Teaching Neuro*Images*: T2 hypointense thalami in infantile GM1 gangliosidosis

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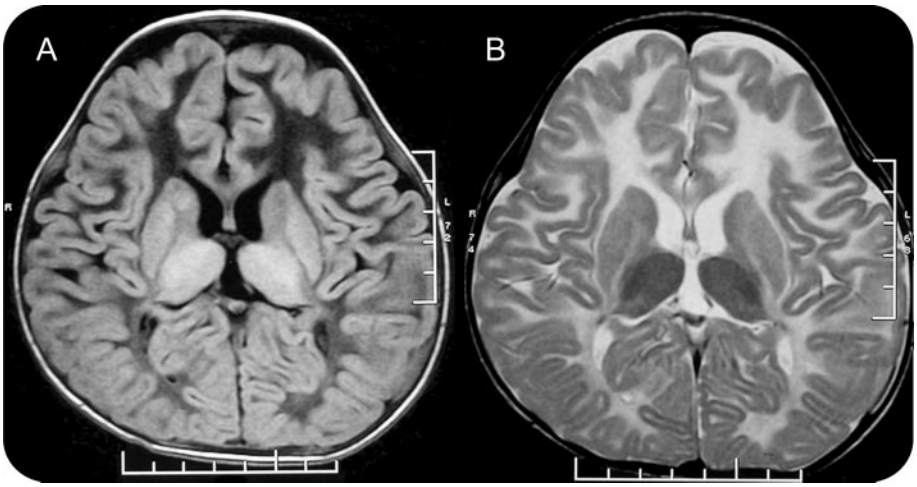
Figure MRI of brain



Axial T1-weighted images (A) and T2-weighted images (B) at the level of thalami show diffuse dysmyelination of hemispheric white matter with bilaterally symmetric thalamic signal change which appear hyperintense on T1-weighted and hypointense on T2-weighted images. Additionally, the thalami and basal ganglia show subtle nonhomogeneous mixed low and high signals on both images.

A 13-month-old boy was evaluated for seizures and developmental regression. He was well until the age of 9 months. Then he developed generalized tonic seizures and progressive loss of ability to sit, hold neck, interact, and smile. Examination revealed coarse facies, bilateral macular cherry red spots, and hepatosplenomegaly. A suggestive MRI of brain (figure) and absent β-galactosidase enzyme activity in leukocytes confirmed the diagnosis of GM1 gangliosidosis.

Accumulation of GM1 gangliosides in the neurons can increase tissue viscosity. This possibly leads to hypointensity of thalami on T2-weighted images. Similar changes in the thalami have been reported in other lysosomal storage disorders.[1]

AUTHOR CONTRIBUTIONS

N.S. and S.S. performed the clinical/diagnostic workup of the child and reviewed the literature. M.K. and S.G. were in charge of the case overall and approved the manuscript.

REFERENCE

1. Autti T, Joensuu R, Aberg L. Decreased T2 signal in the thalami may be a sign of lysosomal storage disease. Neuroradiology 2007;49:571–578.

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