Teaching NeurolImages: Hypomyelinating leukodystrophy with generalized dystonia

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Figure Hypomyelination with atrophy of basal ganglia and cerebellum

Axial T2 (A), T2 fluid-attenuated inversion recovery (B), and T1-weighted (C) images show diffuse symmetrical white matter hyperintensity (arrowheads), small to absent putamen (arrows), and isointense signal of white matter suggestive of hypomyelination (thick arrows); sagittal T2-weighted image (D) shows atrophy of corpus callosum (arrowhead) and cerebellar vermis (arrow).

A 16-year-old boy, born of nonconsanguineous parentage, had an uneventful birth history and mild global developmental delay. He had progressive gait disturbances since 2 years of age. Examination revealed generalized dystonia, scanning speech, pendular nystagmus, cerebellar ataxia, and spasticity. MRI brain showed features of hypomyelination with atrophy of the basal ganglia and cerebellum (H-ABC) (figure).

Next-generation exome sequencing showed a heterozygous mutation in exon 4 of *TUBB4A* (c.731G>A), confirming the diagnosis. The presence of generalized dystonia and atrophic/absent putamen are the 2 features that distinguish H-ABC from the other hypomyelinating leukodystrophies, such as Pelizaeus-Merzbacher disease, Salla disease, and Tay syndrome.^{1,2}

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Author contributions

S. Vinayagamani: study design, acquisition of data, and drafting of the manuscript. S.S. Nair: interpretation of data and critical revision of the manuscript. S. Sundaram: study concept, design, and critical revision of the manuscript.

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References

1. van der Knaap MS, Naidu S, Pouwels PJ, et al. New syndrome characterized by hypomyelination with atrophy of the basal ganglia and cerebellum. *AJNR Am J Neuroradiol* 2002;23:1466–1474.
2. Hamilton EM, Polder E, Vanderver A, et al. Hypomyelination with atrophy of the basal ganglia and cerebellum: further delineation of the phenotype and genotype-phenotype correlation. *Brain* 2014;137:1921–1930.

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