IMAGES IN METABOLIC MEDICINE

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Focal cerebellar infarction as an initial sign of mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes

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Abstract

Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) is a type of mitochondrial disorder and stroke-like lesions are observed prominently in the brain magnetic resonance imaging. Those stroke-like lesions of MELAS patients are usually located in the posterior quadrants and do not correspond to typical vascular territories. This case illustrates that focal cerebellar infarction can be the sole initial sign of MELAS.

KEYWORDS

ataxia, cerebellum, dysarthria, MELAS, mitochondria

Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS) is a type of mitochondrial disorder often presented with symptoms such as seizures, headaches, diabetes mellitus, muscle weakness, sensorineural hearing loss, myopathy, and sudden neurological deficits.¹ Among several mutations known to be associated with MELAS, the m.3243A>G mutation in the mitochondrial transfer RNA Leu^(UUR) gene is one of the most common type. Stroke-like lesions are observed prominently in the brain magnetic resonance imaging (MRI) of MELAS patients.²

A 5-year-old boy with no previous medical history presented with dysarthria and mild right-sided ataxia. A focal infarction in the right posterior cerebellum was noted in the brain MRI (Figure 1). His plasma lactate level (7.2 mmol/L) indicated lactic acidosis. Evaluation workup for the mitochondrial disease was conducted and m.3243A>G mutation was identified. After time, dysarthria and ataxia improved but he did not completely recover. Focal seizures and stroke-like episodes developed 3 years after while dys-arthria and ataxia persisted. The second brain MRI revealed new cortical stroke-like lesions and a cerebellar lesion (Figure 2).

This is the first report describing focal cerebellar infarction as a presenting sign of MELAS. The patient of this case showed dysarthria and ataxia with focal cerebellar infarction shown on MRI. The cerebellar lesion on MRI was very small considering his symptoms, but together with clinical circumstances we concluded that this is the responsible lesion for dysarthria and ataxia of the patient. One possible explanation is that actual cerebellar involvement is larger than the lesion grossly manifested on MRI. Those stroke-like lesions of MELAS patients are usually located in the

Ara Ko and Sang-Jun Lee contributed equally to this study.



FIGURE 1 Initial brain MRI. A, B, Normal cerebral appearance on T2 images. C, D, Diffusion restriction focus in the right cerebellum on diffusion-weighted images suggesting acute infarction (arrow)



FIGURE 2 Follow-up brain MRI. A, B, Multiple scattered foci of cortical hyperintensities at both temporal, parietal, and occipital lobes on T2 images, suggesting multifocal cortical infarctions (arrows). C, Subtle diffusion restriction foci at right cerebellar hemispheric inferior portion on diffusion-weighted images (arrows). D, Increased signal lesion in fluid-attenuated inversion-recovery images of the right inferior cerebellum (arrow)

posterior quadrants and do not correspond to typical vascular territories.¹ However, there are some patients with mitochondrial encephalopathies associated with the m.3243A>G mutations who have deep gray matter changes on MRI without stroke-like lesions.² Also, there are various reported cases showing cerebellar involvement in MELAS patients concomitantly with other cortical involvement or appearing during disease course.^{3–5} This case further illustrates that focal cerebellar infarction can be the sole initial sign of MELAS.

All procedures performed in studies involving human participant were in accordance with the ethical standards of the institutional and national research committee. Informed consent was obtained from the patient included in the study.

CONFLICTS OF INTEREST

None

Ethical approval: All procedures performed in studies involving human participant were in accordance with the ethical standards of the institutional and national research committee.

Informed consent: Informed consent was obtained from the patient included in the study.

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