NEURO*IMAGES*

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

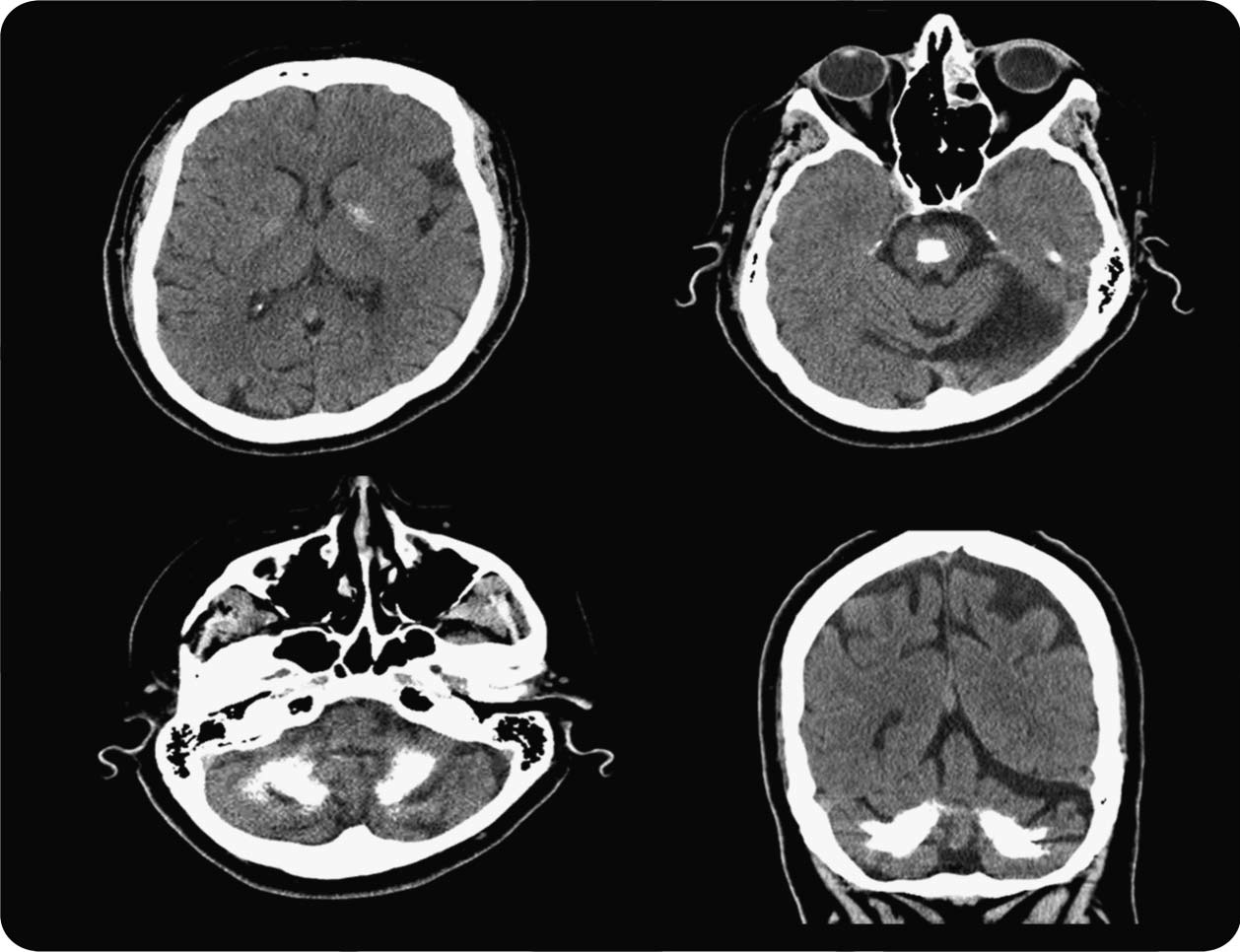


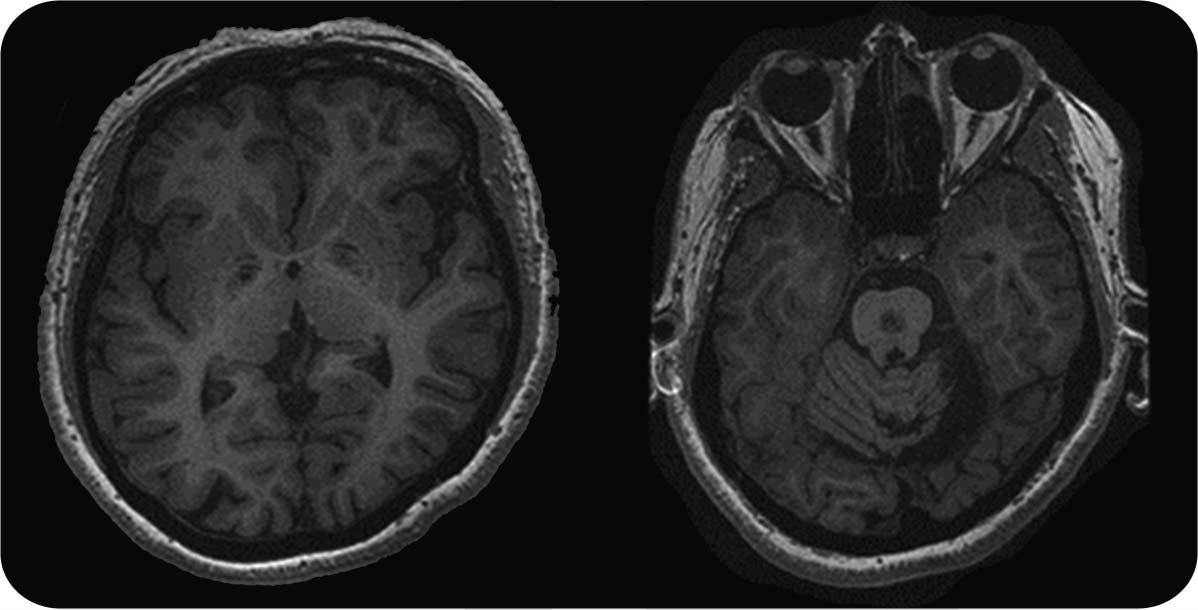
Figure 1 CT head demonstrates profound cerebellar and pontine calcifications, less so in the globus pallidus

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 hemi- spheres, and midpons, compared to the globus pallidus (figure 1). T1-weighted MRI demonstrated hypo- intense 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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