Novel Application of 7T MRI in CSF1R-Related Leukoencephalopathy

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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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