A 2-year-old boy presented with developmental regression, progressive stiffening of limbs, and strabismus since the age of 8 months. A child of consanguineous parents, he had a similarly affected older brother. Nerve conduction studies were suggestive of an axonal sensorimotor neuropathy. A diagnosis of infantile neuroaxonal dystrophy (INAD) was concluded based on a suggestive MRI (figure) and the detection of a pathogenic homozygous variant in the PLA2G6 gene (c.T2370G).

INAD belongs to the family of PLA2G6-associated neurodegeneration.¹ In a child with infantile neuroregression, the peculiar changes in the brainstem and corpus callosum in the presence of cerebellar atrophy serve as a guide to further genetic testing for this disorder.²

**Author contributions**

S. Kesavan: patient management, literature review, initial draft manuscript preparation. I.K.S.: patient management, literature review, initial draft manuscript preparation. S.R.D.: patient management, literature review, initial draft manuscript preparation. L.S.: concept and design of the study, critical review of manuscript, final approval of the version to be published. S.V.:
analysis of the radiologic data, critical review of manuscript, final approval of the version to be published. J.K.S.: concept and design of the study, critical review of manuscript, final approval of the version to be published. N.S.: clinician-in-charge, concept and design of the study, critical review of manuscript for important intellectual content, final approval of the version to be published.

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