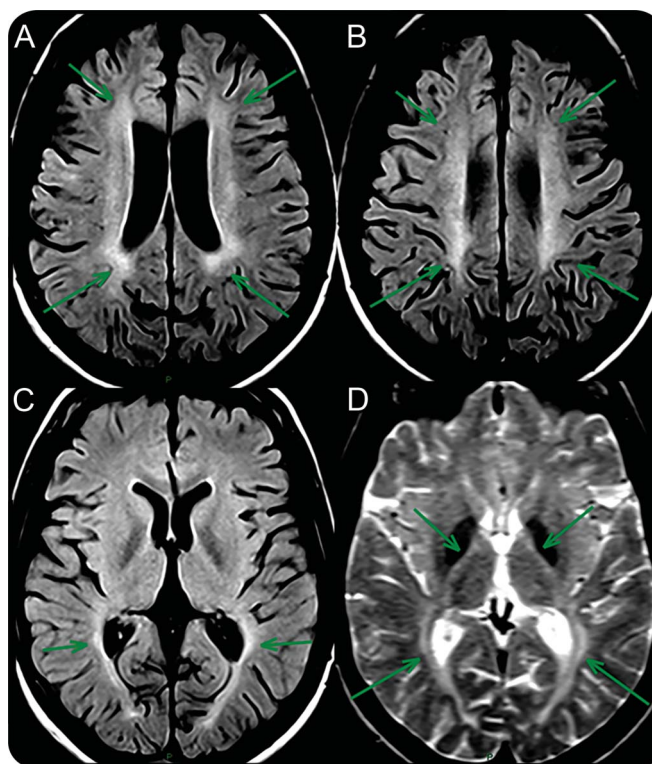


Fatty acid 2-hydroxylase deficiency

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.Asp57Glyfs*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.

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,

Supplemental data
at Neurology.org

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. Pedrosa: jlpedroso.neuro@gmail.com

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Neurology 2015;84;960-961

DOI 10.1212/WNL.0000000000001316

This information is current as of March 2, 2015

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