Teaching NeuroImages:

When alopecia and disk herniations meet vascular leukoencephalopathy

CARASIL

Benjamin Roeben, MD
Sabine Uhrig, MD
Benjamin Bender, MD
Matthis Synofzik, MD

Correspondence to
Dr. Synofzik:
matthis.synofzik@uni-tuebingen.de

Figure MRI showing typical confluent leukoencephalopathy, alopecia, and disk herniation

Typical confluent leukoencephalopathy (A, FLAIR) including the capsula externa, thalamus (B, FLAIR), pontine linear hyperintensity (characteristic “arc sign”²) (C, T2-weighted), and both acute (D, diffusion-weighted imaging; E, apparent diffusion coefficient map) and postischemic defects (A–C). Leukoencephalopathy is combined with large disk herniation at C3–4 with myelopathy (G) and premature alopecia (F). FLAIR = fluid-attenuated inversion recovery.

A 26-year-old Turkish patient presented with progressive spastic gait since age 20 years and premature alopecia. Cerebral and spinal MRI revealed diffuse supra- and infratentorial leukoencephalopathy with recurrent subclinical subcortical infarcts and multiple disk herniations with cervical myelopathy (figure).

Targeted panel-sequencing revealed a novel homozygous splice site mutation (c.1005+1G>T; p.?) in *HTRA1*, which is known to cause CARASIL (cerebral autosomal-recessive arteriopathy with subcortical infarcts and leukoencephalopathy),¹ mostly described in Asians so far.

Download teaching slides:
[Neurology.org](https://www.neurology.org)

From the Department of Neurodegenerative Diseases, Hertie Institute for Clinical Brain Research (B.R., M.S.), German Research Center for Neurodegenerative Diseases (B.R., M.S.), and Department of Neuroradiology (B.B.), University of Tübingen; and CeGaT GmbH—Center for Genomics and Transcriptomics (S.U.), Tübingen, Germany.

Go to [Neurology.org](https://www.neurology.org) for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

While diffuse vascular leukoencephalopathy, alopecia, and disk herniations are unspecific frequent findings particularly in elderly patients, the combination signals the characteristic features of underlying CARASIL in young adults.²

AUTHOR CONTRIBUTIONS

Dr. Röben: acquisition of data, design and conceptualization of the study, drafting and revising the manuscript. Dr. Uhrig: execution of genetic analysis, revising the manuscript. Dr. Bender: acquisition and evaluation of MRI data, revising the manuscript. Dr. Synofzik: acquisition of data, design and conceptualization of the study, revising the manuscript.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

B. Röben reports no disclosures relevant to the manuscript. S. Uhrig is an employee of the genetics diagnostics company CEGAT. B. Bender has received travel expenses from Bayer Vital. M. Synofzik has received travel expenses and presentation honoraria from Actelion Pharmaceuticals Ltd. Go to Neurology.org for full disclosures.

REFERENCES

1. Hara K, Shiga A, Fukutake T, et al. Association of HTRA1 mutations and familial ischemic cerebral small-vessel disease. *N Engl J Med* 2009;360:1729–1739. doi: 10.1056/NEJMoa0801560.
2. Nozaki H, Sekine Y, Fukutake T, et al. Characteristic features and progression of abnormalities on MRI for CARASIL. *Neurology* 2015;85:459–463.

Neurology®

Teaching *NeuroImages*: When alopecia and disk herniations meet vascular leukoencephalopathy: CARASIL

Benjamin Roeben, Sabine Uhrig, Benjamin Bender, et al.

Neurology 2016;86:e166-e167

DOI 10.1212/WNL.0000000000002572

This information is current as of April 11, 2016

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/86/15/e166.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2016/04/09/WNL.0000000000002572.DC1
References	This article cites 2 articles, 1 of which you can access for free at: http://n.neurology.org/content/86/15/e166.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): Stroke in young adults http://n.neurology.org/cgi/collection/stroke_in_young_adults Vascular dementia http://n.neurology.org/cgi/collection/vascular_dementia
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 © 2016 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.