Teaching NeuroImages:

Intracranial optic nerve enlargement in infantile Krabbe disease

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Figure Imaging

(A) Axial T2W image shows bilateral symmetric hyperintensity of deep cerebral and peri-dentate nuclei white matter. Highlighted voxel shows region of interest for magnetic resonance spectroscopy with prominent choline and decreased *N*-acetylaspartate peaks. Enlargement of intracranial optic nerves is seen on (B) coronal T2W and (C) sagittal T1W MRI.

A 6-month-old boy presented with a 2-month history of developmental regression, irritability, and opisthotonic posturing. MRI demonstrated T2 hyperintensity in bilateral cerebral and peri-dentate cerebellar white matter, internal capsules, and pyramidal tracts, with hypertrophy of the intracranial optic nerves. Magnetic resonance spectroscopy (MRS) showed elevated choline peak and choline to *N*-acetylaspartate ratio (figure).

Krabbe disease is an autosomal recessive leukodystrophy caused by deficiency of galactocerebroside β -galactosidase. Accumulation of globoid cells results in optic nerve enlargement,¹ while MRS findings reflect widespread demyelination and neuronal degeneration.² Differential diagnosis of optic nerve enlargement includes glioma, nerve sheath meningioma, dural ectasia, and neurofibromatosis

type I (NF-1).¹ Absence of hyperirritability and skin lesions can help to differentiate NF-1 and Krabbe disease.

AUTHOR CONTRIBUTIONS

Dr. Shreyansh Shah: designing, drafting, and revising the manuscript. Dr. Elizabeth Freeman: clinical examination of the patient. Dr. Varina Wolf: clinical examination of the patient and revising the manuscript. Dr. Santosh Murthy: designing the manuscript. Dr. Timothy Lotze: interpretation of imaging study and clinical examination, revising the manuscript.

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Neurology[®]

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S. Shah, E. Freeman, V. Wolf, et al.
Neurology 2012;78:e126
DOI 10.1212/WNL.0b013e3182563bad

This information is current as of May 14, 2012

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