A case of anti-IgLON5 disease with a complex presentation mimicking other movement disorders

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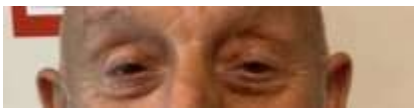
Introduction Anti-IgLON5 disease is an uncommon, recently characterized autoimmune neurological syndrome, with tremendous heterogeneity in clinical manifestations, although movement disorders often occur. As a potentially treatable disorder, the diagnosis should be timely.

Objective To report a case of anti-IgLON5 disease, providing valuable tips for differential diagnosis with other movement disorders.

Case report We observed a 72-year-old man with a ten-year history that started with anxiety-depression syndrome and successive mild cognitive impairment (MCI). As the main movement disorder, he progressively developed gait impairment with recurrent falls. In parallel, he complained of general slowness and weakness, occasional pain, and cramps. Lastly, oculomotor dysfunction, RBD-like sleep disturbances, and dysphagia emerged. Neurological examination showed impaired tandem gait with mild ataxic features, bulbar signs (dysarthria and dysphagia), severe vertical and horizontal gaze limitation, slow repetitive movements in the right upper limb, and lower limb fasciculations. Brain MRI and EEG were normal. Nerve conduction studies and electromyography revealed sensory axonal neuropathy and fasciculations. Stable increased serum CPK levels (600-800 UI/L) resulted, while other blood tests were normal. Polysomnography demonstrated RBD and severe chronic obstructive sleep apnea. CSF analysis showed increased proteins and CSF/serum albumin ratio; amyloid-beta and tau peptide levels were normal. A panel of autoantibodies was assessed in the serum and CSF, including anti-CASPR2, anti-VGKC, anti-LGI-1, anti-IgLON5, providing positivity for anti-IgLON5.

A. Impaired tandem gait with mild ataxic features



B. Vertical and horizontal gaze limitation and mild ptosis

Conclusions Anti-IgLON5 disease may have a very heterogeneous presentation. [1, 3]. In this case, we noticed potential overlap with other neurodegenerative movement disorders (e.g., PSP, synucleinopathies, multisystem degenerative disorders) that should increase awareness about such a treatable disorder. The occurrence of a complex motor syndrome associated with peculiarities in sleep disturbances and ophthalmoparesis and the involvement of PNS might solicit appropriate investigations to identify patients suitable for etiological treatments early.

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