Teaching NeuroImages:

Facial grimacing and sensorineural hearing loss in a woman with cirrhosis of the liver

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Figure Neuroimaging in hypermanganesemia with dystonia, polycythemia, and cirrhosis

(A) Facial grimacing noted on examination. (B) Sagittal and (C-E) axial brain MRI disclose hyperintense signal in basal ganglia (white arrow), pontine tegmentum (*), and cerebellum (white arrowhead) in T1-weighted images.

A 29-year-old woman presented with 5-year history of gait instability and hearing loss. Medical history disclosed cirrhosis of the liver and polycythemia. Examination showed bradykinesia, facial grimacing, cervical dystonia, and ataxia. Neuroimaging showed hyperintense signal in basal ganglia on T1-weighted images (figure). Blood tests showed 18.5 g/dL of hemoglobin and 1,500 nmol/L of serum manganese (reference <320 nmol/L). Genetic analysis revealed previously described pathogenic^{1,2} homozygous mutation c.266T>C(p.Leu89Pro) in the *SLC30A10* gene.

Hypermanganesemia with dystonia, polycythemia, and cirrhosis is an inherited metabolic disorder of manganese metabolism that presents with chronic liver disease, polycythemia, and neurologic manifestations such as movement disorders, ataxia, pyramidal disturbances, and axonal polyneuropathy with hyperintensity of basal ganglia on T1-weighted images, making differential diagnosis with acquired hepatocerebral degeneration and chronic parenteral nutrition.^{1,2}

AUTHOR CONTRIBUTIONS

Case report project conception (Dr. Sgobbi de Souza, Dr. Bortholin, Dr. Bocca Vieira de Rezende Pinto), organization (Dr. Sgobbi de Souza, Dr. Bortholin), and execution (Dr. Sgobbi de Souza, Dr. Bortholin), writing of the first draft (Dr. Sgobbi de Souza, Dr. Bortholin, and Dr. Bocca Vieira de Rezende Pinto), and manuscript review and critique (Dr. Bocca Vieira de Rezende Pinto and Dr. Souza Bulle Oliveira).

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

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