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Neurosurgery

A RARE CASE OF CAUDAL REGRESSION SYNDROME : A CASE REPORT

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ABSTRACT

Summary Background: Caudal regression syndrome is a rare, neural tube defect characterized by development of the caudal aspect of the vertebral column and the spinal cord. Although its precise etiology is not known, toxic, ischemic, or infectious etiologies before the 4th week of gestation are thought to be associated with this syndrome.it results in neurological deficits ranging from bladder and bowel involvement to severe sensory and motor deficits in the lower limbs.

Case Report: We present a 11 months old female child with caudal regression syndrome, clinical findings are swelling in the lumbosacral region with bilateral foot deformity and MRI lumbosacral findings are complete sacral agenesis, myeleomeningocele,tethered cord,left renal agenesis,conus lipoma,hydrosysringomyelia, lipoma of filum terminale, spina bifida and scoliosis. Patient underwent meningomyelocele repair, lipoma excision and detethering of cord.

Conclusions: Caudal regression syndrome is a rare entity with a known association with maternal diabetes. It is characterized by sacrococcygeal dysgenesis with an abrupt termination of a blunt- ending spinal cord. Ultrasound and fetal MRI can be used to make prenatal diagnosis. Early detection and prompt treatment is very important to decrease the risk of complications and thus to improve the diagnosis.

KEYWORDS : Lumbosacral Region, Prenatal Diagnosis, Diabetes Mellitus

Background :

Caudal regression syndrome is a rare congenital abnormality resulting from a developmental failure of a segment of the vertebral column and spinal cord. The function of the residual spinal cord is inversely related to the severity of the morphologic derangement. The majority of cases are sporadic, although there is evidence for a partial genetic contribution. Maternal hyperglycemia is the most important recognized teratogen. Caudal regression syndrome occurs in up to 1% of pregnancies of diabetic mothers with up to 22% of cases associated with Type I or Type II maternal diabetes mellitus (DM). Severe forms can cause early neonatal death due to cardiac, renal and respiratory problems. Prenatal ultrasound and fetal MRI can be used for antenatal diagnosis, while radiographs and MRI of the vertebral column are imaging modalities used in children and adults.

Case Report:

A 11 month old female child admitted with history of swelling in the back and lower limb deformity since birth. MRI spine showed complete sacral agenesis, spina bifida at L3,L4,L5, tethered cord upto L5 level, 1.6*1.3 cm myelocele in lumbosacral region, left renal agenesis with compensated enlargement of right kidney, conus lipoma, lipoma of filum terminale, scoliosis and syrinx of 3.4 cm length in lumbar segment of spinal cord. Xray foot showed bilateral CTEV.

Figure Ia. Clinical picture of the Child with CRS

Figure Ib. Lumbosacral lipomeningomyelocele .

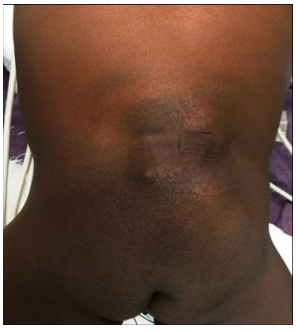


Figure 2a.Intra op picture shows lipomeningomyelocele.

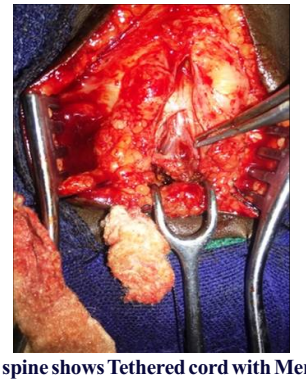


Figure 3a. MRI spine shows Tethered cord with Meningomyelocele.

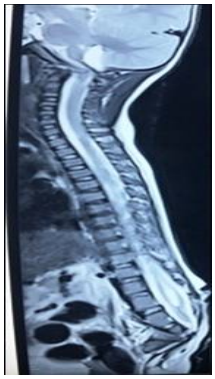


Figure 3b. MRI Myelogram shows cord changes.

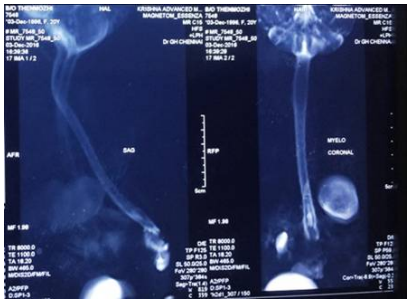


Figure 4a. Syrinx in Lumbar region .

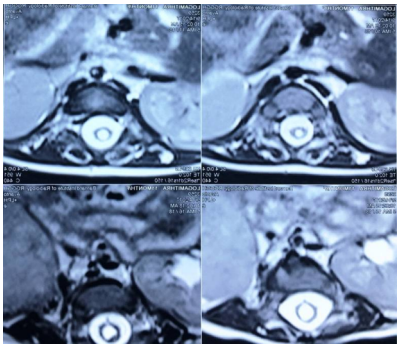


Figure 4b. CT spine shows Sacral Agenesis.

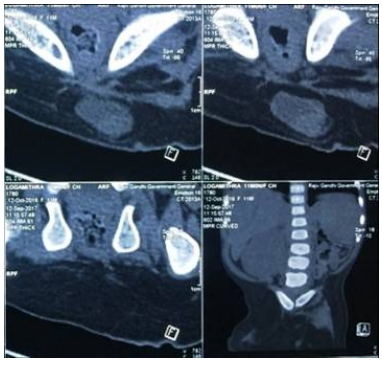


Figure 5a. Xray pelvis shows sacral agenesis.

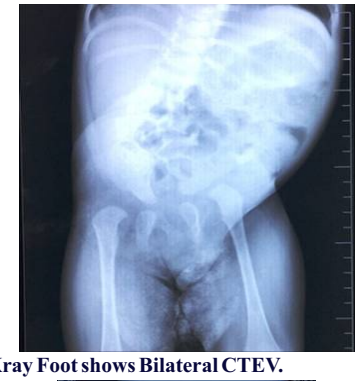
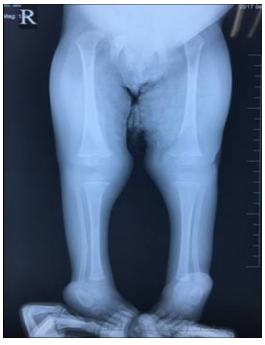


Figure 5b. Xray Foot shows Bilateral CTEV.



Patient underwent detethering of cord, excision of lipoma of filum terminale and myelocele repair. Histopathological examination reported as lipoma and tethered cord.

Post op period uneventful and patient was discharged on 10th post op day after suture removal, advised regular followup and referred to ortho op for CTEV management.

Discussion:

Caudal regression syndrome is rare, with an estimated incidence of 1-2.5 in every newborns. The vast majority of cases are sporadic, however, familial cases occasionally occur. An association with VACTERL and Currarino triad syndromic complexes has been reported.CRS may present with a broad range of symptoms like neurogenic bladder, anorectal malformations, sensory motor paresis, sacral agenesis, foot deformities and gait abnormalities. CRS results from an insult in early pregnancy (<4 week of gestation) [1]. Hyperglycemia, infection,toxic and ischemic insults have been implicated [2].

Two possible aetiologies are suspected.

1. Disturbance of the primary neurulation process .
2. Derailment of the process of degeneration and differentiation of an initially normally developed primary and secondary neural tube.

Other associated anomalies includes renal agenesis, anal atresis, spinal anomalies, tethered cord, cardiovascular anomalies and pulmonary hypoplasia. Matrenal diadetes and polyhydramnios are associated with CRS. Radiological features include lumbosacral vertebral body dysgenesis, truncated blunt spinal cord terminating above the expected level and severe canal narrowing rostral to last intact vertebra [3]. Diagnosis can usually be made in the 2nd or 3rd trimester with prenatal ultrasound, which demonstrates sacrococcygeal dysgenesis with a high and abrupt termination of the spinal cord with blunt-ending conus medullaris. Ultrasound is also helpful in evaluating other associated anomalies such as renal anomalies including agenesis, cystic dysplasia and caliectasis, and gastrointestinal anomalies such as duodenal atresia[4]. Fetal MRI can also be used to make the diagnosis in difficult cases[5]. Due to incomplete penetration of the ultrasound caused by ossified vertebrae, MRI is the imaging modality of choice for the diagnosis of any type of vertebral and spinal cord pathologies in adults[6]. This is also true for caudal regression syndrome, in which MRI demonstrates findings as described above. Additionally, MRI is also helpful in demonstrating the double-bundle arrangement of nerve roots. MR neurography is a helpful tool in the evaluation of the nerve root abnormalities[7] . Antenatal ultrasound may show a "shield sign"- opposed iliac bones in absence of sacral vertebrae., fetal extremities may be seen in a "crossed legged tailor"position or a "budhha" position.

Renshaw classified the spectrum of caudal regression syndrome into five types (Table 1).

S.No	Type	Characteristic features
1.	Type I	Total or partial unilateral sacral agenesis
2.	Type II	Partial/complete lumbar and total sacral agenesis; the iliac blades joining with lateral aspects of the lowest vertebra
3.	Type III	Partial/complete lumbar and total sacral agenesis associated with fuse ilia; or iliac amphiarthrosis acting as a base for caudal end of the most distal vertebra
4.	Type IV	Soft tissues of the lower limbs are fused
5.	Type V	Sirenomelia associated with single femur and single tibia

Imaging allows differentiation of two broad groups of patients with CRS, Group 1 – the conus medullaris is blunt and terminates above normal level with dilated central canal, these patients have major sacral deformities.

Group 2 –the conus medullaris elongated and tethered by a thickened filum terminale or intra spinal lipoma and ends below the normal level, neurological disturbances are more severe in this group.

The main differential diagnosis of caudal regression syndrome is sirenomelia. However, sirenomelia usually is characterized by a more

pronounced caudal dysgenesis with fused lower extremities. The presence of abnormally small abdominal aorta, two-vessel umbilical cord, and renal agenesis or severe dysgenesis with accompanying severe oligohydramnios and pulmonary hypoplasia in sirenomelia makes it more severe and fatal. Prenatal diagnosis is important so that appropriate patient counseling can be provided and postnatal interventions performed[8]. The treatment depends upon clinical symptoms, including the degree of neurological deficits. The main goals of treatment include maintaining and improving renal, cardiac, pulmonary and GI function, preventing renal infection and achieving continence. Urinary incontinence is treated with anticholinergic agents, which decreases the detrusor muscle tone, increases the bladder capacity, and thus decreases intravesicular pressure and urinary frequency[9]. Orthopedic intervention is necessary to correct the associated malformations. Physical therapy can help to prevent secondary deformities, skin ulcers and assists in improving quality of life. Surviving infants usually have normal mental function but do require extensive urologic and orthopedic assistance. Their long-term morbidity consists mostly of a neurogenic bladder dysfunction resulting in progressive renal damage and disabling neuromuscular deficits of the lower extremities[10].

Morbidity is mainly of genitourinary and neuromuscular complications and management options require the coordinated efforts of a team of specialists like neurosurgeons, neurologists, urologists,orthopedists and cardiologists.The incontinence required a continence control system and an imperforated anus , a permnant colostomy is mandatory.

Conclusions:

Caudal regression syndrome is a rare entity with a known association with maternal diabetes. It is characterized by sacrococcygeal dysgenesis with an abrupt termination of a blunt- ending spinal cord. Ultrasound and fetal MRI can be used to make prenatal diagnosis. Early detection and prompt treatment is very important to decrease the risk of urinary incontinence, recurrent urinary tract infections, renal impairment, and the development of a neuropathic bladder, and thus, to improve the prognosis.

Conflict of interest:

The authors declare that they have no conflict of interest relevant to the publication of this article.

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