

Teaching NeuroImages: A 48-year-old woman with MELAS

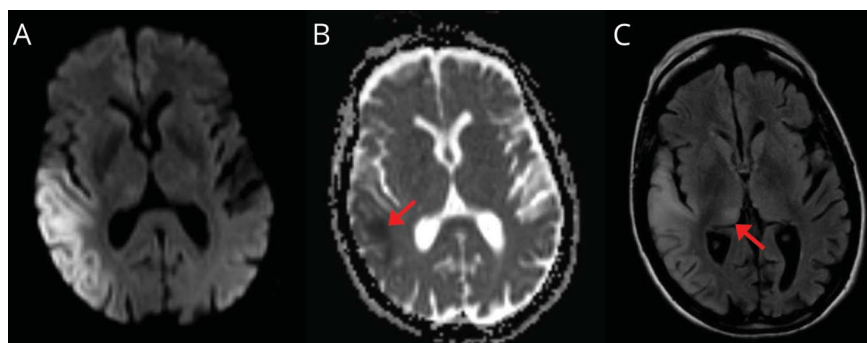
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Correspondence

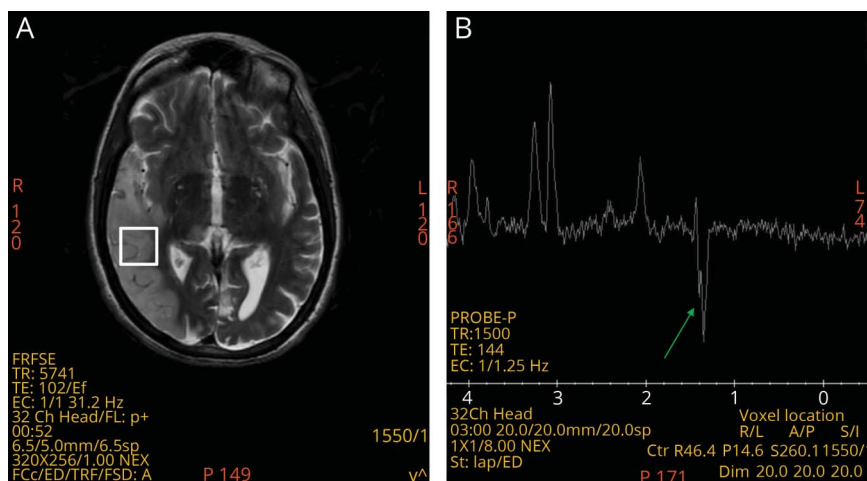
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Figure 1 Brain MRI demonstrates a cortically based region of slow diffusion (A) in the right parietal and right temporal lobes with a small area of darkness on apparent diffusion coefficient (B, arrow), which did not correlate fully with the extent of diffusion restriction



There was corresponding T2 fluid-attenuated inversion recovery (FLAIR) hyperintensity along with mild effacement of the associated sulci (C). In addition, there was abnormal T2 FLAIR signal in the right medial thalamus (C, arrow).

Figure 2 Magnetic resonance spectroscopy (A) within the region of slow diffusion (B) demonstrates an inverted doublet at 1.3 ppm (arrow) consistent with a lactate peak and anaerobic metabolism



A 48-year-old woman with sensorineural hearing loss and diabetes presented with seizures and headache. She was thin and short-statured. Examination revealed encephalopathy, left homonymous hemianopia, and hemineglect. MRI showed slowed diffusion in right hemisphere, which did not follow a clear vascular distribution and without apparent diffusion coefficient correlate, inconsistent with infarction (figure 1).¹ Magnetic resonance spectroscopy revealed

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inversion of lipid lactate peak (figure 2). She had elevated pyruvate but normal lactate. EMG demonstrated myopathy without denervation. Genetic testing showed m.3243A>G pathogenic variant of the *MT-TL1* gene. This mutation is found in more than 80% of patients with mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes and always coexists with wild-type DNA.²

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Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Role	Contribution
Jonah P. Zuflacht, MD	Department of Neurology, Beth Israel Deaconess Medical Center	Author	Designed and conceptualized case report, drafted the manuscript, cared for the patient
Antonia N. Pusso, MD	Department of Neurology, Beth Israel Deaconess Medical Center	Author	Designed and conceptualized case report, provided key edits to the manuscript, cared for the patient

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