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Segmental spinal dysgenesis: A rare congenital spinal malformation

Dear Sir,

Segmental spinal dysgenesis (SSD) is a rare congenital spinal anomaly characterized by localized dysgenesis or agenesis of the lumbar or thoracolumbar spine and spinal cord. Patients usually present with spastic paraparesis and features of neurogenic bladder. Diagnosis of this rare condition is important as it usually does not benefit from surgery, unlike tethered cord syndrome.

A 4-year-old boy presented with inability to walk without support and increased frequency of micturition. On examination, the child was conscious, cooperative, and playful with normal speech for age, actively moving upper limb with paucity of movement of lower limbs. He was able to sit independently but could walk only with support. Lower limbs were hypertonic on passive movement whereas the hip and knee joints were predominantly kept in flexion and external rotation. Both the feet were kept in valgus position. Deep tendon reflexes were brisk in both upper and lower limbs. X-ray hip was normal. There was no evidence of developmental dysplasia of the hip. Clinically there was paraparesis with neurogenic bladder.

He was delivered by normal vaginal delivery with no history of neonatal hypoxia. The baby cried immediately after birth. Developmental milestones such as neck holding at 6 months, sitting with support at 7 months, standing with support >1 year were suggestive of the gross motor developmental delay. The child was unable to stand without support at presentation.

The patient was referred for MRI. The ultrasound of the abdomen was normal. There was no renal anomaly or features of vesicoureteral reflux. MRI of spine was done in a 1.5 T Siemens scanner. MRI revealed an abnormal narrow segment of spinal cord from mid D12 to mid-L1 level. Caudal to the abnormal segment, the spinal cord and conus appeared relatively expanded. No nerve roots were seen arising from the narrowed segment of the spinal cord. Long-segment syrinx was seen in the spinal cord above the abnormal segment extending up to the upper dorsal level. There was a partial fusion of L2 and L3 vertebral bodies [Figure 1]. There was no hemivertebra or butterfly vertebra.

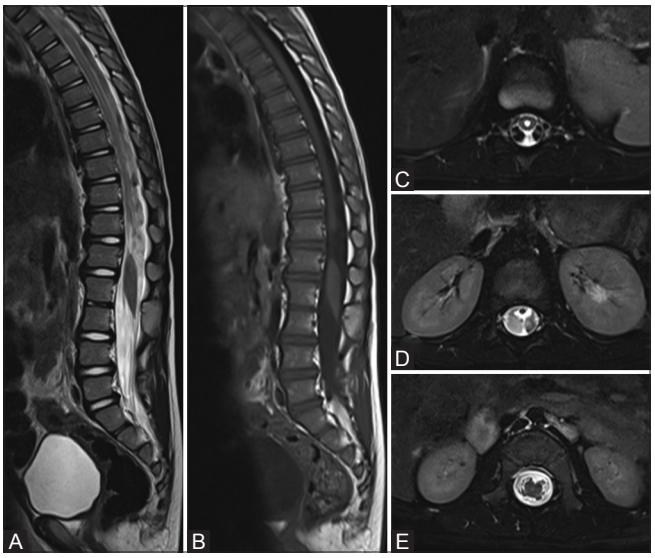

Figure 1 (A-E): MRI dorso-lumbar spine: Sagittal T2WI (A) and Sagittal T1WI (B) shows a significantly thinned segment of lower dorsal spinal cord (from mid D12 to mid L1) with mildly bulky spinal cord and conus caudal to it. Axial fat sat T2WI (C), (D and E) above, at and below the level of spinal cord narrowing respectively. Normal cord caliber with syrinx in (C), significantly thinned spinal cord with no nerve root at (D) and mildly bulky distal cord (E)

SSD is a rare anomaly characterized by localized dysgenesis of the lumbar or thoracolumbar spine, congenital kyphoscoliosis, and focal abnormalities of the spinal cord and nerve roots.[1] Spastic paraparesis and neurogenic bladder are the most common clinical presentations. Spinal dysraphism, vertebral anomalies, and deformities of lower extremities are usually associated. MRI is the modality of choice for diagnosis of this condition as the cord as well as vertebral abnormalities can be well detected. Imaging finding is variable according to the level and extent of the abnormality. Characteristically, there is a markedly thinned cord segment devoid of nerve roots, a normal upper spinal cord and bulky, thickened, and low-lying lower cord caudally.[2] Mostly the upper spinal cord is normal. Syrinx although rare has been described and was mentioned in two out of 10 cases in one series.[1]

SSD may mimic cord tethering or caudal regression syndrome if it involves the terminal segment of the cord. It is important to differentiate SSD from caudal regression syndrome as SSD is less likely to benefit from untethering as the neurologic abnormalities are related to hypoplasia or absence of roots or an entire segment of the spinal cord. Surgery is indicated when there is compression of the spinal cord due to vertebral abnormalities.

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Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
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References

1. Tortori-Donati P, Fondelli MP, Rossi A, Raybaud CA, Cama A, Capra V. Segmental spinal dysgenesis: Neuroradiologic findings with clinical and embryologic correlation. AJNR Am J Neuroradiol 1999;20:445-56.
2. Fondelli MP, Cama A, Rossi A, Piatelli GL, Tortori-Donati P. Segmental spinal dysgenesis: MRI findings in 7 cases (abstr). Neuroradiology 1996;38:89.

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