Teaching NeuroImage: Primary Familial Brain Calcification in *SLC20A2* Genotype

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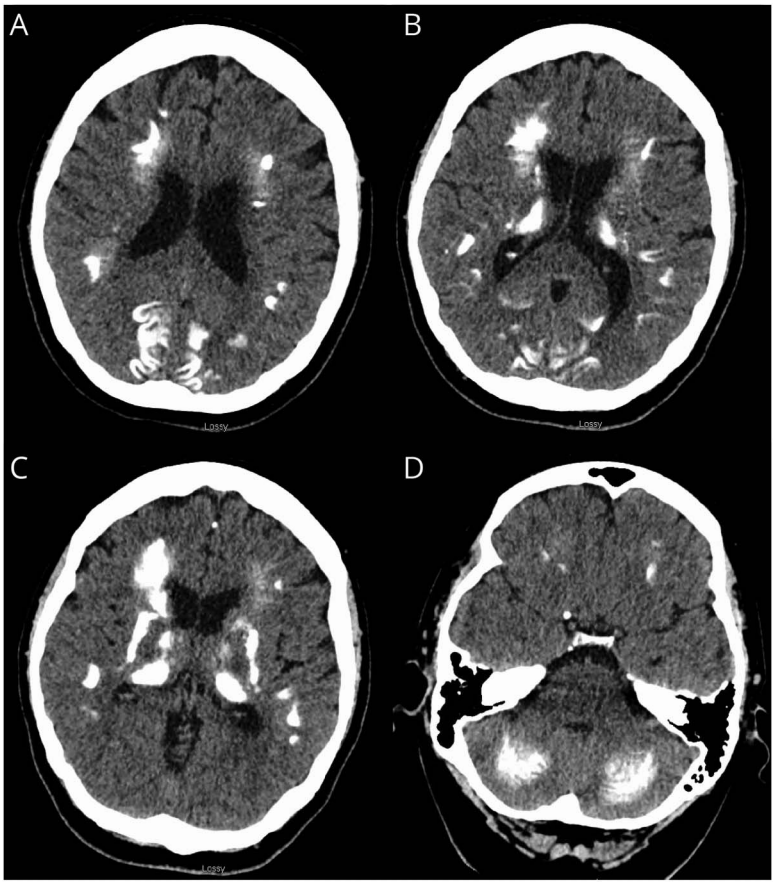
Figure 1 Axial CT Head Images Revealed Bilateral Dense Calcification Throughout the Basal Ganglia, Thalami, Subcortical, and Deep White Matter



A 52-year-old woman presented with a 4-year history of parkinsonism characterized 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 variation. This genotype is associated with calcifications that typically

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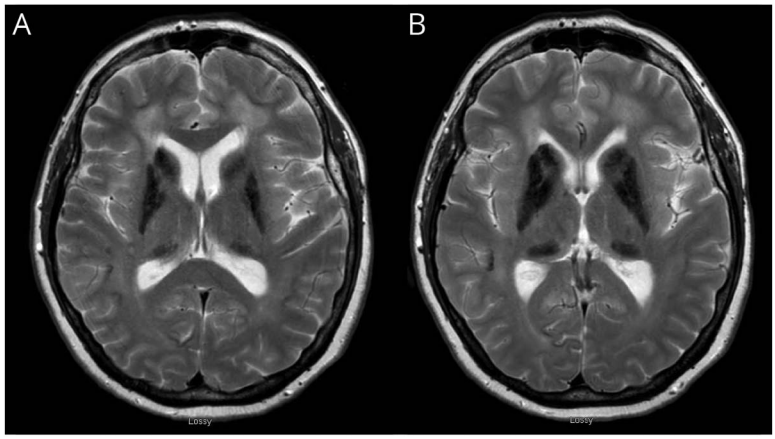
Figure 2 Axial MRI Brain Scan Susceptibility Weighted Imaging (SWI) Sequence Revealed Widespread Calcifications



involve the basal ganglia, thalamus, and cerebellum.[1,2] Patients may be asymptomatic, experience parkinsonism, or less commonly dystonia.[1]

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Name	Location	Contribution
Mary Clare McKenna, MRCP	St. James's Hospital, Dublin 8, Ireland	Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data
Janice Redmond, MD	St. James's Hospital, Dublin 8, Ireland	Drafting/revision of the manuscript for content, including medical writing for content; Major role in the acquisition of data; Study concept or design; Analysis or interpretation of data
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