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 \textit{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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