# Teaching Video NeuroImage: Paroxysmal Nocturnal Dyskinesias

## A Characteristic Feature of *ADCY5* Mutation

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*Neurology*® 2021;97:e1062. doi:10.1212/WNL.0000000000011920

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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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Video

**Teaching slides**
links.lww.com/WNL/B360

## Study Funding
The authors report no targeted funding.

## Disclosure
The authors report no disclosures. Go to Neurology.org/N for full disclosures.

## Appendix Authors

| Name | Location | Contribution |
|---|---|---|
| **Tamara Pringsheim, MD** | University of Calgary, Canada | Acquisition of data, drafted the manuscript for intellectual content |
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*Neurology* 2021;97;e1062 Published Online before print April 7, 2021
DOI 10.1212/WNL.0000000000011920