

Neurology Publish Ahead of Print
DOI: 10.1212/WNL.0000000000012411

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

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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.

Contributions:

John Sollee: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data

Amy Waldman: Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data

Number of characters in title: 60

Abstract Word count: 97

Word count of main text: 97

References: 2

Figures: 1

Tables: 0

Neuroimage Legend Count: 47

Supplemental: Tracked changes for revision.

Search Terms: [120] MRI, [155] Leukodystrophies, [227] All Pediatric

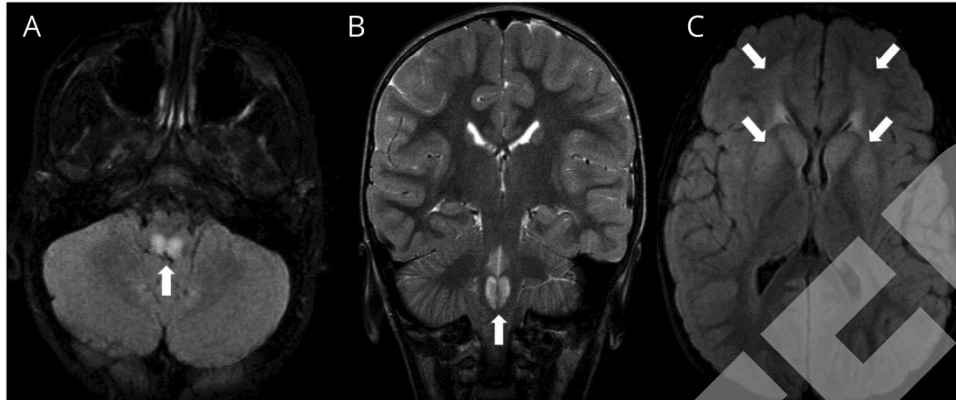
Study Funding: The authors report no targeted funding

Disclosures: J. Sollee reports no disclosures relevant to the manuscript. A. Waldman reports no disclosures relevant to the manuscript.

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>

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. 2002 Nov 15 [updated 2020 Nov 12]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. *GeneReviews*[®] [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2020.

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Neurology published online June 22, 2021

DOI 10.1212/WNL.0000000000012411

This information is current as of June 22, 2021

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