DIAGNOSTIC PITFALLS IN ATYPICAL PRIMARY LATERAL SCLEROSIS

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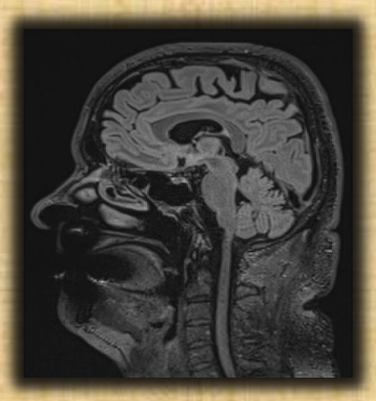
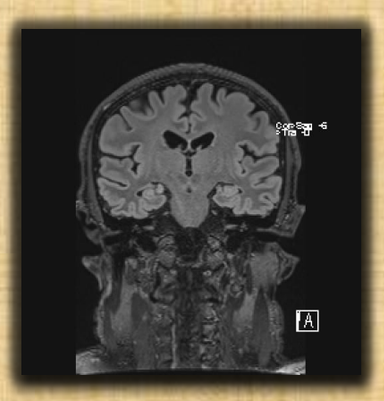
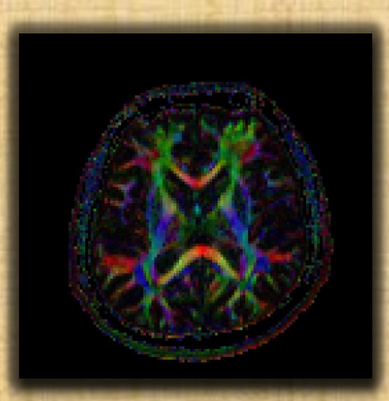
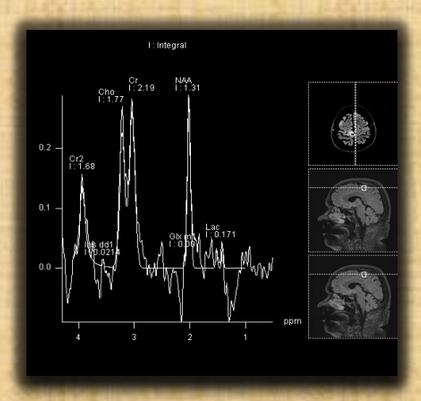
Background and aims

- PLS progressive - upper motor neuron dysfunction, in the absence of clinical signs of lower motor neuron involvement or family history suggestive of hereditary spastic paraplegia
- rare disorder ➪ approximately 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, and emotional liability.

Case description

- 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
- MRI was performed with normal results and the EMG showed enlarged motor unit potentials.
- In 2017, he was addmited 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-swift protocol 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 tipical form of presentation.

References:

1. Singer MA, Kojan S, Barohn RJ, et al. Primary Lateral Sclerosis: Clinical and Laboratory Features in 25 Patients. J Clin Neuromuscul Dis. 2005; 7(1):1–9. [PubMed: 19078775]
2. Pringle CE, Hudson AJ, Munoz DG, et al. Primary lateral sclerosis. Clinical features, neuropathology and diagnostic criteria. Brain : a journal of neurology. 1992; 115(Pt 2):495–520. [PubMed: 1606479]
3. Tartaglia MC, Rowe A, Findlater K, et al. Differentiation between primary lateral sclerosis and amyotrophic lateral sclerosis: examination of symptoms and signs at disease onset and during follow-up. Archives of neurology. 2007; 64(2):232–236. [PubMed: 17296839]
4. Brown WF, Ebers GC, Hudson AJ, et al. Motor-evoked responses in primary lateral sclerosis. Muscle & nerve. 1992; 15(5):626–629. [PubMed: 1584255]
5. Kwan JY, Meoded A, Danielian LE, et al. Structural imaging differences and longitudinal changes in primary lateral sclerosis and amyotrophic lateral sclerosis. Neuroimage (Amst). 2012; 2:151– 160.
6. Muller HP, Unrath A, Huppertz HJ, et al. Neuroanatomical patterns of cerebral white matter involvement in different motor neuron diseases as studied by diffusion tensor imaging analysis. Amyotrophic lateral sclerosis : official publication of the World Federation of Neurology Research Group on Motor Neuron Diseases. 2012; 13(3):254–264.
7. Gordon PH, Cheng B, Katz IB, et al. Clinical features that distinguish PLS, upper motor neurondominant ALS, typical ALS. Neurology. 2009; 72(22):1948–1952. [PubMed: 19487653]