

Teaching NeuroImages: MRI findings in an infant with cavitating leukoencephalopathy

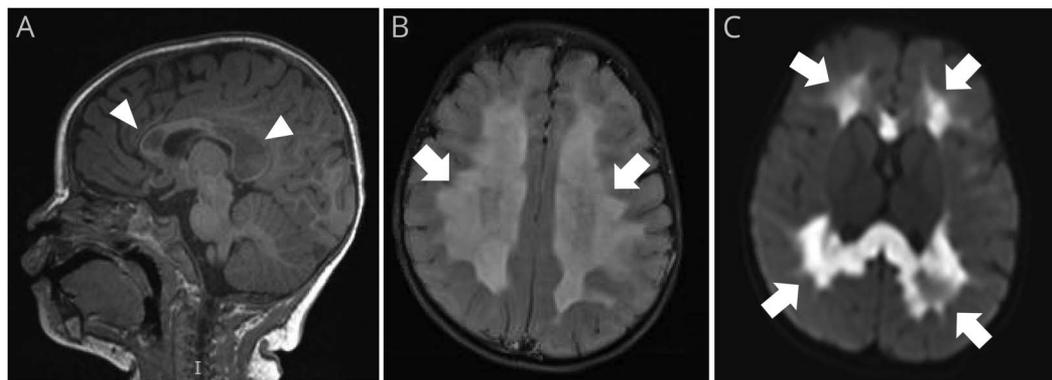
Annie Hong, MD, Peter Assaad, MD, and Shefali Karkare, MD

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Correspondence

Dr. Hong
ahong2@northwell.edu

Figure MRI brain



Sagittal T1-weighted image shows patchy leukoencephalopathy with cavitation affecting the corpus callosum and centrum semiovale, with relative sparing of underlying U-fibers and gray matter (A; arrowheads). Bilateral, symmetric T2 hyperintense white matter lesions on axial T2 fluid-attenuated inversion recovery did not enhance with contrast (B; arrows). Axial diffusion-weighted images demonstrate diffusion restriction (C; arrows).

An 11-month-old previously healthy girl presented with irritability, weakness, and hypotonia following a febrile illness. Brain MRI revealed white matter signal abnormalities with diffusion restriction and cavitation of the corpus callosum (figure). Combined mitochondrial gene panel confirmed cavitating leukoencephalopathy secondary to a pathogenic variant, p.R386C, in the *NDUFV1* gene, which encodes complex I.¹ Cavitating leukoencephalopathy is a neurodegenerative disorder associated with genetic mutations of the mitochondrial complex proteins, characterized by acute neurologic deficits and progressive or intermittent clinical deterioration. Patients may have variable response to megavitamins, steroids, or IV immunoglobulin.² She was treated with megavitamins and regained psychomotor milestones with no further events.

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Appendix Authors

| Name | Location | Role | Contribution |
|----------------------------|-----------------------------------|----------------------|---|
| Annie Hong, MD | Cohen Children's Medical Center | Corresponding author | Designed and conceptualized study, analyzed the data, drafted the manuscript for intellectual content |
| Peter Assaad, MD | Long Island Jewish Medical Center | Author | Interpreted the data, revised the manuscript for intellectual content |
| Shefali Karkare, MD | Cohen Children's Medical Center | Author | Interpreted the data, revised the manuscript for intellectual content |

From the Division of Child Neurology, Department of Pediatrics (A.H., S.K.), and Department of Radiology (P.A.), Cohen Children's Medical Center, New Hyde Park, NY. Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

Disclosure

The authors report no disclosures relevant to the manuscript.
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