Tethered cord syndrome resembling Charcot-Marie-Tooth disease in closed spinal dysraphism

Síndrome da medula ancorada mimetizando doença de Charcot-Marie-Tooth no disrafismo oculto da coluna

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A 23-year-old man presented with progressive difficulty walking since childhood. Examination showed pes cavus, hammer toes, and peroneal atrophy (Figure 1). Electroneuromyography revealed chronic denervation with an axonal pattern, and Charcot-Marie-Tooth disease was suspected. Urinary retention had started 3 years previously. Spinal cord magnetic resonance imaging (MRI) showed a lipomeningomyelocele at the lumbosacral region with spinal cord tethering (Figure 2). The skin of the lumbosacral region was normal. Surgery was performed without immediate improvement.

Tethered cord syndrome due to occult spinal cord dysraphism is a diverse clinical entity, and cutaneous manifestations may be absent. Pes cavus and hammer toes are rare but can be present and strongly resemble Charcot-Marie-Tooth disease1-3. The emergence of urinary symptoms is a hallmark for the diagnosis of cauda equina syndrome instead of neuropathy.

Figure 1. Note pes cavus, hammer toes, and peroneal atrophy, resembling features observed in Charcot-Marie-Tooth disease.

Figure 2. (A) Sagittal T2-weighted MRI of the spine revealed lumbosacral lipomeningomyelocele with a tethered spinal cord. (B) Sagittal T1-weighted MRI of the spine without contrast showed an expansive hyperintense lesion, typically observed in lipomas.
References

