Teaching NeuroImages: MRI guides genetics

Leukoencephalopathy with brainstem and spinal cord involvement (LBSL)

A-D and F axial and E sagittal images show inhomogeneous signal abnormalities in the cerebral white matter (A), corticospinal tracts (B), superior cerebellar peduncles, medial lemniscus (C), pyramids, inferior cerebellar peduncles, cerebellar white matter (D), lateral corticospinal tracts, dorsal columns (E and F), and elevated lactate in magnetic resonance spectroscopy (G), thus meeting all major and some minor MRI criteria for LBSL (leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation).1

From the Department of Neurodegenerative Diseases (J.S., L.S., M.S.), Hertie-Institute for Clinical Brain Research, and German Research Center for Neurodegenerative Diseases (DZNE) (J.S., L.S., M.S.), University of Tübingen, Germany; and Department of Child Neurology (M.S.v.d.K.), VU University Medical Center, Amsterdam, the Netherlands.

Go to Neurology.org for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.
A 12-year-old German girl presented with progressive spastic-ataxic gait and impaired fine motor skills starting from early childhood. MRI revealed striking T2-signal abnormalities in the cerebral and cerebellar white matter, pyramidal tracts and dorsal columns of the spinal cord, as well as lactate elevation in magnetic resonance spectroscopy, indicating leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL) (figure). Sequencing of \textit{DARS2}\(^1\) revealed compound heterozygous frame shift and premature stop mutations (established c.228-11C>G, novel c.617_663del).

Although LBSL is rare, these characteristic MRI findings\(^1\) help to identify patients with LBSL who have unexplained ataxia, spasticity, or leukoencephalopathy and facilitate straightforward genetic diagnostics. This is particularly important because certain features of this disease might be treatable.\(^2\)

\textbf{AUTHOR CONTRIBUTIONS}

Dr. Schicks: design and conceptualization of the study, drafting the manuscript. Dr. Schöls: conceptualization of the study, revising the manuscript. Dr. van der Knaap: execution of genetic analysis, revising the manuscript. Dr. Synofzik: acquisition of data, design and conceptualization of the study, revising the manuscript.

\textbf{STUDY FUNDING}

No targeted funding reported.

\textbf{DISCLOSURE}

J. Schicks reports no disclosures. L. Schöls received research grants of the Deutsche Forschungsgemeinschaft (SCHO754/4-1 and SCHO754/5-1), grants of the German Research Council (BMBF) to Leukonet (01GM0644) and mitoNET (01GM0864), and E-RARE grants to EUROSFA (01GM0807) and RISC (01GM0820). He further received funding from the HSP-Selbsthilfegruppe Deutschland e.V. M. van der Knaap reports no disclosures. M. Synofzik received a research grant by the Volkswagen Foundation, a travel grant by the Movement Disorders Society, and honoraria from Fresenius Kabi and Actelion Pharmaceuticals. Go to Neurology.org for full disclosures.

\textbf{REFERENCES}

Teaching NeuroImages: MRI guides genetics: Leukoencephalopathy with brainstem and spinal cord involvement (LBSL)
Julia Schicks, Ludger Schöls, Marjo S. van der Knaap, et al.

Neurology 2013;80:e176-e177
DOI 10.1212/WNL.0b013e31828cf846

This information is current as of April 15, 2013

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/80/16/e176.full

References
This article cites 2 articles, 1 of which you can access for free at:
http://n.neurology.org/content/80/16/e176.full#ref-list-1

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
Cerebellum
http://n.neurology.org/cgi/collection/cerebellum
Gait disorders/ataxia
http://n.neurology.org/cgi/collection/gait_disorders_ataxia
Leukodystrophies
http://n.neurology.org/cgi/collection/leukodystrophies
Mitochondrial disorders
http://n.neurology.org/cgi/collection/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