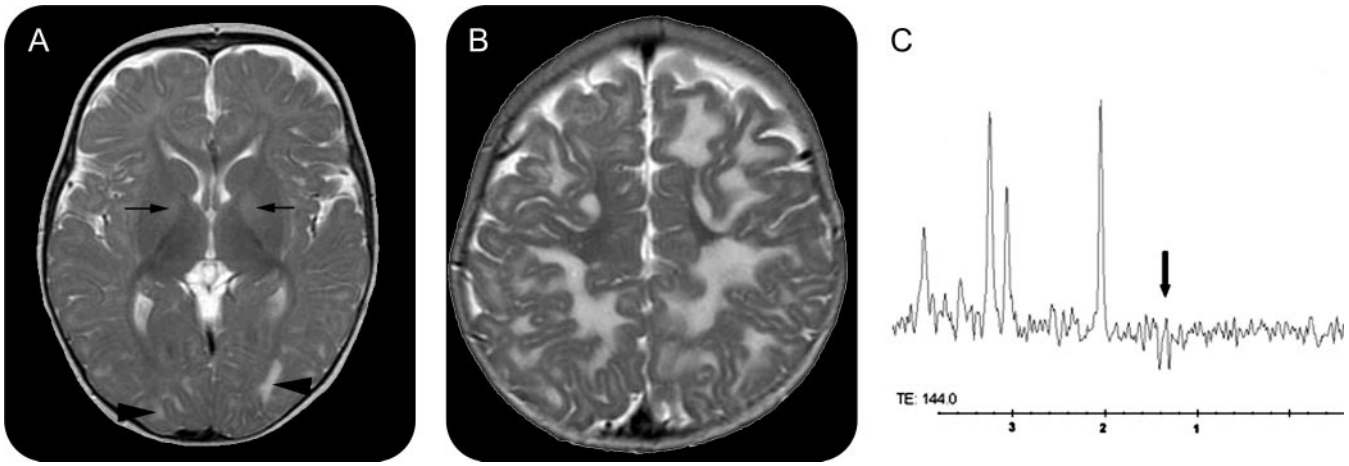
Head bobbing due to succinic semialdehyde dehydrogenase deficiency

Figure Magnetic resonance imaging



(A) Axial T2 MRI at level of basal ganglia shows abnormal high signal in the globus pallidus bilaterally (arrows). Abnormal high signal is also seen in the white matter posteriorly (arrowheads). (B) Axial T2 MRI above the level of the ventricles shows diffuse abnormal high signal in the white matter. (C) Magnetic resonance spectroscopy at long echo time (TE = 144) centered over left thalamus and basal ganglia shows inverted double peak at 1.3 ppm characteristic of lactate.

A 9-month-old boy had a short history of "yes-yes" head bobbing (video on the *Neurology*® Web site at www.neurology.org). Mild global delay prompted investigation. Imaging (figure, A–C), biochemical, and molecular findings (table e-1) were consistent with succinic semialdehyde dehydrogenase (SSADH) deficiency.

SSADH deficiency, a γ-aminobutyric acid degradation disorder, may be associated with movement disorder, including late-onset paroxysmal dystonia responsive to vigabatrin,[1] but head bobbing has not been reported. In SSADH deficiency, head bobbing may be due to increased γ-hydroxybutyrate affecting diencephalic extrapyramidal pathways via the thalamic dorsomedial nucleus.[2] Other disorders associated with "yes-yes" head bobbing include spasmus nutans (associated with nystagmus), rhombencephalosynapsis (cerebellar malformation), and bobble-head doll syndrome (third ventricular lesions).

Declan J. O'Rourke, MRCPI, Stephanie Ryan, FFR RCSI, FRCSI, Mary D. King, FRCPI, FRCPCH, Dublin, Ireland

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Address correspondence and reprint requests to Prof. Mary D. King, Department of Neurology, Children's University Hospital, Temple Street, Dublin 1, Ireland mary.king@cuh.ie

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