

[PICTURES IN CLINICAL MEDICINE]

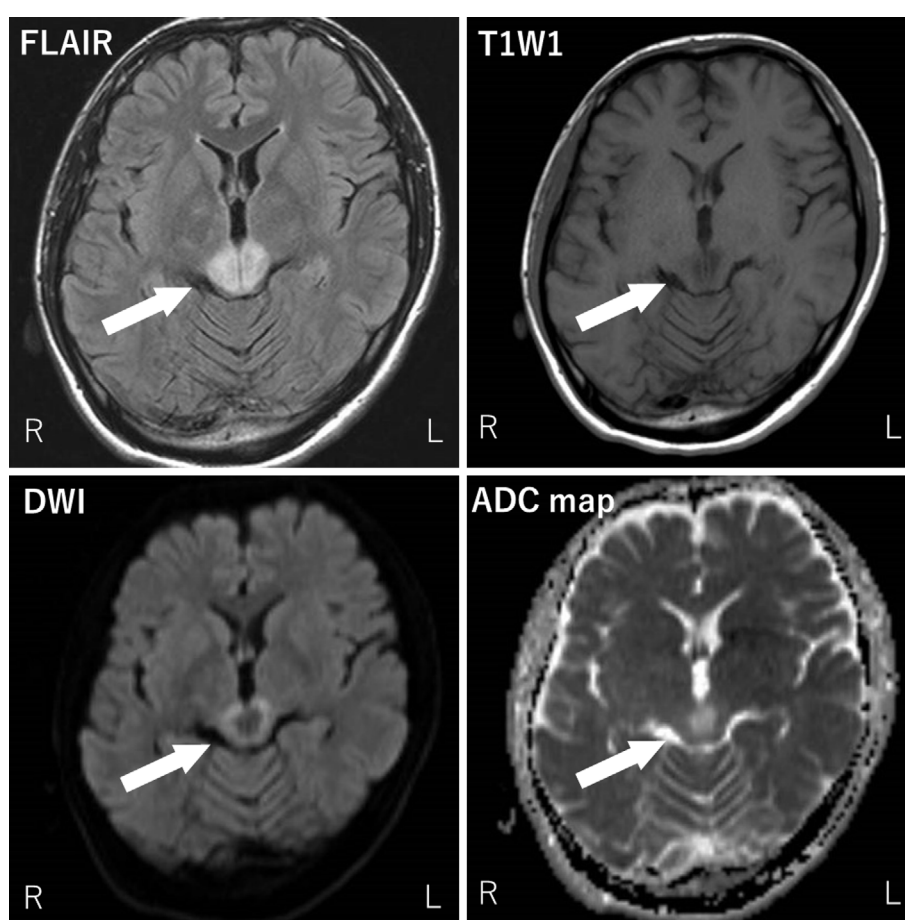
Adult-Onset Leigh Syndrome Due to an m.13513G>A Mutation

Hiroaki Hirosawa¹, Takamasa Nukui¹, Kyo Noguchi² and Yuji Nakatsuji¹

Key words: Leigh syndrome, mitochondrial disease, renal failure

(Intern Med 61: 1627-1628, 2022)

(DOI: 10.2169/internalmedicine.8445-21)



Picture 1.

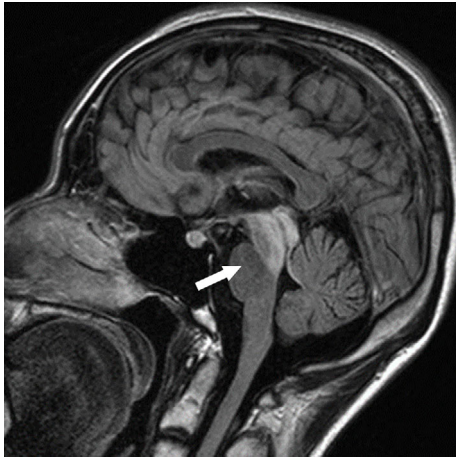
A 32-year-old man was admitted to our hospital with a 6-month history of progressive ptosis, eye movement disturbance, dysuria, and dyschezia. He had hearing loss and renal failure from 23 years of age. He presented with bilateral adduction weakness and gaze nystagmus. His pupils were iso-

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-

¹Department of Neurology, Faculty of Medicine, University of Toyama, Japan and ²Department of Radiology, Faculty of Medicine, University of Toyama, Japan

Received: August 7, 2021; Accepted: September 2, 2021; Advance Publication by J-STAGE: October 19, 2021

Correspondence to Dr. Yuji Nakatsuji, nakatsuj@med.u-toyama.ac.jp



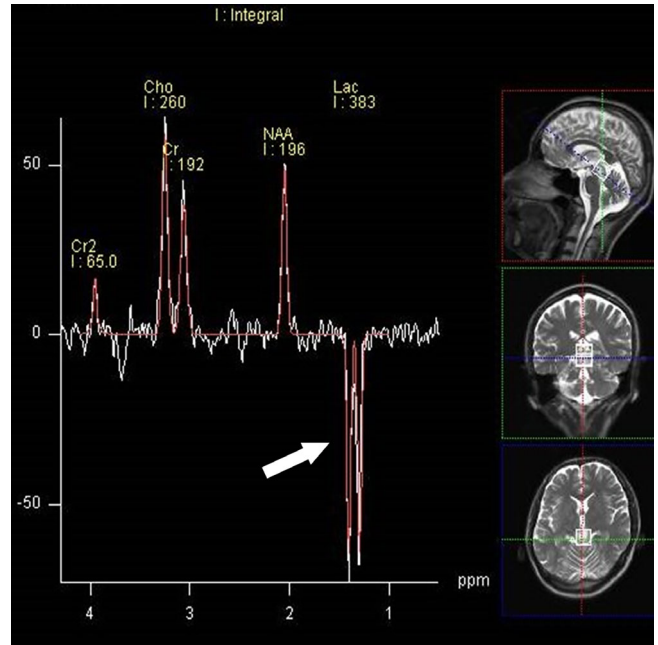
Picture 2.

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 revealed a lactate peak in the lesion (Picture 3, arrow). A genetic analysis revealed an m.13513G>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



Picture 3.

features and biochemical and DNA abnormalities. *Ann Neurol* **39**: 343-351, 1996.

The Internal Medicine is an Open Access journal distributed under the Creative Commons Attribution-NonCommercial-NoDerivatives 4.0 International License. To view the details of this license, please visit (<https://creativecommons.org/licenses/by-nc-nd/4.0/>).