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**Research Paper**

# Caudal Regression Syndrome- An Interesting Case Report

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***ABSTRACT:*** *Caudal regression syndrome (CRS) is a rare congenital disorder, characterized by abnormal development of structures in the caudal region of the embryo like lower lumbar and sacral vertebrae, urogenital and lower gastrointestinal system. It is due to abnormal development of mesoderm. Trauma, nutritional deficit, toxic substances, and genetics (mutations) are considered as the etiology of CRS. In this paper, we report a 10–year-old boy with caudal regression syndrome. Patient presented with urinary and fecal incontinence with club feet. In this patient, caudal regression syndrome was demonstrated on imaging.*

***Key words:*** *Sacral agenesis, Club foot, Urinary incontinence.*

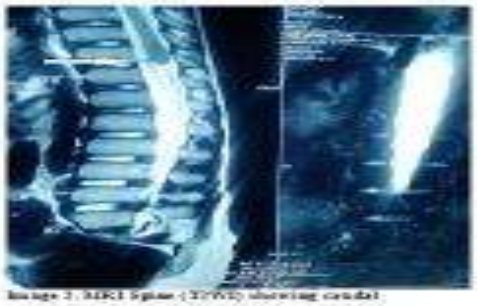
## I. INTRODUCTION

Caudal regression syndrome is a rare syndrome with an incidence of 1:25000 live births, characterised by malformations of the structures derived from the caudal region (or mesoderm) of the embryo that is, the urogenital system, the hindgut, caudal spine and spinal cord, and the lower limbs. It is much more common among overt diabetic mothers.[1, 2] Severe forms are commonly associated with cardiac, renal and respiratory problems, which are responsible for early neonatal death. Now a days magnetic resonance imaging (MRI) is the gold standard for diagnosis.[3, 4] The decision for surgical intervention depends upon the extent of neurological deficits. There is a definite but incomplete association of the syndrome with diabetes mellitus (1%). Teratogens and 7q mutation have been suggested as other factors.[5] We report a 10–year-old boy with caudal regression syndrome. Patient presented with urinary and fecal incontinence with club foot. In this patient, caudal regression syndrome was demonstrated on imaging (MRI).

## II. CASE REPORT

A 16-year-old boy presented to us with urinary and fecal incontinence with Club feet and syndactyly (Image 1). There was no history of maternal diabetes or teratogenic substance intake during gestation with negative familial history. The post-natal period was uneventful. The developmental milestones were normal. On neurological examination tone normal, power proximal 4+/5 and distal 4/5. DTRs all present except ankle which is absent. Sensory examination was normal. There was no cognitive decline and on mini mental scoring, scored 30 out 30. On Echocardiogram, no cardiac anomaly detected. Ultrasonography of abdomen and pelvis was normal. MRI of the spine was performed with sagittal and axial T1WI, T2WI along with MR myelography. Lumbosacral MRI scan showed termination of the spinal cord at the T11 level and narrowing of the canal at the S1 level (Image 2). On this basis the patient diagnosed as caudal regression syndrome. No fracture lines were found at bony structures, but the sacral agenesis was shown on MRI films. The spinal cord fit the classic caudal regression syndrome imaging by lumbar MRI. On micturating cystourethogram (MCU) detrusor hyperreflexia was shown.

**Image 1. Patient with club feet and syndactyly.**



## III. DISCUSSION

Duhamel first proposed caudal regression syndrome, based on his observation that sirenoid monsters have fusion of their lower limbs or synmelia.[6] It is generally diagnosed prenatally on in immediate postnatal period that may requires surgical intervention. Patient may have minimal deficits to severe paralysis depending upon the neural involvement. It occurs more frequently in the offspring of diabetic versus non-diabetic mothers. Trauma, nutritional deficit, toxic agents and genetics are the other factors suggested in the etiology.[3, 7] Our patient is an offspring of a non-diabetic mother and presented to us in late childhood period. Had a normal post-natal period with bladder and bowel incontinence since birth. He had recurrent falls probably due to motor weakness and club feet. The motor weakness and sensory involvement was not prominent. He didn't have cardiac or gastrointestinal defects. Patient is mentally normal. The superiority of lumbosacral MRI over Myelography and myelo-CT is generally accepted today.[4, 8] In our patient on MRI spinal cord ends at lower border of T11 with agenesis of sacral and coccygeal bones.

## IV. CONCLUSION

In our patient, there was no history of maternal diabetes or teratogenic substance intake during gestation with negative familial history. The diagnosis was overlooked by the primary physician and was confirmed by imaging. Hence, we suggest that presence of bladder and bowel dysfunction along with congenital anomalies in childhood period should point towards possibility of caudal regression syndrome.

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