Teaching NeuroImage: Primary Familial Brain Calcification in SLC20A2 Genotype

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A 52-year-old woman presented with a 4-year history of parkinsonism characterised by hypomimia, bradykinesia, right-hand rest tremor, reduced right arm swing and short stride length. CT head (Figure 1) and MRI brain (Figure 2) showed bilateral dense calcification throughout the basal ganglia, thalami, cerebellum, subcortical and deep white matter. Genetic testing revealed a pathogenic heterozygous deletion (NM_001257180.1: c.1794+1del) in the splicing region of the SLC20A2 gene, confirming a diagnosis of autosomal dominant primary familial brain calcification. It subsequently transpired that her brother with cervical dystonia carried the same genetic mutation. This genotype is associated with calcifications that typically involve the basal ganglia, thalamus and cerebellum\(^1\). Patients may be asymptomatic, experience parkinsonism, or less commonly dystonia\(^1\).
Figure Legend:

Figure 1: Axial computed tomography (CT) head images revealed bilateral dense calcification throughout the basal ganglia, thalami, subcortical and deep white matter.

Figure 2: Axial MRI brain scan susceptibility weighted imaging (SWI) sequence reveals widespread caudate, pulvinar, cerebellar, orbitofrontal and occipital calcifications.
References:


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