Novel Application of 7T MRI in CSF1R-Related Leukoencephalopathy

Author(s): Jaroslaw Dulski, MD, PhD\textsuperscript{1,2,3}; Erik H Middlebrooks, MD\textsuperscript{4}; Zbigniew K Wszolek, MD\textsuperscript{1}

Corresponding Author: Zbigniew K Wszolek, wszolek.zbigniew@mayo.edu

Affiliation Information for All Authors: 1. Department of Neurology, Mayo Clinic, Jacksonville, Florida; 2. Division of Neurological and Psychiatric Nursing, Faculty of Health Sciences, Medical University of Gdansk, Gdansk, Poland; 3. Neurology Department, St Adalbert Hospital, Copernicus PL Ltd., Gdansk, Poland; 4. Department of Radiology, Mayo Clinic, Jacksonville, Florida

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A 24-year-old female underwent screening for a family history of early-onset dementia and CSF1R-related leukoencephalopathy in her mother. The patient had no symptoms, and the neurological examination was normal. Three Tesla (3T) brain MRI revealed few scattered, nonspecific subcortical and periventricular white matter lesions (WMLs) without atrophy (Figure 1A). Genetic testing confirmed p.Leu786Ser mutation in the CSF1R gene. Repeat 7T MRI better depicted WMLs (Figure 1B) and revealed characteristic frontal lobes calcifications (Figure 2). The improved sensitivity of 7T MRI benefited treatment decision-making, and the patient was referred to a clinical trial. Treatment decisions in CSF1R-related leukoencephalopathy lack a consensus algorithm but rely on various clinical and imaging features. The added diagnostic accuracy of 7T MRI may guide treatment through better sensitivity of disease severity and progression and earlier diagnosis through added specificity. Prompt diagnosis is crucial and may qualify patients for ongoing investigational drug trials or hematopoietic cell transplantation.

Legends

Fig 1. (A) Coronal 3T FLAIR image showing a small left frontal white matter hyperintensity (arrow). (B) Matching coronal 7T double-inversion recovery (DIR) better depicting the lesion (arrow). DIR, typically a low signal-to-noise ratio (SNR) image, is greatly enhanced at 7T giving better contrast ratio and SNR compared to 3T FLAIR.
Figure 2. (A,B) Matching axial 3T susceptibility-weighted images do not clearly depict the calcifications, a known limitation of 3T MRI in CSF1R-related leukoencephalopathy. (C,D) High-resolution, axial 7T T2*-weighted images show bilateral white matter calcifications (arrows) in a “stepping-stone” configuration typical of CSF1R-related leukoencephalopathy.

References


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Jaroslaw Dulski, Erik H Middlebrooks and Zbigniew K Wszolek
Neurology published online September 30, 2022
DOI 10.1212/WNL.0000000000201450

This information is current as of September 30, 2022

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