



Letter

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LETTER TO THE EDITOR

Teaching case 1-2020 – ADDENDUM: Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia due to a novel *CSF1R* mutation – An unusual cause of dementia

Sigrid Klotz¹, Franz Riederer^{2,6}, Nora Hergovich³, Thomas Schlager³, Lara Steinkellner⁴, Elisabeth Fertl³, Christoph Baumgartner², Matias Wagner^{7,8}, Alexander Zimprich⁵, and Ellen Gelpi¹

¹Division of Neuropathology and Neurochemistry, Department of Neurology, Medical University of Vienna, Austria, ²Neurological Center Rosenhügel and Karl Landsteiner Institute for Epilepsy, Research and Cognitive Neurology, ³Department of Neurology, Clinic Landstraße, ⁴Department of Oncology, Clinic Hietzing, ⁵Department of Neurology, Medical University of Vienna, Vienna, Austria, and ⁶University of Zurich, Faculty of Medicine, Department of Neurology, Zurich, Switzerland, ⁷Institute for Neurogenomics, Helmholtz Zentrum, Munich, Germany, and ⁸Institute of Human Genetics, Technical University Munich, Munich, Germany

Sir, – In the issue “Vol. 39 – No. 1/2020” of *Clinical Neuropathology*, we described the neuropathological features in a male patient in his fifties with an early-onset dementia. We entitled the article “Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia – An unusual cause of dementia [1]”.

At the time of the publication, genetic testing was not available. Fortunately, we have now been able to analyze an archival blood sample and to perform whole exome sequencing (Institute of Human Genetics, Technical University Munich, Germany). A novel heterozygous missense variant c.2546T>C, p. (Phe849Ile) in the *CSF1R* (colony stimulating factor-1 receptor, NM_005211.3) gene was detected. This variant is considered as “likely pathogenic” according to the American College of Medical Genetics and Genomics (ACMG) criteria [2].

Our genetic findings can confirm that the described case was a “definite” case of adult-onset leukoencephalopathy with axonal spheroids and pigmented glia due to *CSF1R* mutation [3].

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Conflict of interest

The authors declare no conflict of interest.

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- [1] Klotz S, Riederer F, Hergovich N, Schlager T, Steinkellner L, Fertl E, Baumgartner C, Zimprich A, Gelpi E. Adult-onset leukoencephalopathy with axonal spheroids and pigmented glia – An unusual cause of dementia. *Clin Neuropathol*. 2020; 39: 4-6.
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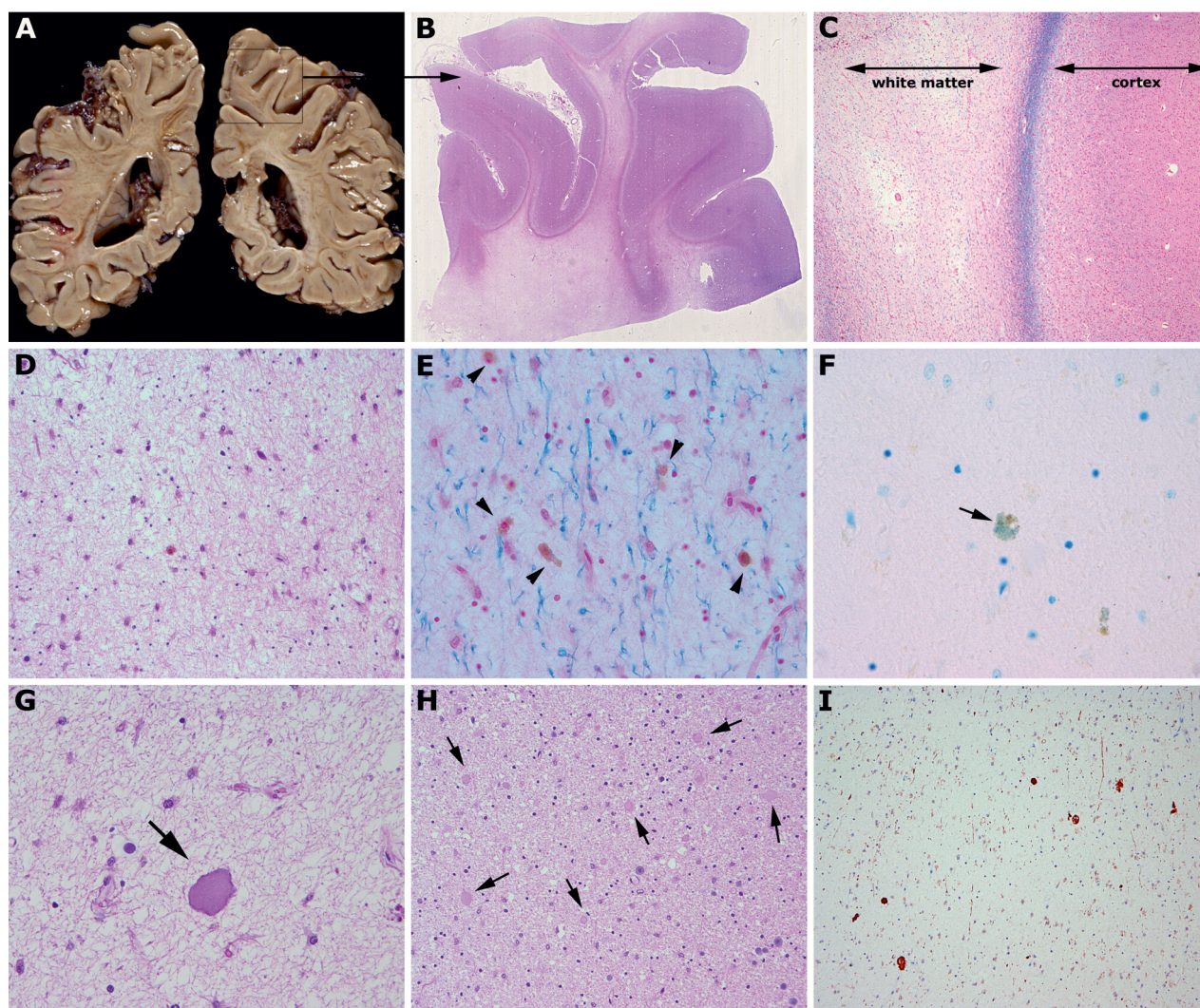


Figure 1. A: Coronal sections through the brain hemispheres show prominent white matter rarefaction with yellowish discoloration. B, C: Klüver-Barrera stain reveals the severe leukoencephalopathy with relative preservation of U-fibers. D: At higher magnification, there is prominent rarefaction of white matter with loss of oligodendrocytes and reactive astrocytes. These show partly brownish pigment in the cytoplasm (E, arrows) without metachromasia (F, toluidine blue). G, H, I: Presence of abundant and partly large axonal spheroids (arrows) that are also well identified with antibodies against neurofilaments (I).

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Correspondence to
Ellen Gelpi, MD, PhD, EFN
Division of Neuropathology and Neurochemistry,
Department of Neurology,
Medical University of Vienna,
AKH 4J, Währinger Gürtel 18 – 20,
1090 Vienna, Austria
ellen.gelpi@meduniwien.ac.at

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