Teaching NeuroImages: Rapidly progressive leukoencephalopathy in mitochondrial complex I deficiency

Fabian Baertling, MD
Jörg Schaper, MD
Ertan Mayatepek, MD
Felix Distelmaier, MD

Correspondence to
Dr. Distelmaier:
Felix.Distelmaier@med.uni-
duesseldorf.de

Figure Brain imaging in *NDUFS1* mitochondrial complex I mutation

(A) Axial T2-weighted MRI of the brain demonstrates bilateral cystic white matter lesions in the centrum semiovale. (B) Corresponding sagittal T1-weighted MRI. (C, D) Follow-up MRI after 3 months reveals confluent demyelination of the entire supratentorial white matter.

At age 8 months, an infant girl displayed rapid developmental regression. Family history, birth, and initial development were unremarkable. After hospital admission, cerebral MRI showed bilateral cystic lesions in the centrum semiovale. Follow-up imaging after 3 months demonstrated a dramatic progression in these alterations with demyelination of the supratentorial white matter (figure). Biochemical and genetic analyses confirmed isolated mitochondrial complex I deficiency due to an *NDUFS1* mutation (encoding NADH-dehydrogenase-ubiquinone Fe-S protein 1; see also reference 1, patient 1). Of note, leukoencephalopathy

is uncommon in mitochondrial complex I mutations but may be a feature of *NDUFS1* defects.²

AUTHOR CONTRIBUTIONS

Fabian Baertling: drafting and revising the manuscript for intellectual content. Jörg Schaper: drafting and revising the manuscript for intellectual content. Ertan Mayatepek: drafting and revising the manuscript for intellectual content. Felix Distelmaier: drafting and revising the manuscript for intellectual content.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

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

Download teaching slides:
www.neurology.org

From the Department of General Pediatrics, Neonatology and Pediatric Cardiology, University Children's Hospital (F.B., E.M., F.D.), and Department of Diagnostic and Interventional Radiology (J.S.), Heinrich-Heine-University, Düsseldorf, Germany.

REFERENCES

1. Distelmaier F, Koopman WJ, van den Heuvel LP, et al. Mitochondrial complex I deficiency: from organelle dysfunction to clinical disease. *Brain* 2009;132:833–842.
2. Lebre AS, Rio M, Faivre d'Arcier L, et al. A common pattern of brain MRI imaging in mitochondrial diseases with complex I deficiency. *J Med Genet* 2011;48:16–23.

Neurology®

Teaching *NeuroImages*: Rapidly progressive leukoencephalopathy in mitochondrial complex I deficiency

Fabian Baertling, Jörg Schaper, Ertan Mayatepek, et al.

Neurology 2013;81:e10-e11

DOI 10.1212/WNL.0b013e31829a339b

This information is current as of July 8, 2013

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/81/2/e10.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2013/07/07/81.2.e10.DC1
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/81/2/e10.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 Pediatric http://n.neurology.org/cgi/collection/all_pediatric Leukodystrophies http://n.neurology.org/cgi/collection/leukodystrophies Mitochondrial disorders; see Genetics/Mitochondrial disorders http://n.neurology.org/cgi/collection/mitochondrial_disorders_see_genetics-mitochondrial_disorders 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.