

# Teaching Video NeuroImages: Characteristic head jerks in congenital oculomotor apraxia due to Joubert syndrome

Friederike Borngräber, MD, Yangfan Peng, MD, Florian Ostendorf, MD, Andrea A. Kühn, MD, and Christos Ganos, MD

*Neurology*® 2019;93:e1125-e1126. doi:10.1212/WNL.0000000000008109

## Correspondence

Dr. Ganos  
christos.ganos@charite.de

## MORE ONLINE

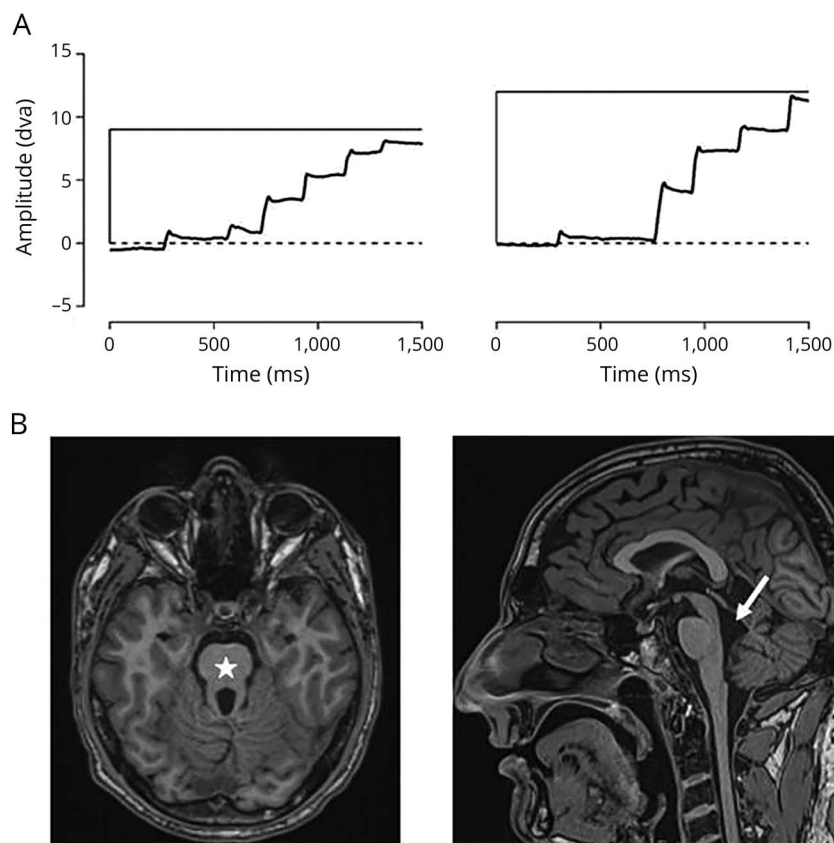
### → Teaching slides

[links.lww.com/WNL/A951](https://links.lww.com/WNL/A951)

### ▶ Video

A 29-year-old man presented in our clinic due to repetitive head jerks since his first year of life, which were previously diagnosed as tics. Examination revealed difficulties initiating voluntary saccades in the horizontal plane and disturbed hand–eye coordination with compensatory head thrusts (video and figure, A). The diagnosis of congenital oculomotor apraxia was made. Brain MRI revealed cerebellar vermis hypoplasia with elongated superior peduncles (“molar tooth sign”; figure, B) characteristic for Joubert syndrome, a rare autosomal-recessive ciliopathy with genetic

**Figure** Video-oculographic recording and T1-weighted transversal and sagittal brain MRI



(A) Video-oculographic recording of exemplary saccades to a visual target, jumping rightward from initial fixation to a peripheral position (9/12 degrees of visual angle [dva]) at time “0,” demonstrating prolonged latencies and hypometria of horizontal visually guided saccades. (B) T1-weighted transversal and sagittal brain MRI show hypoplasia of the cerebellar vermis with elongated superior cerebellar peduncles (molar tooth sign) (star) as well as enlargement of the fourth ventricle (arrow).

From the Movement Disorder and Neuromodulation Unit (F.B., Y.P., A.A.K., C.G.), Department of Neurology (F.O.), Charité-Universitätsmedizin Berlin; and Berlin Institute of Health (F.B.), Germany.

Go to [Neurology.org/N](https://Neurology.org/N) for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

heterogeneity.<sup>1</sup> Joubert syndrome is a common etiology of congenital oculomotor apraxia, a condition first described by Cogan,<sup>2</sup> who considered compensatory head jerks diagnostic.

### Author contributions

F. Borngräber: drafting/revising the manuscript, data acquisition, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, acquisition of data. Y. Peng: drafting/revising the manuscript, data acquisition, analysis or interpretation of data, accepts responsibility for conduct of research and final approval. F. Ostendorf: drafting/revising the manuscript, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, acquisition of data. A.A. Kühn: drafting/revising the manuscript, accepts responsibility for conduct of research and final approval, study supervision. C. Ganos: drafting/revising the manuscript, data acquisition, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval, study supervision.

### Acknowledgment

The authors thank the patient for participation and support.

### Study funding

F. Borngräber is supported by the BIH-Charité Clinician Scientist Program of the Charité-Universitätsmedizin Berlin and the Berlin Institute of Health. Y. Peng and F. Ostendorf report no funding. A.A. Kühn is a consultant to Boston Scientific and has received honoraria for speaking from Medtronic, Boston Scientific, and Abbott, as well as grants from Medtronic, all outside the submitted work. C. Ganos holds research grants from the VolkswagenStiftung (Freigeist Fellowship) and the German Parkinson Society and was also supported by the Deutsche Forschungsgemeinschaft (DFG; GA2031/1-1 and GA2031/1-2).

### Disclosure

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](http://Neurology.org/N) for full disclosures.

### References

1. Parisi M, Glass I. 1993–2019: Joubert syndrome. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. *GeneReviews*®. Seattle: University of Washington; 2003.
2. Cogan D. A type of congenital ocular motor apraxia presenting jerky head movements. *Am J Ophthalmol* 1953;36:433–441.

# Neurology®

## Teaching Video NeuroImages: Characteristic head jerks in congenital oculomotor apraxia due to Joubert syndrome

Friederike Borngräber, Yangfan Peng, Florian Ostendorf, et al.

*Neurology* 2019;93:e1125-e1126

DOI 10.1212/WNL.00000000000008109

**This information is current as of September 9, 2019**

<b>Updated Information &amp; Services</b>	including high resolution figures, can be found at: <a href="http://n.neurology.org/content/93/11/e1125.full">http://n.neurology.org/content/93/11/e1125.full</a>
<b>References</b>	This article cites 1 articles, 0 of which you can access for free at: <a href="http://n.neurology.org/content/93/11/e1125.full#ref-list-1">http://n.neurology.org/content/93/11/e1125.full#ref-list-1</a>
<b>Subspecialty Collections</b>	This article, along with others on similar topics, appears in the following collection(s): <b>All Education</b> <a href="http://n.neurology.org/cgi/collection/all_education">http://n.neurology.org/cgi/collection/all_education</a> <b>Clinical neurology examination</b> <a href="http://n.neurology.org/cgi/collection/clinical_neurology_examination">http://n.neurology.org/cgi/collection/clinical_neurology_examination</a>
<b>Permissions &amp; Licensing</b>	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: <a href="http://www.neurology.org/about/about_the_journal#permissions">http://www.neurology.org/about/about_the_journal#permissions</a>
<b>Reprints</b>	Information about ordering reprints can be found online: <a href="http://n.neurology.org/subscribers/advertise">http://n.neurology.org/subscribers/advertise</a>

*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 © 2019 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

