A 25-year-old Brazilian man presented with an 8-year history of progressive myoclonic epilepsy. Familial history disclosed a Japanese paternal grandmother with late-onset parkinsonism-dementia and a paternal uncle with atypical parkinsonism. Examination showed myoclonic jerks, ataxia, and brisk tendon reflexes. Brain MRI showed diffuse leukodystrophy (figure). Genetic testing (supplemental material on the *Neurology*® Web site at Neurology.org) exhibited 70/35 CAG expansions in the *ATN1* gene, diagnostic of dentatorubral-pallidoluysian atrophy (DRPLA).

DRPLA is a polyglutamine neurodegenerative disorder with a wide spectrum of manifestations ranging from ataxia, choreoathetosis, and dementia in adults to epilepsy and intellectual disability in children.1 The presence of leukodystrophy on suspicion of progressive myoclonic epilepsy should raise the possibility of DRPLA.2

**AUTHOR CONTRIBUTIONS**


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**DISCLOSURE**

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

**REFERENCES**


Neuroimaging findings and family history in this family. (A–D) Axial and coronal T2-weighted brain MRIs showing diffuse hyperintensity in the cerebellar and supratentorial white matter. (E) Mendelian genetic pedigree showing the propositum (III.1, black arrow), paternal grandmother with late-onset parkinsonism-dementia phenotype (I.2), and a paternal uncle with atypical parkinsonism and cerebellar ataxia (II.1).
Teaching NeuroImages: Leukodystrophy and progressive myoclonic epilepsy disclosing DRPLA
Paulo Victor Sgobbi de Souza, Gabriel Novaes de Rezende Batistella, Wladimir Bocca Vieira de Rezende Pinto, et al.
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