Teaching NeuroImages: Imaging phenotype of myoclonic epilepsy with ragged-red fibers

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Figure Imaging and magnetic resonance spectroscopy

Sagittal fluid-attenuated inversion recovery (FLAIR) image (A) shows abnormal hyperintense signal in the mesencephalon, posterior aspect of pons, and medulla oblongata (arrows). Axial FLAIR (B) shows hyperintensity extending superiorly to the medial aspect of thalami (curved arrows). Axial T2 (C) depicts extension of signal abnormality inferiorly to posterolateral aspect of medulla oblongata (circle). Axial diffusion-weighted imaging sequence (D) demonstrates mild restricted diffusion in the tegmentum of midbrain (arrow). Axial T1 postcontrast image (E) depicts enhancement in the periaqueductal gray matter (circle). Spectroscopy long echo time (144) (F) shows elevated lactate at 1.3 ppm (star) and decrease in *N*-acetylaspartate at 2.0 ppm (curved arrow).

A 17-year-old girl presented with subacute decline in ambulation, ataxia, generalized weakness, dysphagia, and asymmetric hearing loss. MRI findings include abnormal signal in medial thalami, mesencephalon, posterior pons, and medulla oblongata (figure). Magnetic resonance spectroscopy showed elevated lactate and decrease in *N*-acetylaspartate (figure, F). DNA isolated from muscle biopsy showed A8344G mutation of mitochondrial DNA (tRNA [Lys] gene), associated with 80% of patients with myoclonic epilepsy with ragged-red fibers (MERRF). MERRF is a rare mitochondrial disorder with variable onset and clinical presentation. Neuroradiologic findings of MERRF are reported rarely, with brainstem and cerebellar degeneration being the main feature of MERRF.¹

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Name	Location	Contribution
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Appendix (continued)

Name	Location	Contribution
Ashrith R. Kandula	Nemours A.I. DuPont Hospital for Children, Wilmington, DE	Concept and design, revision
Kathleen Schenker, MD	Nemours A.I. DuPont Hospital for Children, Wilmington, DE	Concept and design, supervision

Reference

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