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Caudal Regression Syndrome: A Rare Case Report

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Abstract

Caudal regression syndrome is a rare 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 occur in early gestation. It's a sporadic disorder. Maternal diabetes, genetic predisposition and vascular hypo perfusion have been shown to be associated with its pathogenesis. Here we report a newborn of diabetic mother diagnosed as caudal regression syndrome type II. Treatment for this patient is multidisciplinary. So, after giving necessary supportive treatment we discharged the patient with proper advice of his further management to improve the prognosis.

Key words : *Caudal Regression Syndrome, Rare Disorder, Maternal Diabetes.*

Introduction:

Caudal regression syndrome or caudal dysplasia is a rare syndrome. Caudal regression syndrome represents a spectrum of congenital malformations ranging from agenesis of the lumbosacral spine to the most severe cases of sirenomelia with lower extremity fusion, abnormal development of sacrum, lower lumbar vertebrae and major vessel anomalies.[1]The etiology of this syndrome is not well-known. Maternal diabetes, genetic predisposition and vascular hypo perfusion have been suggested as possible causative factors. However in infants of diabetic mothers increased chances of congenital anomalies exist. Approximately 15% to 20% of the patients have diabetic mothers and almost 1% of children of diabetic mothers are affected by this disorder.[2]

Case History:

Baby of Fatema, a male newborn admitted in the NICU due to pre-term (33 weeks) with LBW (2.1 Kg). The baby was delivered by LUCS. His mother had H/O of diabetes mellitus and H/O two previous spontaneous abortion in last two years. On examination we found the baby was pink in color with all the vitals were within normal limit. But the baby had some gross congenital anomalies like buttock looks flat and dimpled, leg seems short, bowed and presented with club foot in both side (Fig 1). The reflex and activity of the baby was moderate. Infantogram of this baby showed lower and mid lumber vertebral bodies were absent, small posterior portion of L3 was seen.The bony sacrum and coccyx were absent. Both

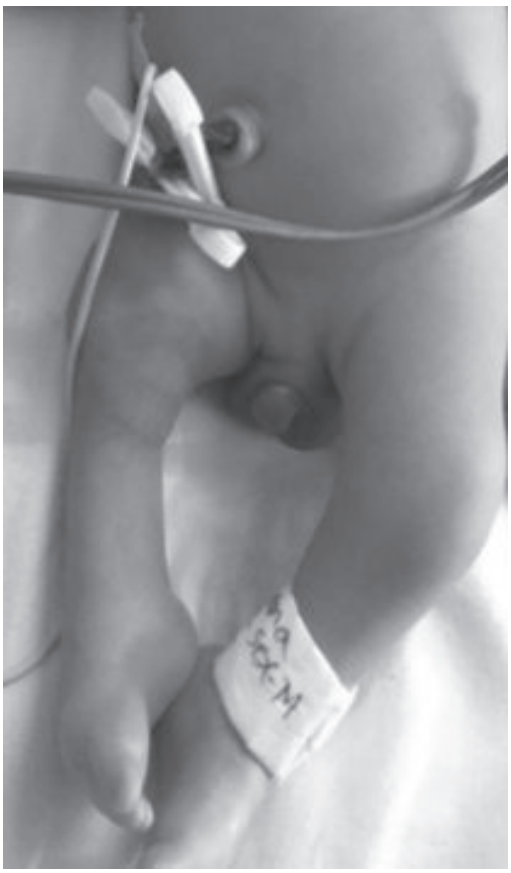

Fig.-1: *Limb deformity in newborn with CRS*

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iliac wings were positioned midline close to each – other with hypo plastic femurs and deformity of both feet (Fig 2). Skeletal findings suggestive of caudal regression syndrome (type- II). USG of Spine showed congenital absence of spine, coccyx, lower and mid lumber bodies. Non- demonstrated conus medullaries, dorsal protrusion and deformity of mid lumber spine Findings consistent with caudal regression syndrome. The baby was treated with supportive treatment. Our plan was to do a MRI and to consult with orthopedic surgeon. But they didn't want to continue the treatment due to personal problems. So we discharged the baby on request with proper advice for his further management.

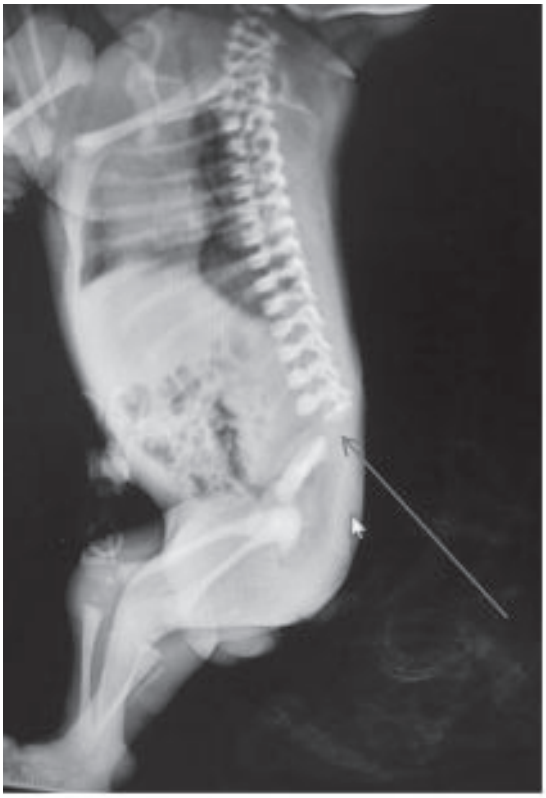

Fig 2: *Infantogram of this baby showed congenital agenesis of sacrum, coccyx, lower and mid lumber vertebral bodies. The bony sacrum and coccyx were absent. Both iliac wings were positioned midline close to each –other with hypo plastic femurs with club feet. Findings suggestive of Caudal Regression Syndrome (Type II).*

Discussion

Caudal regression syndrome (CRS) is a rare entity. It was first described by Duhamel et al.[3] It is seen in approximately 1 in 7500 live births. It is an uncommon malformation in the general population, but occurs in about 1 in 350 infants of diabetic mothers, representing an increase of about 200-fold over the rate seen in the general population.[4] No sex predilection is seen with equal incidence in males and females.[3] CRS is a syndrome characterized by a series of lower vertebral anomalies that should be associated with pelvic bone deformity. [5]Associations reported with CRS mostly include genitourinary, anorectal, vertebral, and cardiopulmonary anomalies and may also include VACTERL syndrome (abnormality of vertebrae, anus, cardiovascular system, trachea, esophagus, renal system, and limb buds) like imperforate anus, malformed external genitalia, bilateral renal aplasia and increased incidence of ectopic kidneys, pulmonary hypoplasia leading to Potter's face, club foot and in the most extreme of the case is when there is extreme external rotation and fusion of lower limbs – condition known as sirenomelia.[6]

This congenital syndrome is known to occur in early gestation, at around 4th week. Although the exact mechanism for this syndrome has not been established, it was thought that during embryogenesis, some insult to caudal eminence leading to failure of canalisation and retrogressive differentiation leading to partial or complete sacral agenesis.[7]

There is a genetic basis accredited to this congenital condition. It is believed to be inherited in dominant form. Possible mutation is described in HLBX9 homeobox gene on chromosome 7. This gene is also expressed in the pancreas Therefore, a possible association between diabetic hyperglycemia and caudal regression is proposed. The relationship and interdependence of developing caudal nervous, spinal, hindgut, and mesonephric elements involved in the closure of the neural tube result in the development of neural, distal vertebral, anorectal, renal, and genital abnormalities that produce CRS. However, structures that are developmentally distant from these caudal elements, such as the brain, proximal spine, and spinal cord, are not generally involved in CRS.[8]

Patients of CRS can further be classified according to Pang D et al [9] method into five different class of lumbosacral agenesis. In this type I class includes patients with total sacral agenesis with involvement of lumbar vertebrae. In type II, total sacral agenesis is present but lumbar vertebrae are spared. In type

III, subtotal sacral agenesis is present with presence of at least S1 vertebra. In these patients ilia articulate with sides of rudimentary sacrum maintaining its normal transverse diameter. Type IV includes those patients with hemi-sacrum and Type V includes patients with fully formed sacrum but with coccygeal agenesis. Our patient belongs to type II. Clinically, most common presentation is neurogenic urinary bladder dysfunction. Motor deficit is more in comparison to sensory deficit. In fact, in most patients there is combined sensory-motor paresis with motor deficit predominating the symptom.Features suggestive of caudal regression syndrome were narrow hips, hypoplastic gluteal muscles and shallow intergluteal cleft. Some patients show severe lower limb paralysis. Radiographic findings are mild lumbosacral segmentation anomalies to severe agenesis of lumbosacral vertebrae.

Magnetic resonance imaging (MRI) may also be performed to assess the degree of certain anomalies such as spinal defects.

Treatment may require the coordinated efforts of a team of specialists. Pediatricians, neurosurgeons, neurologists, urologists, orthopedist surgeons, cardiologists, nephrologists and other health care professionals may need to systematically and comprehensively plan an effective plan for child's treatment.[10]

Prognosis of patient is variable depending upon severity. Affected infants may require complex medical care, certain surgical intervention to reduce the morbidity and orthopedics procedure for limb anomalies. Early intervention is important in ensuring that children with caudal regression syndrome reach their highest potential.

Conclusion

These patients have normal intelligence and therefore lead otherwise normal lives except for neuromuscular deficits of the lower limbs. Correction of these abnormalities helps the child to be independent with a better quality of life.

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