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Diastematomyelia: An unusual cause of chronic back pain in adulthood

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Abstract

Diastematomyelia, or split cord malformation, is a rare congenital spinal anomaly characterized by a longitudinal division of the spinal cord into two hemi cords, separated by a fibrous or bony septum. Typically diagnosed in childhood, adult presentation is infrequent and more common in females. This case report details a 23-year-old woman from a rural region in Northern Pakistan who has suffered from chronic lower back pain since childhood, with a notable exacerbation over the past year. Clinical examination revealed lumbar scoliosis, a dimple at the natal cleft, leg length discrepancy and right foot equinovarus deformity. MRI findings confirmed diastematomyelia, highlighting scoliosis, multilevel segmentation defects, spina bifida and a bony spur splitting the spinal canal. The patient exhibited signs suggestive of tethered cord syndrome and was referred for neurosurgical evaluation. This case underscored the importance of diastematomyelia in adults, particularly in those with chronic back pain and associated neurological symptoms. Early diagnosis and appropriate surgical interventions are crucial to prevent neurological deterioration and improve long term outcomes. The report also reviews the literature on clinical features, diagnostic approaches and management strategies for this condition, emphasizing the need for awareness among healthcare providers.

Keywords: Back pain; Diastematomyelia; Split cord malformation; Neural tube defect; Dysraphism.

Abbreviations: MRI: Magnetic Resonance Imaging; CT: Computed Tomography.

Background

Diastematomyelia, also called split cord malformation, is a congenital spinal condition characterized by a longitudinal split in the spinal cord into two distinct hemicords by a fibrous, cartilaginous, or osseous septum. This condition belongs to the spectrum of spinal dysraphism that is usually diagnosed prenatally or during childhood, often seen in thoracolumbar region. Adult presentation is unusual and is more common in females [1]. We present a case of a young female with diastematomyelia who presented to primary care with complaint of chronic back pain. In this report clinical and radiological features, as well as treatment options, are discussed based on pertinent literature review.

Case presentation

A 23-year-old woman presented to the family medicine clinic with a longstanding history of low back pain, which had been

present since childhood but had significantly worsened over the past year. She was from a rural area in Northern Pakistan, where access to established healthcare facilities was limited, delaying appropriate medical evaluation. The pain was localized to the lower back and radiated bilaterally down her legs. She described it as pricking in nature and rated its severity as 5 out of 10 on her worst days. Over time, she noticed increasing difficulty in walking after prolonged activity, accompanied by observable changes in her gait. There were no symptoms of bowel or bladder dysfunction. Employed as a nurse, her job required frequent patient lifting, which had become increasingly challenging due to her symptoms. She had previously been treated as a case of mechanical back pain with analgesics and muscle relaxants, but without significant improvement. Her past medical and surgical history was unremarkable, and she was unaware of any family history of congenital anomalies. On inspection she had lumber scoliosis, a dimple at the site of the natal cleft (without tuft of hair). There was leg length discrepancy on the right side and right foot equinovarus deformity. Range of movement at the spine was normal however right hip external rotation was restricted. Neurological examination revealed normal sensation, muscle power and tone in both lower limbs. Deep tendon reflexes were present and symmetrical.

MRI lumber spine was ordered for this patient (Figures 1-3).

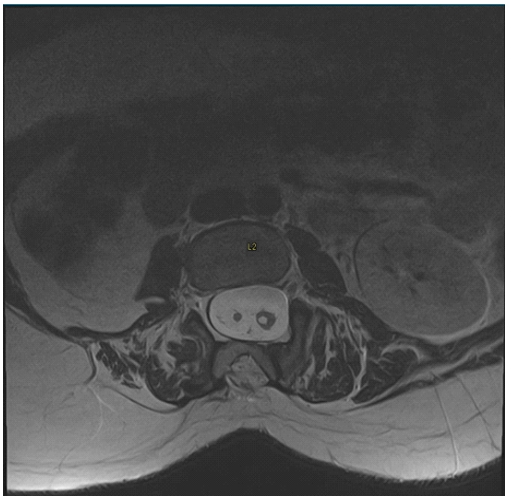

Figure 1: MRI: T2-weighted image, axial plane, level L2: a single dural sac containing both hemicords.

The MRI was reported as Scoliosis of lumbar spine identified with convexity towards right side. Multilevel segmentation defect identified in distal lumbosacral spine as evident by block and butterfly vertebra. Defective posterior elements identified in sacrum. Spina bifida identified at L4-L5 level and distally with deficient posterior elements. A bony spur was identified at L4 vertebra splitting the spinal canal into two. A linear horizontal fibrotic scarring was seen in subcutaneous tissue extending from L4 posterior elements up to subcutaneous tissues. Distal spinal cord below T10 was showing dilatation of central canal suggestive of hydromyelia. Distal cord seemed to split into two at the level of bony spur and recombined at L5 level with possible tethering. There was significant atrophy of posterior paraspinal muscles as well.

The patient was advised to see a neurosurgeon for surgical opinion but she couldn't see one due to financial constraints.

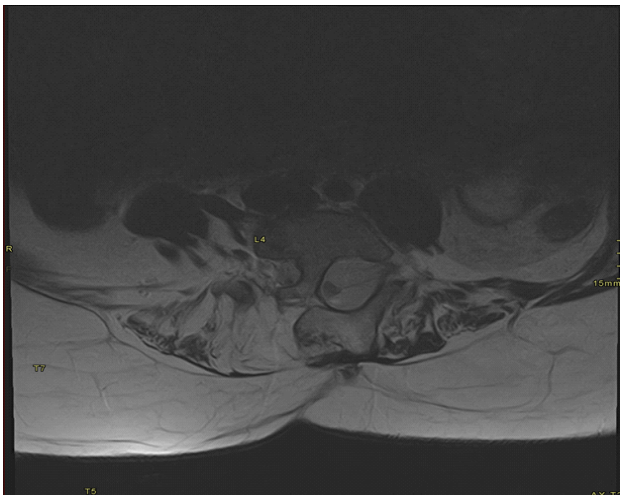

Figure 2: MRI: T2-weighted image, axial plane, level L4: A bony spur splitting the spinal canal into two.

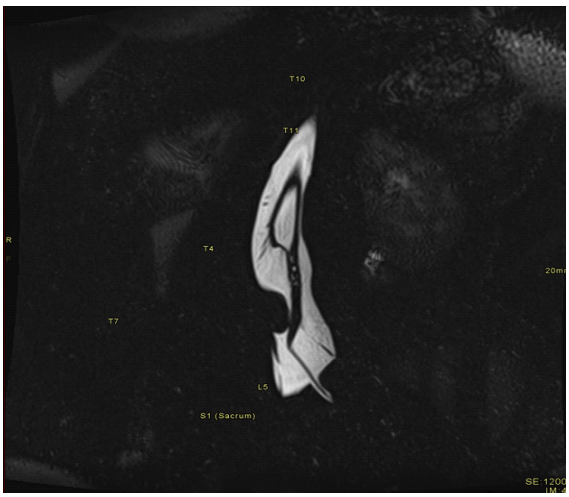

Figure 3: Myelogram: Distal spinal cord below T10 shows dilatation of central canal suggestive of hydromyelia. Distal spinal cord recombined at L5.

She was suggested for customized physiotherapy focusing on her core stabilization, postural alignment and gait training. Safe lifting techniques, shoe lift, pain control medications and routine follow up suggested. The patient returned for several scheduled follow-up visits to receive intravenous analgesic therapy as part of the ongoing management of her chronic back pain. Throughout the course of these visits, her neurological examination remained stable with no new deficits noted, indicating no progression of neurological involvement to date.

Discussion

The term Diastematomyelia is derived from the ancient Greek word 'diastema' meaning cleft. It is a rare congenital anomaly that is believed to occur during the gastrulation stage of development due to abnormal movement and separation of precursor cells [2]. Prenatal sonographic diagnosis of this anomaly is usually possible in the early to mid-third trimester [3]. It was first described in 1837 by French researcher C.P. Ollivier, who used the term 'diastematomyelia' [4]. A new classification and term 'split cord malformation' (SCM) was introduced by D. Pang, who distinguished two types of diastematomyelia [5]. Type 1, the classic presentation where both the hemicords are contained within a single dural envelope and patients are usu-

ally symptomatic, presenting with scoliosis and tethered cord syndrome. Type 2 is the milder form where both hemicords are contained within their own separate dura and patients may have mild symptoms and even be asymptomatic. Based on MRI findings in our case—presence of a bony spur with the spinal cord splitting and recombining—the patient most likely had Type I diastematomyelia, which is associated with more significant symptoms due to tethering.

The condition typically presents in children but occasionally goes undiagnosed until adulthood. Most patients diagnosed with diastematomyelia are children under 7 [6]. Among newborns, the presence of cutaneous stigmata of spinal dysraphism (e.g. hairy patch, dimple, lipoma, hemangioma) or an associated meningocele brings the defect to attention. As described in the literature this condition is associated with various neurologic and orthopaedic abnormalities including foot deformities, muscular atrophy, scoliosis, limb shortening, paraesthesia, bowel bladder incontinence, reflex abnormalities, progressive weakness, lower back pain and gait disturbances [7].

With modern imaging techniques, various types of spinal dysraphism are being diagnosed with increasing frequency. Bony malformations are generally recognized on plain x-rays and more clearly appreciated on CT scan. This disorder must be taken into account in differential diagnosis in patients with X-rays revealing defects of the spine in the form of fusion of vertebrae, hemivertebrae or butterfly vertebrae, especially when there are additional skin lesions on the back. MRI is the modality of choice as it can better elucidate a split spinal cord, degree of neurologic compression and the presence of other associated abnormalities. Myelogram is a useful adjuvant.

Asymptomatic patients do not require surgical treatment. However, they should be observed closely for progressive neurological deterioration as it is known that the condition can worsen. Surgical intervention is warranted in patients who present with new onset neurological signs and symptoms or have a history of progressive neurological manifestations that can be related to this abnormality. The surgical procedure required includes decompression of neural elements and removal of bony spur. This may be accomplished with or without resection and repair of the duplicated dural sacs. Performing it is preferred since the dural abnormality may partly contribute to the "tethering" process responsible for the symptoms of this condition.

Conclusion

Diastematomyelia, although uncommon, poses significant challenges due to its potential to cause tethering of spinal cord, progressive neurological deficit and various associated anomaly. Early recognition of this condition, by prenatal ultrasound or MRI, and appropriate management can help minimize neurological deficits and allow affected individuals to seek treatment sooner, thus improving long-term outcomes. Increased awareness of this condition among primary care providers is essential, particularly in resource-limited settings especially for pain management strategies, provision of orthotic support where needed, physiotherapy, psychosocial support and timely referral for surgical intervention.

Declarations

Informed consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images.

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Conflict of Interest: None

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