Teaching Video NeuroImages: Feeding dystonia in chorea-acanthocytosis

Martin Paucar, MD
Per-Åke Lindestad, MD,
PhD
Ruth H. Walker,
MBChB, PhD
Per Svenningsson, MD,
PhD

Correspondence to
Dr. Paucar:
martin.paucar-arce@karolinska.se

Figure 1 Around 10% acanthocytes (arrows) on a blood smear

A 39-year-old man was evaluated for personality change, involuntary movements, and eating difficulties. Examination demonstrated feeding dystonia, dysarthria, limb dystonia, and chorea (video on the *Neurology*[®] Web site at Neurology.org). Transaminases and creatine kinase levels were elevated. Additional investigation revealed acanthocytes on blood smear, myopathy, and caudate nucleus atrophy (figures 1, 2, e-1, and e-2). Western blot revealed absent chorein, and a genetic test found him to be compound heterozygote for novel *VPS13A* gene mutations (c.266dupT and deletion of exons 52, 53, 55, and 58), establishing a diagnosis of chorea-acanthocytosis.¹ He was treated with botulinum toxin injections in genioglossus, which significantly improved eating and speaking.

AUTHOR CONTRIBUTIONS

Dr. M. Paucar and Dr. P.-Å. Lindestad: study concept, data collection, and writing of the manuscript. Dr. R.H. Walker and Prof. P. Svenningsson: study concept and editing of the manuscript.

ACKNOWLEDGMENT

Western blot analysis for chorein was performed by G. Kwiatkowski and Dr. Benedikt Bader with the financial support of the Advocacy for Neuroacanthocytosis Patients and of the ERA-net E-Rare consortium EMINA (European Multidisciplinary Initiative on Neuroacanthocytosis; BMBF 01GM1003) in the labs of Profs. Hans Kretschmar/Armin Giese (Neuropathology) and Adrian Danek (Neurology) at Ludwig-Maximilians-Universität, Munich, Germany.

STUDY FUNDING

Stockholm County Council.

DISCLOSURE

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

REFERENCE

1. Schneider SA, Aggarwal A, Bhatt M, et al. Severe tongue protrusion dystonia: clinical syndromes and possible treatment. *Neurology* 2006;67:940–943.

Supplemental data
at Neurology.org

Download teaching slides:
Neurology.org

From the Departments of Neurology (M.P., P.S.) and Otolaryngology (P.-Å.L.), Karolinska University Hospital; Department of Clinical Neuroscience (N.P., P.S.), Karolinska Institutet, Stockholm, Sweden; Department of Neurology (R.H.W.), James J. Peters Veterans Medical Affairs Center, Bronx; and Department of Neurology (R.H.W.), Mount Sinai School of Medicine, New York, NY.

Figure 2 Gomori staining of left tibialis anterior muscle displaying variation in fiber size, central nuclei (thin arrows), and splitting (thicker arrow). These features are compatible with myopathy

Neurology®

Teaching Video *NeuroImages*: Feeding dystonia in chorea-acanthocytosis

Martin Paucar, Per-Åke Lindestad, Ruth H. Walker, et al.

Neurology 2015;85:e143-e144

DOI 10.1212/WNL.0000000000002108

This information is current as of November 9, 2015

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/85/19/e143.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2015/11/07/WNL.0000000000002108.DC2 http://n.neurology.org/content/suppl/2015/11/07/WNL.0000000000002108.DC1 http://n.neurology.org/content/suppl/2015/11/07/WNL.0000000000002108.DC3
References	This article cites 1 articles, 1 of which you can access for free at: http://n.neurology.org/content/85/19/e143.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 Basal ganglia http://n.neurology.org/cgi/collection/basal_ganglia Botulinum toxin http://n.neurology.org/cgi/collection/botulinum_toxin
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 © 2015 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.