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Case Report

A rare presentation of segmental spinal dysgenesis: Clinical and radiological findings ☆

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ABSTRACT

Segmental spinal dysgenesis is a rare and complex congenital condition affecting the dorso-lumbar spine, characterized by focal spinal cord dysgenesis and kypho-scoliotic deformity. It arises due to notochord malformation during embryogenesis. The case in question involves a 2-year-old female child. She presented to the outpatient department of our hospital with a history of inability to walk and increased frequency of micturition. The patient's mother had no antenatal visits. Upon examination, the patient was found to have a scoliotic deformity. Magnetic resonance imaging (MRI) of the spine revealed an absence of the spinal cord and spinal nerves from the T5 to L2 levels. A relatively thick spinal cord was visible from the L2 to L4 level. There was a complete absence of the spinal canal at the D10 and D11 levels, along with dorsal levoscoliosis. Segmental anomalies of the vertebrae were also noted in the dorsal spine. Additionally, imaging showed features of neurogenic bladder and mild left hydroureteronephrosis. The child underwent rehabilitation and surgical correction of the scoliosis.

Introduction

Segmental spinal dysgenesis (SSD) is a complex congenital malformation of the spine characterized by segmental agenesis or dysgenesis of the lumbar or thoracolumbar vertebrae and the underlying spinal cord [1]. Typically, the cervical spinal cord is normal, while the dysgenetic segment in the dorsal and lumbar regions is thinned or absent, with no nerve roots. The spinal cord distal to the dysgenetic segment is often thickened [2]. This condition is associated with kypho-scoliotic deformities and segmental vertebral anomalies [3]. SSD results from a defect in the embryogenesis of the notochord and is frequently cited as an example of embryological amnesia [4].

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Although SSD is classically described as involving the thoracic and lumbar spine, some authors have reported cases with involvement of the cervical spinal cord.

Case presentation

The index case is a 2-year-old female who presented to the outpatient department of a tertiary private hospital in Kathmandu with complaints of an inability to walk and increased frequency of micturition since birth. The child hails from a remote district of Nepal and belongs to a low socio-economic background. Her mother did not consult any obstetrician during the antenatal period, nor did she take any medications during this time. The child was delivered vaginally at home with the assistance of a trained birth attendant. Apart from walking, all other developmental milestones were normal. She can move her hands and upper body normally and can speak about 50 words in her mother tongue.

On examination, her vital signs were stable. There was levoscoliosis and spastic paralysis of both lower limbs, which were small due to disuse atrophy. The deep tendon reflexes of the knees and ankles were reduced. An anteroposterior X-ray of the spine revealed levoscoliosis centered at the dorsal vertebrae, along with segmental anomalies, most notably a butterfly vertebra at T7 (Fig. 1). Magnetic Resonance Imaging (MRI) of the brain and spine revealed a complex spinal malformation, including the T7 butterfly vertebra. Partial fusion of the posterior elements of the D9-D11 vertebrae was observed on the right side (Fig. 3). The spinal canal was markedly narrowed from the D9 to D12 vertebrae, with complete obliteration at the D10 and D11 levels (Fig. 3). An empty dural sac with no spinal cord was seen between the D5 to D9 and D12 to the upper border of the L2 vertebra (Figs. 2, 3 and 4). A relatively thick spinal cord was present at the L2 to L4 levels (Fig. 4). No exiting nerves were observed at the D7-D8 and D11-D12 levels. Additionally, there was mild levoscoliosis of the dorsal spine centered at the D9/D10 vertebrae. Partial sacralization of the L5 vertebra was also noted. The urinary bladder was partially visualized, with evidence of overdistention. The luminal outline of the visualized bladder was irregular (Fig. 3). Mild hydroureteronephrosis was noted on the left side, with no significant renal parenchymal thinning (Fig. 4). The MRI of the brain revealed no obvious abnormalities. Other diagnostic workups, including blood tests, echocardiography, ultrasonography of the abdomen, and ophthalmic and audiological examinations, were normal. Genetic testing did not reveal any specific syndrome. Urine investigations revealed cystitis.

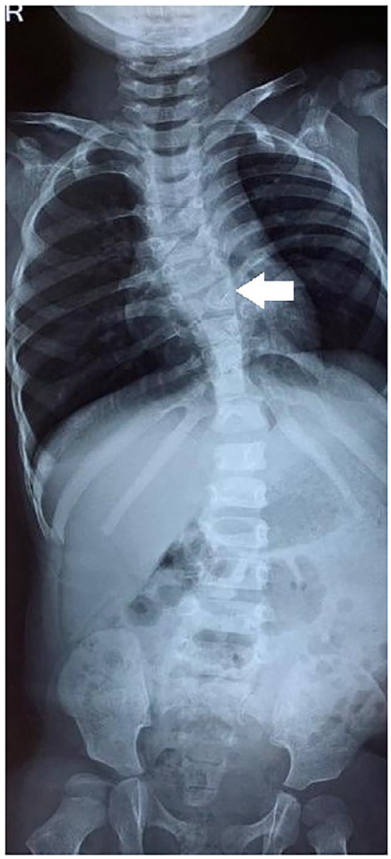

Fig. 1 – Antero-posterior radiograph of spine. There is mild levoscoliosis centered in dorsal spine. D7 butterfly vertebra is seen.

Based on the patient's history and clinical examination, the following differential diagnoses should be considered:

a) Segmental spinal dysgenesis
b) Tethered cord
c) Caudal regression syndrome

MRI is the key tool for diagnosing SSD. It is crucial to differentiate tethered cord from SSD, as the treatment approaches for these 2 conditions are entirely different. Surgical untethering is highly beneficial in cases of tethered cord. However, surgery is generally not indicated in SSD, as the symptoms are due to hypoplasia of the spinal cord and nerve roots. Surgery is only considered in SSD if there is spinal cord compression caused by vertebral abnormalities.

Since the diagnosis of SSD was straightforward on MRI, mainly supportive treatment and physiotherapy were planned. To assess the remaining function of the spinal cord, somatosensory evoked potentials (SSEP) were conducted on all 4 limbs. The results confirmed that there was no nervous conduction through the segments affected by the malformation. A scoliotic correction and arthrodesis surgery were performed after one month. Cystitis was treated with a course of antibiotics. The patient's guardians were educated on self-catheterization techniques and the importance of voiding at regular intervals. Physiotherapy exercises were initiated twice a week, which slightly improved the motor ability of the lower limbs. The patient became able to flex her knees and hips with stimulation and could turn sideways with assistance. She was provided with a custom-made wheelchair, which greatly enhanced her mobility and quality of life. She remains under regular follow-up.

Discussion

Tortori-Donati et al. [1] and Scott et al. [5] were the first researchers to describe and document Segmental Spinal Dysgenesis (SSD). According to them, SSD is a localized deformity of

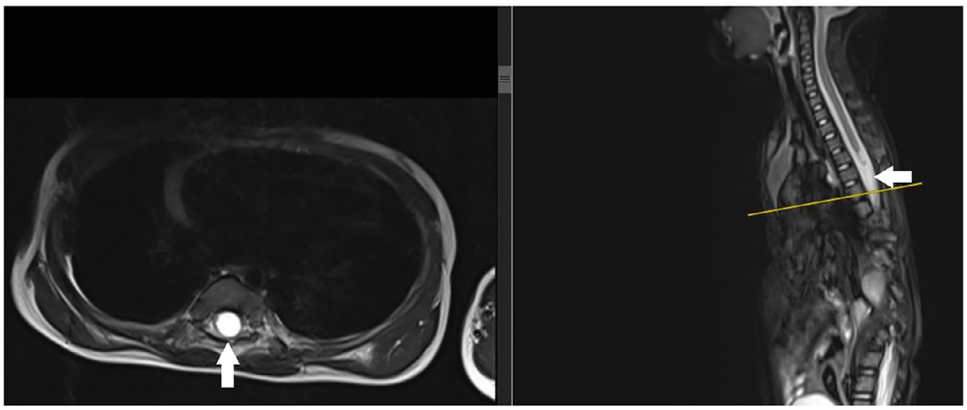

Fig. 2 – T2 Axial and T2 sagittal images at D5-D9 level. Empty dural sac is seen at D5-D9 level with absence of spinal cord and nerve roots.

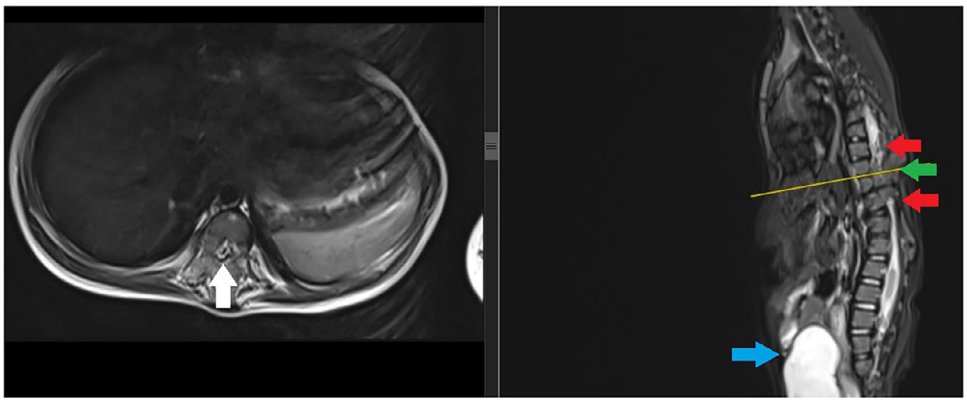

Fig. 3 – T2 Axial and T2 sagittal images at D10-D11 level. The spinal canal is markedly narrowed at D9-D12 level with complete obliteration of the canal at D10-D11 level. Spinal cord and nerve roots are not visualized at these levels. Also note partially fused posterior elements of D9-D11 vertebrae and partially visualized, overdistended bladder.

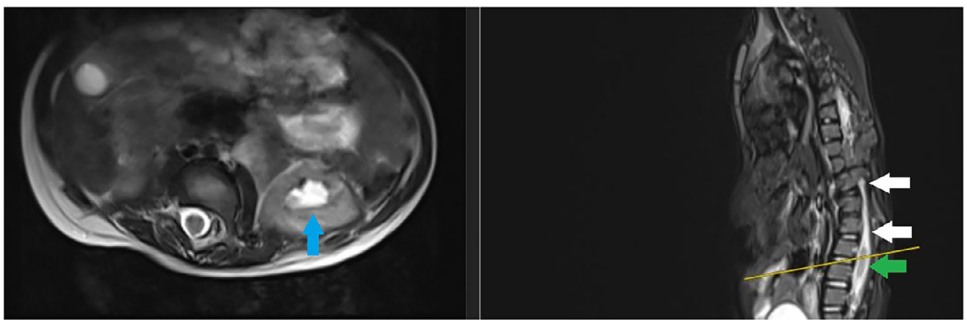

Fig. 4 – T2 Axial and T2 sagittal images at D12- L4 level. There is empty dural sac and absence of cord at D12 to upper border of L2 vertebra. Thickened spinal cord is seen at L2 to L4 vertebra. Also note mild left hydronephrosis.

the thoracolumbar or lumbar spine, characterized by dysgenesis or agenesis of the underlying spinal cord and nerve roots. Researchers like Bristol et al. [6] and Amarnath Chellathurai et al. [2] have also described cases of SSD involving the cervical spinal cord. There is conflicting opinion in the literature about the definition of SSD. We can adopt the defining criteria proposed by Amarnath Chellathurai et al. [2], which include: a) Congenital paraparesis or paraplegia accompanied by lower limb abnormalities. b) Multiple vertebral formation and segmentation anomalies, with or without (kypho)scoliosis. c) An

absent or malformed segment of the spinal cord and underlying nerve roots, affecting any spinal region from cervical to sacral. d) The ability to visualize the segment of the spinal cord distal to the interruption.

Amarnath Chellathurai et al. [2] have categorized SSD into 2 types:

- **Type I SSD:** Associated with mild kyphosis and a thickened spinal cord that ends abruptly with a low-lying caudal cord segment, without significant retro-spinal protrusion, gibbus deformity, or spinal canal narrowing.
- **Type II SSD:** Associated with severe kyphoscoliosis, gibbus deformity, spinal cord thinning at the gibbus apex, and severe spinal canal narrowing.

SSD is linked to defective embryogenesis. The defect in gastrulation is responsible for the development of Type I SSD [2]. When paired analgen fail to fuse in certain areas or remain separate to develop independently, the migration of chordamesoderm to the ectoderm-endoderm surface is abruptly halted, preventing the spinal cord distal to this point from developing. This failure of chordamesoderm migration also results in malformation of the vertebrae, spinal canal, and nerve roots. Type II SSD develops during early spine formation (3-6 weeks) due to segmentation and resegmentation issues, leading to vertebral anomalies like hemivertebrae and block vertebrae [2]. These anomalies can compress the spinal cord by altering vertebral shape, number, and canal structure, resulting in severe narrowing of the spinal canal in all patients. This condition, arising during gestation, is known as a congenital vertebral defect.

The symptoms of SSD are evident immediately after birth [7]. Depending on the degree of spinal cord aplasia or dysgenesis, motor defects can range from minor to paraplegia. Deep tendon reflexes in affected areas are reduced. Deformities of the lower limbs and neurogenic bladder are often associated with SSD [8]. A neurogenic bladder results in overflow incontinence and vesicoureteral reflux. SSD is often seen in association with other forms of closed spinal dysraphism, such as diastematomyelia, filar lipoma, and kidney malformations like horseshoe kidney [3].

Our case represents Type I SSD. Although there is severe spinal canal narrowing at the D9-D12 vertebrae, there is no kyphosis or gibbus deformity. There is a complete absence of the spinal cord from the D5 to D9 level and from D12 to the upper border of the L2 vertebra.

The management of SSD is mainly supportive and involves a combined approach from neurology, neurosurgery, orthopedics, and urology. If there is spinal cord compression, it is advisable to release the compression to prevent further deterioration [9]. If there is tethering of the cord along with SSD, untethering can prevent further deterioration. Surgical arthrodesis stabilizes the spine and prevents progressive deformity. The optimal timing of surgery is a matter of debate. Some authors advocate for urgent arthrodesis, while others suggest allowing enough time for vertebral growth and osseous bed maturation. Opinions on timing vary, with some advocating surgery after 5-6 months and others after 2-3 years [10]. Corsets and braces are useful until surgery is performed. Scoliosis correction surgery is also helpful and is usually performed along with arthrodesis. Physiotherapy exercises can improve motor function in the paralyzed areas. Managing the neurogenic bladder is crucial to prevent vesicoureteral reflux and kidney damage. Infections should be treated with antibiotics. Patients should be taught self-catheterization and encouraged to void the urinary bladder frequently to prevent reflux nephropathy [8].

Conclusion

Segmental spinal dysgenesis is an uncommon and intricate congenital disorder linked to abnormal notochord development during embryogenesis. MRI serves as the key diagnostic tool for identifying this condition. While treatment is generally supportive rather than curative, surgical intervention may be beneficial in select cases. This case report emphasizes the clinical presentation and imaging features of Type I SSD.

Author contributions

Sharma Paudel: Conceptualization, supervision. Prajwal Dahal: manuscript writing. Sabina Parajuli: Software, manuscript writing.

Patient consent

Written informed consent for publication of their clinical details and/or clinical images were obtained from the patient's father. A copy of the consent form is available for review by the Editor of this journal.

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