CASO CLÍNICO/CASE REPORT

Lipomyeloschisis: A Rare Finding

Lipomielosquise: Um Achado Raro

Márcio Duarte [1,*], Bruno Abreu [1], José Prado [1], Marcelo Silva [1]
1-WEBIMAGEM, São Paulo, Brasil

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***Autor Correspondente / Corresponding Author:**
Márcio Luís Duarte
WEBIMAGEM Telerradiologia
Avenida Marques de São Vicente, 446,
São Paulo – São Paulo, Brazil
marcioluisduarte@gmail.com

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Abstract

Introduction: Dysraphisms, especially involving caudal spine, are the most frequent congenital malformations of the spine and spinal cord. Lipomyeloschisis is a common subtype of closed spinal dysraphism with lipoma. On magnetic resonance imaging (MRI), the lesions are isointense with subcutaneous fat in all sequences – they give high signal on both T1- and T2-weighted images. Computed tomography with 3D reconstruction allows detailed evaluation of bones, vertebral anomalies and osseous hamartoma, and helps plan the surgery in conjunction with MRI findings. Neuroimaging can accurately depict both the lesion and the exact location within the spinal canal, so prompting the precise diagnosis, not clinically suspected, even in elderly patients. Neural tube defects are preventable to some extent by proper consumption of folic acid. The newer treatment modalities with stem cells though demonstrate some improvement.

Case Report: Two-year-old boy with spinal malformation from birth on neurological follow-up. At the physical examination, the patient had a normal neuropsychomotor evaluation. *Magnetic resonance imaging* (MRI) demonstrated the medullary cone extending to the vertebral body of L4 – tethered cord syndrome –, with thickening of the *filum terminale*, with a lipomatous structure on the posterior aspect of the spinal canal. This set of findings suggested the diagnosis of lipomyeloschisis.

Conclusion: We report a case of lipomyeloschisis, a neurological disease poorly reported, caused by problems in neurulation process of the neural tube, with variable severity, but with accurate diagnosis by magnetic resonance imaging.

Resumo

Introdução: Disrafismos, especialmente os que envolvem a coluna caudal, são as malformações congénitas mais frequentes da coluna vertebral e da medula espinhal. A lipomielosquise é um subtipo comum de disrafismo fechado da coluna vertebral com lipoma. Na ressonância magnética (RM), as lesões são isointensas com a gordura subcutânea em todas as seqüências – alto sinal nas imagens ponderadas em T1 e T2. A tomografia computadorizada com reconstrução em 3D permite uma avaliação detalhada dos ossos, anomalias vertebrais e hamartoma ósseo, além de ajudar a planejar a cirurgia em conjunto com os achados da RM. A neuroimagem pode detectar, com precisão, tanto a lesão quanto a localização exata no canal espinhal, de modo que o diagnóstico não clinicamente suspeitado é preciso, mesmo em pacien-

tes idosos. Os defeitos do tubo neural são evitáveis até certo ponto pelo consumo adequado de ácido fólico. As novas modalidades de tratamento com células-tronco demonstram algumas melhorias.

Caso Clínico: Menino de dois anos de idade com malformação medular desde o nascimento em acompanhamento neurológico. Ao exame físico apresenta exame neuropsicomotor normal. A RM demonstra cone medular estendendo-se até a altura do corpo vertebral de L4 – síndrome da medula presa –, com espessamento do *filum terminale*, apresentando-se ancorado a estrutura lipomatosa no aspecto posterior do canal espinhal. O conjunto de achados sugere lipomielosquise.

Conclusão: Expomos um caso lipomielosquise, doença neurológica pouco relatada, causada por problemas na neurulação do tubo neural e de gravidade variável, porém de preciso diagnóstico pela ressonância magnética.

Introduction

Neural tube defects (NTDs) are the result of disturbance in the neurulation process.[1] Neurulation, through a coordinated series of events, gives rise to the neural plate, neural folds and the neural tube, which eventually differentiates and develops into the future brain and spinal cord.[1] Most NTDs arise in anterior and posterior neuropores as they close last.[1]

NTDs can be opened or closed depending upon exposed or closed neural tissue.[1] Open NTDs (ONTDs) are the result of primary neurulation and may involve any area of the central nervous system (CNS).[1] Closed NTDs are due to defects in secondary neurulation and are mostly confined to the spine.[1]

Case Report

A two-year-old boy, born at 41 weeks of gestation by normal delivery and with no relevant pre or perinatal records, is under regular neurological follow-up, due to a postnatal diagnosis of a spinal malformation. His mother referred that the ultrasonographic tests performed during pregnancy were normal, denied trauma and the consumption of any drug during that period. At physical examination, no focal deficits were observed and neuropsychomotor evaluation was normal. A spine magnetic resonance imaging (MRI) was obtained, showing the medullary cone extending to the vertebral body of L4 – defining a tethered cord syndrome –, with thickening of the *filum terminale* (0.3 cm), with a lipomatous structure measuring 3.0 x 0.5 x 0.8 cm on the posterior aspect of the spinal canal. This set of findings suggested the diagnosis of lipomyeloschisis (**Figs. 1 and 2**).

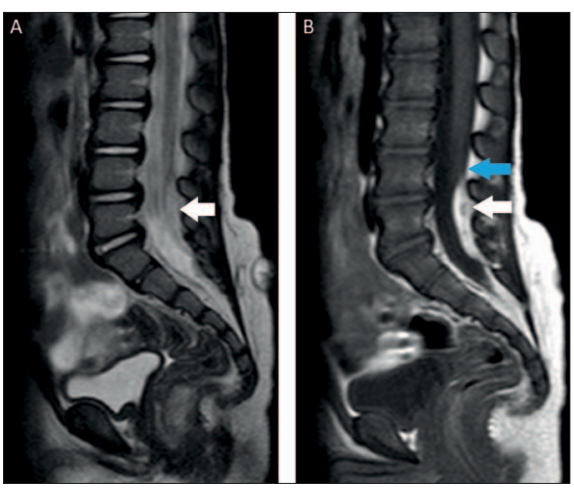

Figure 1. Sagittal section MRI in T2 sequence in A and T1 sequence in B, both without contrast, demonstrating medullary cord extending up to a L4 vertebral body (blue arrow) – tethered cord – with a lipoma between the dura and the bony wall in the extradural space (white arrow).

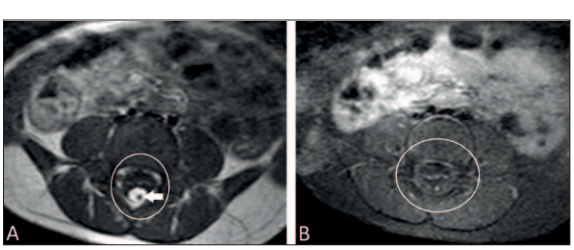

Figure 2. Axial section MRI in T1 sequence without contrast in A and T1 FAT SAT sequence with contrast in B demonstrating a lipoma between the dura and the bony wall in the extradural space (white circle) without contrast enhancement and the intracanal lipoma (white arrow).

Discussion

Dysraphisms, especially involving caudal spine, are the most frequent congenital malformations of the spine and spinal cord.[2] Lipomyeloschisis is a common subtype

of closed spinal dysraphism with lipoma.[2-4] Lipoma is a monophyllic mass originating from the mesoderm.[2] Embryologically, spinal lipomas result from early dysjunction between neuroectoderm and cutaneous ectoderm; the surrounding mesenchyme creeps between and adheres to the primitive ependyma, which induces it to transform into fat.[2]

The lipoma may extend upward into the spinal canal between the dura and the bony wall in the extradural space.[3] It may even enter the open central canal of the spinal cord and pass upward to form apparently isolated intradural lipoma at higher levels.[3] Aberrant tissues such as cartilage, striated muscle, neural cells, and bone may be present within the subcutaneous lipoma.[3,5]

Clinical presentation of these lesions is variable, depending upon size and location of the lipoma.[2] Paraparesis, sensory changes, urinary incontinence and pain are frequent presenting complaints.[2]

MRI is indispensible to characterize the type of dysraphism, cord and nerve root compression, presence of low-lying tethered cord, and dorsal dermal sinus—all of which could be the cause of symptom in this setting.[3] On MRI, the lesions are isointense with subcutaneous fat in all sequences – they give high signal on both T1- and T2-weighted images.[2]

The early diagnosis of this clinical condition is important since it requires surgical correction. And the sooner it is done, in a child's life, the less functional impact it can generate. Even if the child does not have, until now, neurological symptoms suggestive of low spinal dysfunction, this may happen over time. Therefore, a successful surgical correction, at an early stage of life, can obviate a significant number of future complications. ■

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