Fatigable ptosis as an initial presentation of adult-onset Leigh syndrome

A 20-year-old man presented with bilateral fatigable ptosis for 1 month. On examination, there was bilateral incomplete ptosis, which deteriorated during upward gaze and improved at rest (figure, A and B). Tests for myasthenia gravis were all negative. Brain MRI showed symmetric hyperintensities at periaqueductal gray matter on T2- and diffusion-weighted images (figure, C). CSF lactic acid was elevated. Mitochondrial genome test demonstrated a homoplasmic T9176C mutation in the MT-APT6A gene, known as pathogenic mutation of Leigh syndrome.1 In our patient, fatigable ptosis may be ascribed to the dysfunction at centrally located synapse between the nuclear complex of the third nerve and supranuclear pathways.2

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