Extensive striatal, cortical, and white matter brain MRI abnormalities in Wilson disease

A 16-year-old boy presented with progressive dysarthria and gait and behavior disorders. The diagnosis of Wilson disease was made, based on Kayser-Fleischer rings, hypocupremia, hypoceruloplasminemia, and increased 24-hour urinary copper, and confirmed by molecular analysis (homozygous state, p.[Glu1382*]; [Glu1382*]). Brain MRI demonstrated diffuse bilateral cortical and subcortical abnormalities (figure). Chelator therapy (D-penicillamine) produced partial improvement, although the patient developed epileptic seizures, presumably due to the cortical involvement. Wilson disease with extensive cortical-subcortical lesions is rare, but should be considered as a possible etiology of diffuse leukoencephalopathy with cystic evolution.

Jean-Marc Trocello, MD, PhD, France Woimant, MD, Souleiman El Balkhi, PhD, Jean-Pierre Guichard, MD, Joel Poupon, PhD, Philippe Chappuis, PhD, Francois Feillet, MD, PhD

From the French National Reference Centre for Wilson’s Disease (J.-M.T., F.W.), Biological Toxicology Laboratory (S.E.B., J.P.), Neuroradiology (J.-P.G.), and Laboratoire de Biochimie et Biologie Moléculaire (P.C.), AP-HP, Hôpital Lariboisière, Paris; and National Reference Centre for Inborn Errors of Metabolism (F.F.), INSERM U954, Nancy, France.

Author contributions: Dr. Trocello: design, conceptualization, drafting the manuscript. Dr. Woimant: design, conceptualization, drafting the manuscript. Dr. El Balkhi: interpretation of data, revising the manuscript. Dr. Guichard: interpretation of data, revising the manuscript. Dr. Poupon: interpretation of data, revising the manuscript. Dr. Chappuis: interpretation of data, revising the manuscript. Dr. Feillet: conceptualization and revising the manuscript.

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. Trocello: jean-marc.trocello@hrl.aphp.fr


Extensive striatal, cortical, and white matter brain MRI abnormalities in Wilson disease

Jean-Marc Trocello, France Woirmant, Souleiman El Balkhi, et al.

*Neurology* 2013;81;1557

DOI 10.1212/WNL.0b013e3182a95883

This information is current as of October 21, 2013

- **Updated Information & Services**: including high resolution figures, can be found at: http://n.neurology.org/content/81/17/1557.full
- **References**: This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/81/17/1557.full#ref-list-1
- **Citations**: This article has been cited by 3 HighWire-hosted articles: http://n.neurology.org/content/81/17/1557.full##otherarticles
- **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 Movement Disorders http://n.neurology.org/cgi/collection/all_movement_disorders
  - Gait disorders/ataxia http://n.neurology.org/cgi/collection/gait_disorders_ataxia
  - Metabolic disease (inherited) http://n.neurology.org/cgi/collection/metabolic_disease_inherited
  - MRI http://n.neurology.org/cgi/collection/mri
- **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 © 2013 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.