

Teaching Video NeuroImage: Stop-Motion Chorea in PURA Syndrome

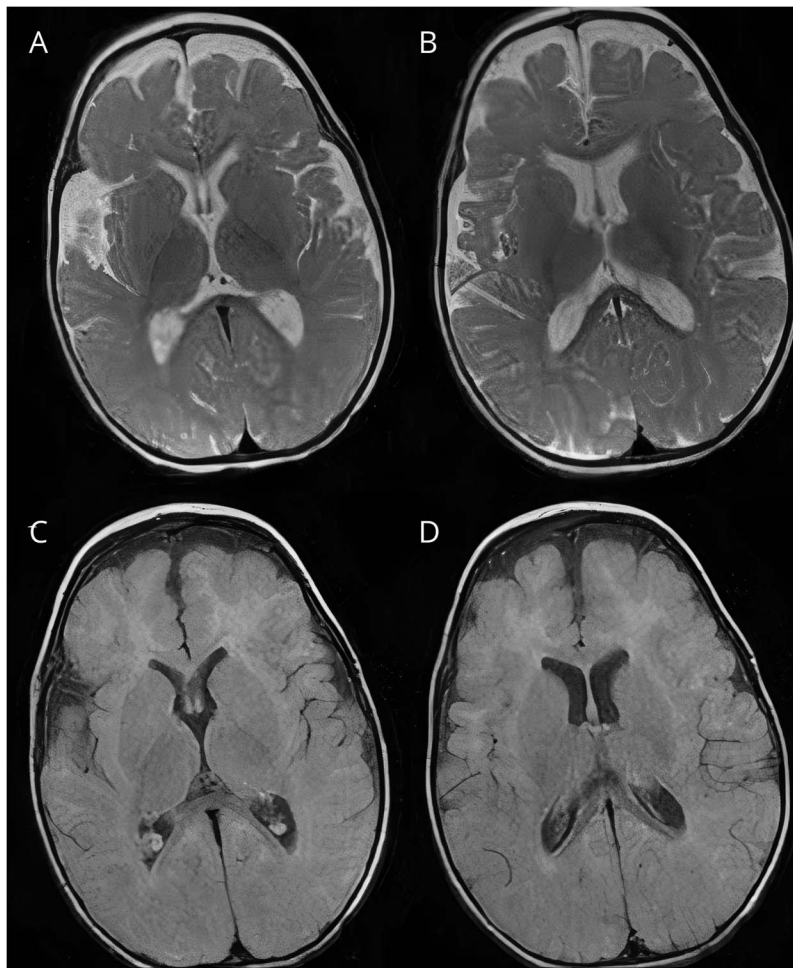
Gustavo Leite Franklin, MD, PhD, Eli Paula Bacheladenski, MD, Danielle C. B. Rodrigues, MD, and Ana C.S. Crippa, MD, PhD

Neurology® 2023;100:492-493. doi:10.1212/WNL.0000000000201605

Correspondence

Dr. Franklin
gustavolf_88@hotmail.com

Figure Brain MRI



(A) and (B): Axial T2-weighted brain MRI; (C) and (D): Axial T1-weighted brain MRI, all showing volumetric reduction bilaterally, notably in frontal lobe.

A 5-year-old girl with abnormal facial features, strabismus, horizontal nystagmus, hypotonia, and a history of hypersomnolence, seizures and developmental delay began to experience a generalized complex movement disorder. Clinically, there was a mixed hyperkinetic movement disorder, consisting of chorea, dystonia, myoclonus, and hand stereotypies. The presence of generalized jerks, interposed with those complex movements, resembled a stop-motion animation (Video 1), similar to the animation technique in which objects are physically manipulated in small increments and

MORE ONLINE

 Video

Teaching slides

links.lww.com/WNL/C556

From the Internal Medicine Department (G.L.F.), Pontifical Catholic University of Paraná; and Neurology Pediatric Unit (E.P.B., D.C.B.R., A.C.S.C.), Universidade Federal do Paraná, Curitiba (PR), Brazil.

Go to Neurology.org/N for full disclosures.

photographed frame by frame. Brain MRI showed mild frontal cortical atrophy (Figure). Genetic investigation was performed, and CGH array was performed, finding a pathogenic variant arr[GRCh37]5q31.2q31.3(139033279_140058893)x1 in PURA gene, compatible with PURA syndrome.¹ The presence of complex hyperkinetic movement disorders in infants with global developmental delay may be an important clue to diagnose PURA syndrome. Affected patients may be misdiagnosed with dyskinetic cerebral palsy if genetic studies are not pursued.²

Study Funding

The authors report no targeted funding.

Disclosure

All authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

Publication History

Received by *Neurology* May 7, 2022. Accepted in final form October 11, 2022. Submitted and externally peer reviewed. The handling editor was Associate Editor Roy Strowd III, MD, Med, MS.

Appendix (continued)

Name	Location	Contribution
Eli Paula Bacheladenski, MD	Neurology Pediatric Unit, Universidade Federal do Paraná, Curitiba (PR), Brazil	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; analysis or interpretation of data
Danielle C. B. Rodrigues, MD	Neurology Pediatric Unit, Universidade Federal do Paraná, Curitiba (PR), Brazil	Drafting/revision of the manuscript for content, including medical writing for content; study concept or design; analysis or interpretation of data
Ana C. S. Crippa, MD, PhD	Neurology Pediatric Unit, Universidade Federal do Paraná, Curitiba (PR), Brazil	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; analysis or interpretation of data

Appendix Authors

Name	Location	Contribution
Gustavo Leite Franklin, MD, PhD	Internal Medicine Department, Pontifical Catholic University of Paraná, Curitiba (PR), Brazil	Drafting/revision of the manuscript for content, including medical writing for content; major role in the acquisition of data; study concept or design; analysis or interpretation of data

References

- Johannesen KM, Gardella E, Gjerulfsen CE, et al. PURA-related developmental and epileptic encephalopathy: phenotypic and genotypic spectrum. *Neurol Genet.* 2021; 7(6):e613.
- Reijnders MRF, Janowski R, Alvi M, et al. PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. *J Med Genet.* 2018;55(2):104-113.

The *Neurology*[®] Null Hypothesis Online Collection...

Contributing to a transparent research reporting culture!



The *Neurology* journals have partnered with the Center for Biomedical Research Transparency (CBMRT) to promote and facilitate transparent reporting of biomedical research by ensuring that all biomedical results—including negative and inconclusive results—are accessible to researchers and clinicians in the interests of full transparency and research efficiency.

Neurology's Null Hypothesis Collection is a dedicated online section for well conducted negative, inconclusive, or replication studies. View the collection at: [NPub.org/NullHypothesis](https://www.npub.org/NullHypothesis)

Neurology®

Teaching Video NeuroImage: Stop-Motion Chorea in PURA Syndrome
Gustavo Leite Franklin, Eli Paula Bacheladenski, Danielle C. B. Rodrigues, et al.
Neurology 2023;100;492-493 Published Online before print December 20, 2022
DOI 10.1212/WNL.0000000000201605

This information is current as of December 20, 2022

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/100/10/492.full
References	This article cites 2 articles, 2 of which you can access for free at: http://n.neurology.org/content/100/10/492.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Movement Disorders http://n.neurology.org/cgi/collection/all_movement_disorders Chorea http://n.neurology.org/cgi/collection/chorea Myoclonus http://n.neurology.org/cgi/collection/myoclonus
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 © 2022 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

