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Teaching Neuro*Images*: An extremely rare cause of treatable acute encephalopathy

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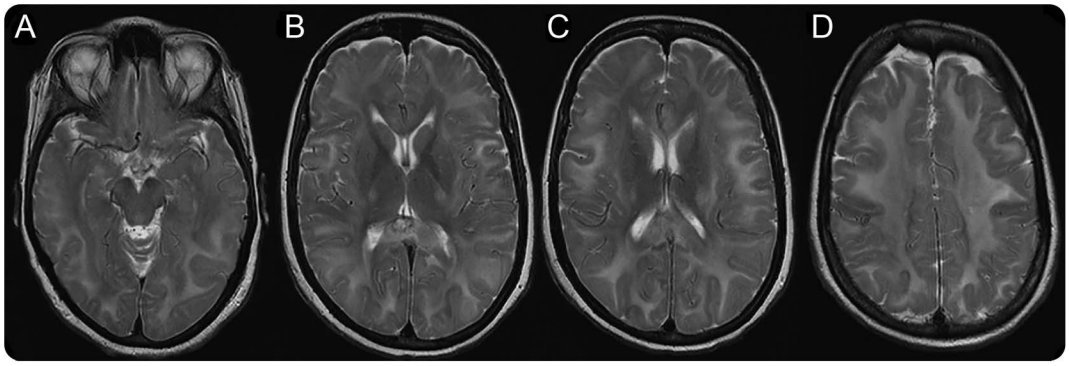
Figure Neuroimaging findings of thiamine transporter deficiency



Neuroimaging studies in *SLC19A3* gene mutations. (A–D) Axial brain MRI discloses marked diffuse hyperintensity of profound and subcortical brain white matter and corticospinal tracts hyperintensity in the posterior limb of the internal capsule in T2-weighted images.

A 32-year-old Brazilian man presented with subacute encephalopathy and gait instability. Examination showed generalized dystonia and lethargy. Brain MRI showed diffuse symmetrical leukoencephalopathy without changes in the basal ganglia (figure). Genetic analysis revealed heterozygous mutations c.74dupT and c.980-14A<G in the *SLC19A3* gene. There was marked neurologic improvement after high doses of thiamine replacement.

Inherited defects of thiamine metabolism and transport are a rare group of neurometabolic diseases resulting from enzyme deficiencies or transporter dysfunctions giving rise to heterogeneous neurologic manifestations such as biotin-responsive basal ganglia disease, episodic ataxia, acute encephalopathy, and lethal microcephaly with clinical improvement after thiamine replacement.[1,2]

AUTHOR CONTRIBUTIONS

Dr. Souza: case report project conception, organization, and execution, writing of the first draft. Dr. Bortholin: case report project conception, organization, and execution, writing of the first draft. Dr. Pinto: case report project conception and execution, writing of the first draft, manuscript review and critique. Dr. Oliveira: manuscript review and critique.

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DISCLOSURE

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