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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From the Departments of Neuroradiology (R.S., H.R.A.) and Neurochemistry (R.C.), National Institute of Mental Health and Neurosciences, Bangalore, India.

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