Teaching Video NeuroImage: Carbamazepine Improves Gait Initiation in Autosomal Recessive Myotonia Congenita

Yasuhiro Fuseya, MD, PhD, Nana Ishikawa, MD, Ryogen Sasaki, MD, PhD, and Hirofumi Yamashita, MD, PhD

Neurology® 2022;98:e328. doi:10.1212/WNL.00000000000013010

Correspondence

Dr. Yamashita
hirofumi@kuhp.kyoto-u.ac.jp

MORE ONLINE

Teaching slides

links.lww.com/WNL/B639

Videos

A 60-year-old man presented with muscle stiffness. He had tended to fall since childhood. His parents were first cousins, and his relatives had no symptoms. Examination showed myotonia in ocular, hand, and limb muscles, followed by improvement with repeated activity (warm-up phenomenon; Video 1). Cold exposure did not aggravate myotonia. His lower legs showed muscle hypertrophy. Genetic tests showed a homozygous p.M560T mutation in the *CLCN1* gene, which encodes skeletal muscle chloride channel 1, consistent with autosomal recessive myotonia congenita (Becker disease).¹ Although treatment with mexiletine or phenytoin did not improve his symptoms, carbamazepine (350 mg/d) did, especially the walking disturbance (Video 2). Although the warm-up phenomenon is sometimes also observed in sodium channel myotonia,² it is characteristic of myotonia congenita and an important clue for the diagnosis.

Study Funding

The authors report no targeted funding.

Disclosure

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

Appendix Authors

Name	Location	Contribution
Yasuhiro Fuseya, MD, PhD	Department of Molecular and Cellular Physiology, Kyoto University, Kyoto, Japan	Managed the patient and drafted the manuscript and video
Nana Ishikawa, MD	Akahoshi Neurology Clinic, Osaka, Japan	Managed the patient
Ryogen Sasaki MD, PhD	Department of Neurology, Kuwana City Medical Center, Mie, Japan	Performed a genetic test
Hirofumi Yamashita, MD, PhD	Department of Neurology, Japanese Red Cross Wakayama Medical Center, Wakayama, Japan	Managed the patient and drafted the manuscript and video

References

1. Zhang J, George AL, Jr., Griggs RC, et al. Mutations in the human skeletal muscle chloride channel gene (*CLCN1*) associated with dominant and recessive myotonia congenita. *Neurology*. 1996;47:993-998.
2. Trip J, Drost G, Ginjaar HB, et al. Redefining the clinical phenotypes of non-dystrophic myotonic syndromes. *J Neurol Neurosurg Psychiatry*. 2009;80:647-652.

From the Departments of Molecular and Cellular Physiology and Neurology (Y.F.), Graduate School of Medicine, Kyoto University Hospital; Akahoshi Neurology Clinic (N.I.), Osaka; Department of Neurology (R.S.), Kuwana City Medical Center, Mie; and Department of Neurology (H.Y.), Japanese Red Cross Wakayama Medical Center, Wakayama, Japan.

Go to 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.

Neurology®

Teaching Video NeuroImage: Carbamazepine Improves Gait Initiation in Autosomal Recessive Myotonia Congenita

Yasuhiro Fuseya, Nana Ishikawa, Ryogen Sasaki, et al.

Neurology 2022;98:e328 Published Online before print October 21, 2021

DOI 10.1212/WNL.0000000000013010

This information is current as of October 21, 2021

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/98/3/e328.full
References	This article cites 2 articles, 2 of which you can access for free at: http://n.neurology.org/content/98/3/e328.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Education http://n.neurology.org/cgi/collection/all_education All Movement Disorders http://n.neurology.org/cgi/collection/all_movement_disorders Clinical neurology examination http://n.neurology.org/cgi/collection/clinical_neurology_examination Ion channel gene defects http://n.neurology.org/cgi/collection/ion_channel_gene_defects Muscle disease http://n.neurology.org/cgi/collection/muscle_disease
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.