Teaching *NeuroImages*: MRI findings in carbamoyl phosphate synthetase 1 deficiency

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Figure 1 MRI findings in insular cortices and deep frontal gyri

Coronal fluid-attenuated inversion recovery images show hyperintensity of the insular cortices (closed arrows) and deep frontal gyri (open arrows) on both sides adjacent to the subcortical U-fibers (A, B).

Figure 2 MRI findings in caudate head and putamen

Axial proton density (A) and coronal fluid-attenuated inversion recovery (B) images show linear hyperintensities in bilateral putamina (closed arrows) and caudate head (open arrows).

A girl with carbamoyl phosphate synthetase 1 (CPS-1) deficiency, symptomatic since the newborn period, underwent liver transplantation at 7 months. She later developed ataxia and truncal dystonia. MRI brain at age 2 years showed hyperintensities in proton density and fluid-attenuated inversion recovery images involving the insular cortices, deep frontal gyri (figure 1, A and B), caudate nucleus, and putamen (figure 2, A and B). These findings are characteristic of proximal urea cycle disorders, namely CPS-1 deficiency,^{1,2} and

are unlikely due to myelinolysis, pontine or extrapontine. The above findings on an MRI in an encephalopathic child give a clue to the diagnosis of a hyperammonemic disorder.

AUTHOR CONTRIBUTIONS

Dr. Nunley conducted background research and was primary author for the manuscript. Dr. Ghosh assisted with conceptualization and editing of the manuscript.

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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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