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Magnetic Resonance Imaging and Spectroscopy of the Brain in MELAS Syndrome

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A 40-year-old woman presented to the neurology department with headache that had continued for 5 days and the sudden onset neurological disorders. The patient and her two teen daughters had no medical history of note, but her mother had a history of repeated stroke-like episodes and had died at age 63. The neurological findings of the patient were right homonymous hemianopia, agnosia, acalculia, dyslexia, and dysgraphia. Laboratory tests revealed no abnormalities including in the serum lactate level (15.2 mg/dL; reference value, 3.7-16.3 mg/dL) or the serum pyruvate level (0.58 mg/dL; reference value, 0.3-0.9 mg/dL). Spinal fluid examination revealed no white or red blood cells, with a protein level of 23 mg/dL, and a glucose level of 58 mg/dL. Electrocardiogram revealed normal sinus rhythm and nonspecific ST-T changes. The patient then underwent brain magnetic resonance imaging (MRI) (Fig. 1A-C). Fluid-attenuated inversion recovery imaging (Fig. 1A) and diffusion-weighted imaging (DWI) (Fig. 1B) revealed a high signal intensity lesion in the left occipital lobe, which was isointense on the apparent diffusion coefficient (ADC) map (Fig. 1C). Vasogenic edema due to the breakdown of the blood-brain barrier was suspected from the MRI findings. Additionally, proton MR spectroscopy (MRS) (Fig. 1D) showed an elevated lactate peak and a decreased N-acetyl aspartate/creatine ratio in the lesion, which suggested increased anaerobic glycolysis. Genetic analysis of the mitochondrial DNA from a blood sample revealed a m.3243A > G point mutation most commonly associated with mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS). After high-dose corticosteroid therapy, the patient received coenzyme Q10, taurine, thiamine, vitamin C, and antiepileptic drugs. The symptoms of the patient gradually improved, and no new symptoms of MELAS appeared during the 24 months of follow-up.

MELAS is a multisystemic mitochondrial disorder inherited matrilineally. Although early symptoms of the disease are nonspecific, brain MRI demonstrated stroke-like lesions in approximately 90% of the patients with MELAS syndrome.[1] DWI and ADC mapping are useful to distinguish MELAS from acute ischemic stroke. In the ischemic lesions of the brain, the signal on the ADC map is decreased by cytotoxic edema, whereas vasogenic edema in MELAS lesions generally does not reduce the signal on the

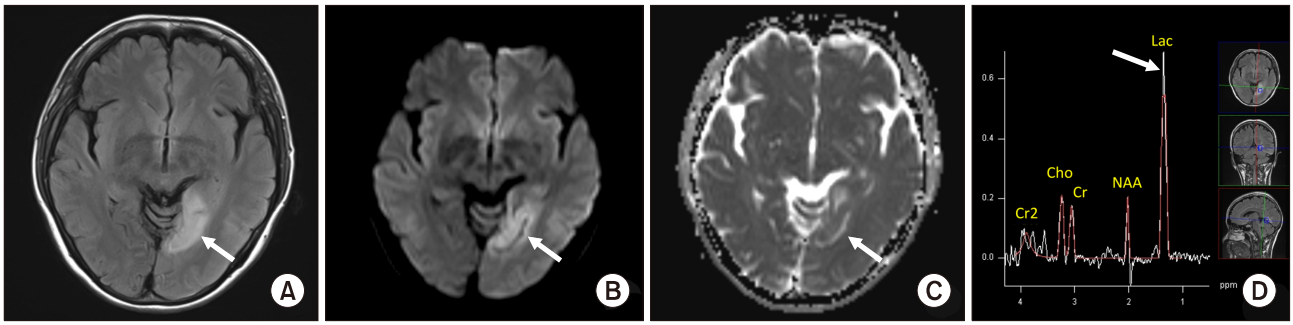

FIG. 1. Magnetic Resonance imaging revealed a left occipital lesion (small arrows), which was hyperintense on fluid-attenuated inversion recovery imaging (A) and diffusion-weighted imaging (B) and isointense on apparent diffusion coefficient map (C). Proton magnetic resonance spectroscopy (D) revealed a markedly elevated lactate peak at 1.3 ppm (large arrow) and a decreased N-acetyl aspartate/creatine ratio in the left occipital lesion.

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ADC map.[2] Furthermore, MRS shows markedly elevated lactate peaks in MELAS lesions.[2,3] In molecular tests, the m.3243A > G point mutation is present in more than 80% of MELAS cases.[4] The treatments aim at supporting mitochondrial energy production with L-arginine, coenzyme Q10, taurine, and multiple vitamins, but the overall prognosis is poor.[5]

CONFLICT OF INTEREST STATEMENT

None declared.

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