Teaching Video NeurolImages: Rippling muscle disease with caveolin myopathy

Pierre R. Bourque, MD, Ari Breiner, MD, John Brooks, and Jodi Warman Chardon, MD

Neurology® 2018;91:e1726-e1727. doi:10.1212/WNL.0000000000006427

Correspondence

Dr. Bourque
pbourque@toh.ca

Figure Moderate hypertrophy of the quadriceps, but normal muscle bulk otherwise

A 43-year-old man described adolescence-onset abnormal muscle contractions. On examination, there was hypertrophy (figure) with normal power of thigh muscles but mild weakness of distal leg muscles (Medical Research Council grade 4+/5). Muscle percussion and stretching induced electrically silent slow rolling muscle contractions (video 1). He had mildly elevated creatine kinase (600 U/L). Family history was consistent with autosomal dominant rippling

MORE ONLINE

▶ **Video**

→ **Teaching slides**

links.lww.com/WNL/A723

From the Department of Medicine, Division of Neurology (P.R.B., A.B., J.B., J.W.C.), The Ottawa Hospital; The Ottawa Hospital Research Institute (P.R.B., A.B., J.W.C.); and Department of Genetics (P.R.B., A.B., J.W.C.), Children's Hospital of Eastern Ontario, Ottawa, Canada.

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.

muscle disease.¹ A heterozygous variant was documented in coding exons of the caveolin 3 gene (CAV c. 83A>T; p.Asp28Val).

In addition to RMD, allelic caveolin disorders include benign hyperCKemia, distal myopathy, limb girdle muscular dystrophy 1C, and isolated hypertrophic cardiomyopathy.²

Author contributions

P.R. Bourque: clinical data acquisition, manuscript preparation. A. Breiner: critical review of manuscript. J. Brooks: video

editing, manuscript review. J. Warman Chardon: genetic analysis, critical review of manuscript.

Study funding

No targeted funding reported.

Disclosure

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

References

1. Torbergson T. Rippling muscle disease: a review. *Muscle Nerve* 2002; 11:S103–S107.
2. Galbiati F, Minetti C, Lisanti MP. Caveolinopathies: mutations in caveolin-3 cause four distinct autosomal dominant muscle diseases. *Neurology* 2004;62:538–543.

Neurology®

Teaching Video NeuroImages: Rippling muscle disease with caveolin myopathy

Pierre R. Bourque, Ari Breiner, John Brooks, et al.

Neurology 2018;91:e1726-e1727

DOI 10.1212/WNL.0000000000006427

This information is current as of October 29, 2018

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/91/18/e1726.full
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/91/18/e1726.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Genetics http://n.neurology.org/cgi/collection/all_genetics Clinical neurology examination http://n.neurology.org/cgi/collection/clinical_neurology_examination 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 © 2018 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.