[PICTURES IN CLINICAL MEDICINE]

Adult-Onset Leigh Syndrome due to an m.13513G>A Mutation

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Picture 1.

A 32-year-old man was admitted to our hospital with a 6-month history of progressive ptosis, eye movement disturbance, dysuria, and dyschezia. He had hearing loss and renal failure from 23 years of age. He presented with bilateral adduction weakness and gaze nystagmus. His pupils were iso-

coric and reacted sluggishly to light. Laboratory tests revealed high lactate and pyruvate levels in the cerebrospinal fluid. Brain magnetic resonance imaging revealed high-intensity signals in the midbrain and hypothalamus on fluid-attenuated inversion recovery and an apparent diffusion co-

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Picture 2.

Picture 3.

efficient map. The central lesion showed low-intensity signals on T1-weighted imaging, and the peripheral lesion showed high-intensity signals on diffusion-weighted imaging (Picture 1, 2, arrows). Magnetic resonance spectroscopy re-

vealed a lactate peak in the lesion (Picture 3, arrow). A genetic analysis revealed an m.13,513 G>A mutation in the mitochondrial DNA, and he was diagnosed with Leigh syndrome. Adult-onset Leigh syndrome rarely presents with basal ganglia lesions, unlike childhood Leigh syndrome (1).

The authors state that they have no Conflict of Interest (COI).

Reference

1. Rahaman S, Blok RB, Dahl HH, et al. Leigh syndrome: clinical features and biochemical and DNA abnormalities. *Ann Neurol* **39**: 343-351, 1996.

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