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When Occult Dysraphism meets high-flow vascular malformation: A rare paraspinal AVM in Spina Bifida Occulta

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Abstract
Spina bifida occulta (SBO) is the mildest form of closed spinal dysraphism and is characterized by incomplete fusion of the posterior vertebral elements without involvement of the spinal cord or meninges. It is frequently asymptomatic and often detected incidentally. However, associated structural anomalies may coexist.

We describe the case of a 21-year-old male with known beta-thalassemia who presented with a slowly enlarging swelling over the lower back. MRI of the spine revealed a defect in the posterior vertebral arch at the S2 level along with bifid spinous processes at L3 and L4, consistent with the findings of spina bifida occulta. In addition, a sizable high-flow vascular lesion was noted within the posterior subcutaneous tissues and paraspinal muscles, extending from L1 to S3. The lesion demonstrated numerous serpiginous signal voids on routine sequences and showed heterogeneous enhancement following contrast administration. Subsequent Doppler ultrasound evaluation demonstrated prominent arterial inflow and venous outflow channels, supporting the diagnosis of a paraspinal arteriovenous malformation. There was no evidence of extension into or communication with the thecal sac.

This case underscores the value of contrast-enhanced MRI in conjunction with Doppler ultrasound in accurately distinguishing closed forms of spinal dysraphism from open neural tube defects, as well as in detailed assessment of associated vascular abnormalities.

Keywords: Spina bifida occulta, closed spinal dysraphism, paraspinal arteriovenous malformation, magnetic resonance imaging, vascular anomaly

Introduction

Spinal dysraphism encompasses a range of developmental abnormalities that arise from incomplete or abnormal closure of the neural tube during early fetal life. Among these, spina bifida occulta represents the mildest variant and is characterized by failure of fusion of the posterior vertebral elements, with intact skin covering and normally formed neural structures. The lumbosacral spine is most frequently affected, and many cases are identified incidentally during imaging performed for unrelated reasons.

MRI serves as the primary imaging modality for assessing potential intraspinal associations, including tethered cord, lipomatous lesions, dermal sinus tracts, and vascular anomalies. Paraspinal arteriovenous malformations, although uncommon, are high-flow vascular entities that demand accurate radiological characterization to guide therapeutic decision-making and procedural planning.

Case Report

A 21-year-old male, known case of spina bifida with beta-thalassemia, presented with complaints of swelling over the lower back since childhood, with recent increase in size. There was no history of trauma, discharge, bowel or bladder dysfunction, or lower limb neurological deficits. Clinical examination did not reveal cutaneous stigmata of spinal dysraphism.

MRI of the whole spine was performed using T1-weighted, T2-weighted, STIR, diffusion-weighted, and post-contrast sequences.

Imaging Findings

Posterior Soft Tissues

A large, well-defined lesion measuring approximately 3.5 × 15.1 × 19.6 cm was identified in the posterior midline and paramedian region extending from L1 to S3 levels. The lesion was iso- to hypointense on T1-weighted images and hyperintense on T2/STIR sequences with multiple serpiginous flow voids. Heterogeneous enhancement was observed on post-contrast images without diffusion restriction. No communication with the thecal sac was demonstrated.

Lumbar Spine

A posterior vertebral arch defect was noted at S2, along with bifid spinous processes at L3 and L4. Diffuse low marrow signal intensity was observed, likely related to chronic marrow hyperplasia secondary to beta-thalassemia.

Spinal Canal

The conus medullaris terminated normally at the L1 level. No evidence of tethered cord, intradural mass lesion, or syringomyelia was identified.

Screening Doppler Ultrasound

Color Doppler examination demonstrated multiple tortuous vascular channels with arterial feeders and venous drainage patterns, confirming a high-flow arteriovenous malformation.

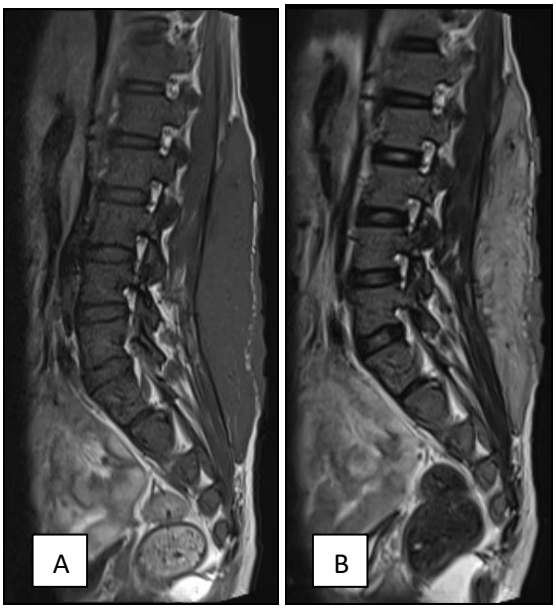

Fig 1: T1 (A) hypointense and T2 (B) hyperintense mass lesion involving the subcutaneous and intramuscular plane of posterior midline and paramedian region of the lower back extending from L1 to S3 vertebral level

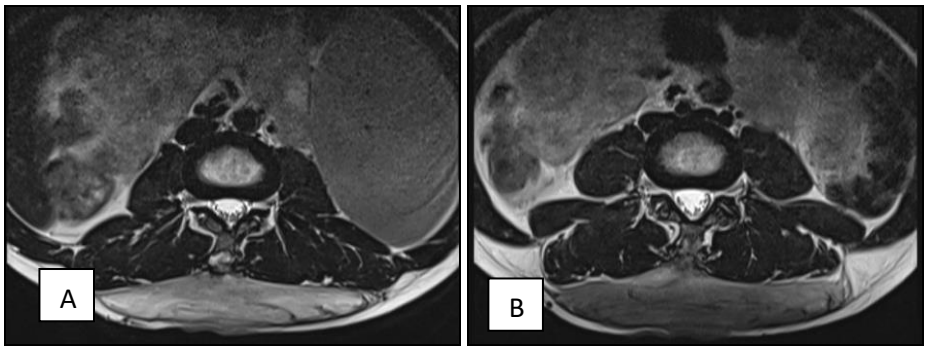

Fig 2: T1 weighted axial images showing hypointense mass lesion involving the subcutaneous and intramuscular plane of posterior midline and paramedian region of the lower back at L3-4 (A) and L4-5 (B)

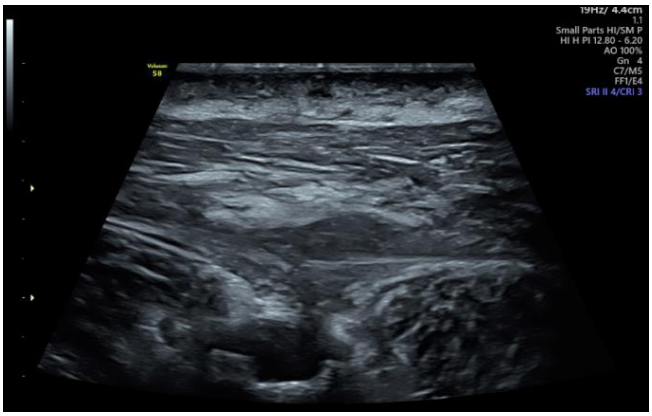

Fig 3: Ill defined heteroechoic lesion in the subcutaneous plane of posterior midline and paramedian region of lower back

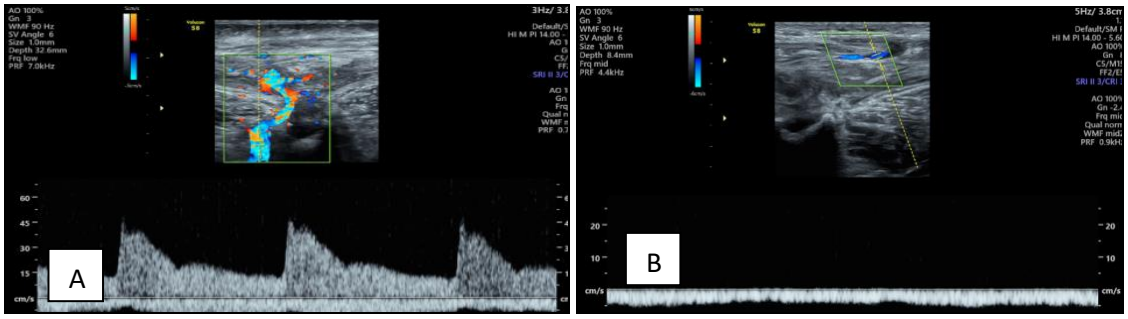

Fig 4: Colour doppler ultrasound showing arterial (A) and venous (B) flow within the mass lesion

Discussion

Spina bifida occulta arises due to failure of complete fusion of the posterior vertebral arches during early embryologic development. In contrast to open forms of spinal dysraphism, the neural tissue remains normally positioned, and there is no protrusion of the meninges or spinal cord through the defect.

MRI remains the preferred modality for comprehensive evaluation of spinal dysraphism, as it provides precise visualization of the conus medullaris, spinal canal contents, and any associated structural abnormalities. In the present case, the conus was normally positioned, thereby excluding features suggestive of tethered cord syndrome.

Paraspinal arteriovenous malformations are rare high-flow vascular lesions resulting from abnormal direct communications between arterial and venous channels without an intervening capillary bed. On MRI, they are typically identified by multiple serpiginous signal voids and prominent enhancement following contrast administration. Doppler sonography further aids in confirming the high-flow nature of these lesions by demonstrating arterial inflow and rapid venous drainage patterns.

Conclusion

This case illustrates the characteristic imaging findings of spina bifida occulta in conjunction with an uncommon coexistence of a sizable posterior paraspinal arteriovenous malformation. The combined use of contrast-enhanced MRI and Doppler ultrasound is crucial for precise lesion characterization, confirmation of the high-flow vascular component, and appropriate therapeutic planning

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