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.

Study funding
No targeted funding reported.

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

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Teaching NeuroImages: Hypomyelinating leukodystrophy with generalized dystonia
S. Vinayagamani, Sruthi S. Nair and Soumya Sundaram
Neurology 2020;94;e335-e336
DOI 10.1212/WNL.0000000000008827

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