Teaching NeuroImages: Substantia nigra T2 hyperintensities in a man with Leber hereditary optic neuropathy

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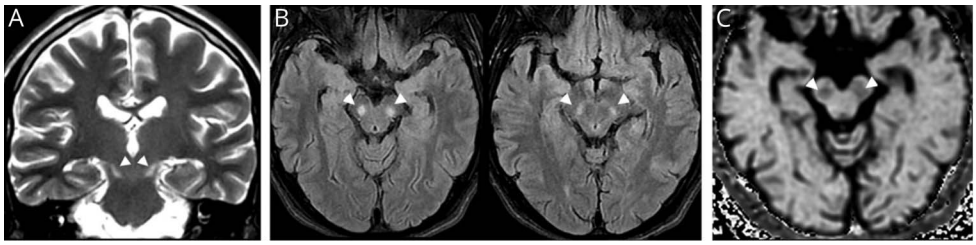
Figure MRI scans show symmetric substantia nigra T2 hyperintensities



Coronal T2 MRI (A) and axial fluid-attenuated inversion recovery MRI (B) show bilateral symmetric substantia nigra T2 hyperintensities (arrowheads). These lesions do not show restricted diffusion (arrowheads) on axial diffusion-weighted imaging (C).

A 29-year-old man with alcoholism presented with sudden progressive central visual loss in the right eye. Visual acuity was 20/300 (right) and 20/20 (left). Other findings included right relative afferent pupillary defect and bilateral hyperemic nonedematous optic discs concerning for OD retrobulbar optic neuritis. MRI head and orbit revealed symmetric T2 hyperintensities in the substantia nigra, without optic nerve hyperintensities (figure). Subsequently he lost vision in his left eye. Given that his maternal uncle developed bilateral optic neuritis in his 20s, genetic testing for Leber hereditary optic neuropathy was performed, finding a point mutation (m.11778G>A) in mitochondrially encoded NADH dehydrogenase 4. The unusual gray matter hyperintensities in this case favored a metabolic rather than demyelinating etiology.

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Appendix Authors

Name	Location	Role	Contribution
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Charles Smith, MD	University of Kentucky	Author	Drafting and revision, literature review, imaging acquisition and formatting
Padmaja Sudhakar, MD	University of Kentucky	Author	Drafting and revision

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