Teaching Video NeurolImage: Paroxysmal Nocturnal Dyskinesias

A Characteristic Feature of *ADCY5* Mutation

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

Neurology® 2021;97:e1062. doi:10.1212/WNL.00000000000011920

Correspondence

Dr. Pringsheim,
tmprings@ucalgary.ca

MORE ONLINE

Video

Teaching slides

links.lww.com/WNL/B360

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.

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
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

References

1. Meneret A, Roze E, Maranci JB, et al. Sleep in *ADCY5*-related dyskinesia: prolonged awakenings caused by abnormal movements. *J Clin Sleep Med*. 2019;15(7):1021-1029.
2. Meneret A, Gras D, McGovern E, Roze E. Caffeine and the dyskinesia related to mutations in the *ADCY5* gene. *Ann Intern Med*. 2019;171(6):439.
3. de Almeida Marcelino AL, Mainka T, Krause P, Poewe W, Ganos C, Kühn AA. Deep brain stimulation reduces (nocturnal) dyskinetic exacerbations in patients with *ADCY5* mutation: a case series. *J Neurol*. 2020;267(12):3624-3631.

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.

Go to Neurology.org/N for full disclosures.

Neurology®

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

Tamara M. Pringsheim, Nicholas Cothros and Emmanuel Roze
Neurology 2021;97:e1062 Published Online before print April 7, 2021
DOI 10.1212/WNL.00000000000011920

This information is current as of April 7, 2021

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/97/10/e1062.full
References	This article cites 3 articles, 0 of which you can access for free at: http://n.neurology.org/content/97/10/e1062.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Clinical Neurology http://n.neurology.org/cgi/collection/all_clinical_neurology All Movement Disorders http://n.neurology.org/cgi/collection/all_movement_disorders All Pediatric http://n.neurology.org/cgi/collection/all_pediatric
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2021 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.