


ARTICLES

- e110 Novel *HSPB1* mutation causes both motor neuropathy and distal myopathy
D.J. Lewis-Smith, J. Duff, A. Pyle, H. Griffin, T. Polvikoski, D. Birchall, R. Horvath, and P.F. Chinnery
[OPEN](#)
- e114 Heterozygous mutations in *HSD17B4* cause juvenile peroxisomal D-bifunctional protein deficiency
D.J. Amor, A.P.L. Marsh, E. Storey, R. Tankard, G. Gillies, M.B. Delatycki, K. Pope, C. Bromhead, R.J. Leventer, M. Bahlo, and P.J. Lockhart
[OPEN](#)
- e113 Peripheral neuropathy in patients with CPEO associated with single and multiple mtDNA deletions
D. Lehmann, M.E. Kornhuber, C. Clajus, C.L. Alston, A. Wienke, M. Deschauer, R.W. Taylor, and S. Zierz
[OPEN](#)
- e112 Muscle involvement in limb-girdle muscular dystrophy with GMPPB deficiency (LGMD2T)
S.T. Oestergaard, T. Stojkovic, J.R. Dahlqvist, C. Bouchet-Seraphin, J. Nectoux, F. Leturcq, M. Cossée, G. Solé, C. Thomsen, T.O. Krag, and J. Vissing
[OPEN](#)
- e116 *SORL1* mutations in early- and late-onset Alzheimer disease
M.L. Cuccaro, R.M. Carney, Y. Zhang, C. Bohm, B.W. Kunkle, B.N. Vardarajan, P.L. Whitehead, H.N. Cukier, R. Mayeux, P. St. George-Hyslop, and M.A. Pericak-Vance
[OPEN](#)

- e118 Germline and somatic mutations in the *MTOR* gene in focal cortical dysplasia and epilepsy
R.S. Møller, S. Weckhuysen, M. Chipaux, E. Marsan, V. Taly, E.M. Bebin, S.M. Hiatt, J.W. Prokop, K.M. Bowling, D. Mei, V. Conti, P. de la Grange, S. Ferrand-Sorbets, G. Dorfmueller, V. Lambrecq, L.H.G. Larsen, E. Leguern, R. Guerrini, G. Rubboli, G.M. Cooper, and S. Baulac
[OPEN](#)
- e120 De novo *FGF12* mutation in 2 patients with neonatal-onset epilepsy
I. Guella, L. Huh, M.B. McKenzie, E.B. Toyota, E.M. Bebin, M.L. Thompson, G.M. Cooper, D.M. Evans, S.E. Buerki, S. Adam, M.I. Van Allen, T.N. Nelson, M.B. Connolly, M.J. Farrer, and M. Demos
[OPEN](#) 

CLINICAL/SCIENTIFIC NOTES




- e115 FHF1 (FGF12) epileptic encephalopathy
S. Al-Mehmadi, M. Splitt, for DDD Study group, V. Ramesh, S. DeBrosse, K. Dessoify, F. Xia, Y. Yang, J.A. Rosenfeld, P. Cossette, J.L. Michaud, F.F. Hamdan, P.M. Campeau, and B.A. Minassian, for CENet Study group
[OPEN](#)
- e109 Epidermolysis bullosa simplex with muscular dystrophy associated with *PLEC* deletion mutation
V.C. Alvarez, S.T. Penttilä, V.L. Salutto, B. Udd, and C.G. Mazia
[OPEN](#)
- e117 Copy number analysis reveals a novel multiexon deletion of the *COLQ* gene in congenital myasthenia
W. Wang, Y. Wu, C. Wang, J. Jiao, and C.J. Klein
[OPEN](#)

e119 Phenotypic convergence of Menkes and Wilson disease
B. Bansagi, D. Lewis-Smith, E. Pal, J. Duff, H. Griffin, A. Pyle, J.S. Müller, G. Rudas, Z. Aranyi, H. Lochmüller, P.F. Chinnery, and R. Horvath

[OPEN](#)

e121 Mitochondrial cytopathy with common MELAS mutation presenting as multiple system atrophy mimic
A.-K. Pröbstel, A. Schaller, J. Lieb, J. Hench, S. Frank, P. Fuhr, L. Kappos, and M. Sinnreich

[OPEN](#)

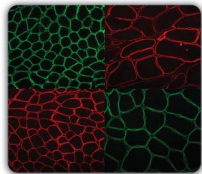
 Podcast  Video  LOE classification  LOE recommendation



tinyurl.com/NeurologyNG



twitter.com/GreenJournal



Cover image: Merosin (clone 22B2) expression (green) and VIA4-specific glycosylation (red) on immunofluorescence-stained muscle sections. Stylized by Kaitlyn Aman Ramm, *Neurology*[®] Editorial Assistant. See “Muscle involvement in limb-girdle muscular dystrophy with GMPPB deficiency (LGMD2T)”.

Neurology[®] Genetics

Neurology.org/ng • Online ISSN: 2376-7839
Volume 2, Number 6, December 2016



VISION: *Neurology[®] Genetics* will be the premier peer-reviewed journal in the field of neurogenetics.

MISSION: *Neurology: Genetics* will provide neurologists with outstanding original contributions that elucidate the role of genetic and epigenetic variation in diseases and biological traits of the central and peripheral nervous systems.

Editor

Neurology: Genetics

Stefan M. Pulst, MD, Dr med, FAAN
Professor and Chair, Department of Neurology
University of Utah
Salt Lake City, UT
ngjournal@neurology.org
Specialties: Genetics, movement disorders

Editor-in-Chief

Neurology[®]

Robert A. Gross, MD, PhD, FAAN
Professor of Neurology and of Pharmacology and Physiology
Strong Epilepsy Center
University of Rochester Medical Center
Rochester, NY
journal@neurology.org

Editorial Inquiries

Tel: 612-928-6400
Toll-free: 800-957-3182 (US)
Fax: 612-454-2748
ngjournal@neurology.org

Academy Officers

Terrence L. Cascino, MD, FAAN, President
Ralph L. Sacco, MD, MS, FAAN, President Elect
James C. Stevens, MD, FAAN, Vice President
Aaron E. Miller, MD, FAAN, Secretary
Lisa M. Shulman, MD, FAAN, Treasurer
Timothy A. Pedley, MD, FAAN, Past President

Executive Office, American Academy of Neurology

Catherine M. Rydell, CAE, Executive Director/CEO
201 Chicago Ave
Minneapolis, MN 55415
Tel: 612-928-6100

Editorial Office

Patricia K. Baskin, MS, Executive Editor
Kathleen M. Pieper, Managing Editor, *Neurology*
Lee Ann Kleffman, Managing Editor, *Neurology: Genetics*
Sharon L. Quimby, Managing Editor, *Neurology[®] Clinical Practice*
Morgan S. Sorenson, Managing Editor, *Neurology[®] Neuroimmunology & Neuroinflammation*
Cynthia S. Abair, MA, Senior Graphics Editor
Robert J. Witherow, Senior Editorial Associate
Karen Skaja, Senior Editorial Associate
Andrea R. Rahkola, Editorial Associate
Kaitlyn Aman Ramm, Editorial Assistant
Kristen Swendsrud, Editorial Assistant

Publisher

Wolters Kluwer
Baltimore, MD

Publishing Staff

Kim Jansen, Executive Publisher
Alexandra Lazerow, Production Team Leader, AAN Journals
Steve Rose, Editorial Assistant
Stacy Drossner, Production Associate

Neurology[®] Genetics

Neurology.org/ng • Online ISSN: 2376-7839
Volume 2, Number 6, December 2016



Editor

Neurology[®] Genetics

Stefan M. Pulst, MD, Dr med, FAAN
Professor and Chair, Department of Neurology
University of Utah
Salt Lake City, UT
ngjournal@neurology.org
Specialties: Genetics, movement disorders

Editor-in-Chief

Neurology[®]

Robert A. Gross, MD, PhD, FAAN
Professor of Neurology and of Pharmacology and Physiology
Strong Epilepsy Center
University of Rochester Medical Center
Rochester, NY
journal@neurology.org

Deputy Editors

Neurology

Bradford B. Worrall, MD, MSc, FAAN
Professor of Neurology and Public Health Sciences
University of Virginia
Charlottesville, VA

Neurology: Genetics

Nicholas Elwood Johnson, MD
University of Utah
Salt Lake City, UT
Specialties: Muscle diseases, patient-centered outcomes

Associate Editors

Neurology: Genetics

Alexandra Durr, MD, PhD
Hôpital de la Salpêtrière
Paris, France
Specialties: Spastic paraplegia, Huntington disease, ataxia

Massimo Pandolfo, MD, FAAN
Hôpital Erasme
Université Libre de Bruxelles
Brussels, Belgium
Specialties: Epilepsy, Friedreich ataxia, stem cells

Raymond P. Roos, MD, FAAN
University of Chicago Medical Center
Chicago, IL
Specialties: Motor neuron disease, prion disease, neurovirology/neuroimmunology

Jeffery M. Vance, MD, PhD
University of Miami
Miami, FL
Specialties: Parkinson, Alzheimer disease, inherited neuropathies

Biostatistics

Richard J. Kryscio, PhD
University of Kentucky
Lexington, KY

Christopher A. Beck, PhD
University of Rochester
Rochester, NY

Sue Leurgans, PhD
Rush University Medical Center
Chicago, IL

Level of Evidence Evaluations

Gary S. Gronseth, MD, FAAN
University of Kansas
Kansas City, KS

Podcasts

Ted M. Burns, MD
University of Virginia
Charlottesville, VA

Andrew M. Southerland, MD, MSc, Deputy Podcast Editor
University of Virginia
Charlottesville, VA

Ombudsman

David A. Drachman, MD, FAAN
University of Massachusetts Medical School
Worcester, MA

Scientific Integrity Advisor

Robert B. Daroff, MD, FAAN
Case Western Reserve University
Cleveland, OH

Level of Evidence Review Team

Melissa J. Armstrong, MD, Gainesville, FL
Richard L. Barbano, MD, PhD, FAAN, Rochester, NY
Richard M. Dubinsky, MD, MPH, FAAN, Kansas City, KS
Jeffrey J. Fletcher, MD, MSc, Ann Arbor, MI
Gary M. Franklin, MD, MPH, FAAN, Seattle, WA
David S. Gloss II, MD, MPH&TM, Charleston, WV
John J. Halperin, MD, FAAN, Summit, NJ
Jason Lazarou, MSc, MD, Toronto, Ontario, Canada
Steven R. Messé, MD, FAAN, Philadelphia, PA
Pushpa Narayanaswami, MBBS, DM, FAAN, Boston, MA
Alex Rae-Grant, MD, Cleveland, OH

Publication Information

Neurology[®] is a registered trademark of the American Academy of Neurology (registration valid in the United States).

Neurology[®] Genetics (eISSN 2376-7839) is an open access journal published online for the American Academy of Neurology, 201 Chicago Ave, Minneapolis, MN 55415, by Wolters Kluwer, 16522 Hunters Green Parkway, Hagerstown, MD 21740-2116. Business offices are located at Two Commerce Square, 2001 Market St., Philadelphia, PA 19103. Production offices are located at 351 West Camden Street, Baltimore, MD 21201-2436. © 2016 American Academy of Neurology.

Neurology[®] Genetics is an official journal of the American Academy of Neurology. Journal website: Neurology.org/ng, AAN website: AAN.com

Copyright and Permission Information: Please go to the journal website (www.neurology.org/ng) and click the “©Request Permissions” icon for the relevant article. Alternatively, send an email to healthpermissions@wolterskluwer.com. General information about permissions can be found here: http://www.lww.com/webapp/wcs/stores/servlet/content_resources_permissions-journals_11851_-1_12551.

Translation Rights & Licensing queries: Silvia Serra, Global Director of Rights, Licensing, Permissions, Wolters Kluwer Citi Building, 25 Canada Square, E14 5LB, England, UK. tel: 144-(0)203-1976643; translationrights@wolterskluwer.com.

Disclaimer: Opinions expressed by the authors and advertisers are not necessarily those of the American Academy of Neurology, its affiliates, or of the Publisher. The American Academy of Neurology, its affiliates, and the Publisher disclaim any liability to any party for the accuracy, completeness, efficacy, or availability of the material contained in this publication (including drug dosages) or for any damages arising out of the use or non-use of any of the material contained in this publication.

Advertising Sales Representatives: Wolters Kluwer, 333 Seventh Avenue, New York, NY 10001. Contacts: Eileen Henry, tel: 732-778-2261, fax: 973-215-2485, eileen.henry@wolterskluwer.com and Hilary Druker, cell: 609-304-9187, fax: 215-754-4912, hilary.druker@wolterskluwer.com. In Europe: Avia Potashnik, Wolters Kluwer, tel: +44 207 981 0722; +44 7919 397 933 or e-mail: avia.potashnik@wolterskluwer.com.

Careers & Events: Monique McLaughlin, Wolters Kluwer, Two Commerce Square, 2001 Market Street, Philadelphia, PA 19103, tel: 215-521-8468, fax: 215-521-8801; monique.mclaughlin@wolterskluwer.com.

Reprints: Meredith Edelman, Commercial Reprint Sales, Wolters Kluwer, Two Commerce Square, 2001 Market Street, Philadelphia, PA 19103, tel: 215-555-1212 (office), 215-356-2721 (mobile); meredith.edelman@wolterskluwer.com; reprintsolutions@wolterskluwer.com.

Special projects: US & Canada: Alan Moore, Wolters Kluwer, Two Commerce Square, 2001 Market Street, Philadelphia, PA 19103, tel: 215-521-8638, alan.moore@wolterskluwer.com. International: Silvia Serra, Global Director of Rights, Licensing, Permissions, Wolters Kluwer Citi Building, 25 Canada Square, E14 5LB, England, UK. tel: 144-(0)203-1976643; translationrights@wolterskluwer.com.

Neurology[®] Genetics

2 (6)
Neurol Genet 2016;2;

This information is current as of January 1, 2016

Updated Information & Services	including high resolution figures, can be found at: http://ng.neurology.org/content/2/6.full.html
Permissions & Licensing	Information about reproducing this article in parts (figures,tables) or in its entirety can be found online at: http://ng.neurology.org/misc/about.xhtml#permissions
Reprints	Information about ordering reprints can be found online: http://ng.neurology.org/misc/addir.xhtml#reprintsus

Neurol Genet is an official journal of the American Academy of Neurology. Published since April 2015, it is an open-access, online-only, continuous publication journal. Copyright . All rights reserved. Online ISSN: 2376-7839.

