RESIDENT & FELLOW SECTION

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# Teaching NeuroImages: Biotin-responsive basal ganglia disease

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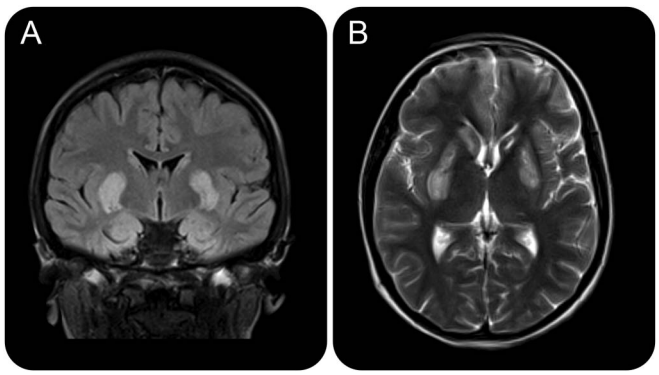
**Figure 1** Coronal and axial T2-weighted MRI at presentation




Coronal fluid-attenuated inversion recovery (A) and axial T2-weighted (B) MRI slices at presentation showing relatively symmetric hyperintensity in the corpora striata, and apparent cystic changes in the left head of caudate.

A 15-year-old Indian girl presented with dysphagia, oropharyngeal dystonic movements, brisk reflexes, and unsteadiness in tandem walking over years.

MRI revealed T2-weighted hyperintensity in the corpora striata (figure 1). Following treatment, imaging at 2 months and 2 years showed improvement in the corpus striatal hyperintensity with residual cystic damage (figure 2). Muscle biopsy was normal.

Genetic testing confirmed biotin-responsive basal ganglia disease, which is secondary to mutations in the *SLC19A3* thiamine transporter gene, usually in Middle Eastern and Indian patients.[1,2] Untreated, it can progress to dystonia, quadriparesis, and coma.[1] Biotin and thiamine therapy significantly improved the patient's clinical condition.[2]

## AUTHOR CONTRIBUTIONS

J.S.: study design and concept, analysis and interpretation of data/images, drafting the manuscript. R.L.: analysis and interpretation of data/images, drafting and revising the manuscript. I.D.: study design and concept, analysis and interpretation of data/images, drafting and revising the manuscript, study supervision.

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## DISCLOSURE

J. Schwarting and R. Lakshmanan report no disclosures relevant to the manuscript. I. Davagnanam is funded by the NIHR UCL/UCLH Biomedical Research Centre. Go to Neurology.org for full disclosures.

## REFERENCES

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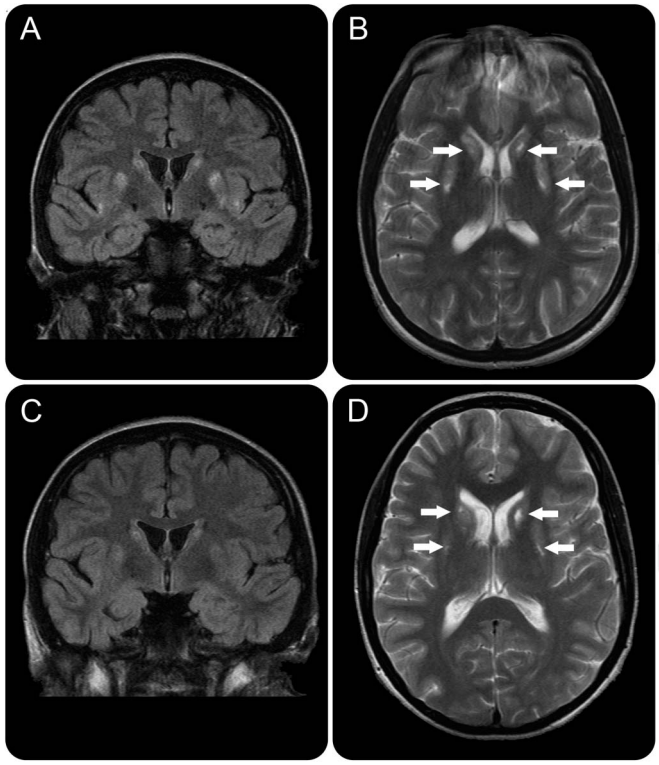
**Figure 2** Coronal and axial T2-weighted MRI 2 months and 2 years after treatment




Coronal and Axial T2-weighted MRI slices showing symmetrical hyperintensities in the corpora striata at 2 months (A and B) and 2 years (C and D) after biotin and thiamine treatment, demonstrating a progressive reduction in the corpus striatal hyperintensity with residual small areas of cystic damage (white arrows).