Teaching NeuroImages: Rare Adult-Onset Genetic Leukoencephalopathy

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Figure 1 Core Features in T2, T2-Fluid-Attenuated Inversion Recovery (FLAIR), and T1 MRI Sequences

Axial T2-weighted images (A, B) show confluent symmetric frontal-predominant white matter hyperintensities sparing subcortical U-fibers (white arrowheads), hypointense in T1-weighted sequences (C, black arrowheads). Sagittal T2-FLAIR (D) shows corpus callosum hyperintensities and atrophy (arrow).

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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Figure 2 Persistent Abnormalities in MRI Diffusion Sequences

Diffusion-weighted images (A, B) show deep white matter diffusion restriction with corresponding low apparent diffusion coefficient (black arrows), persistent in similar sequences taken 4 months later (C, D).

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

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Appendix Authors

Name	Location	Contribution
Pavankumar Rudrabhatla, MD	Department of Neurology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India	Major role in acquisition of data and drafting of manuscript

Appendix (continued)

Name	Location	Contribution
Sekar Sabarish, MD	Department of Imaging Sciences and Interventional Radiology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India	Major role in acquisition of data and interpretation of imaging data
Harikrishnan Ramachandran, MD	Department of Neurology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India	Study concept and design
Sruthi S. Nair, MD, DM	Department of Neurology, Sree Chitra Tirunal Institute for Medical Sciences and Technology, Thiruvananthapuram, Kerala, India	Study concept and design, and critical revision of manuscript

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