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

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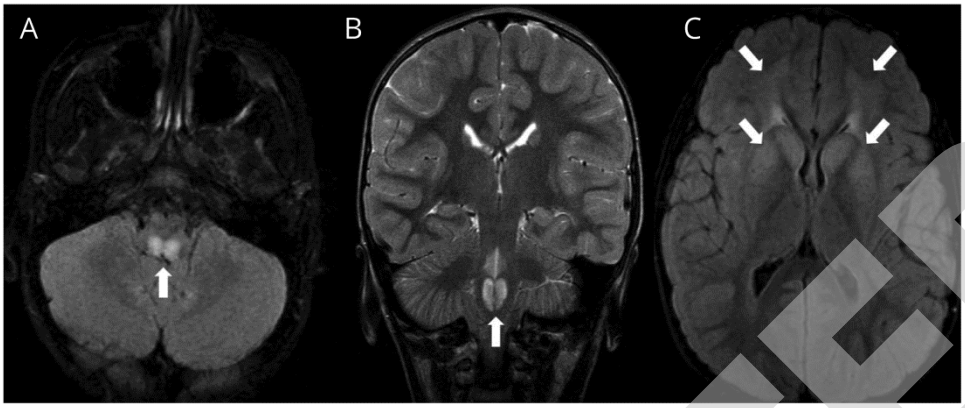
A 6-year-old boy presented with dysphagia, vomiting, and weight loss. Early developmental milestones were notable for mild gross motor and speech delay. Hypotonia was present on examination. A brain MRI revealed bilateral enhancing dorsal medullary lesions (Figure, contrast not shown). The differential 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

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