A 47-year-old man presented to clinic with progressive right-hand bradykinesia and shuffling gait for 8 months. He was diagnosed with parkinsonism. Brain MRI demonstrated diffusion-weighted imaging (DWI)–hyperintense signal involving bilateral corona radiate (figure 1) and several lacunae with extensive white matter hyperintensity lesions involving bilateral lateral periventricular regions without enhancement. (C and D) Diffusion-weighted imaging-positive lesions without hypointensity appearance on apparent diffusion coefficient. (E) Hypointensity lesions on rostrum of corpus callosum (white arrow) and predominant splenial atrophy (black arrowheads).
T2-weighted MRI showed white matter changes (figure 2). Colony-stimulating factor 1 receptor (CSF1R) gene sequencing revealed the pathogenic variant c.2381T>C(p.Ile794Thr), confirming the diagnosis of hereditary diffuse leukoencephalopathy with spheroids. The patient was treated with levodopa and selegiline with minimal response.2

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Disclosure
The authors report no disclosures relevant to the manuscript. Go to Neurology.org/N for full disclosures.

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References
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