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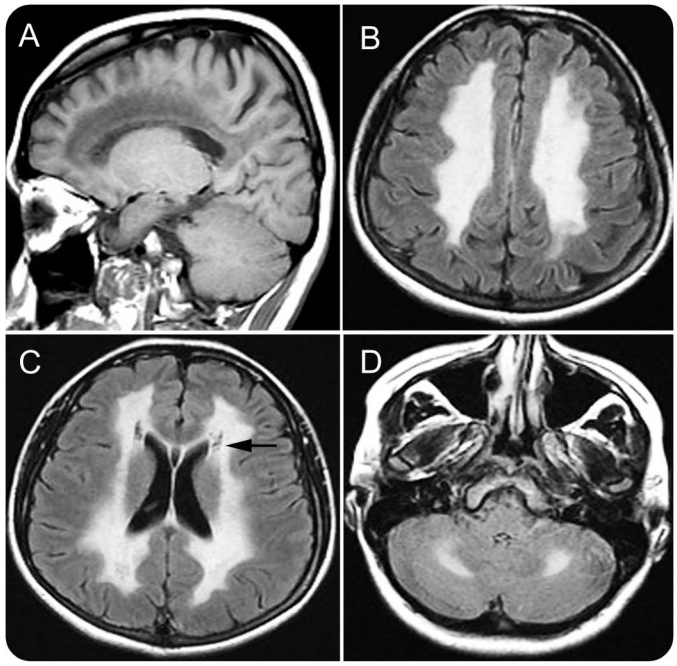
Teaching NeuroImages: Vanishing white matter ovarioleukodystrophy

Shibani S. Mukerji, MD, PhD
Florian S. Eichler, MD

Correspondence to
Dr. Mukerji:
smukerji@partners.org

Figure Brain imaging findings



Hypointense signal is observed on sagittal T1-weighted MRI (A). Axial T2/fluid-attenuated inversion recovery sequences shows extensive, symmetric, confluent high signal intensity in the frontoparietal (B), periventricular (C), and cerebellar regions (D). Periventricular low signal intensity similar to CSF signal behavior is suggestive of cystic degeneration (arrow).

A 21-year-old Pakistani woman with a history of 2 seizures at 20 months of age and consanguineous parents was diagnosed with premature ovarian failure and hyperprolactinemia. Her neurologic examination was normal. Brain imaging showed confluent white matter T2/fluid-attenuated inversion recovery hyperintensities with periventricular cavitations (figure). Genetic testing showed homozygosity for a p.Lys273Arg missense mutation in the *EIF2B2* gene (NM_014239.3: c.818A>G), which had previously been reported as disease-causing,[1] and confirmed vanishing white matter (VWM) ovarioleukodystrophy. At 2-year follow-up, she remains neurologically asymptomatic with stable imaging. There is phenotypic variation in VWM disease and neurologic deterioration is associated with stressors (head trauma, fever).[1,2]

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AUTHOR CONTRIBUTIONS

Dr. Mukerji conceptualized and designed the study, interpreted the data, and drafted the manuscript. Dr. Eichler interpreted the data and revised the manuscript for intellectual content.

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DISCLOSURE

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

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From the Department of Neurology (S.S.M., F.S.E.), Massachusetts General Hospital and Harvard Medical School, Boston; Center for Rare Neurological Diseases (F.S.E.), Massachusetts General Hospital, Boston.

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Shibani S. Mukerji and Florian S. Eichler

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