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Teaching NeuroImages: Subacute encephalopathy in a young woman with *THTR2* gene mutation

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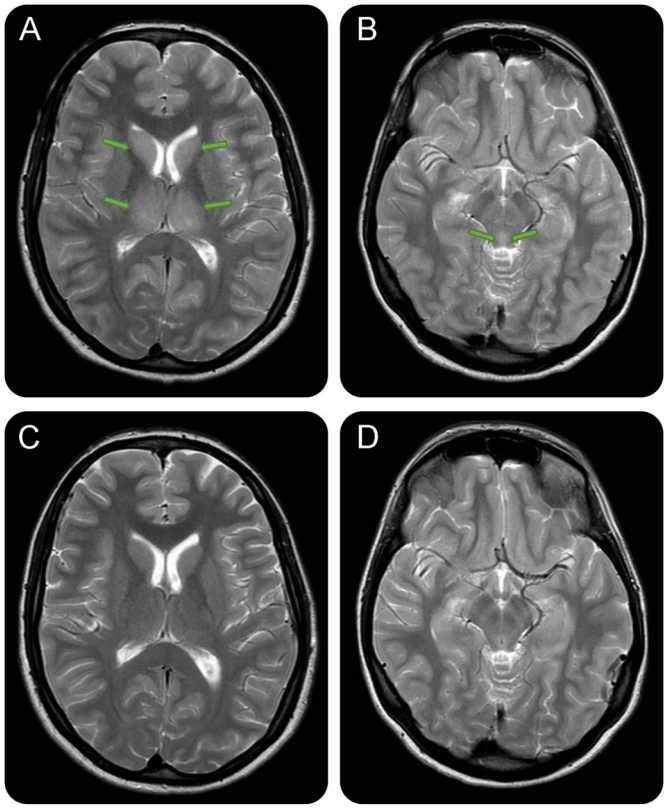
Figure Brain abnormalities on MRI



Initial axial brain MRI findings. T2-weighted images show symmetrical high signal intensities in the bilateral paramedian thalami and caudate heads (A) and in the periaqueductal gray matter (B) (arrows). Increased diffusion-weighted imaging signal and increased apparent diffusion coefficient values were seen in these brain regions. There was no gadolinium enhancement. Follow-up MRIs show near-complete resolution of the lesions after 35 days of thiamine treatment (C, D).

A 21-year-old woman presented with coma after 5 days of fever, gait ataxia, and somnolence. Brain MRI showed lesions in medial thalami, caudate heads, and periaqueductal region (figure). Hyperlactatemia was present; serum thiamine levels were normal.

Thiamine IM 600 mg daily led to complete recovery within 3 days. Sequencing analysis of thiamine transporter 2 (*THTR2*) gene revealed a base deletion, at homozygote state, in intron 3 in position c.980-4 upstream at donor/acceptor site of exon 4, predicted by computational algorithms to be deleterious.

In young adults, *THTR2* dysfunction may foster subacute encephalopathy with typical radiologic manifestations that is promptly responsive to thiamine administration.[1,2]

AUTHOR CONTRIBUTIONS

Elia Sechi: principal author/corresponding author. Alberto Addis: revised the manuscript for content. Giulia Fadda: revised the manuscript for content. Luigi Minafra: analyzed and interpreted the data. Valentina Bravatà: analyzed and interpreted the data.

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GianPietro Sechi: analyzed and interpreted data, revised the manuscript for content.

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DISCLOSURE

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