A young man with symptomatic epilepsy and right hemianopia

Family affair

A 32-year-old man presented with generalized epileptic seizures and right homonymous hemianopia. MRI detected a left parieto-occipital lesion suspicious for lymphoma (figure 1); biopsy showed inflammatory demyelinating white matter lesions suggestive of multiple sclerosis (figure 2, A–C). Hemianopia and neuro-

Cranial MRI revealed an irregular, T2-hyperintense, partly gadolinium (Gd)-enhancing left parieto-occipital lesion extending into the corpus callosum.

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Brain biopsy showed inflammatory white matter lesions with increased cellularity and reactive gliosis (A), (confluent) demyelination (B), and macrophages that showed LFB-positive myelin degradation products within their cytoplasm (C).
imaging remained unchanged 12 weeks after high-dose IV steroids. Seizures responded to levetiracetam. A maternal uncle had neurologic impairment leading to infant death; blood screening for very-long-chain fatty acids established the diagnosis of X-adrenoleukodystrophy. Because histologic findings are nonspecific and asymmetric focal brain lesions are a rare manifestation, family history was the key to the diagnosis, considering that severity and age at onset of X-adrenoleukodystrophy may vary.1

Louisa von Baumgarten, MD, Munich; Sabine Pfeifenbring, MD, Göttingen; Nicole Terpolilli, MD, Ulrich Schüller, MD, PhD, Klaus Jahn, MD, PhD, Christian Opherk, MD, Tobias Freilinger, MD, Munich, Germany

Author contributions: Dr. von Baumgarten wrote the original manuscript. Dr. Pfeifenbring analyzed and interpreted the biopsy specimen, drafted figure 2/figure 2 legend, and revised the manuscript. Dr. Terpolilli acquired the biopsy specimen, revised the manuscript, and made substantial intellectual contribution. Dr. Schüller analyzed and interpreted the biopsy specimen and revised the manuscript. Dr. Jahn revised the manuscript and made substantial intellectual contributions. Dr. Opherk revised the manuscript and made substantial intellectual contributions. Dr. Freilinger drafted and revised the manuscript and made substantial intellectual contributions.

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Correspondence & reprint requests to Dr. von Baumgarten: Louisa.vonBaumgarten@med.uni-muenchen.de


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