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Case Report

Segmental spinal dysgenesis with open spinal dysraphism and Chiari II features, case report[☆]

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ARTICLE INFO

Article history:

Received 18 June 2020

Revised 19 July 2020

Accepted 20 July 2020

Keywords:

Spinal cord

Chiari II

Magnetic resonance imaging

SSD

Open spinal dysraphism

ABSTRACT

Segmental spinal dysgenesis (SSD) is a complex spinal anomaly characterized by localized dysgenesis of the lumbar or thoracolumbar spine, and severe congenital kyphosis or kyphoscoliosis. We describe a newborn who presented with severe congenital paraplegia and a lumbar mass. Magnetic resonance imaging confirmed SSD type II associated with open spinal dysraphism and intracranial Chiari II features; this association has not been reported. The association modifies the disease management and outcome. The previous classification of SSD could be revisited based on our case.

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Introduction

Segmental spinal dysgenesis (SSD) is a rare congenital anomaly. Patients usually present with kyphosis or kyphoscoliosis and congenital paraplegia with or without club foot deformity [1,2]. Magnetic resonance imaging (MRI) plays a pivotal role to diagnose this entity for early management plan [3]. The correlation between the embryology and imaging features of this entity has been described thoroughly most recently [4]. However, the association between SSD and Chiari II features remains unreported.

Case report

A full-term neonate born by spontaneous vaginal delivery after an uneventful pregnancy showed poor Apgar scores. Examination showed severe lumbar kyphosis with a lumbar mass and club foot deformity, and we observed macrocephaly (head circumference at the 95th percentile) and paraplegia.

He underwent urgent brain and whole-spine MRI, which revealed severe lumbar kyphosis with a gibbus deformity associated with a posterior midline bony defect in the thoracolumbar region. The spinal cord showed significant thinning of a segment in the mid-thoracic region at the level of the posterior spinal defect. We observed the neural placode attached to the skin surface consistent with myelocele (Fig. 1A), as well as absence of the spine distally along with a thickened conus

[☆] AIRP approval has been obtained through our institution.

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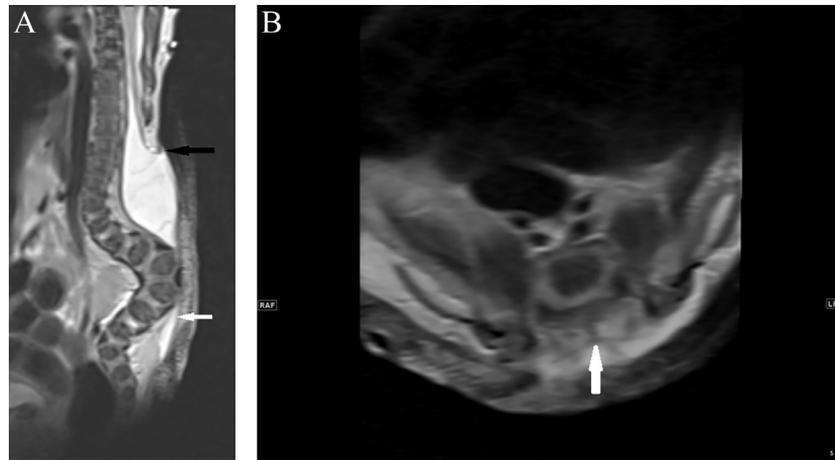


Fig. 1 – (A): Sagittal T1WI of the spine demonstrates significant thinning of the spinal cord segment in the mid-thoracic region at the level of the posterior spinal defect (black arrow). The neural placode attached to the skin surface consistent with myelocele, below the level of kyphosis there is thick distal spine segment filling the thecal sac (white arrow). **(B)** Axial T2WI below the level of the kyphosis shows thickened distal cord (white arrow).



Fig. 2 – Sagittal T1WI of the brain demonstrates small posterior fossa with tonsillar herniation, tectal beaking, and supratentorial hydrocephalus.

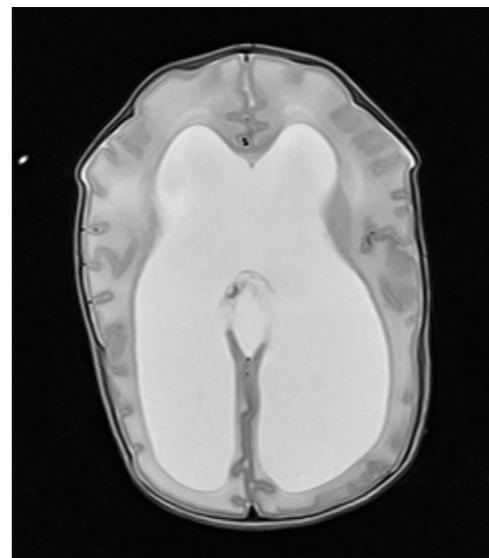


Fig. 3 – Axial T2WI of the brain demonstrates ventriculomegaly with absent septum pellucidum.

caudal to the gibbus deformity (Fig. 1B). The patient's brain MRI revealed a small cranial fossa with vermian herniation and tectal plate beaking (Fig. 2). Significant supratentorial hydrocephalus was observed with absence of the septum pellucidum (Fig. 3).

Furthermore, he had bilateral medially rotated pelvic kidneys below the level of the kyphotic segment of the spine (L1-2) with mild hydronephrosis (Fig. 4).

The patient was diagnosed with SSD with associated hydrocephalus and a Chiari II anomaly and was referred to a neurosurgeon for correction of the open myelocele and ventricular shunt placement.

Discussion

SSD was first described by Scott et al in 1988 [1] as a complex closed spinal dysraphism characterized by localized spinal cord agenesis or dysgenesis involving the lumbar or thoracolumbar region with absent nerve roots and severe congenital kyphosis or kyphoscoliosis [2]. SSD is a rare disease entity and reported before with no other associated brain anomalies.

Our patient showed open spinal dysraphism with Chiari II features; this complex presentation is associated with poor prognosis.

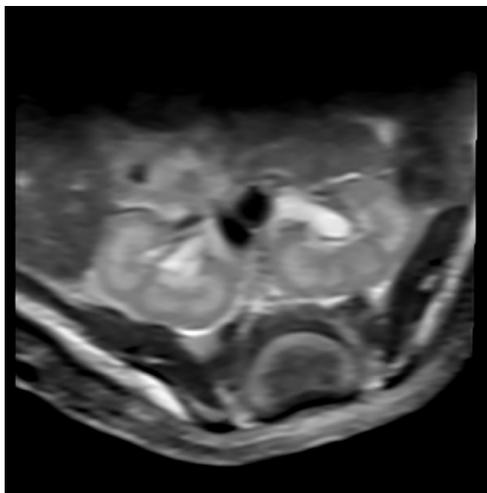


Fig. 4 – Axial T2WI of the lower lumbar spine shows bilateral medially rotated pelvic kidneys below the level of the kyphotic segment of the spine (L1-2) with mild hydronephrosis.

SSD refers to a complex closed spinal dysraphism associated with certain diagnostic criteria, including congenital paraparesis/paraplegia with lower extremity abnormalities, congenital kyphoscoliosis, or kyphosis with or without vertebral anomalies, absent or malformed segments of the spinal cord and underlying nerve roots, and visualization of the spinal cord segment distal to the interrupted cord [1,2]. Previous studies have described cervicodorsal junction involvement in a few cases [3,4].

Horseshoe or pelvic kidneys are known to be associated with SSD and are attributed to failure of ascent of the kidneys secondary to mechanical interference caused by the deformed spine. The condition is usually asymptomatic; however, it may manifest with ureteropelvic junction obstruction, hydronephrosis, or nephrolithiasis [5].

Recently, Chellathurai et al [4] categorized SSD as follows: Type I SSD associated with mild kyphosis and a thickened spinal cord that ends abruptly with a low-lying caudal cord segment and sparing of the spinal canal and Type II SSD associated with severe kyphoscoliosis, gibbus deformity, spinal cord thinning at the gibbus apex, and severe spinal canal sparing.

Our patient presented with type II SSD with severe kyphosis and spinal cord thinning at the apex of the gibbus deformity along with myelocele (a form of open spinal dysraphism), associated with a Chiari II anomaly.

The main embryological defect in SSD includes defective notochord formation during early gestational life (2-5 weeks), which affects both neural tube and vertebral body formation, resulting in a morphologically hypoplastic or absent cord cephalad to the bony abnormality with present of the distal

segment. Our patient showed concomitant anomalous primary neurulation secondary to defective closure of the neural tube, resulting in open spinal dysraphism represented by myelocele in which the placode is flush with the skin surface. Notably, myelocele constitutes <2% of all cases of open spinal dysraphism [6].

Vermian herniation may occur secondary to cerebrospinal fluid leakage from the open neural tube or from spinal cord tethering at the level of the defect [7]. Supratentorial hydrocephalus may occur secondary to external compression of the aqueduct by a beaking deformity of the mesencephalic spur [8].

In our view, the previous classification of SSD could be revisited to consider addition of open spinal dysraphism and Chiari II features to the classification, which could be considered a third type of anomaly associated with this entity.

Teaching point

SSD is a rare, complex spinal anomaly observed in neonates. Radiologists should perform thorough evaluation in suspected cases for early detection of SSD in association with a Chiari II anomaly because early diagnosis alters the management and prognosis of this condition.

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