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CAUDAL REGRESSION SYNDROME: A RARE AND INTERESTING CASE REPORT

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BACKGROUND

Caudal Regression Syndrome (CRS) or Caudal dysgenesis syndrome (CDS) is a rare disorder characterized by maldevelopment of the caudal half of the body with variable involvement of the gastrointestinal, genitourinary, skeletal and nervous systems[1,2,3,4,5]. It includes a wide range of congenital anomalies with abnormal features ranging throughout the body including caudal spine and spinal cord, the hindgut, the urogenital system and the lower limbs, with a wide variation in the spectrum of severity. An incidence rate of 1-3 newborns per 100,000 live births is reported with the prevalence rate higher amongst infants of diabetic mothers. A multidisciplinary management approach is needed, and many patients may require a staged surgical approach depending on the clinical features present and degree of disability. Here we describe a patient with Caudal regression syndrome that presented to our hospital.

Case Presentation

We present a case of an eleven month male child, 1st born of second degree consanguineous marriage who was referred to our neurosurgical unit with maldevelopment of bilateral lower limbs and abnormal attitude of the upper and lower limbs. On initial assessment, the patient was found to have underdeveloped lower limbs, bilateral club feet (Figure 1), hypospadias (Figure 2), underdeveloped anus (Figure 3), an absent sacrum and scoliosis. Patient also had history of dribbling of urine and fecal incontinence. A bony hard mass of approximately 3x2 cm was felt over the lumbar spine at L4 level. There were bilateral dimples in the gluteal region signifying maldeveloped pelvis (Figure 4). The upper limbs were noted to be in an adducted and internally rotated position and lower limbs were flexed. The patient had crossed feet (Figure 1) and prefers to sit with pressure over the hips (Buddha position). The anus was patulous with no sphincter activity (Figure 3). The patient also had a left undescended testis. On neurodevelopmental assessment the patient had mild to moderate language, social, cognitive and fine motor deficits and moderate to severe motor deficit especially related to ambulation.

Figure 1: Crossed Feet, Underdeveloped Lower Limbs And Bilateral Club Feet

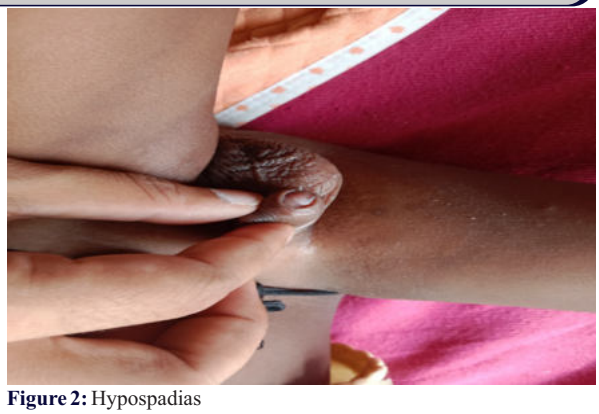


Figure 2: Hypospadias

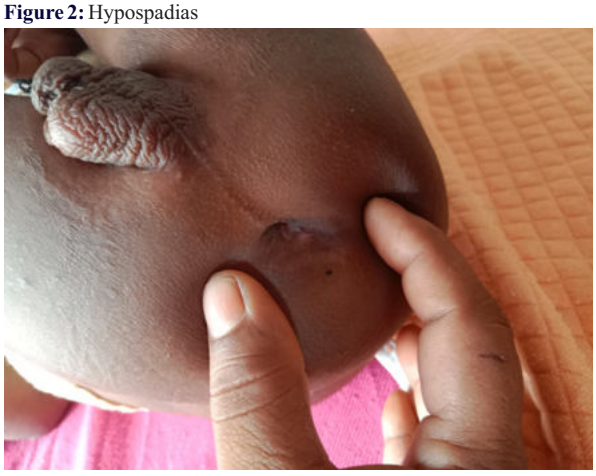


Figure 3: Patulous anus devoid of sphincter activity

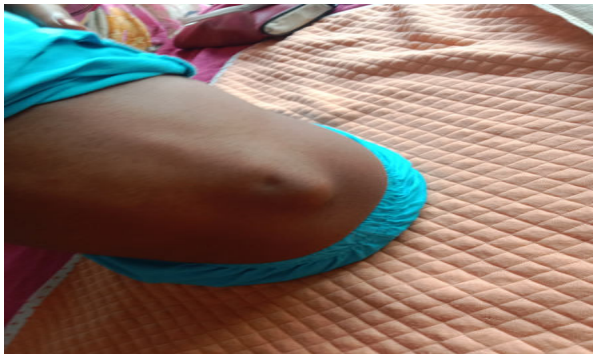
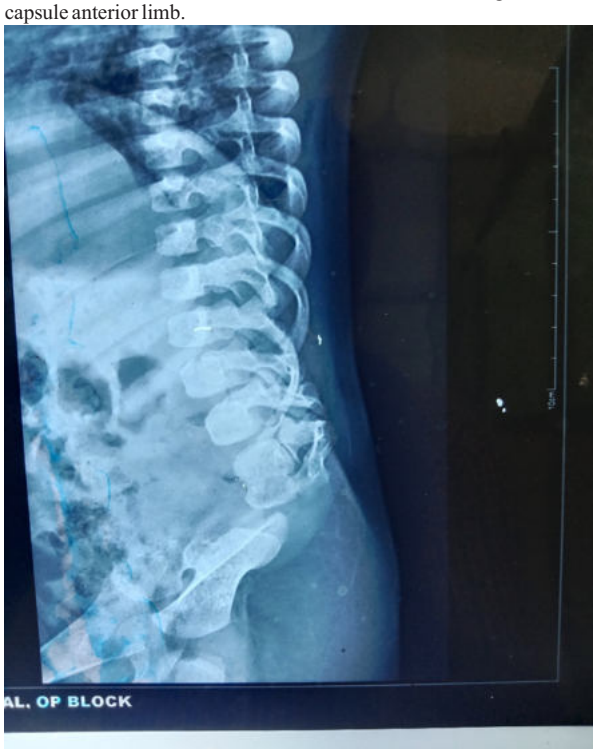


Figure 4: Bony Swelling Over The Back

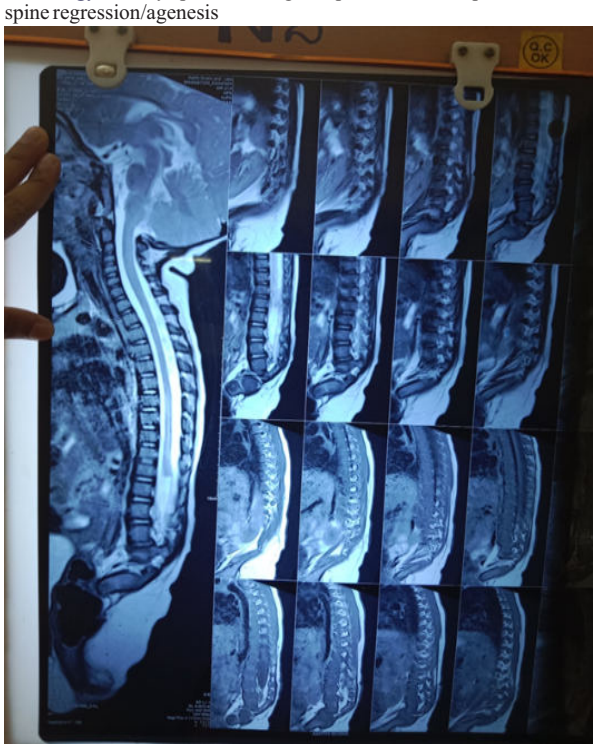
The mother is a known case of Type 2 Diabetes Mellitus who was on IV insulin during the antenatal period. The mother gives history of intake of native herbal medications for infertility. During antenatal period there was a history of reduced fetal movements and the prenatal ultrasound done showed hypoplastic lower limbs. There was no exposure to teratogens, illicit drugs or alcohol. The baby was delivered

via LSCS in view of breech presentation with a birth weight of 2.8kg.

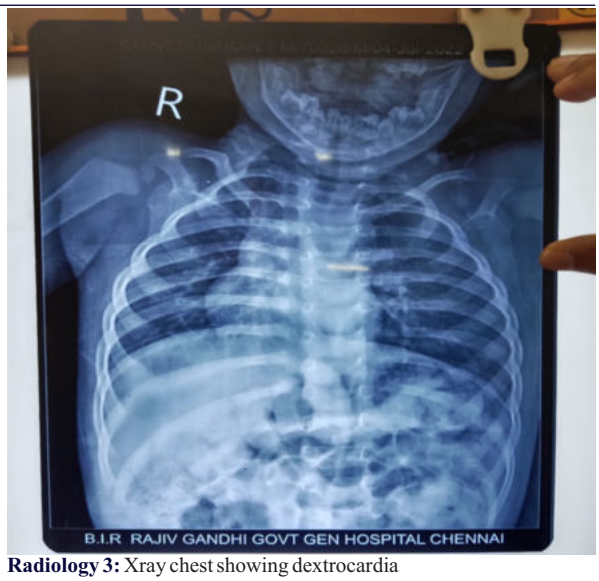
An Xray Spine taken shows complete sacral and partial lumbar spine regression/agenesis (Radiology 1) .Magnetic resonance imaging (MRI) of the spine done revealed a complete sacral and partial lumbar spine regression/agenesis. L1 and L2 were visualized and L3 was partially visualized with spinal cord showing syrinx in the dorsal cord. The dorsal cord ended at D10 level, all of which suggests a type 1 Caudal regression Syndrome (Radiology 2). A chest roentgenogram revealed dextrocardia (Radiology 3). Spinal ultrasound (US) revealed absence of the sacrum with the conus having a blunt end at the tip at the T12-L1 disc space. Abdominal and renal USG were within normal limits. MRI brain done showed a chronic infarct in the right internal capsule anterior limb.



Radiology 1: Xray spine showing complete sacral and partial lumbar spine regression/agenesis



Radiology 2: MRI spine with features of caudal regression



Radiology 3: Xray chest showing dextrocardia

After a multidisciplinary consultation, the patient was planned for diagnostic laparoscopy for the undescended testis, correction of club foot and correction of hypospadias. The patient was lost to follow-up.

DISCUSSION

Caudal Regression Syndrome , also known as caudal dysplasia, sacral dysgenesis or regression, congenital sacral agenesis, sacro-coccygeal dysgenesis and caudal dysplasia sequence is a rare disorder characterized by maldevelopment of the caudal half of the body with variable involvement of the gastrointestinal, genitourinary, skeletal and nervous systems[1,2,3,4,5]. It is noted to be more prevalent in infants of diabetic mothers documented at up to 1 in 350 live births and between 20% and 25% of mothers of infants with CDS have insulin-dependent diabetes mellitus[6,7]. A related condition, sirenomelia sequence (mermaid syndrome) or symmelia, previously thought to be a more severe form of CRS, now a distinct entity, is characterized by fusion of the legs and a variable combination of the other abnormalities [8,9] . The prevalence of sirenomelia is about 1 in 100,000 live births[10] . The Currarino triad or sequence is another related, yet distinct condition and includes anorectal atresia or ectopia, coccygeal and partial sacral agenesis, and a pre-sacral mass lesion such as anterior meningocele, lipoma or dermoid cyst[11,12].

Family history and maternal diabetes mellitus are two of the risk factors for this disorder. The abnormal embryologic development of the caudal mesoderm occurs within the first 4 weeks of embryonic development [13]. The exact pathogenesis of this syndrome is poorly understood. Suggested hypotheses include maternal exposure to cocaine or alcohol ,vascular steal theory or hypo-perfusion, fetal hypoxemia and amino acid imbalances. A primary deficiency of the caudal mesoderm is implicated in CRS. Presence of an aberrant umbilical artery (persistent vitelline artery) and the single midline lower limb in Sirenomelia, suggests vascular steal as a more probable etiology. Multiple genetic factors such as mutations in the VANGL1 gene on chromosome 1p13, the CELSR1 and the HLXB9 gene on chromosome 7q36 in cases of the Currarino syndrome, have been implicated[14,15,16,17].

The clinical features in this patient included bilateral clubfeet, The clinical manifestations of this patient included bilateral clubfoot, underdeveloped lower limbs, underdeveloped anus, hypospadias , an absent sacrum and scoliosis in addition to a history of maternal diabetes. CRS is a varied combination of abnormalities involving multiple systems. The common factor to these abnormalities relates to defects in the development of the caudal mesoderm and the structures that it ultimately forms. Common findings include flexion contractures of the knees and hips, pelvic deformity, syn- or poly-dactyly, anorectal malformations, abdominal wall defects, gut malrotation, intestinal atresia, renal agenesis or dysplasia, absent bladder, transposition of external genitalia, hypospadias, myelomeningocele and hydrocephalus [1].

The Renshaw and Pang classification is used for the CRS, while the

Pang, Kjaer and Stocker and Heifetz classification is used for sirenomelia [18,19,20,21] .Renshaw described CRS based on type of defect and articulation between bones. Type 1 has total or partial unilateral sacral agenesis; type II has variable lumbar and total sacral agenesis and the ilia articulates with the sides of the lowest vertebra; type III has variable lumbar and total sacral agenesis and the caudal end plate of the lowest vertebra rests above fused ilia or an iliac amphiarthrosis; type IV has fusion of soft tissues in both lower limbs; type V, also known as sirenomelia, has fused bones of lower limbs[21] . Stocker and Heifetz classified sirenomelia in 7 types: I would have all thigh and leg bones the type II has a single fibula, the type III has an absent fibulae, the type IV haspartially fused femurs and fused fibulae, the type V has partially fused femurs along with absent fibulae, the type VI has a single femur and a single tibia, and type VII has a single femur and absent tibiae [1,20] . The above mentioned patient has Type 1 CRS.

Management of CRS varies from patient to patient depending on the abnormalities present. Most recent is the use of growth hormones to improve distal innervaton [1,22] . Morbidity is mainly from genitourinary and neuromuscular complications requiring a coordinated multidisciplinary effort from neurosurgeons, neurologists, urologists, orthopedicians and cardiologists [23] .

A rare phenomenon, Caudal regression syndrome invariably results in long term morbidity and a difficult life for both the child as well as the parents. An improved understanding of the disease process and newer methods of management would definitely go a long way.

ABBREVIATIONS

CRS- Caudal Regression Syndrome

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