

Spinocerebellar ataxia type 3 presenting simultaneously with motor neuron disease and cerebellar ataxia

Ataxia espinocerebelar tipo 3 apresentando-se simultaneamente com doença do neurônio motor e ataxia cerebelar

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A 66-year-old man reported an eight-year history of progressive ataxia and one-year of weakness, fasciculation and muscle atrophy (Figure 1). Examination disclosed

hypermetric saccades, diffuse fasciculations and absent deep tendon reflexes. Family history was remarkable for autosomal dominant ataxia (Figure 2). MRI showed cerebellar atrophy.

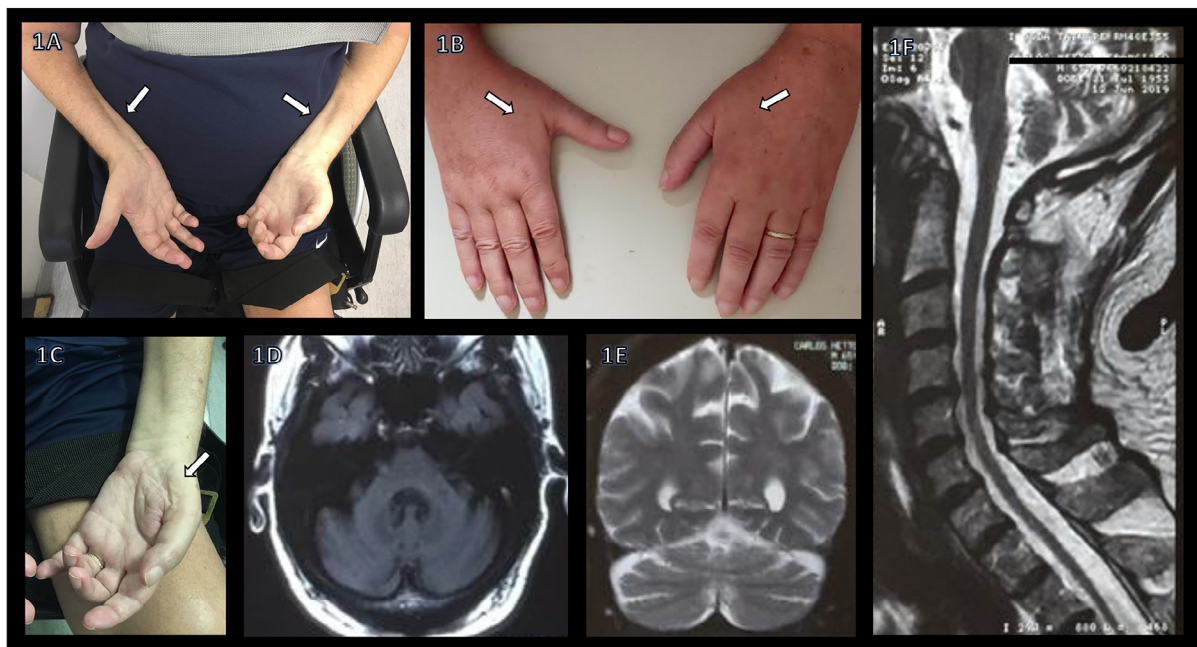


Figure 1. Index patient with spinocerebellar ataxia type 3 and motor neuron disease presenting with forearm flexor muscle atrophy (1A), split hand sign noted as a preferential wasting of first dorsal interosseus (1B) and thenar muscles abductor pollicis brevis (1C). Brain MRI showed global diffuse atrophy, more marked on middle and superior cerebellar peduncles, as on cerebellum (1D-E), and cervical spine MRI showed diffuse cervical spinal cord atrophy with no signal changes (1F).

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Ethical statement: Our Ethics Committee has approved this report. Patient provided informed consent for this publication.

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Genetic testing confirmed SCA3 (66 allelic expansion on ATXN3 gene). EMG disclosed diffuse denervation and confirmed motor neuron disease (MND).

Spinocerebellar ataxias (SCAs) may manifest as MND, particularly SCA2. Although SCA3 usually manifest as peripheral nerve involvement in a neuropathy pattern and

anterior horn degeneration, marked MND is uncommon^{1,2}. SCA3 with simultaneous sporadic amyotrophic lateral sclerosis should also be considered and may be related to accumulation of transactivation-responsive DNA-binding protein 43 (TDP-43) in the lower motor neurons³.

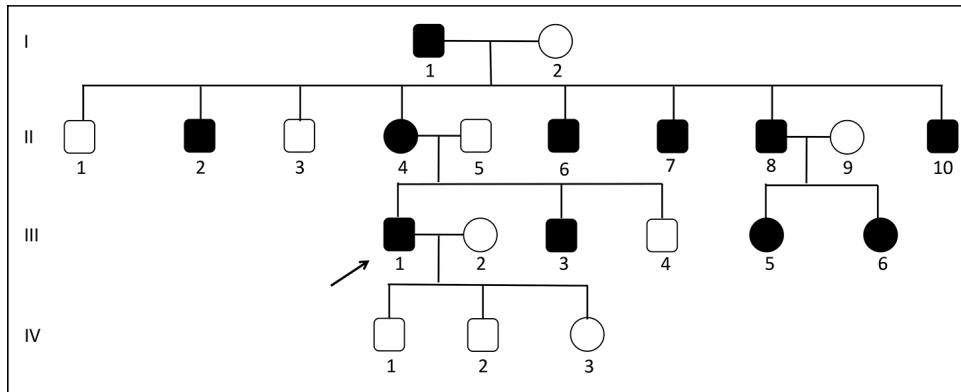


Figure 2. Heredogram. Family with spinocerebellar ataxia type 3 (SCA3).

REFERENCES

1. Pinto S, De Carvalho M. Machado-Joseph disease presenting as motor neuron disease. *Amyotroph Lateral Scler* 2008 Jun;9(3):188-91. <https://doi.org/10.1080/17482960701702603>
2. Moro A, Munhoz RP, Arruda WO, Raskin S, Moscovich M, Teive HA. Spinocerebellar ataxia type 3: subphenotypes in a cohort of brazilian patients. *Arq Neuropsiquiatr* 2014 Sep;72(9):659-62. <https://doi.org/10.1590/0004-282x20140129>
3. Toyoshima Y, Takahashi H. TDP-43 pathology in polyglutamine diseases: with reference to amyotrophic lateral sclerosis. *Neuropathology*. 2014;34(1):77-82. <https://doi.org/10.1111/neup.12053>