Neutral lipid-storage disease with myopathy and Jordan anomaly

Riccardo Zuccarino, MD, Daniel M. Anderson, MD, Carol Holman, MD, Shawna Feely, MS, Laurie Gutmann, MD, and Ludwig Gutmann, MD

Neurology® 2020;95:599-600. doi:10.1212/WNL.0000000000010585

Correspondence

Dr. Zuccarino
riccardozuccarino@yahoo.it

Figure Unstained vacuoles with Giemsa and stained vacuoles with oil red O

Jordan anomaly can be seen on a peripheral blood smear using the Giemsa method (A) or oil red O (B) staining showing lipid accumulation of neutral triglycerides in vacuoles in leukocytes (pictures were both taken at 100×). All patients reported with neutral lipid storage diseases have this finding. The same is seen in myocytes.

A 58-year-old man presented with slowly progressive weakness involving proximal arm, distal leg, and finger extensor muscles. His history included embolic stroke, cardiomyopathy, diabetes, and triglyceridemia. Family history of myopathy was absent, but consanguinity was present. Muscle biopsy and carnitine/acylcarnitine testing were deferred. Heterozygous mutation in *GNE* p.V727L (VUS) and homozygous mutations in *PNPLA2* p.R221P (pathogenic) were present. *PNPLA2* mutations cause neutral lipid storage disease with myopathy (NLSDM¹) and associated cardiomyopathy, metabolic disorders, and leucocyte vacuoles (figure). Jordan anomaly² (leukocytic triglyceride containing vacuoles) on peripheral blood smear confirmed NLSDM. This is a unique abnormality documenting the pathologic basis for NLSDM.

Study funding

No targeted funding reported.

Disclosure

The authors report no disclosures relevant to the manuscript. Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

From the Departments of Neurology (R.Z., D.M.A., S.F., Laurie Gutmann, Ludwig Gutmann) and Pathology (C.H.), University of Iowa Hospitals and Clinics, Iowa City; and Neuro-muscular Omnicentre (NEMO)-Fondazione Serena Onlus (R.Z.), Arenzano, Genoa, Italy.

Go to [Neurology.org/N](https://www.neurology.org/N) for full disclosures.

Appendix Authors

Name	Location	Contributions
Riccardo Zuccarino, MD	University of Iowa, Iowa City; Centro Clinico NEMO, Genoa, Italy	Study concept and design, analysis and interpretation of data, drafting and critical revision of manuscript for intellectual content
Daniel M. Anderson, MD	University of Iowa, Iowa City; Mayo Clinic Health System–Franciscan Healthcare, La Crosse, WI	Study concept and design, analysis and interpretation of data, drafting and critical revision of manuscript for intellectual content
Carol Holman, MD	University of Iowa, Iowa City	Providing images, critical revision of manuscript for intellectual content
Shawna Feely, MS	University of Iowa, Iowa City	Interpretation of data, critical revision of manuscript for intellectual content

Appendix (continued)

Name	Location	Contributions
Laurie Gutmann, MD	University of Iowa, Iowa City	Study concept and design, analysis and interpretation of data, drafting and critical revision of manuscript for intellectual content
Ludwig Gutmann, MD	University of Iowa, Iowa City	Study concept and design, analysis and interpretation of data, drafting and critical revision of manuscript for intellectual content

References

1. Pennisi EM, Arca M, Bertini E, et al. Neutral lipid storage diseases: clinical/genetic features and natural history in a large cohort of Italian patients. *Orphanet J Rare Dis* 2017;12:90.
2. Jordan CH. The familial occurrence of fat containing vacuoles in the leukocytes diagnosed in two brothers suffering from dystrophia musculorum progressiva. *Acta Med Scand* 1953;145:419–423.

Call for Voices: Lived Experiences

The Editors of the *Neurology* specialty site Equity, Diversity, & Inclusion encourage you to submit short first-person accounts (1,000 words or less) of experiences lived within the realm of equity, diversity, and inclusion (EDI) with the goal of informing and enlightening our community on these critical issues. Some topics to consider include, but are not limited to:

- Descriptions of personal experiences that shaped your views of EDI.
- Reflections on the intersection between personal identity and career.
- Discussions at the intersection of EDI and neurology patient care, research, education, advocacy, or policy.

Submit your contributions to journal@neurology.org and include “Voices Submission” in the subject line.

Visit the *Neurology*® Website at Neurology.org/N

- More article-based content on home pages
- Streamlined menus and navigation
- Enhanced blog sections for specialty areas
- Same experience on desktop, tablet, and mobile devices
- Improved article reading experience; links more evident (pdf, analytics, social media)
- *Neurology*® *Clinical Practice* initiative “Practice Current” global surveys will be accessible across sites

Find *Neurology*® on Facebook: <http://tinyurl.com/neurologyfan>

Follow *Neurology*® on Twitter: <https://twitter.com/GreenJournal>

Neurology®

Neutral lipid-storage disease with myopathy and Jordan anomaly

Riccardo Zuccarino, Daniel M. Anderson, Carol Holman, et al.

Neurology 2020;95:599-600 Published Online before print August 5, 2020

DOI 10.1212/WNL.0000000000010585

This information is current as of August 5, 2020

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/95/13/599.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/95/13/599.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 All Neuromuscular Disease http://n.neurology.org/cgi/collection/all_neuromuscular_disease 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 © 2020 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.