

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

ALESSANDRO FINKELSZTEJN; RUI D'AVILA, CLARISSA M BORBA, ANELISE DACAVATÁ SZORTIKA, ALBERTO BATISTA E FRANCINE ZIQUINATTI

Background: Primary progressive form of Multiple Sclerosis (PP-MS) has a challenging diagnosis, since the clinical picture may resemble that of other neurodegenerative diseases, including metachromatic leukodystrophy (MLD), Krabbe disease (KD) and spinocerebellar ataxias (SCAs). Objective: To report a case in which hereditary degenerative disorders have had to be ruled out before the diagnosis of PP-MS was made. 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 MLD and KD 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.