Teaching NeuroImages: Neuroimaging in Adult-Onset Alexander Disease

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Figure MRI of the Cervical Spine and Brain

(A) Sagittal T2-weighted imaging along the midline demonstrates atrophy of the cervical medullary junction (arrow). (B) Axial T2 FLAIR image reveals hyperintense T2 FLAIR signal in the bilateral dentate nuclei (arrow). Medullary atrophy also noted with abnormal signal (arrowhead).

A 35-year-old woman presented with gait disturbance, generalized stiffness, difficulty swallowing, and bladder incontinence for the past 3 years. Neurologic evaluation revealed brisk deep tendon reflexes in all 4 limbs and bilateral Hoffman signs. Her family history was significant for a brother diagnosed through genetic testing with Adult-onset Alexander disease (AOAD) at age 28. MRI head and cervical spine (figure A and B) showed classic signs of AOAD. Genetic testing confirmed glial fibrillary acidic protein gene mutation R239G consistent with AOAD. Although infantile Alexander Disease is more common, these unique MRI findings should raise suspicion of AOAD and prompt consideration for genetic testing.^{1,2}

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Disclosure

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

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Name	Location	Contribution
Neethu Gopal, MBBS	Department of Neurology, Mayo Clinic, Florida	Execution, writing of the first draft, and review
Vivek Gupta, MD	Department of Radiology, Mayo Clinic, Florida	Execution, review, and critique
Lindsay N. Williams, MD	Department of Neurology, Mayo Clinic, Florida	Execution, review, and critique
Sukhwinder J.S. Sandhu, MD	Department of Radiology, Mayo Clinic, Florida	MRI description, execution, review, and critique

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