

Teaching NeuroImages: When MRI is a clue in episodic ataxia

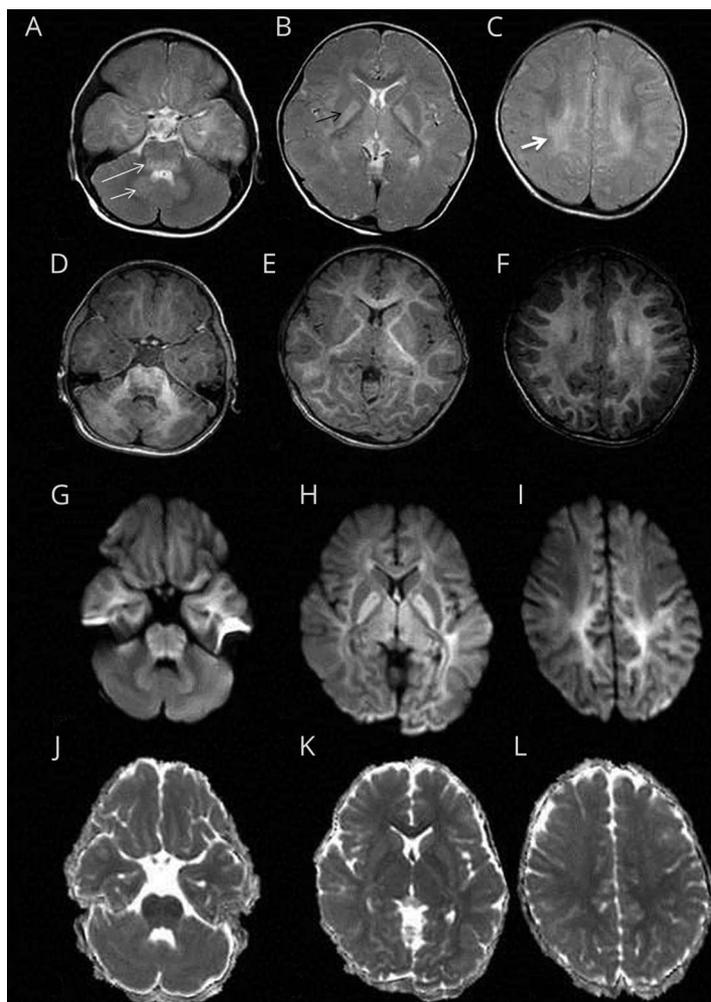
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Figure MRI in maple syrup urine disease



MRI shows hyperintense signal in (A) cerebellar deep white matter including nuclei (short white arrow) and dorsal pons (long white arrow), (B) globus pallidi (black arrow) and thalami, and (C) bilateral cerebral white matter (white arrow) in axial T2 scan. These changes are hypointense on T1 scan (D–F). Diffusion-weighted images (G–I, $b = 1,000$) and apparent diffusion coefficient (J–L) show acute diffusion restriction.

A 3-year-old girl presented with episodic ataxia for the past month. It lasted for 1–2 days continuously after a febrile illness. MRI was suggestive of a neurometabolic disorder (figure, A–L). Plasma valine and leucine and urinary branched-chain aminoacids were

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elevated. Clinical exome revealed a homozygous, missense, pathogenic variation in *BCKDHB* gene (exon 5, chr6: 80878686A>C; p.His191Pro).

Patients with intermittent maple syrup urine disease (MSUD) may develop episodic decompensation during periods of stress secondary to acute leucinuria and abnormal neurotransmitter activity.¹ Bilateral, symmetrical diffusion restriction in myelinated areas of the brain is seen in aminoacidopathies, including MSUD, nonketotic hyperglycinemia, and Canavan disease.²

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Disclosure

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Appendix Authors

Names	Location	Role	Role and contribution
Sumeet R. Dhawan, MD, DM	Postgraduate Institute of Medical Education and Research, Chandigarh, India	Author	Original drafting and revising the manuscript for intellectual content
Arushi Gahlot Saini, MD, DM	Postgraduate Institute of Medical Education and Research, Chandigarh, India	Corresponding author	Clinician-in-charge, conceptualization of the study, critical review of the manuscript for intellectual content

Appendix (continued)

Names	Location	Role	Role and contribution
Sameer Vyas, MD, DM	Postgraduate Institute of Medical Education and Research, Chandigarh, India	Author	Analysis and interpretation of radiologic data, critical review of the manuscript
Savita Verma Attri, PhD, MAMS, FIMSA	Postgraduate Institute of Medical Education and Research, Chandigarh, India	Author	Biochemical analysis, revising the manuscript for intellectual content

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