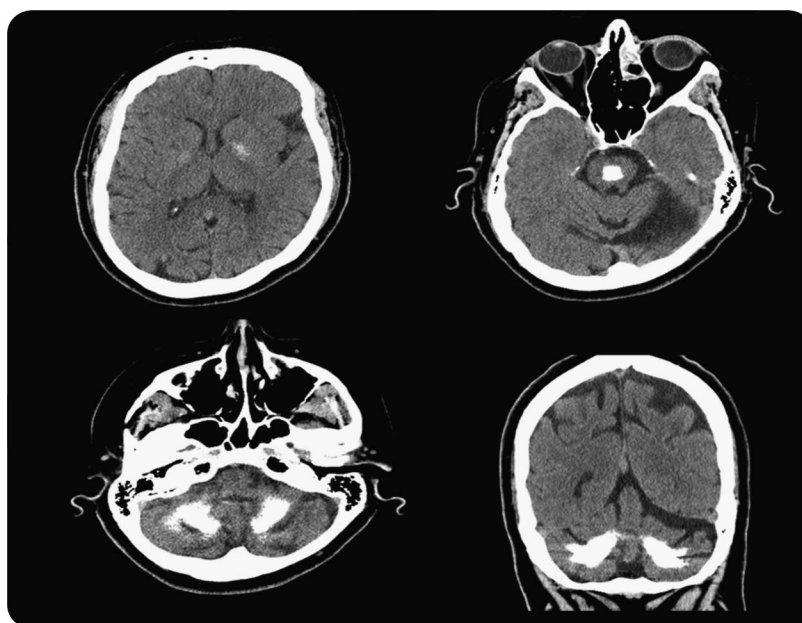


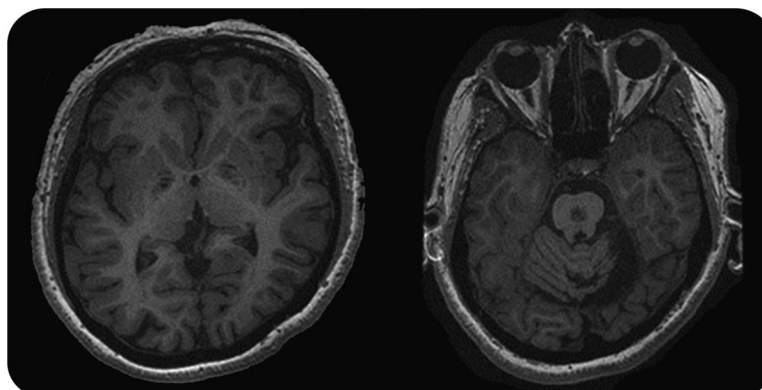
Extensive intracranial calcifications in a patient with a novel polymerase gamma-1 mutation

Figure 1 CT head demonstrates profound cerebellar and pontine calcifications, less so in the globus pallidus



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 20.¹ Mitochondrial disorders² 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.

Figure 2 T1-weighted MRI brain shows hypointense signal in the globus pallidus and midpons



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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.

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