

Diagnostic pitfalls in atypical primary lateral sclerosis

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Background and aims: Primary lateral sclerosis (PLS) is a disorder of progressive upper motor neuron dysfunction, in the absence of clinical signs of lower motor neuron involvement or family history of hereditary spastic paraplegia. PLS is a rare disorder (1–4% of all patients with motor neuron disease). Patients experience stiffness, decreased balance and coordination, and mild weakness, and if the bulbar region is affected, difficulty speaking and swallowing. **Case description:** Our patient is a 47 year old male, without a history of illness, who in 2014 observes a decreased muscular force in his left leg. In 2015, he developed gait disturbances with frequent falls and a paresis in his left arm. At that point, an MRI was performed with normal results and the EMG showed enlarged motor unit potentials. In 2017, he was admitted in our hospital for further investigations. Clinically he presented: left spastic hemiparesis (2/5 MRC brachial and 3/5 MRC crural) with pyramidal signs. Basic laboratory studies, including serum chemistries, serum B12, and complete blood count were normal. Additional studies included testing for Lyme disease, human t-cell lymphocytotropic virus-1, paraneoplastic panel, HIV testing, polyglucosan body disease and CSF evaluation. Serum long-chain fatty acids were evaluated to exclude adrenomyeloneuropathy. The EMG didn't show new changes. A cerebral MRI with spectroscopy and Chemical-shift protocol was performed which showed: atrophy in the right motor cortex, hyperintensity within the corticospinal tract and the Cho/NAA was twice the normal value. **Conclusions:** The diagnosis of PLS was based on the clinical history and neurological exam suggesting insidious onset of slowly progressive upper motor neuron dysfunction in the absence of family history, diagnostic testing or signs suggesting another disorder. The particular aspect of this case is the fact that our patient presented spastic hemiplegia instead of paraplegia, which is the typical form of presentation.