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 Hoffmann 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

**Study Funding**
No targeted funding reported.

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

Appendix Authors

<table>
<thead>
<tr>
<th>Name</th>
<th>Location</th>
<th>Contribution</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neethu Gopal, MBBS</td>
<td>Department of Neurology, Mayo Clinic, Florida</td>
<td>Execution, writing of the first draft, and review</td>
</tr>
<tr>
<td>Vivek Gupta, MD</td>
<td>Department of Radiology, Mayo Clinic, Florida</td>
<td>Execution, review, and critique</td>
</tr>
<tr>
<td>Lindsy N. Williams, MD</td>
<td>Department of Neurology, Mayo Clinic, Florida</td>
<td>Execution, review, and critique</td>
</tr>
<tr>
<td>Sukhwinder J.S. Sandhu, MD</td>
<td>Department of Radiology, Mayo Clinic, Florida</td>
<td>MRI description, execution, review, and critique</td>
</tr>
</tbody>
</table>
Teaching NeuroImages: Neuroimaging in Adult-Onset Alexander Disease
Neethu Gopal, Vivek Gupta, Lindsey N. Williams, et al.
Neurology 2021;96:e814-e815 Published Online before print September 11, 2020
DOI 10.1212/WNL.0000000000010803

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