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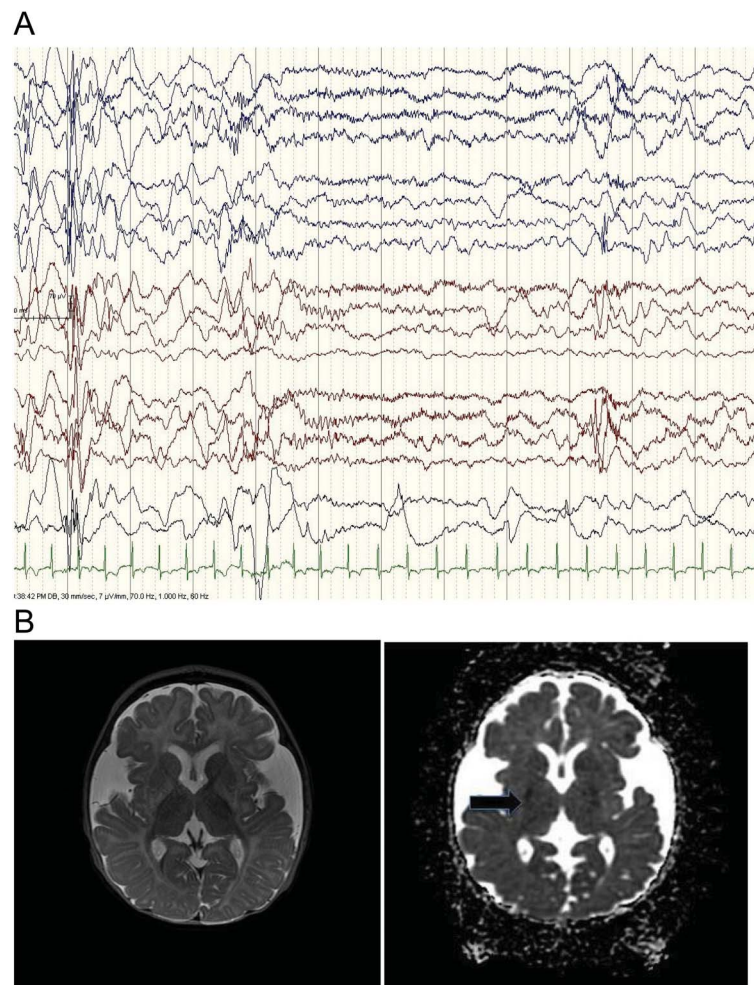
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Teaching NeuroImages: Infant with glutaric aciduria type 1 presenting with infantile spasms and hypersarhythmia

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Figure EEG and brain MRI findings



(A) Hypersarhythmia on EEG. EEG at presentation showed hypersarhythmia: high voltages, disorganization, and frequent multifocal and generalized spikes. (B) Acute basal ganglia injury. T2-weighted image demonstrates widened opercula and increased T2 signal in globi pallidi and dorsal putamina. Apparent diffusion coefficient image shows subtle reduced diffusion in the globus pallidi.

A 7-month-old boy with glutaric aciduria type 1 (GA1) presented with 1 week of clustered flexor spasms. Examination revealed mild axial hypotonia without encephalopathy. Video-EEG monitoring revealed hypersarhythmia and infantile spasms (figure, A). MRI showed acute basal ganglia injury (figure, B). After 3 weeks of prednisolone treatment,

5-month follow-up showed continued resolution of hypersarhythmia and spasms.

GA1 is an autosomal recessive condition due to deficiency of the enzyme necessary for degradation of lysine, tryptophan, and hydroxylysine. Infantile spasms have only been reported once before in GA1.¹ New-onset spasms may be associated with

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suboptimal metabolic control, even in the absence of encephalopathy.²

AUTHOR CONTRIBUTIONS

Nichole Young-Lin: participated in conceptualization of the manuscript, drafted the manuscript and figure legend, and revised the manuscript for intellectual content. Sarah Shalev: selected appropriate EEG images (i.e., interpretation of data) and revised the manuscript for intellectual content. Orit A. Glenn: selected appropriate brain MRI images (i.e., interpretation of data) and revised the manuscript for intellectual content. Marisa Gardner: participated in analysis of results and revised the manuscript for intellectual content. Chung Lee: revised the manuscript for intellectual content. Anthony Wynshaw-Boris: participated in conceptualization of the manuscript and revised the manuscript for intellectual content. Amy A. Gelfand: participated in conceptualization of the manuscript and revised the manuscript for intellectual content.

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