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).

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References
