Teaching Video NeuroImages: Paroxysmal Nocturnal Dyskinesias: A Characteristic Feature of <i>ADCY5</i> Mutation

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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. This is important for clinical practice as ADCY5-related paroxysmal dyskinetic episodes are disabling and can respond to caffeine or deep brain stimulation. This patient experienced a decrease in frequency of dyskinetic episodes with caffeine.
Appendix 1. Authors

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<tr>
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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.
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