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

Figure Photographs and brain MRI of the patient

The photographs show bilateral fatigable ptosis (A), which improves at rest (B). (C) Brain MRI discloses ill-defined and symmetric hyperintensities at periaqueductal gray matter on T2- and diffusion-weighted images.

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

Eun Hye Oh, MD, Song-Hwa Chae, MD, Jae-Wook Cho, MD, Seung Kug Baik, MD, Seo-Young Choi, MD, Kwang-Dong Choi, MD, Jae-Hwan Choi, MD

From the Departments of Neurology (E.H.O., S.-H.C., J.-W.C., J.-H.C.) and Radiology (S.K.B.), Research Institute for Convergence of Biomedical Science and Technology, Pusan National University School of Medicine, Pusan National University Yangsan Hospital; and Department of Neurology (S.-Y.C., K.-D.C.), Pusan National University School of Medicine and Biomedical Research Institute, Pusan National University Hospital, Busan, South Korea.

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Correspondence to Dr. J.-H. Choi: rachelbolan@hanmail.net

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