Teaching Video NeuroImages: Atypical Abnormal Eye Movements in *PNPO*-Related Epilepsy

Sara Pavitt, MD, Amanda G. Sandoval Karamian, MD, Gaurav Chattree, MD, Jenna Klotz, MD, and Shannon Beres, MD

Neurology® 2021;96:e1927. doi:10.1212/WNL.0000000000010861

Correspondence

Dr. Pavitt
sara.pavitt2@ucsf.edu

A full-term female neonate developed focal motor seizures at 1 hour of life, followed by paroxysmal nonepileptic abnormal eye–head movements on day 5 (video 1). CSF revealed mild hypoglycorrhachia, borderline low CSF:serum glucose, and low pyridoxal-5'-phosphate (PSP). Genetic testing showed compound heterozygous mutations in the pyridoxamine 5'-phosphate oxidase (*PNPO*) gene consistent with PSP-dependent epilepsy. Aberrant gaze saccades with head jerks are classically described in GLUT-1 deficiency,¹ though not described in other conditions with associated hypoglycorrhachia. *PNPO*-associated epilepsy has been associated with abnormal eye movements² and should be considered when neonates present with seizures and aberrant saccades with coordinated head movements.

Study Funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

Appendix Authors

Name	Location	Contribution
Sara Pavitt, MD	Stanford Children's Health, Palo Alto, CA	Data collection, drafted the manuscript for intellectual content
Amanda Sandoval Karamian, MD	Stanford Children's Health, Palo Alto, CA	Drafted the manuscript for intellectual content
Gaurav Chattree, MD	Stanford Health Care, Palo Alto, CA	Data collection, revised the manuscript for intellectual content
Jenna Klotz, MD	Stanford Children's Health, Palo Alto, CA	Data collection, revised the manuscript for intellectual content
Shannon Beres, MD	Stanford Children's Health, Palo Alto, CA	Revised the manuscript for intellectual content

References

- Pearson TS, Pons R, Engelstad K, Kane SA, Goldberg ME, De Vivo DC. Paroxysmal eye–head movements in Glut1 deficiency syndrome. *Neurology* 2017;88:1666–1673.
- Mills PB, Surtees RAH, Champion MP, et al. Neonatal epileptic encephalopathy caused by mutations in the *PNPO* gene encoding pyridox(am)ine 5'-phosphate oxidase. *Hum Mol Genet* 2005;14:1077–1086.

From Stanford Children's Health (S.P., A.G.S.K., J.K., S.B.) and Stanford Health Care (G.C.), Palo Alto, CA.

Go to Neurology.org/N for full disclosures.

MORE ONLINE

Video

Teaching slides

<http://links.lww.com/WNL/B232>

Neurology®

Teaching Video NeuroImages: Atypical Abnormal Eye Movements in *PNPO*-Related Epilepsy

Sara Pavitt, Amanda G. Sandoval Karamian, Gaurav Chattree, et al.
Neurology 2021;96:e1927 Published Online before print September 10, 2020
DOI 10.1212/WNL.0000000000010861

This information is current as of September 10, 2020

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/96/14/e1927.full
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/96/14/e1927.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Epilepsy/Seizures http://n.neurology.org/cgi/collection/all_epilepsy_seizures Metabolic disease (inherited) http://n.neurology.org/cgi/collection/metabolic_disease_inherited Neonatal http://n.neurology.org/cgi/collection/neonatal Neonatal seizures http://n.neurology.org/cgi/collection/neonatal_seizures
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 © 2020 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.