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Spinal cord atrophy in spinocerebellar ataxia type 1

Atrofia da medula espinhal na ataxia espinocerebelar do tipo 1

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A 50-year-old man presented with 9-years history of progressive ataxia. His father had undiagnosed ataxia. Examination showed ataxia and pyramidal signs. Brain MRI: olivopontocerebellar atrophy; spine MRI: global spinal cord atrophy (Figure). Genetic test confirmed spinocerebellar ataxia type-1 (SCA1).

SCA1 is characterized by cerebellar ataxia with variable degrees of ophthalmoplegia, pyramidal signs, and peripheral neuropathy[1]. Spinal cord atrophy was described in other SCA subtypes, but not in SCA1[2]. Prominent pyramidal signs and spinal cord atrophy in SCA1 may be explained by long tracts involvement, as in hereditary spastic paraplegia[3]. Spinal cord atrophy must be considered in neuroimaging features related to SCA1.

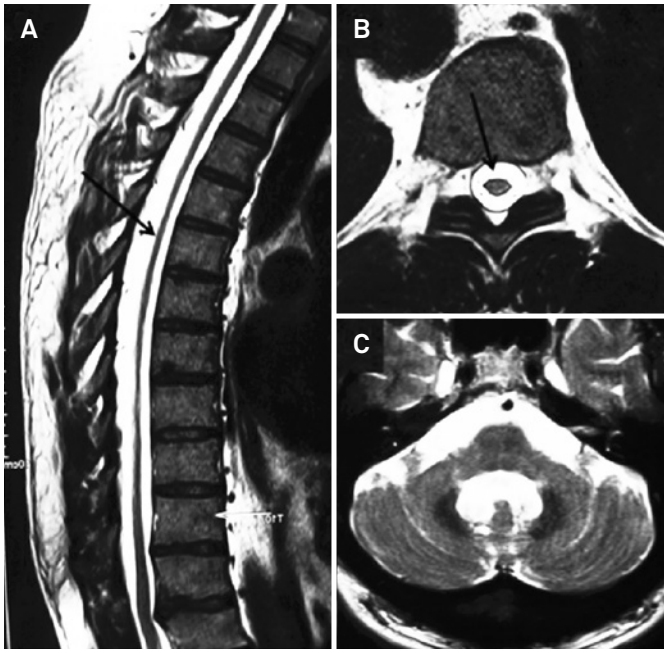

Figure. A. Sagittal T2-weighted spine MRI demonstrates global spinal cord atrophy; B. Axial T2-weighted spine MRI shows a marked reduced transverse diameter of the thoracic spinal cord; C. Axial T2-weighted brain MRI discloses moderate olivopontocerebellar atrophy.

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