Teaching NeuroImages: Biotin-responsive basal ganglia disease

Julian Swarting
Rahul Lakshmanan,
FRANZCR
Indran Davagnanam,
FRCR

Correspondence to
Julian Swarting:
julian.swarting@med.
uni-muenchen.de

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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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).

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