Teaching *NeuroImages*: Leukodystrophy and progressive myoclonic epilepsy disclosing DRPLA

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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

P.V.S. Souza: conception, organization, and execution of the case report project, writing of the first draft. G.N.R. Batistella: conception, organization, and execution of the case report project, writing of the first draft. W.B.V.R. Pinto: conception, organization, and execution of the case report project, writing of the first draft. A.S.B. Oliveira: writing of the first draft, review and critique of the manuscript.

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DISCLOSURE

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

REFERENCES

1. Licht DJ, Lynch DR. Juvenile dentatorubral-pallidoluysian atrophy: new clinical features. *Pediatr Neurol* 2002;26:51–54.
2. Sunami Y, Koide R, Arai N, Yamada N, Mizutani T, Oyanagi K. Radiologic and neuropathologic findings in patients in a family with dentatorubral-pallidoluysian atrophy. *AJNR Am J Neuroradiol* 2011;32:109–114.

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Ethical statement: Full consent was obtained from the patient's parents and family for the case report publication.

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).

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