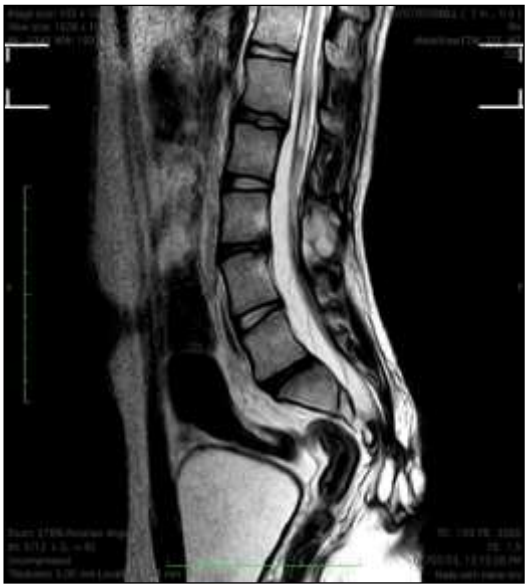

Image 5

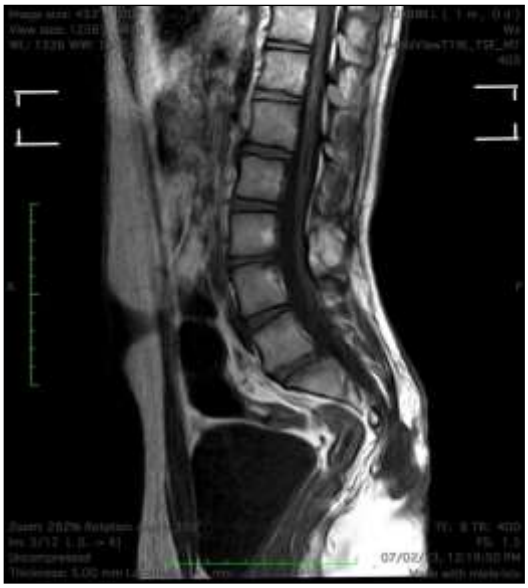

Image 6

Images 5 & 6: Image 5: Sagittal T2W and Image 6: Sagittal T1W show a multi-septated cystic lesion in the sacral region covered with skin.

Further MRI of whose spine was done which revealed that the spinal-cord was low lying and extends to the level of S2 vertebral body and was fused with a multiseptated skin covered cystic lesion measuring approximately 17.4 mm X 10.3 mm in size through a defect in the posterior sacrum. Sacrum was deformed with presence of only two to three sacral segments and non visualisation of coccyx. A sagittal cleft is also seen in L5 and S1 vertebral bodies. Extending from the level of upper border of L2 vertebral body, the spinal cord appears vertically split into two hemi-cords upto the level of lower body of L5.A syrinx cavity is seen in the spinal cord with involvement of right hemi-cord extending from D12-L5 vertebral body with maximum size of 0.5 cm X 0.4 cm in axial plane. Cervical curvature also reversed.

NCCT of the spine was performed to better access the spine. It was noted that the sacrum was deformed with presence of only two to three sacral segments and non visualisation of coccyx.

Based on MRI and NCCT findings we concluded with the diagnosis of caudal regression syndrome with hypoplastic sacrum with low lying spinal cord with diastematomyelia and syrinx cavity in the right hemi cord was made.

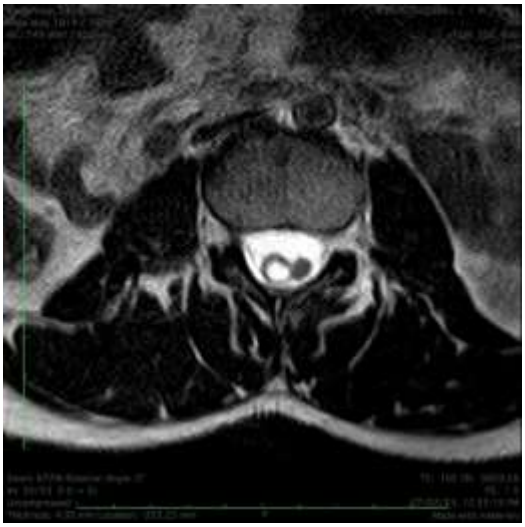

Image 7

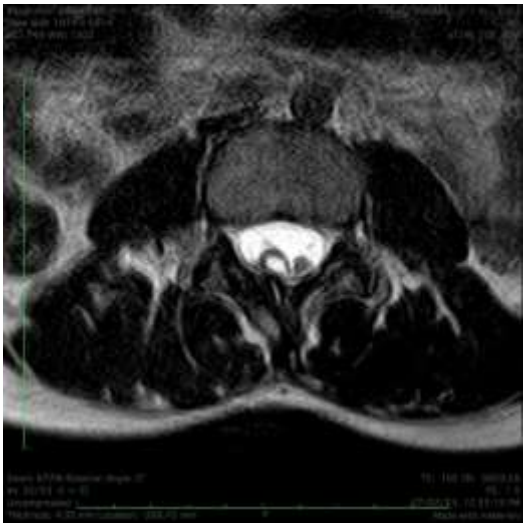

Image 8

Images 7 & 8: Axial T2W images through the lumbar region show vertically split spinal cord with two hemi-cords and associated hyper intense signal intensity in the right hemi-cord.

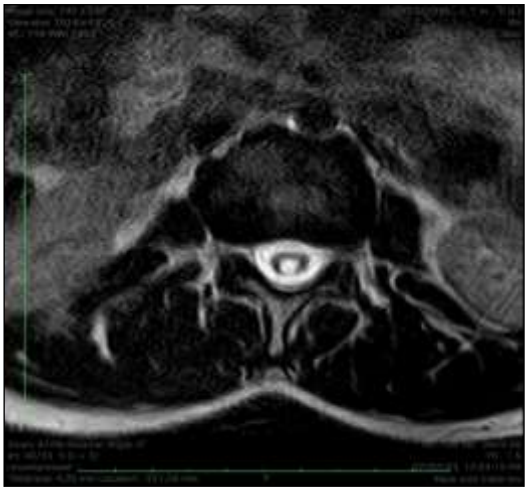

Image 9

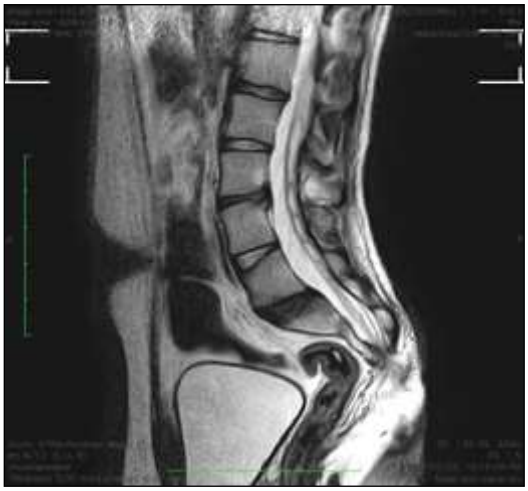

Image 10

Images 9 & 10: Axial T2W and Sagittal T2W images through the lumbar region shows hyperintense signal within the spinal cord.

Discussion

CRS is characterised by failure of development of lumbosacral vertebra and the caudal spinal cord. The severity can range from absent coccyx to complete absence of lumbar and sacral vertebra. It has complex aetiology with implication of both environmental and genetic factors in aetiopathogenesis. [5] Failure of notochord formation during the gastrulation phase of embryogenesis is the underlying defect. [6]

There is overlap of clinical features of caudal regression syndrome with clinical features of sirenomelia and VACTERL (vertebral defects, anal atresia, cardiac

defects, tracheoesophageal fistula, renal anomalies, and limb anomalies), complete systemic evaluation of the patient presenting with clinical feature should be done. [7] The patient in our case had associated left club foot. Cardiac, renal evaluation of the patient was normal, and there were no tracheaesophageal malformations.

Based on the vertebral defect type and nature of attachment of iliac bones to lowest vertebra, caudal regression syndrome can be classified into four distinct types. The classification system was proposed by Renshaw in 1978. [8] Type I CRS is characterized by either partial or complete unilateral sacral agenesis. Type II which is the most common form, shows partial sacral agenesis, with bilaterally symmetrical defect between ilia and either normal or hypoplastic first sacral vertebra. The type III vertebral defect is characterised by total sacral agenesis with associated variable lumbar vertebral agenesis. The two iliac bones are attached to the sides of the lowest vertebra. Type IV being the worst type, shows either fusion of iliac bones or iliac amphiarthrosis in addition to features described in type III. The patient in our case was categorised as type II CRS. [13]

Long-term prognosis is determined by the severity of defect in vertebra and spinal cord and associated malformations. [20] CRS is associated with long-term neurological, orthopaedic, and urologic complications. Although there were associated malformations like diastematomyelia with syrinx, sagittal cleft vertebrae and multiloculated cystic skin covered sacral lesion in our patient he presented with mild neurological and orthopaedic complications like contagious twitching of legs and associated club foot on left side.

conclusion

CRS is a rare entity with a spectrum of congenital malformations and known association with maternal diabetes. It is characterised by sacrococcygeal dysgenesis and an abrupt termination of a blunt-ending spinal cord. MRI is the imaging modality of choice in adults. Early detection and prompt treatment is of utmost importance to decrease the risk of urinary incontinence, recurrent urinary tract infections, renal impairment, the development of a neuropathic bladder, and thus, to improve the prognosis.

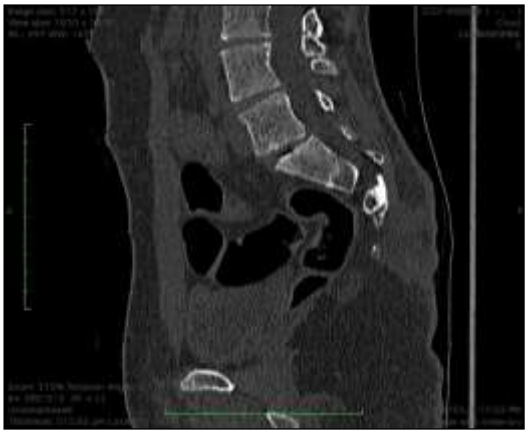

Image 11

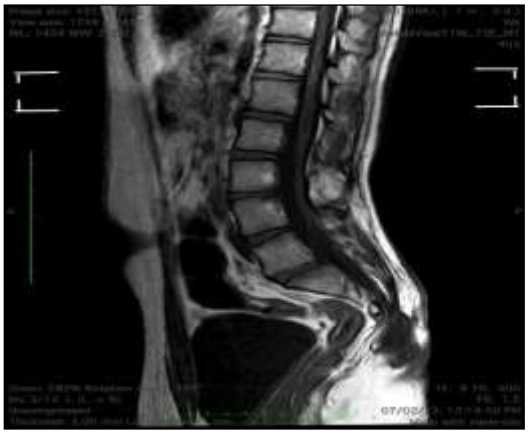

Image 12

Image 11 & 12: Image 11: Sagittal NCCT (Bone window) and Image 12 (Sagittal T1W) through the lumbar region shows non-visualisation of the coccyx with presence of only 2-3 sacral segments.

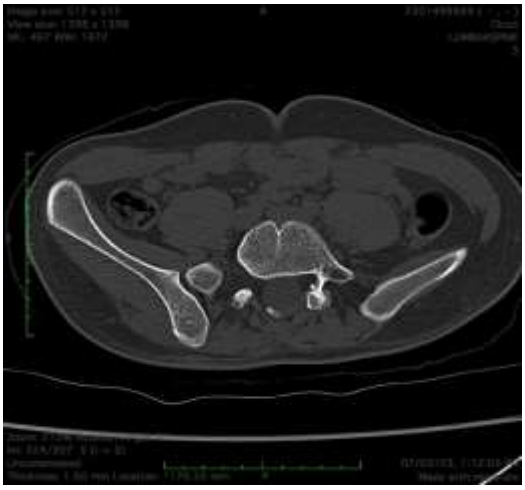

Image 13

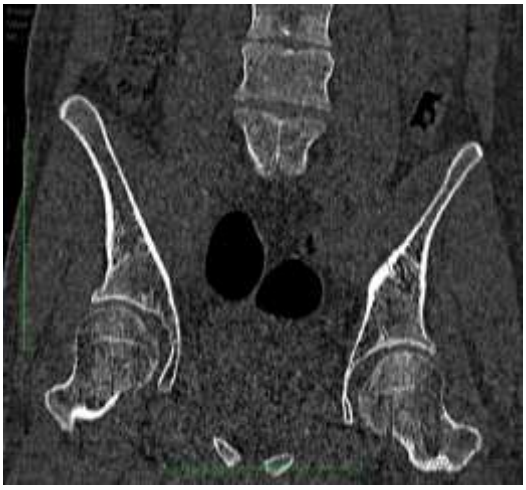

Image 14

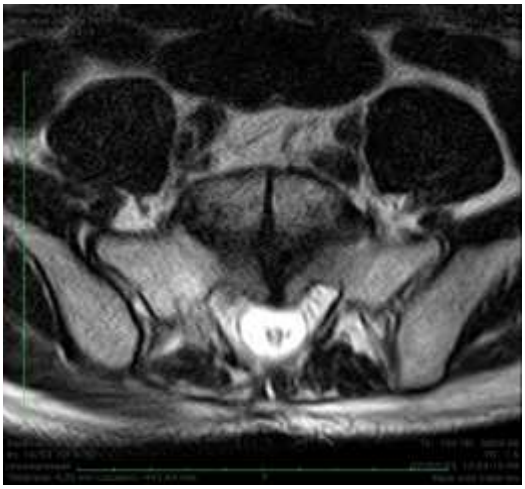

Image 15

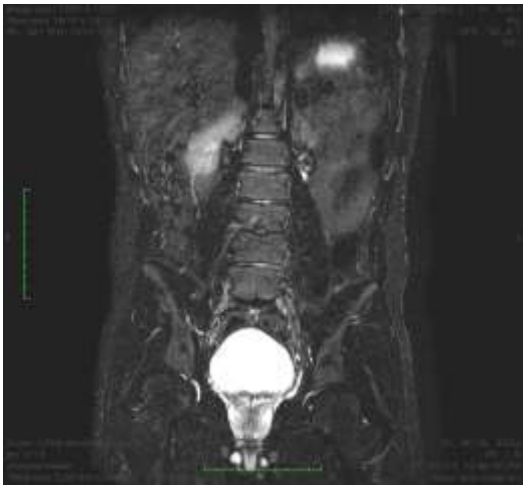

Image 16

Images (13-16): Image 13: Axial T2W image at the level of sacroiliac joints, Image 14 Coronal STIR image of the lower spine, Image 15: Axial NCCT and Image 16: Coronal NCCT at level of L5 vertebra show a sagittal cleft in the midline at the L5-S1 level.

References

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