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

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Figure Imaging

Axial T2 (A, B) and fluid-attenuated inversion recovery (C) images show symmetric confluent hyperintensities in a leopard-like appearance throughout the periventricular white matter, centrum semiovale (sparing subcortical U fibers), genu, and splenium of the corpus callosum. Axial contrast-enhanced T1 images show symmetric enhancement of the bilateral third cranial nerve (D, arrows), bilateral fifth cranial nerve (E, arrows), bilateral sixth cranial nerve (F, arrows), bilateral seventh cranial nerve (G, arrows), and bilateral eighth cranial nerve (H, arrows).

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).^{1,2} 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.

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Disclosure

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Appendix Authors

Name	Location	Role	Contribution
Purabi Sonowal, MD	Children's Hospital of Michigan, Detroit	Author	Study concept and design; acquisition, analysis, drafted the manuscript for intellectual content
Dinesh Lulla, MD	Children's Hospital of Michigan, Detroit	Author	Interpreted the data, revised the manuscript for intellectual content

Appendix *(continued)*

Name	Location	Role	Contribution
Amanda Weber, DO	Children's Hospital of Michigan, Detroit	Author	Interpreted the data, revised the manuscript for intellectual content
Deniz Altinok, MD	Children's Hospital of Michigan, Detroit	Author	Acquisition of data

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