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A Case Report on Caudal Regression Syndrome

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Abstract: ***Caudal regression syndrome is a very rare neural tube disorder, in which the caudal vertebral column and spinal cord develop abnormally, causing substantial sensory and motor deficits, primarily in the legs. An abnormality in the spinal cord and nerve roots interacts with a section of lumbar, lumbosacral, or coccygeal spinal dysgenesis or agenesis. Here, we present a rare case of caudal regression syndrome in a 12 - year - old female child who was brought to the pediatrics department with a known case of neurogenic bladder and bowel dissected with pain in bilateral lower legs which was shock like, on and off (1 - 2 episodes/week), aggravated in cold temperature and bladder disturbances. For which, a neurological opinion was taken. Plain X - ray revealed a partial sacral agenesis, MRI revealed sacralization of L5 vertebral body is seen. On the basis of radiological findings, a diagnosis of the caudal regression syndrome was confirmed.***

Keywords: CRS, MRI, MCUG, PCS and ultrasound KUB

1. Introduction

Caudal regression syndrome (CRS) is a rare congenital disorder. It's estimated that 1 to 2.5 in every 100, 000 newborns is born with this condition. Caudal regression syndrome is characterized by abnormal development of the lower (caudal) end of the spine. A wide range of abnormalities may potentially occur in infants with caudal regression syndrome including abnormal development (agenesis) of the sacrum and coccyx and abnormalities of the lumbar spine. Abnormalities of the lower spine can cause a variety of additional complications including joint contractures, clubfeet and disruption or damage of the end of the spinal cord may occur, potentially causing urinary incontinence. Additional anomalies of the gastrointestinal tract, kidneys, heart, respiratory system, upper limbs and upper portions of the spine can also occur.

2. Causes

The condition is likely caused by the interaction of multiple genetic and environmental factors. One risk factor for the development of caudal regression syndrome is the presence of diabetes in the mother. It is thought that increased blood sugar levels and other metabolic problems related to diabetes may have a harmful effect on a developing fetus, increasing the likelihood of developing caudal regression Syndrome.

- **Mesoderm**: The mesoderm is the middle layer of tissue in a developing embryo that consists of a group of cells that are responsible for building structural parts (bones and organs) of a fetus' body. Around day 28 of pregnancy, genetic changes can affect the development of the mesoderm.
- **Abdominal artery abnormality**: An artery is a tube responsible for sending blood to different parts of your body. For a developing fetus, the artery that brings blood to the lower part of their body is facing a direction other than toward the lower part of their body, which prevents blood from reaching that area.
- **Genetic mutation**: Changes to your genes that occur during conception (when the egg and sperm meet) can cause this condition, specifically genetic mutations in the HLXB9 or the VANGL1[7].

Clinical Presentation

Caudal regression syndrome may present with a broad range of symptoms:

- neurogenic bladder and anorectal malformations
- Sensorimotor paresis (motor deficits > sensory deficits)
- Features of sacral agenesis: narrow hips, hypoplastic gluteal muscles, shallow intergluteal cleft
- Mild foot deformities and gait abnormalities
- Infants with underdeveloped bones in their legs caused by caudal regression syndrome may have an irregular stance that can affect how they walk. This could include:
- Frog - leg position: child's legs bend with their knees pointed outward and their feet in line with their hips.
- Clubfeet
- Calcaneovalgus: child's feet turn outward and upward.

Organ symptoms

Symptoms of caudal regression syndrome can affect the development and function of child's renal system, digestive and urinary tract. Symptoms could include:

- Abnormally shaped kidneys, a missing kidney or the kidneys fuse together. This can cause kidney failure.
- More tubes than necessary take urine from their kidneys to their bladder (ureteral duplication).
- Frequent urinary tract infections.
- Their bladder comes out of an opening in their abdominal wall (bladder exstrophy).
- Bladder nerves that control the function of their bladder don't send signals to their brain to work as expected (neurogenic bladder).

- Lack of bladder or bowel control.
- Constipation.
- Blocked anal opening (imperforate anus).

Caudal regression syndrome can also affect the development and function of child's reproductive organs. Symptoms that affect their reproductive organs include:

- The opening of their urethra is on the underside of their penis (hypospadias).
- Their testes don't descend (cryptorchidism).
- The connection between the lower part of their large intestine (rectum) and their vagina causes bowel contents to leak out and pass through their vagina (rectovaginal fistula).
- Reproductive organs didn't develop (genital agenesis).

Symptoms of caudal regression syndrome can also affect other internal organs in child's body, including:

- Twisting of their large intestine.
- A bulge in their groin or lower abdomen (inguinal hernia).
- Congenital heart conditions.

Diagnosis

Radiographic features imaging appearances can significantly vary depending on the severity of regression. In general, the following may be seen:

- Lumbosacral vertebral body dysgenesis/hypogenesis
- The level of atresia/dysgenesis is usually below L1 and often limited to the sacrum
- Truncated blunt spinal cord terminating above the expected level (wedged - or cigar - shaped conus medullaris)
- Severe canal narrowing rostral to last intact vertebra

Antenatal ultrasound

- A blunted sharp ending distal cord on a longitudinal sonogram is typical
- The conus often ends way above the expected level (sometimes even higher than L1) [5]
- Absent/hypoplastic sacrum
- Hypoplastic extended lower extremities (limbs are separated of sirenomelia)
- May show a "shield sign": opposed iliac bones in absence of sacral vertebrae: typically seen on an axial scan
- Fetal extremities may be seen in a "crossed legged tailor" position or a "Buddha" position
- In an early scan (1st trimester), the crown - rump length may be less than expected for gestational age as an indirect feature.

MRI

- It shows similar features to those on ultrasound but in more detail
- Useful to assess canal stenosis
- A characteristic wedge - shaped cord terminus may be seen
- Imaging allows the differentiation of two broad groups of patients with caudal regression syndrome.

Group 1:

1) The conus medullaris is blunt and terminates above the normal level; there is sometimes an associated dilated central canal or a cerebrospinal fluid - filled cyst at the lower end of the conus.
2) These patients have major sacral deformities.

Group 2:

1) The conus medullaris is elongated and tethered by a thickened filum terminale or intraspinal lipoma and ends below the normal level. Neurologic disturbances are more severe in this group[3].

Management

Children with caudal regression syndrome may need ongoing physical therapy, psychiatric support, and other supportive services. Pediatricians, neurosurgeons, neurologists, urologists, orthopedists, orthopedist surgeons, cardiologists, kidney specialists (nephrologists) and other health care professionals may need to systematically and comprehensively plan an affect child's treatment.2

- Surgery to repair skeletal system symptoms or symptoms that affect your child's bones.
- Surgery to improve the function of your child's genitals, organs, urinary tract and/or bowels.
- Medications to reduce symptoms.
- Using a ventilator to help with breathing.
- Wearing a brace, using prosthetics or assisted mobility devices to help your child move.

Your child may need more than one surgery to manage their symptoms as they grow. Based on your child's symptoms, they might need to start treatment with medications or surgery soon after they're born or during early infancy.

Multiple surgeries may be necessary to treat various urological, spinal and cardiac abnormalities, anal atresia and certain limb deformities associated with caudal regression syndrome. In addition, anticholingergic drugs may be administered to treat urological abnormalities.2

3. Case Report

A 12 - year - old female child was brought to the department of neurosurgery department with a known case of neurogenic bladder and bowel dissected with pain in bilateral lower legs which was shock like, on and off (1 - 2 episodes/week), aggravated in cold temperature, relieved by medication (crocin). she also has complaints of bladder and bowel disturbances for the past days, not able to control over bladder and bowel disturbances or bowel). The child was delivered through LSCS at full term. She was admitted in Rainbow children's hospital with a complaints of urinary tract infection one year ago (klebsiella), on CIC (clean intermittent catheterization) she has dribbling of urine from 2 years of age and incomplete evacuation feaces. She has a past history of spinal surgery at 2 years of age. The patient has undergone detethering of cord and excision of lipoma surgery.

For which, a pediatric neurologist was consulted and advised for MCUG (MICTUATING CYSTO - URETHROGRAM), ultrasound screening scan and X - Ray by the department of

radiology. The ultrasound KUB shows RK 10.7cms (minimally prominent PCS), LK 9.9cms (minimally prominent PCS with cortical scar in lower pole). elongated contour of bladder, variable mild prominent of both lower ureters and diffuse irregular wall thickening with trabeculations. In MCUG, few distended bowel loops are noted and distended bladder is irregular in outline, there are few trabeculations and sacculations noted (bladder hypertropic changes). The X - Ray impression shows sacralization of L5 vertebral body. The sacrum is deformed and not formed on left side from S2 level with posterior elements defect. Tethered low lying cord with the neural placode ending at S2 level into fatty elements.

4. Discussion

The Caudal regression syndrome is a rare congenital malformation. Even though specific etiologic factor is unknown, it is related to maternal diabetes, genetic predisposition and vascular hypoperfusion3. Just as in the case presented, this alteration is characterized by agenesis of the sacrum involving iliac and lumbar vertebrae with their corresponding spinal segments and variable abnormalities in the lower limbs as well as in other organs. Fetal diagnosis tools allow early syndrome detection. Prenatal ultrasound is the most frequently used paraclinical instrument; a key element of prenatal ultrasound is the detailed evaluation of the spine and lower limbs; it also allows for a Caudal regression syndrome diagnosis through the demonstration of an abrupt termination of the lumbar spine and hypoplastic lower limbs.

The ultrasound KUB shows that the right kidney measures 10.7cms. It appears normal in size and shows cortical echoes. No evidence of calculi. Minimally prominent pelvicalyceal system. Left kidney measures 9.9cms. it appears normal in size and shows normal cortical echoes. No evidence of calculi. Minimally prominent pelvicalyceal system. Cortical scar in lower pole of left kidney. Variable mild prominence of both lower ureters. Urinary bladder is elongated contour of bladder. Diffuse irregular wall thickening with trabeculations (fig.1 & 2). Pre void measures 278ml, post void measures 128ml and post CIC measures 128ml (fig.3).

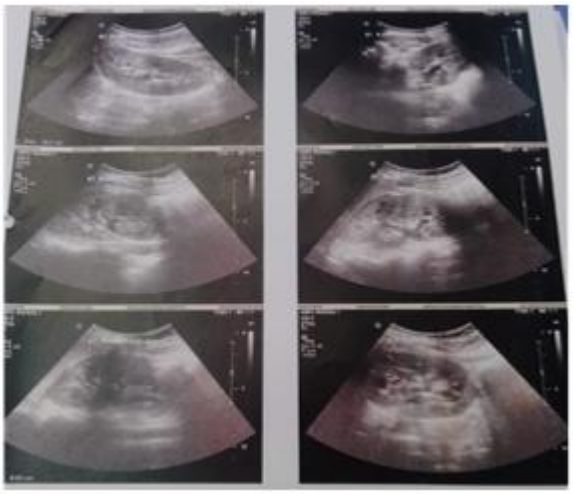

Figure 1

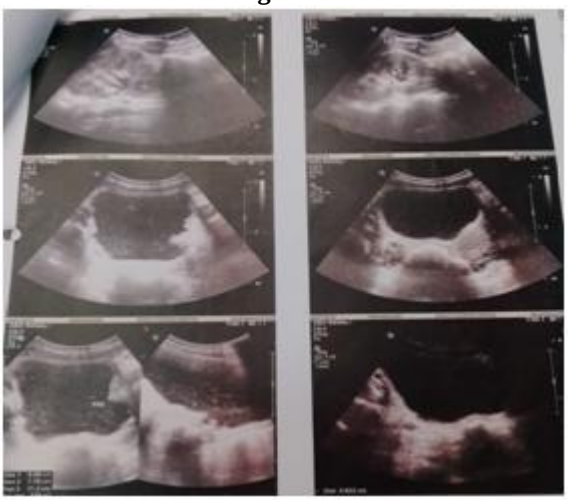

Figure 2

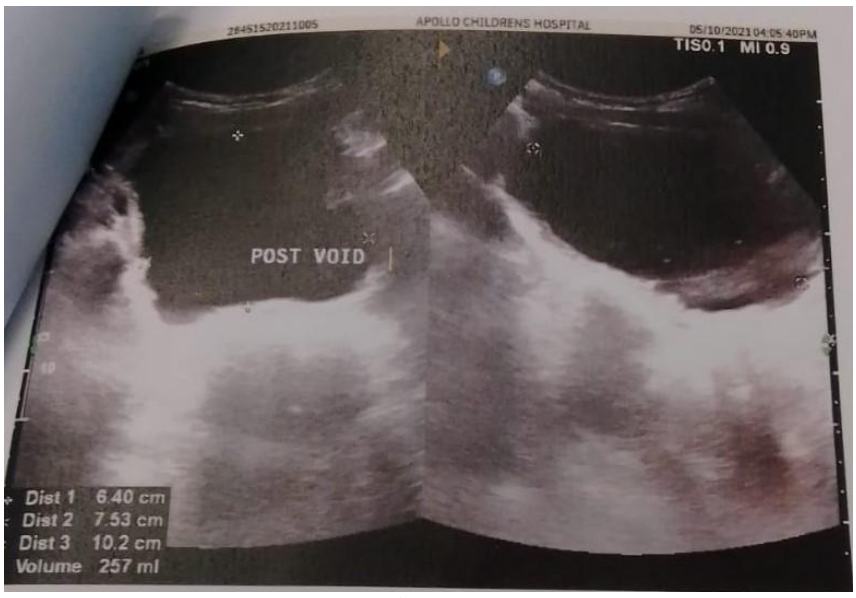

Figure 3

The MRI lumbar spine and spine survey shows sacralisation of L5 vertebral body is seen. The sacrum is deformed and not formed on left side from S2 level with posterior elements defect. Left pyriformis muscle is not clearly seen. There is a loss of normal conus bulge. Tethered low lying cord is seen with the neural placode ending at S2 level into fatty elements. The neural element is extruding outside through the defect and the subcutaneous fat is seen extending into canal. Intact skin covering is also evident. Multilevel syrinx formation is seen in the spinal cord extending from C4 vertebra upto conus medullaris. Maximum diameter measures 7mm at D11 vertebral body. Few T1 hypointense and T2/STIR hyperintense lesions are seen in the body of sacrum on right side. Lumbar vertebral bodies are normal in height. Alignment is preserved. No spondylolisthesis. Lumbar intervertebral disc demonstrate normal signal with preserved disc height.

The MUCG (MICTURATING CYSTO - URETHROGRAM) report shows distended bladder is irregular in outline, there are few trabeculations and sacculations noted (bladder hypertropic changes). Impression shows no vesico ureter reflux and there is no significant post void residual urine noted.

The child was admitted with the above mentioned complaints and findings. After all pre operative investigations, informed consent and pre anaesthetic checkup, the child underwent detethering of cord and excision of lipoma. Under ETGA, child positioned prone over bolsters, parts prepared and draped. Vertical incision made around the first scar to excise the scar and deepened. Bilateral paraspinal muscles elevated subperiosteally. Subcutaneous tissue dissected to reach dura and dura opened verterbrally. Lipoma dissected cut and excised tethered nerve roots released. Hemostasis achieved. Dura closed primarily. Wound closed in layers. Ethilon mattress cutaneous to skin. Post operatively child was treated with antibiotics such as inj. supacef and analgesics such as tramadol. Child was clinically and hemodynamically stable.

The child was found to be hemodynamically stable and hence being discharged with advice of physical activity and the following medications such as tablet Etoshine 120mg, tablet Ibuprofen 400mg TDS, tab Dolo, tab Pan 40mg, tab Bactrim 480mg OD, tab Pregabid, Jusdee, tab Diamox 250mg, tab Lonazep and tab Zincovit.

5. Conclusion

Caudal regression syndrome is a rare complex disorder characterized by abnormal development of the lower (caudal) of the spine. The exact cause is unknown. The severity of the spinal abnormalities and any concomitant symptoms define the prognosis for caudal regression syndrome. The caudal regression syndrome remains associated with structural and systematic problems including genitourinary, gastrointestinal, orthopaedic, neurological, respiratory and cardiac anomalies. Early detection and management is necessary. This needs a careful investigation, evaluation, pre - operative planning and regular followup. The treatment of caudal regression syndrome is symptomatic and is largely supportive.

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[7] Some information also is posted on the following page on the website: https: //my. clevelandclinic. org/health/diseases/23425 - caudal - regression - syndrome