A 32-month-old boy, born at term, presented with progressive developmental regression from 14 months of age. On examination, he had horizontal nystagmus, diminished gag reflex, hypertonicity, and depressed reflexes. Brain MRI revealed symmetric T2/fluid-attenuated inversion recovery confluent hyperintensities in the periventricular white matter, corpus callosum, and centrum semiovale (sparing subcortical U fibers) with enhancement of multiple cranial nerves (figure). The characteristic leopard-like appearance of the hyperintensities prompted further evaluation for metachromatic leukodystrophy (MLD). Arylsulfatase A enzyme activity was found to be low, and the patient had a homozygous pathogenic variant in the ARSA gene (c.465+1G), confirming MLD.

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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Appendix (continued)

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References
Teaching NeuroImages: A rare case of metachromatic leukodystrophy with multiple bilateral cranial nerve enhancement


Neurology 2019;93:e1742-e1743
DOI 10.1212/WNL.0000000000008400

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