A 10-year-old girl with infantile-onset hypotonia and motor delay presented with a 6-year history of paroxysmal episodes of involuntary upward gaze with preserved consciousness (video 1). These episodes occurred multiple times a day and increased during fatigue or fever. At age 3, she had a history of 5 episodes of fasting-induced hypoglycemic seizures. She had bradykinesia and limb dystonia. Her younger brother was similarly affected. A diagnosis of amino acid decarboxylase deficiency was confirmed by a pathogenic homozygous variation in exon 5 (c.475G>A p.Ala159Thr) of dopamine decarboxylase gene. Treatment with levodopa, pyridoxine, folinic acid, and trihexyphenidyl resulted in clinical benefit. Other early-onset neurotransmitter disorders with recurrent oculogyric crises include sepiapterin reductase deficiency, tyrosine hydroxylase deficiency, and dopamine transporter defects.

References
Teaching Video NeuroImages: Oculogyric crises in a 10-year-old girl
Renu Suthar, Naveen Sankhyan and Pratibha Singhi
Neurology 2019;92;e82
DOI 10.1212/WNL.0000000000006698

This information is current as of December 24, 2018