

A 4-Year-Old Girl With Paroxysmal Nocturnal Dyskinesia

Teaching Video NeuroImages

Neurology[®]

Resident & Fellow Section

Vignette

- 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
- 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

Video



Paroxysmal Nocturnal Dyskinesia: A Characteristic Feature of ADCY5 Mutation

- The clinical manifestations were linked to a heterozygous c.1252C>T (p.R418W) pathogenic variant in the *ADCY5* gene
- 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*
- *ADCY5*-related paroxysmal dyskinetic episodes are disabling and can respond to caffeine or deep brain stimulation