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Case Report

A silent malformation: Closed spinal dysraphism in a boy with urinary incontinence ☆

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ABSTRACT

Spinal dysraphisms (SDs) are characterized by a heterogeneous range of congenital spinal anomalies that arise from derivative disturbances in neural tube development. Closed spinal dysraphism, a variant, is less common and typically has a subtle clinical presentation. Timely and correct diagnosis is essential to avert chronic complications. We describe a 12-year-old boy who presented with progressively worsening bladder and bowel incontinence since early childhood. On clinical examination, he was found to have swelling of the sacral region, gross motor impairment, and a slim physique. MRI showed hydro/syringomyelia, tethered cord, lipomeningocele, spina bifida in the sacral region, chronic cystitis and dural ectasia. These test results were consistent with the diagnosis of closed spinal dysraphism. This case highlights the need to consider urinary incontinence and sacral anomalies as manifestations of closed spinal dysraphism. Neuroimaging plays a key role in diagnosis, and especially MRI is the gold standard in detecting detailed structural abnormalities. To improve patient outcomes and quality of life, this paper emphasizes the intricacy of closed spinal dysraphism and the necessity of multidisciplinary management and early intervention.

Introduction

Spinal dysraphisms (SDs) are a collection of disorders of the spinal cord and the spine and are classified as congenital disabilities. These include various kinds of malformations of the spinal cord, which are due to the failure of mesenchymal, bony, and neural tissue to close along the midline. These defects take place during the period of 2-6 weeks of embryonic life [1]. SDs, part of a category of neural tube defects, occur at a population frequency of somewhere between 1 and 3 in every 1000 live births [2]. Burmese children with spinal malformations resulting from disruptions of secondary neurulation also tend to have anorectal or urogenital defects, reflecting the fact that the cloaca is in close embryological scanty with the caudal mass that later develops into the lumbosacral region

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[1,3]. Also, these children often have upper gastrointestinal or respiratory tract malformations because the notochord is involved in the development of the neural tube and the thoracoabdominal viscera [1]. Radiological imaging, in conjunction with the appropriate history and clinical findings, is crucial in confirming the diagnosis of SDs and therefore requires complex knowledge of the embryology of the spinal cord. MRI has evolved over time and remains as a noninvasive tool at hand for diagnosis of SDs [4]. Here under we present a case of an adolescent boy with bladder- bowel incontinence who came to our centre for further evaluation.

Case report

A 12-year-old boy was referred for evaluation of progressively worsening loss of bladder and bowel control. His evacuations had been spontaneous since childhood, and there had been no prior history of retention. His past obstetric history was significant for an FTND (full-term normal delivery) with a 3.5 kg birth weight without NICU admission. On physical examination, a 2 × 2 cm swelling was observed in the lower back.

The swelling on the patient's lower back has increased in size, now measuring 3 × 3 cm. During physical examination, the patient was noted to have a lean build, with a weight of 40 kg and a height of 145 cm, resulting in a BMI of 18, which is on the lower end of the normal range (18.5-24.9). The patient was unable to walk properly, exhibiting a limp in the left leg, indicating gross motor impairment. However, fine motor milestones for the hands were normal, and language and social milestones were achieved up until age, though the patient exhibited slow and hesitant responses. The patient is not currently enrolled in school due to issues with bowel and bladder incontinence.

On general physical examination (GPE), the patient's heart rate (HR), blood pressure (BP), respiratory rate, and temperature were all within normal limits. The patient was oriented to time, place, and person, and was able to answer general questions and perform simple tasks with their hands. On abdominal examination, there was tenderness in the lower abdomen. On local examination of the back, a well-defined swelling was noted over the lower sacral region, covered with skin. The swelling was nonmobile, soft to touch, and mildly tender, with an approximate size of 2 × 2 cm.

On MRI, there is a long segment intramedullary, multiseptated, T2 hyperintensity seen in the cervico-thoracic and lumbar cord measuring 37.4 cm from the mid body of C2 to upper-end plate of L3 vertebra, with spinal cord expansion and effacement of anterior and posterior subarachnoid space in the cervical and dorsal parts, with a maximum diameter of 12.3 mm at C5-C6 disc level suggestive of hydro/syringomyelia (Fig. 1).

In these images also we can see that the multiseptated syringomyelia extends all the way up to the lower endplate of L3 where the conus medullaris is ending, and the filum terminal appears attached to the posterior part of the dural sac suggestive of tethered cord. There is no evidence of tonsillar herniation, ruling out Chiari malformation and the visualized sagittal sections of the brain appeared normal (Fig. 1).

In the coronal sections of the sacrum, there is asymmetry of the sacrum with the right end being normal while the left end shows some defect in the lower part (Fig. 2). There was also high signal fluid containing large bowel on the left side.

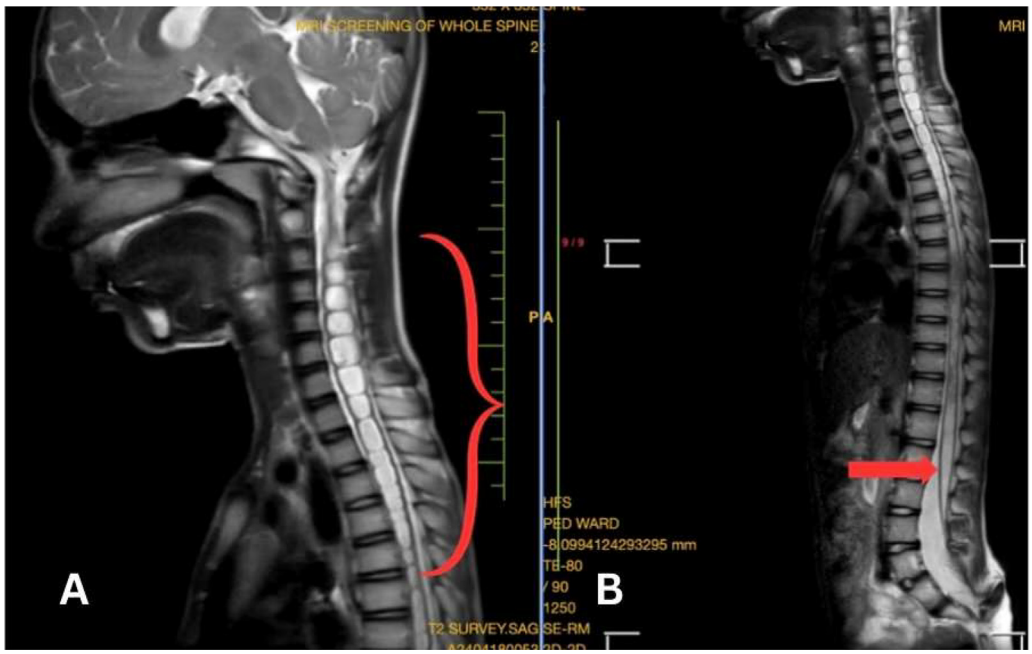

Fig. 1 – T2 weighted sagittal images of the cervical (A) and whole spine (B) showing multiseptated spinal cord in the cervical region giving it a lobulated appearance seen extending into the thoracic spine as well as indicated by the braces which is suggestive of hydro/syringomyelia. In image B the syringomyelia is seen extending all the way down to the lumbar spine where the cord is seen attached to the posterior dural sac which is suggestive of a tethered cord.

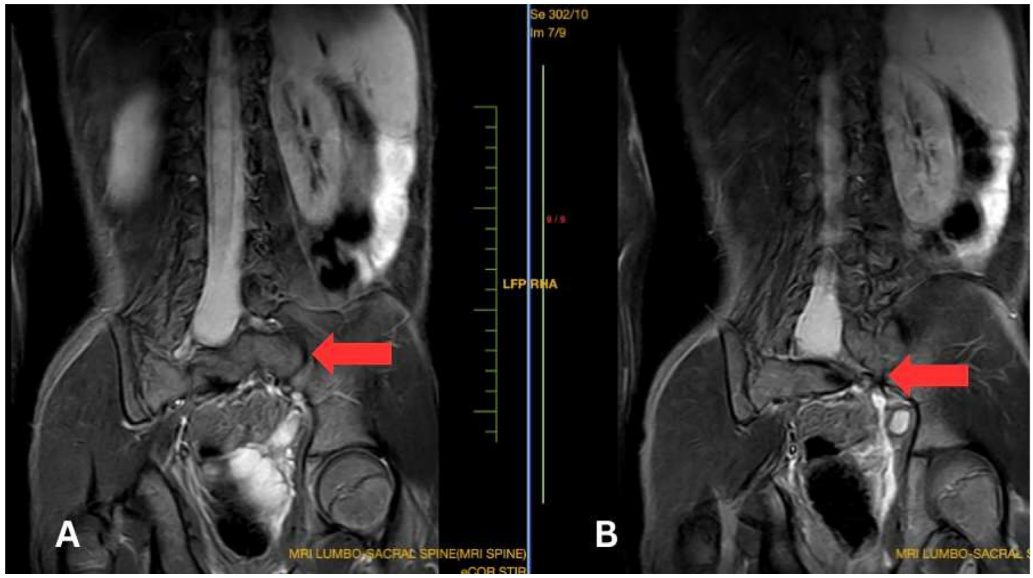

Fig. 2 – Short Tau Inversion Recovery (STIR) coronal images of the sacrum showing loss of normal ala on the left side in A and B causing asymmetry of the pelvis. Large bowel loops containing fluid appearing hyperintense.

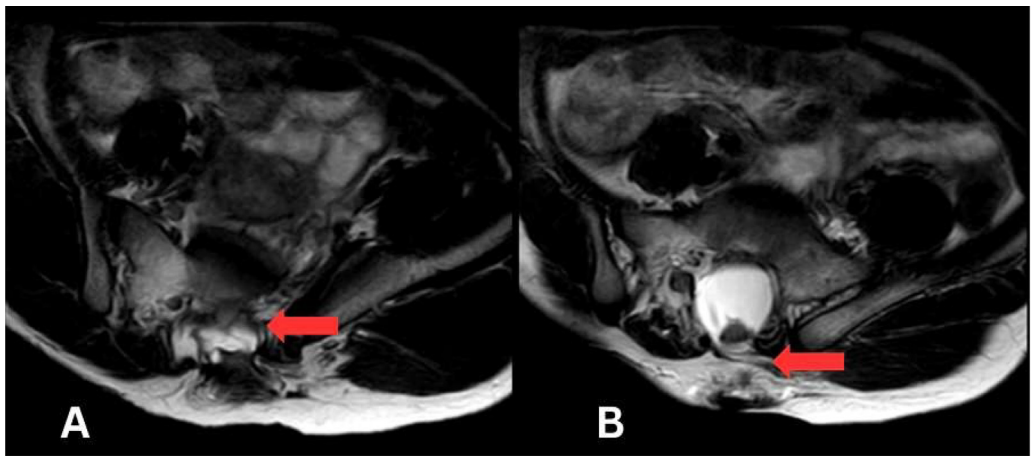

Fig. 3 – Axial T2 weighted images showing bony spina bifida indicated by red arrow in image A through which the thecal sac is seen extruding towards the subcutaneous plane as indicated by red arrow in image B.

Closed spinal dysraphism in the sacral region with the absence of posterior bony elements in sacral segments from L5 to S3 vertebrae is suggestive of spina bifida. T2/PDFS hyperintense structure seen extruding from the above-mentioned defect of size approx. 11 mm at the level of the S2 vertebra into the subcutaneous plane (Fig. 3). The meninges and thecal sac along with tethered conus and cauda equina within protrude dorsally through the bony defect into the subcutaneous space on the left side with overlying subcutaneous fat seen covering this meningocele sac suggestive of lipomeningocele. The myelomeningocele sac measures approx. 2.5 × 1.2 × 1.9 cm in dimension.

Increase in anterior-posterior diameter of dural sac in lumbosacral region with scalloping of posterior vertebral body suggestive of dural ectasia with maximum diameter of almost 3 cm (Fig. 4).

Irregular bladder thickening with multiple sacculations and diverticula suggestive of chronic cystitis causing urinary incontinence (Fig. 5).

The patient was admitted to the neurosurgery department where a shunting surgery was adviced.

Discussion

Bladder bowel incontinence as a presentation of syringomyelia is an unusual presentation of the underlying spinal abnormalities. The presence of isolated spinal abnormalities without associated tonsillar herniation as rare to find. Also the presence of myelocele at the sacral end with tethered cord makes the diagnosis complete.

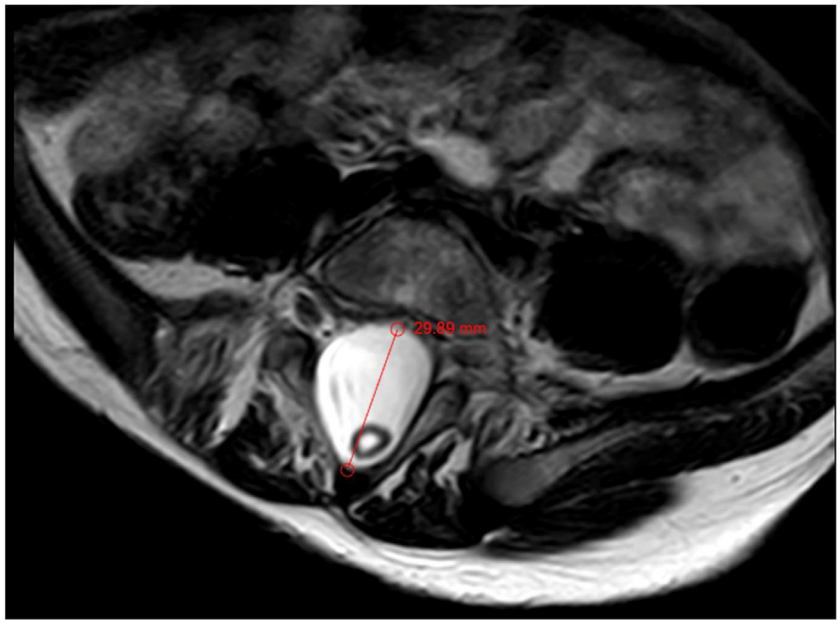

Fig. 4 – Axial T2 weighted image showing an increase in the anteroposterior diameter of the dural sac with a ring-like spinal canal measuring 29.8 mm which is suggestive of dural ectasia.

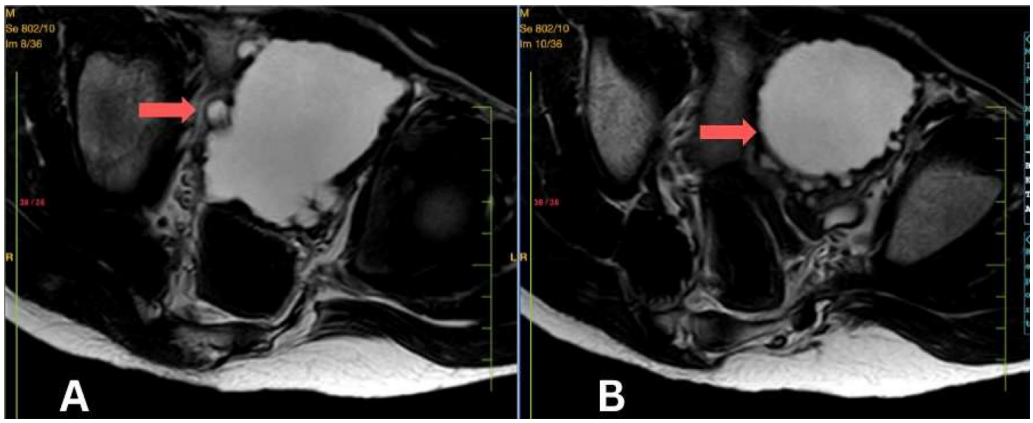

Fig. 5 – Axial STIR images showing multiple sac-like pouches arising from the bladder wall as shown by the red arrow in image A and irregularly thickened wall in image B suggestive of chronic cystitis.

The family of developmental disorders known as spinal dysraphisms (SDs) has a complicated, multivariate etiology that includes nutritional, environmental, and genetic factors [5,6]. The pathophysiology of SDs has been linked to mutations in the genes MTHFR, MTHFD1, MTRR, VANGL1, and VANGL2 as well as single nucleotide polymorphisms (SNPs) in the T locus on chromosome 6q [7]. The development of these diseases is significantly influenced by environmental variables during the periconceptional period, such as maternal obesity, poor nutrition, smoking, hyperhomocysteinemia, sedentary lifestyles, and mental stress. These variables are associated with elevated levels of inflammation and oxidative stress, which are connected to faster telomere shortening and biological aging in mothers [5]. Among these environmental factors, perinatal maternal nutrition stands out as a key determinant of SD prevalence, closely tied to the patient's socioeconomic status. A notable example is the role of folic acid deficiency, which remains one of the most significant contributors to these disorders [8].

There are basically 3 main embryologic stages of spine development [9,10].

1. Gastrulation: The first stage, typically takes place during the second or third week of embryonic development. The process of gastrulation is how a blastula becomes a trilaminar embryo through the development of 3 germ layers: endoderm, mesoderm, and ectoderm.
2. Primary neurulation: The neural plate forms due to interactions between the notochord and overlying ectoderm during the third and fourth weeks. The neural tube is created by the neural plate bending and folding, and it closes like a zipper from front to back and vice versa.
3. Secondary neurulation: The fifth and sixth weeks are when the third and last stage occurs. At this point, the caudal cell

Table 1 – Broad classification of spinal dysraphism with subtypes.

Spinal dysraphism	Subtype	Details
Open spinal dysraphism		
	Myelomeningocele	
	Myelocele	
	Hemi-myelomeningocele	
	Hemi-myelocele	
Closed spinal dysraphism	With subcutaneous mass	
	thoracolumbar region	Lipomyelomeningocele, Lipomyelocele, Terminal Myelocystocele, Meningocele
	Cervical	Cervical myelomeningocele, Cervical Myelocystocele, Meningocele
	Without subcutaneous mass	Split cord malformation, Dorsal dermal sinus, Lipoma (Intradural/Filar), Tight filum terminale, Persistent fourth ventricle, Neurenteric cyst, Caudal regression syndrome, Segmental spinal dysgenesis

mass forms a secondary neural tube. This solid secondary neural tube experiences cavitation in a process known as retrogressive differentiation, which ultimately produces the filum terminale and the tip of the conus medullaris.

Any disruption in these developmental phases may result in defective spinal or spinal cord formation.

The following categories can be used to broadly classify spinal dysraphism (Table 1).

Conclusion

In summary, the MRI findings were conclusive of closed cord dysraphism-spina bifida, lipomeningocele in the sacral region, hydro/syringomyelia of the spinal cord, low-lying conus medullaris with focal tethered cord, dural ectasia and chronic cystitis.

A diverse and intricate collection of anomalies resulting from disruptions in spinal development is known as spinal dysraphism. SD has considerable economic and emotional repercussions and contributes significantly to childhood morbidity due to cognitive impairment and associated brain abnormalities. When diagnosing and evaluating SDs before surgery, neuroimaging is crucial. Nonetheless, SD's imaging qualities could look difficult. As a result, a rational approach based on the integration of clinical, embryological, and neuroimaging data substantially supports the diagnosis in the vast majority of instances.

Patient consent

The authors declare that they have obtained written informed consent from the patient's guardian prior to writing the case report, including permission for publication of all photographs, images, and clinical data included herein.

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