

Teaching NeuroImages: Beaking in the brainstem

A diagnostic clue

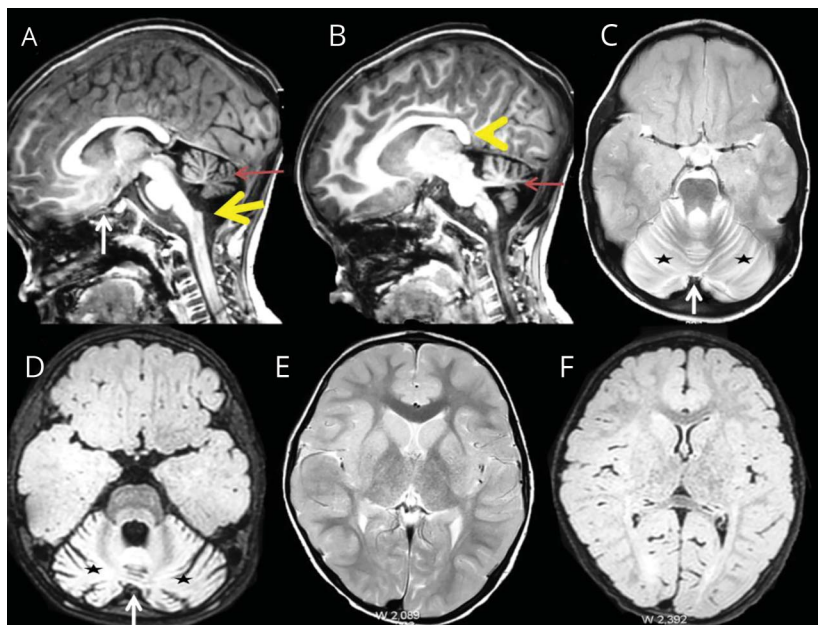
Shivan Kesavan, MD, Indar Kumar Sharawat, DM, Sumeet R. Dhawan, DM, Lokesh Saini, DM, Sameer Vyas, DM, Jitendra Kumar Sahu, DM, and Naveen Sankhyan, DM

Neurology® 2019;92:e2066-e2067. doi:10.1212/WNL.0000000000007374

Correspondence

Dr. Saini
drlokeshsaini@gmail.com

Figure MRI of the brain in a child with *PLA2G6*-associated infantile neuroaxonal dystrophy (INAD)



Midline sagittal T1-weighted sequences demonstrate claval hypertrophy (A, yellow arrow), shallow optic chiasm (A, white arrow), vertically oriented splenium of the corpus callosum (B, arrowhead), and cerebellar atrophy (A and B, red arrow). The T2-weighted (C) and fluid-attenuated inversion recovery (FLAIR) (D) sequences show hyperintense signal changes in bilateral cerebellar hemisphere with prominent folia (star) and inferior vermian atrophy (arrow) in a child with *PLA2G6*-associated INAD. T2-weighted (E) and FLAIR (F) sequences did not show any iron deposition in globus pallidus.

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.:

MORE ONLINE

→ Teaching slides

links.lww.com/WNL/A863

From the Pediatric Neurology Unit, Department of Pediatrics, Advanced Pediatrics Centre (S.K., I.K.S., S.R.D., L.S., J.K.S., N.S.), and Department of Radiodiagnosis and Imaging (S.V.), Post Graduate Institute of Medical Education & Research, Chandigarh, India.

Go to Neurology.org/N for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

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.

Study funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

References

1. Romani M, Kraoua I, Micalizzi A, et al. Infantile and childhood onset PLA2G6-associated neurodegeneration in a large North African cohort. *Eur J Neurol* 2015;22:178–186.
2. Illingworth MA, Meyer E, Chong WK, et al. *PLA2G6*-associated neurodegeneration (PLAN): further expansion of the clinical, radiological and mutation spectrum associated with infantile and atypical childhood-onset disease. *Mol Genet Metab* 2014;112:183–189.

Neurology®

Teaching NeuroImages: Beaking in the brainstem: A diagnostic clue

Shivan Kesavan, Indar Kumar Sharawat, Sumeet R. Dhawan, et al.

Neurology 2019;92:e2066-e2067

DOI 10.1212/WNL.0000000000007374

This information is current as of April 22, 2019

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/92/17/e2066.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/92/17/e2066.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): Developmental disorders http://n.neurology.org/cgi/collection/developmental_disorders 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 © 2019 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

