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# CAUDAL REGRESSION SYNDROME ASSOCIATED WITH RECURRENT URINARY TRACT INFECTION: A CASE REPORT

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**ABSTRACT** Caudal Regression Syndrome (CRS) is a rare congenital malformation of the lower spine and spinal cord. It can range from absent coccyx without neurological sequelae to lumbosacral agenesis, leading to a spectrum of clinical presentations. Genitourinary and gastrointestinal abnormalities are prevalent, including neurogenic bladder and bowel incontinence. Neurogenic bladder can lead to reflux of urine and frequent urinary tract infections (UTIs), damaging the kidneys. Early diagnosis and treatment are the key to preventing irreversible kidney damage. Here we are presenting a 2-year, 8-month female child with recurrent UTIs with left hydroureteronephrosis (HUN) with bladder and bowel incontinence. On evaluation, we diagnosed caudal regression syndrome. The diagnosis was confirmed with an MRI of the spine, and further management was planned.

**KEYWORDS :** caudal regression syndrome, sacral agenesis, Genito-urinary anomalies, neurogenic bladder, urinary tract infection

## INTRODUCTION

CRS, also known as caudal dysplasia and sacral agenesis syndrome, occurs in 0.1-0.25 per 10,000 normal pregnancies. Infants born to diabetes moms have a 200-fold higher incidence (one in 350) (1). Geoffroy Saint-Hilaire and Hohl first characterized Caudal Regression Syndrome in 1852, and Duhmel developed the term in 1960 (2,3). It is caused by a failure to induce caudal elements in the embryo before the 28th day of gestation. The injury occurs along the posterior medial mesodermic axis, preventing the development of the caudal mesoblastic yolk (4,5). The cause could be due to maternal diabetes, genetics, vascular hypoperfusion, or teratogens (6). It can affect the lower limbs, lumbar and coccygeal vertebrae, as well as spinal cord segments.

Common associated findings include orthopedic, neurologic, gastrointestinal, genitourinary, and cardiac defects, as well as imperforate anus, renal dysplasia, and deformed genitalia (3). Renshaw divided individuals into four kinds, with the severity of abnormalities increasing from type 1 to 4(7). Affected children usually have a tiny pelvis, small flat buttocks, hip dysplasia, lower limb hypoplasia, CTEV (congenital talipes equinovarus), delay in gross motor milestones (8). Some children may experience constipation, bladder dyssynergia, or urine incontinence, which can lead to recurrent UTIs.

## CASE STUDY

2year, 8month female child who is developmentally normal, born to 2nd degree consanguineous parents (6th in birth order) with no antenatal risk factors. Parents observed straining while passing urine at 9 days of age. At 3 months of age, the child developed a fever associated with straining and crying during micturition, was diagnosed with a culture-positive UTI, and treated with a sensitive antibiotic. Mother noticed dribbling of urine on standing, straining, and constipation from 9 months to 1 year of age. Till 1 year of age, the child had a total of 3 episodes of culture-positive UTIs that required IV antibiotics; in between, the child is on UTI prophylaxis. Initial USG KUB was normal; at 1 year old, it showed left hydronephrosis (HDN) with a mildly dilated upper ureter. MCUG showed left grade 5 VUR (Vesicoureteral reflux) with hydroureteronephrosis (HUN) (image 1). DMSA showed left diffuse multifocal cortical scarring (image 2). Renal function tests are normal and no hypertension. For VUR, the child underwent ureteric reimplantation and DJ stenting at 1 year and 3 months; the stent was removed after 3 months; MSUG was repeated, which showed no VUR with elongated bladder with small diverticula with irregular bladder wall; but in USG, HUN persisted with significant post-void residual urine. In view of recurrent UTIs even after correcting VUR, the child was referred to us for further evaluation. On examination, the child's weight and height are appropriate for age. No dysmorphic features. There is a small pelvis with small flat buttocks and bilateral buttock dimples with short intergluteal cleft with Mongolian spots, with overflow incontinence and dribbling of urine (image 3). Neurological examination showed absent anal reflex and wasting of lower limb muscles. We suspected neurogenic bladder probably due to lower spinal cord pathology, so investigated with MRI Lumbosacral spine, which revealed partial agenesis of the S3 vertebra with the absence of S4, and S5 vertebra along with coccyx (image 4), with abrupt cut-off of the spinal cord at the level of L1, suggestive of Pang type 3 caudal regression syndrome (image 5). The urodynamic study showed detrusor overactivity. The urologist advised tamsulosin as it improves bladder storage and emptying, and planned for Blocksom vesicostomy. For constipation, the child is on oral PEGLEC powder.

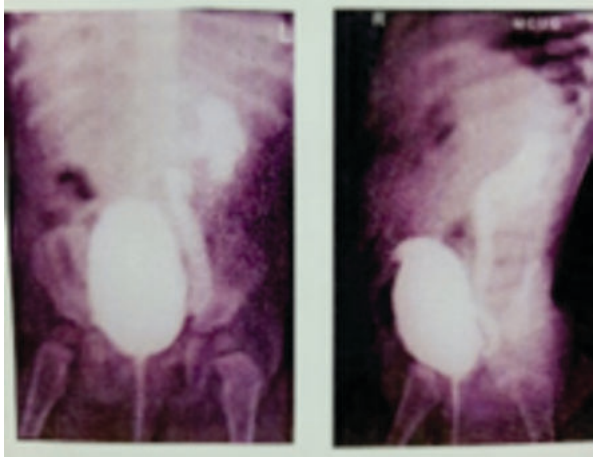

**Image 1.** Left HUN, Grade 5 VUR

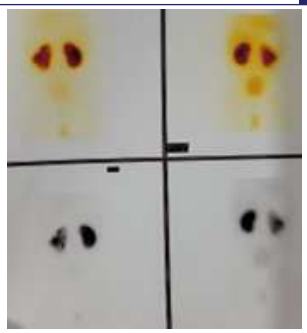

**Image 2.** DMSA scan- Left kidney diffuse multifocal scarring.

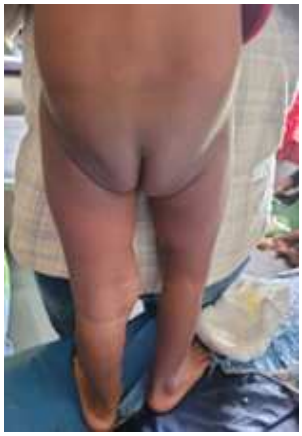

**Image 3.** Small pelvis with small flat buttocks and bilateral buttock dimples with short intergluteal cleft.

Coronal T2 weighted MRI Lumbosacral spine

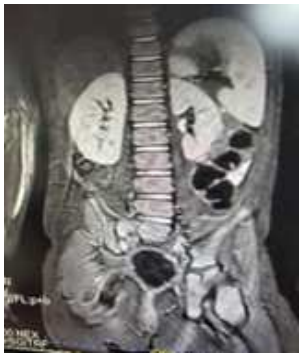

**Image 4.** Sacral Vertebrae: S1- fused with iliac bone on right side, S2- normal. S3- partial agenesis. S4, S5 and coccyx absent.

**Urinary Bladder:** Pear shaped with irregular wall thickening with diverticula- Neurogenic bladder

Sagittal T2 weighted MRI Lumbosacral spine

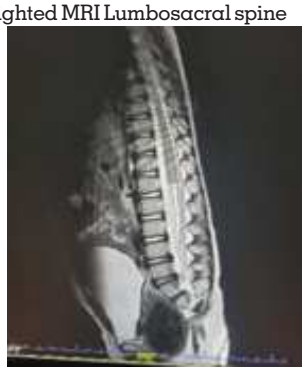

**Image 5.** Absence of conus medullaris with abrupt cutoff of spinal cord at L1 vertebra.

## DISCUSSION

CRS is strongly linked to maternal diabetes [9]. CRS abnormalities are associated with the caudal type homeobox 2 (CDX2) gene [10]. It has been linked to Currarino syndrome, OIES syndrome (omphalocele-exstrophy of the cloaca-imperforate anus-spinal malformations), and VACTERL (vertebral defects, anal atresia, cardiac defects, tracheo-esophageal fistula, renal anomalies, and limb abnormalities) [11,12,13]. Pang et al. divided CRS into five kinds. Type 1 caudal agenesis (CA) including lumbar vertebrae, Type 2 total CA, Type 3 Subtotal CA at least S1 present, Type 4 Hemi sacrum, Type 5 Coccygeal agenesis (14). Our case comes under type 3 Pang CRS.

Clinical signs range from asymptomatic to serious neurological impairments and orthopedic difficulties affecting the lower limbs. Motor and sensory impairment are of lesser severity; paresis is typically more severe than sensory impairment [15]. Muscle paralysis at the S2-S5 cord levels, as well as urethral and anal sphincter dysfunction (7). Patients with spinal cord anomalies at a little higher level, L5-S1 (subtotal or complete sacral agenesis), experience more severe neurological symptoms (16). Absence of more than 1 sacral vertebra leads to neuropathic bladder (17). It is commonly associated with Genito-urinary anomalies (3). Neuropathic bladder with VUR with associated recurrent UTIs leading to kidney damage (18). There is associated fecal incontinence in 33% of cases (19). MRI is an excellent tool for assessing the state of the vertebra, spinal cord, intra- and extra-dural lesions, and related concurrent abnormalities in juvenile CRS patients, as well as directing future CRS care (20). The presence of obvious deformities in the spine and lower limbs, as well as an imperforate anus, results in an early identification of the condition, shortly after delivery. Less severe abnormalities, such as minor sacral underdevelopment, may be discovered later.

## CONCLUSIONS

Infantile onset and recurrent UTIs with urinary incontinence associated with bowel symptoms definitely need to be thoroughly investigated for organic (lower spine) pathology. A detailed general physical examination of the child is very important in such cases, as this child having phenotypic features of sacral hypoplasia with lower limb wasting. It guided us towards CRS. The presence of obvious deformities in the spine and lower limbs, as well as an imperforate anus, results in an early identification of the condition, may be after delivery but less severe abnormalities, such as minor sacral underdevelopment will be missed. Along with neurogenic bladder associated Genito-urinary anomalies prone them to recurrent urinary tract infections.

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