

Teaching Video NeuroImage: Paroxysmal Nocturnal Dyskinesias

A Characteristic Feature of *ADCY5* Mutation

Tamara M. Pringsheim, MD, Nicholas Cothros, MD, PhD, and Emmanuel Roze, MD, PhD

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Correspondence

Dr. Pringsheim,
tmprings@ucalgary.ca

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A 4-year-old girl with a history of axial hypotonia, delayed developmental motor milestones, and hyperkinetic movements presented with increasingly frequent episodes of paroxysmal nocturnal dyskinesia from 12 months of age (video 1). Dyskinesias were also present intermittently during waking hours and appeared worse when she was upset or agitated. Ictal EEG performed during the daytime showed no electrographic seizure. The clinical manifestations were linked to a heterozygous c.1252C > T (p.R418W) pathogenic variant in the *ADCY5* gene (RefSeq accession number NM_183,357). Nighttime videorecording or polysomnography may help to distinguish nocturnal dyskinesia from sleep-related hypermotor seizures and disorders of arousal. The presence of nocturnal dyskinesia should prompt the clinician to perform a molecular analysis of *ADCY5*.¹ This is important for clinical practice as *ADCY5*-related paroxysmal dyskinetic episodes are disabling and can respond to caffeine or deep brain stimulation.^{2,3} This patient experienced a decrease in frequency of dyskinetic episodes with caffeine.

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Appendix Authors

Name	Location	Contribution
Tamara Pringsheim, MD	University of Calgary, Canada	Acquisition of data, drafted the manuscript for intellectual content
Nicholas Cothros, MD, PhD	University of Calgary, Canada	Drafted the manuscript for intellectual content
Emmanuel Roze, MD, PhD	Department of Neurology, Pitié-Salpêtrière Hospital, Sorbonne University and the Assistance Publique-Hôpitaux de Paris, France	Interpreted the data, revised the manuscript for intellectual content

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From the Department of Clinical Neurosciences (T.M.P., N.C.), University of Calgary, Canada; and Department of Neurology (E.R.), Pitié-Salpêtrière Hospital, Sorbonne University and the Assistance Publique-Hôpitaux de Paris, France.

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