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

John Sollee, BS, and Amy Waldman, MD, MSCE

Neurology® 2021;97:e2050-e2051. doi:10.1212/WNL.00000000000012411

Correspondence

Dr. Waldman
waldman@chop.edu

Figure Dorsal Medullary Lesions in Alexander Disease

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 [arrow heads] and basal ganglia [arrows]) are present on axial FLAIR images.¹

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. 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 hypernasal 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 and dysphagia).

Study Funding

The authors report no targeted funding.

Disclosures

J. Sollee reports no disclosures relevant to the manuscript. A. Waldman reports no disclosures relevant to the manuscript. Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

MORE ONLINE

Teaching slides

links.lww.com/WNL/B454

From the Division of Neurology (J.S., A.W.), Children's Hospital of Philadelphia, PA; Warren Alpert Medical School of Brown University (J.S.), Providence, RI; and Departments of Neurology and Pediatrics (A.W.), Perelman School of Medicine at the University of Pennsylvania, Philadelphia.

Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

Appendix Authors

Name	Location	Contribution
John Sollee, BS	Warren Alpert Medical School of Brown University, Providence, RI	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; and analysis or interpretation of data
Amy Waldman, MD, MSCE	Division of Neurology, Children's Hospital of Philadelphia, PA	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; and analysis or interpretation of data

References

1. Van der Knaap M, Naidu S, Breiter S. et al. Alexander disease: diagnosis with MR imaging. *Am J Neuroradiol*. 2001;22(3);541-552.
2. Srivastava S, Waldman A, Naidu S. Alexander disease. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, editors. *GeneReviews*. University of Washington, Seattle; 1993-2020. Updated November 12, 2020. <https://www.ncbi.nlm.nih.gov/books/NBK1172/>

Neurology®

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

John Sollee and Amy Waldman

Neurology 2021;97:e2050-e2051 Published Online before print June 22, 2021

DOI 10.1212/WNL.00000000000012411

This information is current as of June 22, 2021

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/97/20/e2050.full
References	This article cites 1 articles, 1 of which you can access for free at: http://n.neurology.org/content/97/20/e2050.full#ref-list-1
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 Leukodystrophies http://n.neurology.org/cgi/collection/leukodystrophies 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
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2021 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.