Teaching NeuroImages: Dorsal Medullary Lesions in Juvenile-Onset Alexander Disease

Author(s):
John Sollee, BS¹, ²; Amy Waldman, MD, MSCE¹, ³

Corresponding Author:
John Sollee
solleejr@email.chop.edu

Affiliation Information for All Authors: ¹. Division of Neurology, Children's Hospital of Philadelphia, Philadelphia, PA; ². Warren Alpert Medical School of Brown University, Providence, RI; ³. Departments of Neurology and Pediatrics, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA

Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.
A 6-year-old boy presented with dysphagia, vomiting, and weight loss. Early developmental milestones were notable for mild gross motor and speech delay. Hypotonia was present on examination. A brain MRI revealed bilateral enhancing dorsal medullary lesions (Figure, contrast not shown). The differential diagnosis included a leukodystrophy or mitochondrial disease. Alexander disease was confirmed genetically (de novo variant in GFAP targeted testing: p.Arg-376-Gly). Typical features also include hyper-nasal speech with subsequent motor difficulties and autonomic dysfunction over time.\(^2\) GFAP sequencing should be considered in patients with unilateral or bilateral dorsal medullary lesions with localizing symptoms (e.g., vomiting, dysphagia).
Figure
Legend: Brain MRI in Alexander disease reveals distinctive hyperintense bilateral dorsal medullary lesions on (A) axial FLAIR and (B) coronal T2-weighted images in a heart-shaped appearance. (C) Additional diagnostic criteria (T2 hyperintense signal abnormality in the frontal white matter and basal ganglia) are present on axial FLAIR images.\(^1\)

Teaching Slides -- [http://links.lww.com/WNL/B454](http://links.lww.com/WNL/B454)

References
Teaching NeuroImages: Dorsal Medullary Lesions in Juvenile-Onset Alexander Disease
John Sollee and Amy Waldman
*Neurology* published online June 22, 2021
DOI 10.1212/WNL.0000000000012411

This information is current as of June 22, 2021

<table>
<thead>
<tr>
<th>Updated Information &amp; Services</th>
<th>including high resolution figures, can be found at: <a href="http://n.neurology.org/content/early/2021/06/22/WNL.0000000000012411.citation.full">http://n.neurology.org/content/early/2021/06/22/WNL.0000000000012411.citation.full</a></th>
</tr>
</thead>
</table>
| Subspecialty Collections      | This article, along with others on similar topics, appears in the following collection(s):  
                               | All Pediatric [http://n.neurology.org/cgi/collection/all_pediatric](http://n.neurology.org/cgi/collection/all_pediatric)  
                               | Leukodystrophies [http://n.neurology.org/cgi/collection/leukodystrophies](http://n.neurology.org/cgi/collection/leukodystrophies)  
                               | MRI [http://n.neurology.org/cgi/collection/mri](http://n.neurology.org/cgi/collection/mri) |
| Permissions & Licensing       | Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:  
                               | [http://www.neurology.org/about/about_the_journal#permissions](http://www.neurology.org/about/about_the_journal#permissions) |
| Reprints                      | Information about ordering reprints can be found online:  
                               | [http://n.neurology.org/subscribers/advertise](http://n.neurology.org/subscribers/advertise) |