Teaching NeuroImage: Primary Familial Brain Calcification in SLC20A2 Genotype

Author(s): Mary Clare McKenna, MRCPI1,2; Janice Redmond, MD1; David Bradley, PhD1; Peter Bede, MD PhD1,2

Corresponding Author: Mary Clare McKenna, mary.mc-kenna.1@ucdconnect.ie

Affiliation Information for All Authors: 1. Neurology Department, St. James’s Hospital, Dublin 8, Ireland; 2. Computational Neuroimaging Group, Biomedical Sciences Institute, Trinity College Dublin, Ireland

Equal Author Contribution:

Contributions:
Mary Clare McKenna: 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: 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
David Bradley: Drafting/revision of the manuscript for content, including medical writing for content; Study concept or design; Analysis or interpretation of data
Peter Bede: 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

Figure Count: 2

Table Count: 0

Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.
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:

Teaching NeuroImage: Primary Familial Brain Calcification in SLC20A2 Genotype
Mary Clare McKenna, Janice Redmond, David Bradley, et al.
Neurology published online September 20, 2022
DOI 10.1212/WNL.0000000000201343

This information is current as of September 20, 2022

Updated Information & Services
including high resolution figures, can be found at:
http://n.neurology.org/content/early/2022/09/20/WNL.0000000000201343.citation.full

Subspecialty Collections
This article, along with others on similar topics, appears in the following collection(s):
All Genetics
http://n.neurology.org/cgi/collection/all_genetics
Basal ganglia
http://n.neurology.org/cgi/collection/basal_ganglia
Parkinson's disease/Parkinsonism
http://n.neurology.org/cgi/collection/parkinsons_disease_parkinsonism
Tremor
http://n.neurology.org/cgi/collection/tremor

Permissions & Licensing
Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
http://www.neurology.org/about/about_the_journal#permissions

Reprints
Information about ordering reprints can be found online:
http://n.neurology.org/subscribers/advertise