NEUROIMAGES

Cavitating leukodystrophy as a manifestation of cerebral involvement in MFN2 neuropathy

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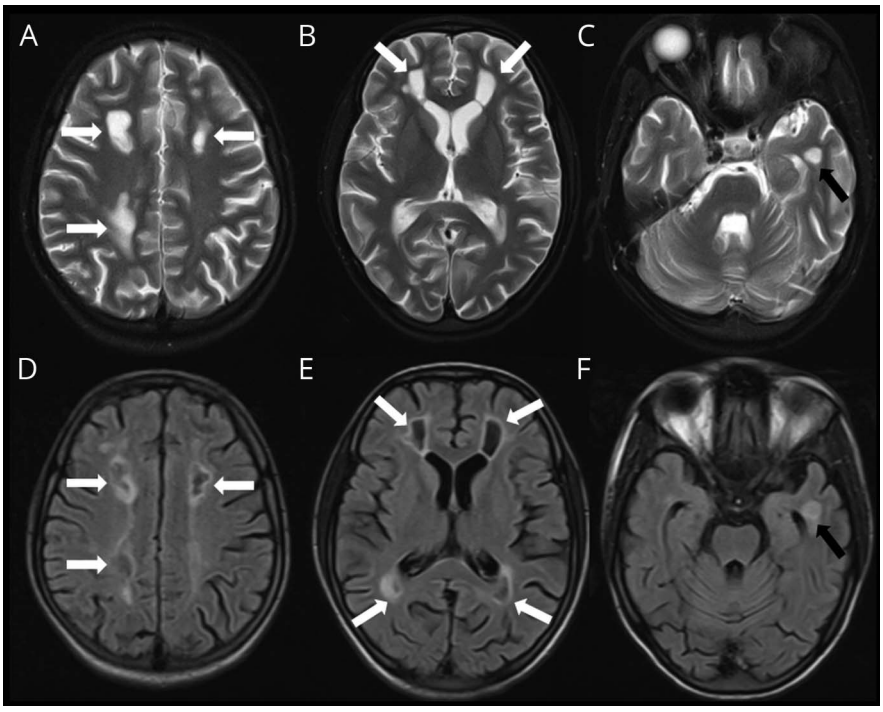
A 16-year-old girl presented with progressive bilateral lower limb weakness for 3 years. Examination revealed tongue fasciculations, muscle wasting, and extensor plantar responses. Nerve conduction studies revealed motor-sensory axonal polyneuropathy. MRI brain showed multifocal cavitating white matter disease with diffusion restriction (figures 1 and 2). Exome sequencing revealed heterozygous missense variation c.775C>T(p.Arg259Cys) in exon 8 of the *MFN2* gene, pathogenic for Charcot-Marie-Tooth disease 2A (CMT2A).

Dominant mutations of *MFN2* (encoding mitochondrial protein mitofusin-2) cause a disorder of mitochondrial DNA maintenance[1] resulting in axonal sensorimotor neuropathy.

Figure 1 MRI brain: T2–weighted and T2–fluid-attenuated inversion recovery (FLAIR)–weighted images



Axial T2-weighted images (A–C) show discrete hyperintense foci in periventricular and deep cerebral white matter. Axial T2-FLAIR images (D and E) show suppression of signal within the lesions suggesting cystic nature (white arrows). A nonsuppressed lesion (F) was seen in the left temporal lobe (black arrow).

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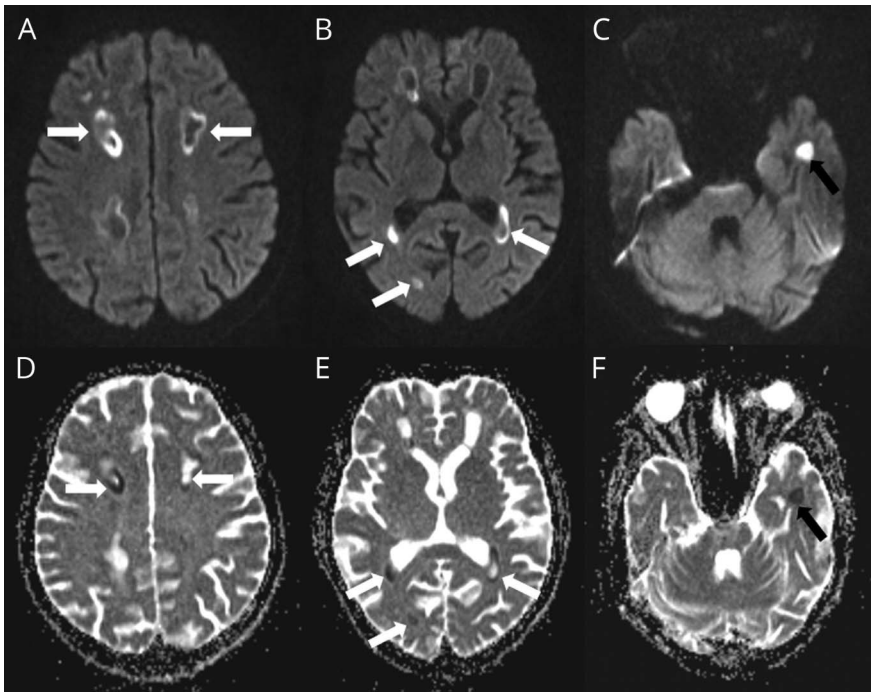
Figure 2 MRI brain: diffusion-weighted imaging



Axial diffusion-weighted images (A–C) and corresponding apparent diffusion coefficient maps (D–F) show diffusion restriction along the walls of some cystic lesions (white arrows) and solid diffusion restriction in the left temporal lobe lesion (black arrows). Spinal cord was normal (images not shown).

Nonspecific white matter alterations are reported in few patients with CMT2A.[2] Multifocal cavitating leukodystrophy may be seen rarely.

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Appendix Authors

Name	Location	Contribution
Ranjith Kumar Manokaran, MD, DM	Division of Paediatric Neurology, Sri Ramachandra Institute of Higher Education and Research (SRIHER), Chennai, India	Conceptualization, data collection, manuscript editing
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Deepti Kewalramani, MD	Department of Pediatric Neurology, BJ Wadia Children Hospital, Mumbai, India	Data collection, manuscript drafting

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