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SPINAL DYSRAPHISM AND ASSOCIATED ANOMALIES: INSIGHTS FROM A CASE SERIES OF FOUR CASES

Radiology

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ABSTRACT

Closed spinal dysraphism (CSD) encompasses a spectrum of congenital anomalies resulting from defective neurulation during embryologic development. These anomalies, characterized by skin- covered defects, often present diagnostic challenges due to subtle clinical signs such as sacral dimples, hairy patches, or subcutaneous masses. Magnetic resonance imaging (MRI) plays a pivotal role in diagnosing these conditions, allowing for early intervention to prevent progressive neurological, musculoskeletal, and urological complications. This case series presents four pediatric patients with various forms of CSD, including tethered cord syndrome, diastematomyelia, lipomyelomeningocele, and spina bifida occulta. Each case illustrates the diverse clinical presentations and MRI findings associated with CSD. Key findings included low-lying conus medullaris, split cord malformations, lipomatous masses, and syrinx formation. Surgical interventions, such as cord untethering and lesion excision, were performed in three cases, resulting in significant improvement or stabilization of symptoms. This series highlights the importance of early recognition of high-risk cutaneous markers, prompt imaging, and timely surgical intervention to prevent irreversible damage. It underscores the critical role of MRI in delineating the anatomy and guiding management. Further research into minimally invasive surgical techniques and prenatal diagnosis is necessary to optimize outcomes for patients with CSD.

KEYWORDS

INTRODUCTION

Spinal dysraphism is a term encompassing a spectrum of congenital anomalies that result from incomplete fusion of the spine during embryologic development. These anomalies can be broadly categorized into open and closed types, with the latter being referred to as occult spinal dysraphism (OSD). Closed-type spinal dysraphism is characterized by the presence of skin-covered spinal anomalies, often making the diagnosis less apparent compared to open types like myelomeningocele. While the clinical manifestations of closed-type spinal dysraphism vary, early detection and intervention are crucial to prevent irreversible neurological, musculoskeletal, and urological complications (1).

Closed spinal dysraphism includes a range of conditions such as tethered cord syndrome, spina bifida occulta, diastematomyelia (split cord malformation), and lipomyelomeningocele. These anomalies are frequently associated with cutaneous markers such as sacral dimples, hairy patches, or subcutaneous masses. Magnetic resonance imaging (MRI) remains the gold standard for the diagnosis of these conditions, providing detailed visualization of spinal abnormalities and their associated anomalies (2).

The pathophysiology of closed spinal dysraphism often involves tethering or anchoring of the spinal cord, which causes it to stretch as the child grows. This results in ischemic changes to the spinal cord and progressive neurological deficits, including motor weakness, sensory loss, and bladder or bowel dysfunction. Early imaging and diagnosis are critical in preventing such complications (3). Despite advances in imaging techniques, the clinical presentation and associated findings in patients with closed spinal dysraphism present diagnostic challenges.

Closed spinal dysraphism is a relatively rare condition, with an estimated incidence of 0.5 cases per 1,000 live births. However, the probability of detecting these anomalies varies based on clinical presentation and associated risk factors. For instance, sacral dimples located more than 2.5 cm from the anus or larger than 5 mm in size have a higher likelihood of being associated with underlying dysraphism (4). Other high-risk cutaneous markers include hemangiomas, hairy patches, and lipomas.

The incidence of occult spinal dysraphism is higher among children with associated anomalies, such as anorectal malformations or cloacal exstrophy. Early diagnosis in such cases is crucial, as delayed intervention can lead to progressive neurological and orthopedic complications, including scoliosis and foot deformities (5).

This case series aims to provide insights into the imaging spectrum of closed-type spinal dysraphism through the presentation of four pediatric cases. By correlating with clinical presentations, this series highlights the role of imaging in the early identification and management of these conditions.

CASE DETAILS

Case 1: Closed-Type Spinal Dysraphism with Tethered Cord.

A 5-day-old female neonate presented with a soft tissue swelling over the lumbosacral region noted since birth. Clinical suspicion of spinal dysraphism was raised. MRI findings were consistent with a closed-type spinal dysraphism, associated with a tethered cord.

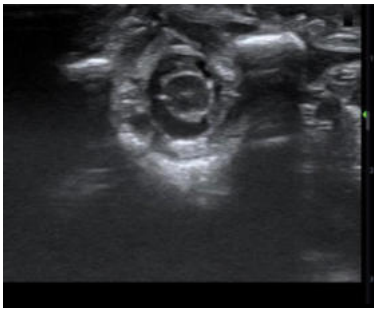

Fig 1: Significant fatty tissue around left aspect of lumbosacral spine. In the lower lumbar and sacral planes, the tissue penetrates the spinal canal. Low position of conus terminalis. Matching defects in the spinal column. spinal dysraphism with tethered cord.

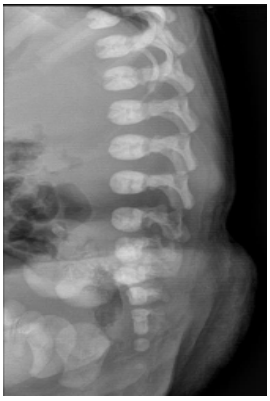

Fig 2: Segmentation defects of sacral vertebra affecting all elements. Fatty mass tangential to left dorsal aspect of spinal column.

Case 2: Spina Bifida Occulta (Closed Spinal Dysraphism).
A 2-year-old male child presented with complaints of chronic constipation. Clinical examination revealed a sacral dimple. Imaging evaluation was suggestive of spina bifida occulta, consistent with closed-type spinal dysraphism.

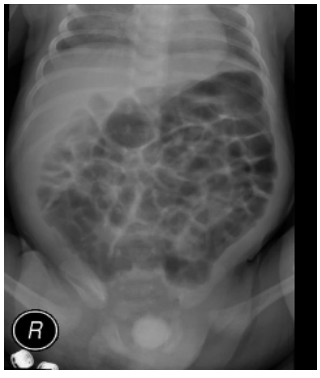

Fig 3: Supine radiograph shows air-filled small and large bowel loops right till rectum. This was imaged when he presented with constipation. Winking owl sign or the absent pedicle sign of left posterior element of L3 vertebra.

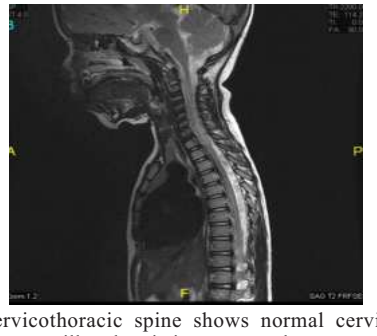

Fig 4: Cervicothoracic spine shows normal cervicomedullary junction. No tonsillar herniation or cord compression Normal posterior fossa. Normal lying conus at L1-2 with central canal dilatation from dorsal cord extending to tip of cord tethering.

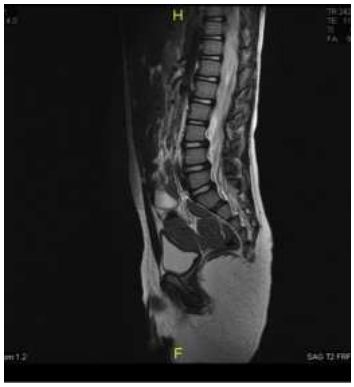

Fig 5: Sagittal lT2 demonstrates dilated central canal of cervicothoracic spinal cord seen mainly in the mid to lower half of thoracic spine. Absent left posterior arch of L3 with an apparent fusion of L2-3 vertebral spinous processes. Central canal dilatation of spinal cord with compression and thinning of dorsal and ventral ganglia. There is no diastematomyelia or paramedian syrinx. Thin element of T1 and T2 linear hyperintense signal is seen in the posterior thecal sac in the lumbosacral segment - consistent with an intrathecal lipomatous component.

Case 3: Lipomyelomeningocele with Tethered Cord.
A 3-year-old male child presented with a history of a lumbosacral mass noted since birth. Clinical and imaging evaluation revealed a lipomyelomeningocele, associated with a low-lying, tethered spinal cord—features consistent with closed spinal dysraphism.

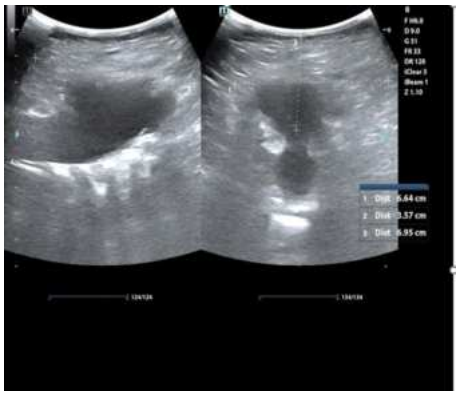

Fig 6: Findings – a large solid cystic mass lesion at the site of swelling measuring – 6.9 x 6.6 x 3.5 cm with internal fluid noted.it is communicating with spinal canal with increased interpedicular distance at the site of the lesion.

Case 4: Diastematomyelia – Type II.
A 2-year-old female child presented with lumbar hypertrichosis and decreased sensation in the right lower limb. MRI of the spine revealed diastematomyelia Type II (single dural sac with split cord), consistent with closed spinal dysraphism.

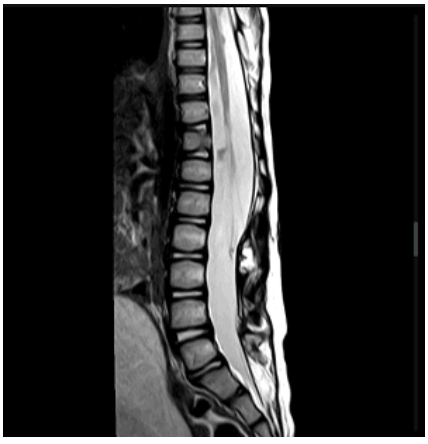

Fig 7 :

MRI findings include:
- Mild lower dorsal scoliosis with left-sided convexity.
- Congenital vertebral block from T8–T9 to T10–T11, with a butterfly vertebra at T11.
- Diastematomyelia Type II extending from T9–T10 to L2–L3, with hemicords located within a single dural sac and no bony or fibrous septum.
- The conus medullaris terminates at the L2–L3 level (within normal anatomical limits).
- Spina bifida occulta involving multiple vertebral levels.
- Mildly distended subarachnoid spaces and thecal sac.

DISCUSSION
- Closed spinal dysraphism (CSD) is a heterogeneous group of congenital anomalies resulting from defective neurulation or

canalization during embryonic development. This case series highlights the diverse clinical and imaging presentations of CSD and emphasizes the critical role of magnetic resonance imaging (MRI) in its diagnosis and management. The four cases discussed provide a representative spectrum of the condition, including tethered cord syndrome, diastematomyelia, lipomyelomeningocele, and spina bifida occulta, each with distinct clinical and radiological features.

- The reported incidence of spinal dysraphism ranges from 0.5 to 2 cases per 1,000 live births, with closed forms being less apparent clinically compared to open forms such as myelomeningocele (3). The clinical manifestations of CSD often vary depending on the specific anomaly and the degree of spinal cord involvement. The conditions are frequently identified in childhood, with symptoms ranging from cutaneous markers, such as dimples or hairy patches, to more severe neurological deficits like gait disturbances, urinary incontinence, and back pain, as seen in the cases presented here.

- A significant feature of CSD is the progressive nature of the neurological deficits caused by tethering of the spinal cord. As the child grows, the tethering exerts traction on the cord, resulting in ischemic changes that lead to sensory, motor, and autonomic dysfunction (2). Early diagnosis and intervention are therefore crucial to prevent irreversible damage.

CONCLUSION

This case series illustrates the diverse presentations and imaging findings of closed spinal dysraphism, highlighting the critical role of MRI in its diagnosis and management. Early recognition of high-risk clinical features and timely surgical intervention are key to preventing long-term complications. Further research is needed to refine diagnostic criteria and optimize treatment strategies, ensuring the best possible outcomes for affected patients.

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