Teaching NeuroImages: Beaking in the brainstem

A diagnostic clue

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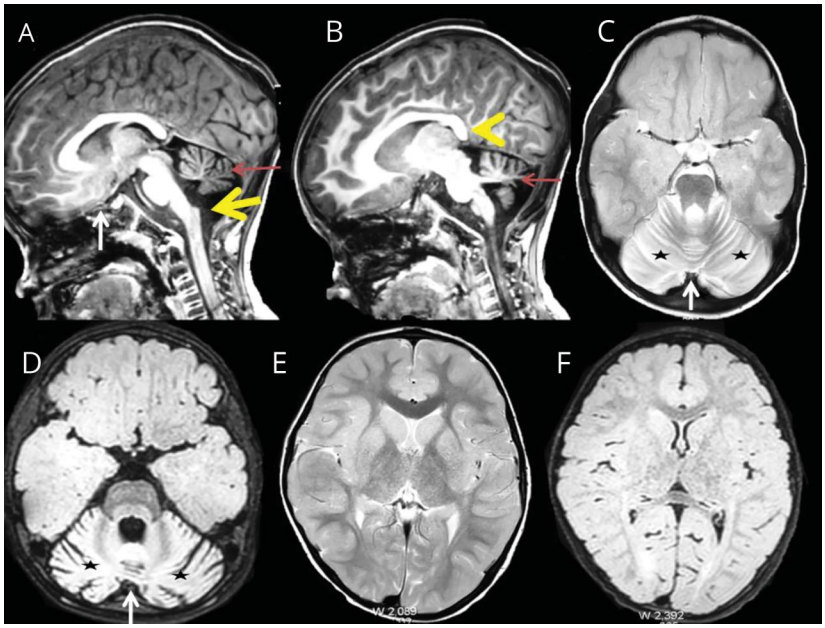
Figure MRI of the brain in a child with *PLA2G6*-associated infantile neuroaxonal dystrophy (INAD)



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]

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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.:

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