A 34-year-old woman presented with a 1-year history of progressive apathy, executive dysfunction, and memory impairment. Examination revealed moderate frontal dysfunction and bipyramidal signs. MRI brain (figure 1) showed a symmetric leukoencephalopathy sparing subcortical U-fibers.

Evaluation for an acquired white matter disease was negative. Next-generation sequencing showed a pathogenic heterozygous missense mutation in exon 18 of CSF1R gene (p.Ile794Thr)

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confirming the diagnosis of adult-onset leukoencephalopathy with axonal spheroids and pigmented glia (ALSP). Inheritance is autosomal dominant or sporadic. Presence of symmetric or asymmetric nonenhancing white matter lesions with persistent diffusion restriction (figure 2) and corpus callosum thinning differentiates ALSP from acquired demyelination.1,2

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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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Appendix (continued)

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References

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