

HEREDITARY NEURODEGENERATIVE DISORDERS AS A DIFFERENTIAL DIAGNOSIS OF PRIMARY PROGRESSIVE MULTIPLE SCLEROSIS: A CASE REPORT

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Place: Multiple Sclerosis Reference Center of Rio Grande do Sul, Hospital de Clínicas de Porto Alegre, Brazil. Case report: A 46-year-old white woman presented with a slowly progressive cerebellar syndrome (ataxia, vertigo, dysarthria, and upper limb tremor), with onset at the age of 39. She also evolved with dysphagia, tetraparesis, hypoesthesia in the four limbs, urinary incontinence, visual impairment and cognitive complaints. Throughout the course of the disease, there was no evidence of relapses. Also, there was no family history of ataxia or consanguinity. Four years after the onset, magnetic resonance imaging (MRI) of the brain demonstrated supra- and infratentorial white matter lesions compatible with demyelination, as well as brain and cerebellar atrophy. Somatosensory evoked potential demonstrated a small delay in the central conduction time, and electroneuromyography was normal. At that time, she was diagnosed with MS, and treatment with beta-interferon was started. Three years later, when she was first seen by the authors, she reported stabilization of the disease, as well as partial remission of the cerebellar symptoms. She underwent methylprednisolone pulse therapy, with no improvement. Investigation for SCAs II, III, VI and VII, as well as vasculitides and Wilson's disease, was negative. Due to the atypical MS evolution, the possibility of metachromatic leukodystrophy and Krabbe disease was also suspected, but arylsulfatase A, urinary sulfatides and galactocerebrosides were normal. Hence, PP-MS was the final diagnosis. Conclusions: SCAs, MLD and KD are some of the diseases which must be included in the differential diagnosis of PP-MS.