Fatty acid 2-hydroxylase deficiency

Clinical features and brain iron accumulation

Figure Brain MRI discloses white matter changes and iron accumulation

Axial fluid-attenuated inversion recovery (FLAIR)-weighted brain MRI shows confluent periventricular hyperintense signal and deep white matter changes (A, B) (arrows); axial FLAIR and T2 spin-echo images disclose symmetric hyperintense signal in the posterior internal capsules and optic radiations and hypointense globus pallidi consistent with iron accumulation (C, D) (arrows).

A 21-year-old woman presented with progressive spastic paraplegia, dysarthria, and strabismus since 7 years of age (video on the *Neurology*[®] Web site at Neurology.org). Brain MRI disclosed white matter changes and iron accumulation (figure). Whole exome sequencing detected in fatty acid 2-hydroxylase (*FA2H*) gene 2 variants never reported: c.169_170insGCGGGCCAGG (p.Asp57Gly*66), leading, if translated, to a truncated protein, and c.117C>A (p.Phe39Leu), predicted by computational algorithms to be deleterious.

FA2H deficiency is responsible for SPG35, a rare autosomal recessive complicated hereditary spastic paraplegia.^{1,2} Strabismus, dysarthria, and spastic paraplegia with brain MRI showing iron accumulation and white matter changes are common in SPG35 and may suggest the diagnosis.^{1,2} Molecular analysis is necessary to confirm this unusual condition.

Supplemental data at Neurology.org

José Luiz Pedroso, MD, PhD, Benjamin W. Handfas, MD, Agessandro Abrahão, MD, Fernando Kok, MD, PhD, Orlando G.P. Barsottini, MD, PhD, Acary S. Bulle Oliveira, MD, PhD

From the Universidade Federal de São Paulo (J.L.P., A.A., O.G.P.B., A.S.B.O.); Hospital Israelita Albert Einstein (B.W.H.); and Mendelics Genomic Analysis (F.K.), São Paulo, Brazil.

Author contributions: J.L. Pedroso: case report conception, organization, and execution, manuscript review and critique. B.W. Handfas: case report organization and execution, manuscript review and critique. A. Abrahão: case report organization and execution, manuscript review and critique. F. Kok: genetic evaluation, manuscript review and critique. O.G. Barsottini: case report conception, organization,

and execution, writing of the first draft, manuscript review and critique. A.S.B. Oliveira: case report conception, organization, and execution, manuscript review and critique.

Study funding: No targeted funding reported.

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

Correspondence to Dr. Pedroso: jlpedroso.neuro@gmail.com

1. Dick KJ, Al-Mjeni R, Baskir W, et al. A novel locus for an autosomal recessive hereditary spastic paraplegia (SPG35) maps to 16q21-q23. *Neurology* 2008;71:248–252.
2. Dick KJ, Eckhardt M, Paisán-Ruiz C, et al. Mutation of *FA2H* underlies a complicated form of hereditary spastic paraplegia (SPG35). *Hum Mutat* 2010;31:E1251–E1260.

Enjoy Big Savings on NEW 2015 AAN Practice Management Webinar Subscriptions

The American Academy of Neurology offers 14 cost-effective Practice Management Webinars which you can attend live or listen to recordings posted online. AAN members can purchase one webinar for \$149 or subscribe to the entire series for only \$199—a big savings from the 2015 nonmember price of \$199 per webinar or \$649 for the subscription. Register today for upcoming webinars, access recorded webinars, and see the rest of the 2015 schedule at AAN.com/view/pmw15:

- January 20: Now is the Time: Getting Paid for Chronic Care Coordination
- February 10: Coding for Neurodiagnostic Procedures Made Easy
- March 3: Case Studies: Neurologists Succeeding in New Health Care Models
- March 24: Improving Your Referral Network

Introducing Stackly

Organize. Disseminate. Collaborate. Discover.

With simple tools for collecting, organizing, and sharing research materials, Stackly is the new center of your research world.

- Share what you find and discover what others share
- Collaborate with colleagues on topics that matter to you most

Stackly will integrate with your web browser and work directly with all Neurology® Journals as well as other publication sites. Access Stackly on the home page and to the right of each article. Visit <http://www.neurology.org/site/includefiles/homepage/stackly.xhtml> to learn more and start stacking for FREE.

Neurology®

Fatty acid 2-hydroxylase deficiency: Clinical features and brain iron accumulation

José Luiz Pedroso, Benjamin W. Handfas, Agessandro Abrahão, et al.

Neurology 2015;84;960-961

DOI 10.1212/WNL.0000000000001316

This information is current as of March 2, 2015

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/84/9/960.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2015/02/28/WNL.0000000000001316.DC1
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/84/9/960.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 MRI http://n.neurology.org/cgi/collection/mri Spastic paraplegia http://n.neurology.org/cgi/collection/spastic_paraplegia
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.