Teaching NeuroImages: Autosomal dominant leukodystrophy in a sporadic case

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Figure MRI characteristics in autosomal dominant leukodystrophy

Fluid-attenuated inversion recovery sequences show symmetric hyperintense signals involving the deep white matter of both hemispheres with relative sparing of the periventricular regions (A, arrows) and hyperintensity of cerebellar peduncles and pontine nuclei (B, arrows). T1-weighted images show diffuse spinal cord atrophy (C).

A 50-year-old man with a 5-year history of progressive genitourinary dysautonomia noticed recent leg stiffness. Neurologic examination showed mild spastic paraparesis. MRI showed an unusual pattern of white matter changes in the brain and spinal cord atrophy (figure). Electroneurography, CSF, and blood/urine metabolic studies had normal results. Despite a negative family history, the patient's features were suggestive of autosomal dominant leukodystrophy (ADLD, MIM #169500).¹ Molecular testing confirmed this diagnosis, documenting a duplication in *LMNB1*, coding for lamin B1.²

ADLD is an adult-onset leukodystrophy with early presentation of dysautonomia preceding cerebellar and pyramidal signs. MRI is the best imaging technique to diagnose ADLD.¹

AUTHOR CONTRIBUTIONS

Dr. Brunetti: study concept and design, drafting the manuscript, accepts responsibility for conduct of research, and acquisition of data.

Dr. Ferilli: analysis or interpretation of data, accepts responsibility for conduct of research, and acquisition of data. Dr. Nociti: analysis or interpretation of data, accepts responsibility for conduct of research. Dr. Silvestri: interpretation of data, accepts responsibility for conduct of research, study supervision, revising the manuscript, and final approval.

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DISCLOSURE

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

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