A 55-year-old woman presented to our center with an almost lifelong action tremor, associated with peripheral neuropathy, progressive sensorineural hearing loss, and a strong family history of tremor. CT of the brain was notable for extensive intracranial calcifications, much more prominent in the dentate nucleus, cerebellar hemispheres, and midpons, compared to the globus pallidus (figure 1). T1-weighted MRI demonstrated hypointense signal in the aforementioned areas (figure 2). Polymerase gamma-1 (POLG1) gene analysis revealed a novel heterozygous sequence variant at c3239G>c; p.Ser1080Thr. Similar diffuse intracranial calcification can be seen in a variety of disorders including idiopathic basal ganglia calcifications and spinocerebellar ataxia 20. Mitochondrial disorders are a well-recognized cause; however, to our knowledge this is the first time that such extensive intracranial calcium deposits have been described in a patient with a POLG1 mutation.
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Author contributions: Dr. Sidiropoulos: study concept and design. Dr. Sidiropoulos: acquisition of data. Drs. Sidiropoulos, Moro, Lang: analysis and interpretation of data. Drs. Moro, Lang: critical revision of the manuscript for important intellectual content. Drs. Moro, Lang: study supervision.

Study funding: No targeted funding reported.

Disclosure: C. Sidiropoulos reports no disclosures. E. Moro has received honoraria from Medtronic for consulting services and lecturing and has received research support from St. Jude Medical, CurePSP, CIHR, and educational grant support from Medtronic. A. Lang has served as an advisor for Abbott, Allon Therapeutics, Astra Zeneca, Avanir Pharmaceuticals, Biogen Idec, Biovail, Boehringer-Ingelheim, Cephalon, Ceregene, Eisai, GSK, Lundbeck A/S, Medtronic, Merck Serono, Merck, Novartis, and Teva; received grants from Canadian Institutes of Health Research, Dystonia Medical Research Foundation, Michael J. Fox Foundation, National Parkinson Foundation, Parkinson Society of Canada, and Ontario Problem Gambling Research Center; and has served as an expert witness in cases related to the welding industry. Go to Neurology.org for full disclosures.

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Neurology 2013;81;197-198
DOI 10.1212/WNL.0b013e31829a3438

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