Teaching Video NeuroImages: Paroxysmal Nocturnal Dyskinesias: A Characteristic Feature of ADCY5 Mutation

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Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.
A four-year-old girl with a past 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, http://links.lww.com/WNL/B361). Dyskinesias were also present intermittently during waking hours and appeared worse when 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_183357). Nighttime videorecording and/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.
Appendix 1. Authors

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<thead>
<tr>
<th>Name</th>
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Teaching Slides - [http://links.lww.com/WNL/B360](http://links.lww.com/WNL/B360)

Video - [http://links.lww.com/WNL/B361](http://links.lww.com/WNL/B361)

References


Video Legend

Video shows a paroxysmal nocturnal bout of chorea and dystonia in a child with ADCY-5 related dyskinesia.
Teaching Video NeuroImages: Paroxysmal Nocturnal Dyskinesias: A Characteristic Feature of ADCY5 Mutation
Tamara M. Pringsheim, Nicholas Cothros and Emmanuel Roze
Neurology published online April 7, 2021
DOI 10.1212/WNL.0000000000011920

This information is current as of April 7, 2021

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