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Teaching Neuro*Images*: MRI in infantile Sandhoff disease

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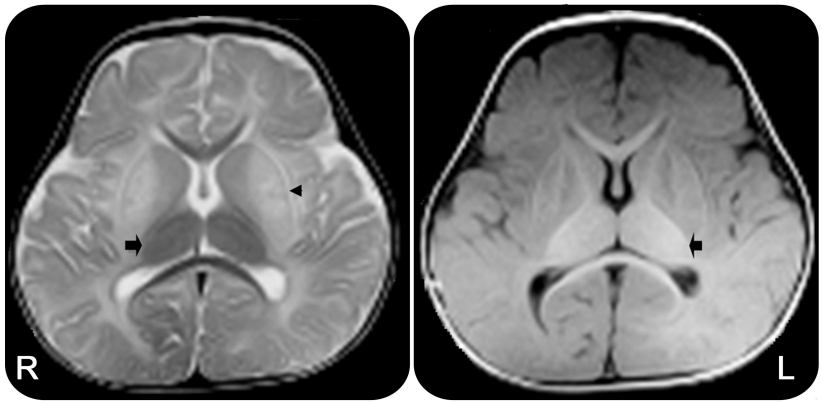
Figure Brain MRI



Axial T2- and T1-weighted images demonstrate bilateral symmetric thalamic T2 hypointensities, which are due to calcification associated with intracellular ganglioside deposition, and T1 hyperintensities (arrows). Bilateral putamina show T2 hyperintensities (arrowhead). There is delayed myelination indicated by T2 hyperintensities in the white matter. Corpus callosum is spared.

A 1-year-old girl, whose parents were second cousins, presented with developmental delay and regression of milestones. She had macrocephaly, generalized hypotonia, brisk reflexes, and hepatosplenomegaly. Ophthalmoscopic examination revealed bilateral macular cherry-red spots. MRI of the brain (figure) demonstrated bilateral symmetric thalamic T2 hypodensities and T1 hyperintensities with delayed myelination. Total hexosaminidase activity of serum was reduced to 86 nmol/h/mL (reference range 350–750 nmol/h/mL), confirming the condition to be Sandhoff disease. This autosomal recessive disorder occurs as a result of deficiency of both β-hexosaminidase A and B, leading to accumulation of GM2 ganglioside.[1,2] Tay-Sachs disease (β-hexosaminidase A deficiency) presents similarly but does not include hepatosplenomegaly.[1,2]

AUTHOR CONTRIBUTIONS

Dr. Seshadri: drafting, revising the manuscript, study concept, interpretation of data, acquisition of data. Dr. Christopher: drafting, revising the manuscript, study concept, interpretation of data, acquisition of data, study supervision. Dr. Arvinda: study supervision.

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