CASE REPORT

Segmental Spinal Dysgenesis

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ABSTRACT:
We report a rare case of segmental spinal dysgenesis in a nine months old boy. The boy was born with flaccid paraplegia and lower limb deformities. Radiological workup revealed severe vertebral and spinal cord anomalies resulting into kyphosis and neurological deficits leading to grave disability and morbidity. The aim of reporting this case is to highlight the role of magnetic resonance imaging in the diagnosis of extent of spinal abnormalities for surgical correction.
Keywords: Segmental spinal dysgenesis, Thoracolumbar kyphosis, Cordanomaly, Magnetic resonance imaging

INTRODUCTION:

Segmental spinal dysgenesis is a rare congenital spinal anomaly characterized by localized agenesis or dysgenesis of the lumbar or thoracolumbar spine,severe congenital kyphosis or kyphoscoliosis and focal abnormalities of the underlying spinal cord and nerve roots.[1]Association with visceral abnormalities has also been reported. The neonate is symptomatic at birth with flaccid or spastic paraparesis or paraplegia and lower limb deformities. The exact cause is not known but studies reveal that these complexmalformationsoccur during gastrulation period of embryonic life. Although plain radiograph and CT scan are important for diagnosis but, MRI type imaging reliably detects spinal cord abnormalities and should always be performed at presentation.[2]

CASE REPORT:

A 9 months old boy was referred to radiology department for evaluation of flaccid paraplegia and lower limb deformities, since birth from pediatric surgical department. Detailed history revealed that the boy was delivered by C-Section at 38 weeks of gestation. Prenatal ultrasound at 38 weeks revealed breech presentation with gross vertebral anomalies at thoracolumbar junction. Vertebral anomalies were not documented in anomaly scan done at 22 weeks of gestation. There was no history of any congenital anomalies in her sibilings. At birth he had hypotonic lower limbs with flexed and abducted hip joints, flexed knees and bilateral clubbed TEV (talipes equinovarus) feet. He had a history of recurrent urinary tract infection for which he was taken to paediatric OPD multiple times. His physical examination demonstrated a kyphotic deformity with a bony spur (gibbus) at thoracolumbar junction. He had flaccid paraplegia and bilateral TEV. However, his higher mental functions revealed no retardation. His base line investigations were normal. Echocardiography revealed a small ASD with left to right shunt.

X-Ray dorsolumbar spine demonstrated acute angle kyphotic deformity with epicenter at LV-2 marked by gibbus apex. The spine appeared separated with complete offset between upper and lower segments. The body of TV-11 appeared hypoplastic with butterfly anomaly however bodies of LV-1 and LV-2 appeared to be fused, block vertebrae. Abdominal ultrasonography revealed bilateralhydro-nephro-ureter but there was no evidence of horseshoe kidney or other renal anomalies. MRI dorso-lumbar spine revealed morphologically abnormal spinal cord, cord in the upper spine ended at TV8-9 IV disc level. Cord in lower spine was much bulkier and nerve rootswere traversing anteriorly in front of cord after originating from it (Figure 1). There was no discernable cord between upper and lower segments at the level of gibbus. There was narrowing of spinal canal with complete obliteration and CSF column cut off at LV-2 level, marked by gibbus apex (Figure 2). Upper four sacral segments were visualized, however fifth sacral and coccygeal segments were not seen, representing partialsacro-coccygeal agenesis. Spinal bifida at LV-3 level noted, however no dermal sinus tract visualized. Based upon MRI findings a final diagnosis of segmental spinal dysgenesis was made. MCUG was performed which revealed neurogenic bladder complicated by bilateral vesicoureteric reflux. This patient presented at the age of nine months and was suggested anterior and posterior arthrodesis for vertebral stabilization and spinal cord decompression by the orthopedic surgeon. Patient's bilateral TEV was surgically corrected and casts were applied. As the patient had associated small sized atrial septal defect, it was left for spontaneous closure.

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Figure:1
MRI L.S spine, T2W seq, axial view revealing bulky lower segment spinal cord with nerve roots traversing anteriorly from the cord

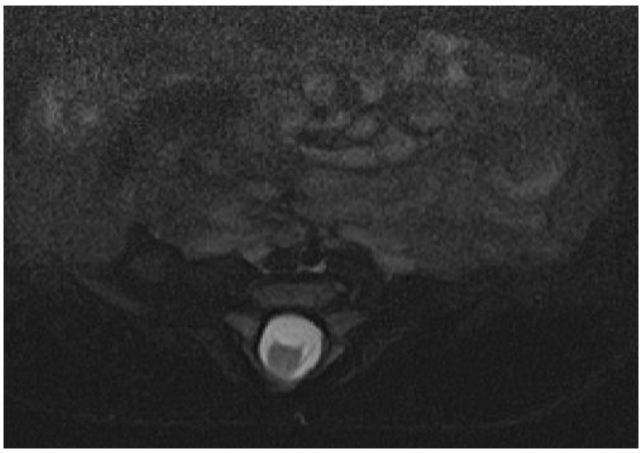
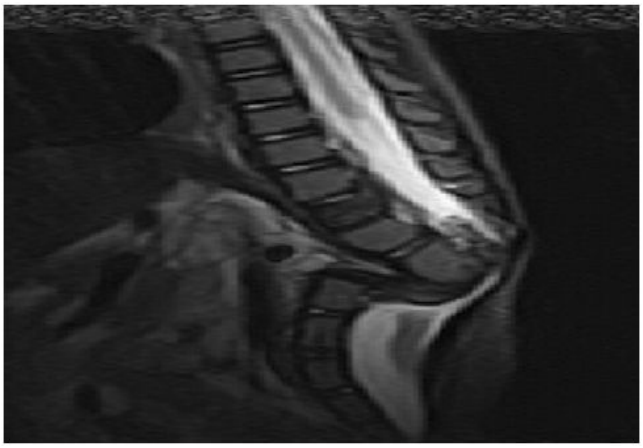


Figure:2
MRI L.S spine, T2W seq, sagittal view revealing spinal canal narrowing and complete CSF column cutoff at LV-2level with gibbus deformity



DISCUSSION:

Segmental Spinal Dysgenesisis (SSD)is characterized by localized deformity of the thoracolumbar, lumbar or lumbosacral spine associated with abnormal development of the underlying spinal cord and nerve roots.[3] Overall incidence of SSD has not been published widely presumably because of its rarity.Few studies have been reported since Scott recognized SSD as an autonomous entity in 1988.[2] Theexact cause of this complicated malformation is not yet known but it is presumed to be due to an insult during gastrulation period of embryonic life, with resultant arrest in development of potentialneuro-ectodermal tissue. Dias suggested that complex dysraphic malformations with associated vertebral and systemic anomalies may represent a disorder of gastrulation.[4] Prenatal diagnosis of SSD is achievable using ultrasound.[5]

Ultrasound features of the case Fratelli reported consisted of mal-alignment of caudal segment of the spine, talipes equinovarus and reduced movements of the knee joint. Because of its embryonicorigin, the condition is present at birth. The clinical picture depends upon the level and severity of malformation. Bony defects include hypoplastic or absent vertebrae, resulting into congenital acute angle kyphosis or kyphoscolosis with epicenter represented as a bony spur or gibbus apex on back. There is spinal canal stenosis at the corresponding level. The condition may be associated with spina bifida or sacro-coccygeal agenesis. Spinal cord abnormalities include normal upper spinal cordand a bulky low lying lower spinal cord segment with an indiscernible cord segment in between. The presence of a cord segment within the caudal spinal canal is a dominant and unique feature of SSD.[6] It is usually associated with renal abnormalities such as horse shoe kidneys or ectopic kidney. Studies have revealed that neurogenic bladder is consistently present and may be complicated by urinary tract infection, vesicoureteric reflux and hydronephrosis,[7] which increase the rate of morbidity and mortality in such patients. Some cases have associated cardiac anomalies like Dextrocardia and/or septal defects.[8]

Patients usually present in infantile period with kyphosis, Kyphoscolosis, paraparesis or paraplegia and lower limb deformities which include bilateral TEV, flexed abducted hip joints and flexed kneesrepresenting Buddha Posture.[9] The bony defects of spine can be demonstrated on plain radiographs but CT scan of dorsolumbar spine is a best modality to reveal bony abnormalities. MR imaging reliably detects spinal cord abnormalities and should always be performed at presentation.[10]

Patients with segmental spinal dysgenesis have an increased risk for the development of severe neurological deficits owing to the associated spinal instability and spinal canal stenosis. MRI plays an important role and can influence the surgical strategy. Early reconstructive surgery is recommended as soon as the diagnosis is made. Spinal cord decompression and vertebral stabilization is pivotal at an early stage to limit the progression of kyphosis and worsening of neurological impairment.

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