Teaching NeuroImages: Alternating ptosis and Marcus Gunn jaw- winking phenomenon with *PHOX2B* mutation

Anna Purna Basu, PhD
Paul Bellis, MBBS
Roger G. Whittaker, PhD
Michael C. McKean, MD
Anita M. Devlin, MD

Correspondence & reprint
requests to Dr. Basu:
anna.basu@ncl.ac.uk

Figure Marcus Gunn phenomenon and alternating ptosis

Left: waking state, not sucking: right ptosis. Right: waking state, sucking. Right-sided Marcus Gunn jaw-winking phenomenon (see video) and temporary left ptosis, which resolves when sucking ceases.

A 2-month-old baby presented with variable right-sided ptosis (without fatigability or maternal myasthenia) and recurrent unexplained central apneas since birth. On sucking, there was a right Marcus Gunn phenomenon with temporary left-sided ptosis (figure; video on the *Neurology*[®] Web site at www.neurology.org). MRI brain and EMG were normal. She was heterozygous for a polyalanine expansion mutation in the *PHOX2B* gene, indicating congenital central hypoventilation syndrome (CCHS).

The Marcus Gunn phenomenon, associated with congenital ptosis, is recognized in CCHS, probably reflecting congenital cranial dysinnervation from disruption of normal *PHOX2B* expression in the oculomotor nucleus.¹ Associated alternating ptosis has once been reported² and could be explained by Hering's law.

AUTHOR CONTRIBUTIONS

All coauthors were involved in clinical care or investigative workup of the patient. A.P.B. and P.B. wrote the article, to which the other authors added their comments.

ACKNOWLEDGMENT

The authors thanks Mr. Tang for assistance with video and photographic material and the family for their consent.

REFERENCES

1. Pattyn A, Morin X, Cremer H, Goridis C, Brunet JF. Expression and interactions of the two closely related homeobox genes *Phox2a* and *Phox2b* during neurogenesis. *Development* 1997;124:4065–4075.
2. Michel G, Villega F, Desprez P, Dollfus H, Speeg-Schatz C. [Ondine's curse and rare oculomotor abnormalities: a case report.] *J Fr Ophtalmol* 2006;29:422–425.

Supplemental data at
www.neurology.org

From Paediatric Neurology (A.P.B., A.M.D.), Paediatric Respiratory Medicine (P.B., M.C.M.), and Neurophysiology (R.G.W.), Royal Victoria Infirmary; and Institute of Neuroscience (A.P.B.), Newcastle University, Newcastle upon Tyne, UK.
The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

Neurology®

Teaching NeuroImages: Alternating ptosis and Marcus Gunn jaw-winking phenomenon with *PHOX2B* mutation

Anna Purna Basu, Paul Bellis, Roger G. Whittaker, et al.

Neurology 2012;79:e153

DOI 10.1212/WNL.0b013e3182703fe3

This information is current as of October 22, 2012

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 Copyright © 2012 by AAN Enterprises, Inc.. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/79/17/e153.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2012/10/21/79.17.e153.DC1
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/79/17/e153.full#ref-list-1
Citations	This article has been cited by 1 HighWire-hosted articles: http://n.neurology.org/content/79/17/e153.full##otherarticles
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Neuro-ophthalmology http://n.neurology.org/cgi/collection/all_neuroophthalmology All Pediatric http://n.neurology.org/cgi/collection/all_pediatric Clinical neurology examination http://n.neurology.org/cgi/collection/clinical_neurology_examination Eyelids http://n.neurology.org/cgi/collection/eyelids Neonatal http://n.neurology.org/cgi/collection/neonatal
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 Copyright © 2012 by AAN Enterprises, Inc.. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.