Teaching Video NeuroImages: Oculogyric crises in a 10-year-old girl

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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.[1] Other early-onset neurotransmitter disorders with recurrent oculogyric crises include sepiapterin reductase deficiency, tyrosine hydroxylase deficiency, and dopamine transporter defects.[2]

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Video

Teaching slides
links.lww.com/WNL/A767

References

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2. Kurian MA, Gissen P, Smith M, Heales SJ, Clayton PT. The monoamine neurotransmitter disorders: an expanding range of neurological syndromes. Lancet Neurol 2011;10:721–733.

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