Teaching NeuroImages: Adult-onset vanishing white matter disease

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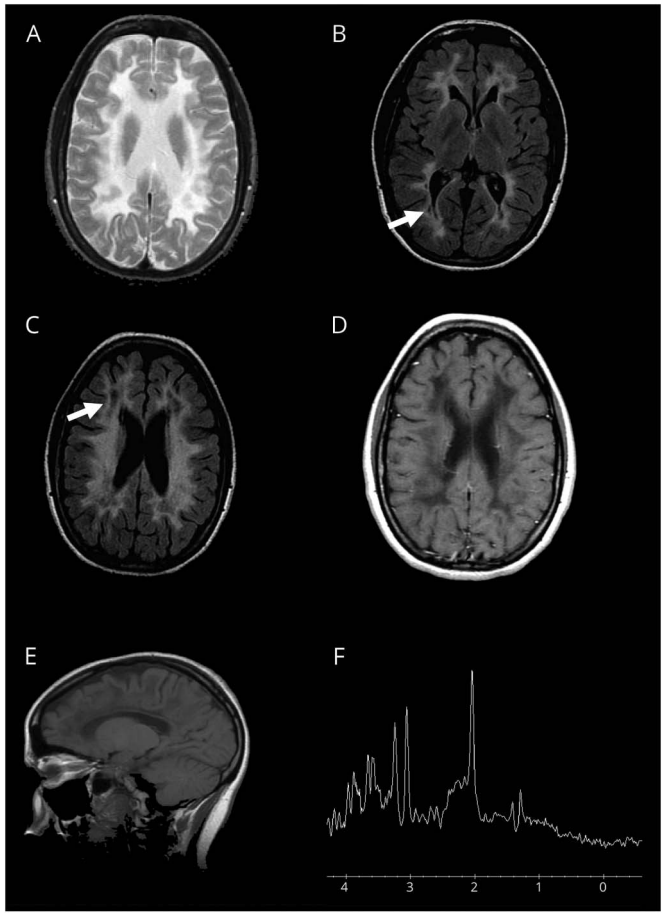
Figure Neuroimaging findings in vanishing white matter disease



(A) MRI brain axial T2: extensive leukoencephalopathy. (B, C) MRI brain axial T2 fluid-attenuated inversion recovery: diffuse abnormality of the cerebral white matter with evidence of partial cystic degeneration (arrows). (D) MRI brain axial T1+ gadolinium: no contrast enhancement. (E) MRI brain sagittal T1: abnormal white matter signal and thinning of the corpus callosum. (F) Magnetic resonance spectroscopy: mildly elevated lactate peak.

An 18-year-old woman with premature ovarian failure presented with focal motor seizures with secondary generalization and headache 2 weeks after a viral infection. She also had a recent psychological stress background. Examination only revealed generalized hyperreflexia. Neuroimaging studies showed bilateral confluent leukoencephalopathy and periventricular cystic degeneration (figure). Genetic testing showed homozygosity for a p.Arg113His missense mutation in the *EIF2B5* gene, confirming the diagnosis of VWM. Phenotypic

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variation of VWM is extremely wide, with episodes of sudden major neurologic degeneration triggered by febrile infections, minor head trauma, or stressful events.[1] MRI findings are usually diagnostic and associated with mutations in *EIF2B1-5* genes.[2]

Author contributions

Rocío-Nur Villar-Quiles: acquisition and interpretation of data, manuscript preparation. Celia Delgado-Suárez: acquisition and interpretation of data, manuscript preparation. Manuela Jorquera-Moya: acquisition and interpretation of data, critical revision of manuscript for intellectual content. Javier Arpa-Gutiérrez: critical revision of manuscript for intellectual content. Gloria Ortega-Suero: critical revision of manuscript for intellectual content.

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Disclosure

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