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 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.

(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.

REFERENCES
Teaching NeuroImages: MRI in infantile Sandhoff disease
Roopa Seshadri, Rita Christopher and H.R. Arvinda
Neurology 2011;77:e34
DOI 10.1212/WNL.0b013e318227b215

This information is current as of August 1, 2011