Teaching *NeuroImages*: MRI guides genetics

Leukoencephalopathy with brainstem and spinal cord involvement (LBSL)

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Figure Brain and spinal cord T2-weighted MRI

A-D and F axial and E sagittal images show inhomogeneous signal abnormalities in the cerebral white matter (A), cortico-spinal 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

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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 *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 findings¹ 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

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.

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