Teaching NeuroImages: MRI findings of biotin-responsive basal ganglia disease before and after treatment

Mnahi Bin Saeedan,
MBBS, MPH
Mohammed Asif Dogar,
MBBS, DABR

Correspondence to
Dr. Bin Saeedan:
Mbinsaeedan@gmail.com

Figure 1 Axial brain fluid-attenuated inversion recovery MRI

Images show bilateral swelling and high signal intensities of the caudate heads and basal ganglia with relative sparing of globus pallidi and thalami (A) and cortical frontal lobes (B).

An 11-year-old boy presented with new generalized tonic-clonic seizures and encephalopathy. MRI revealed bilateral striatum T2 hyperintensity and edema bilateral thalami and frontal lobe cortices T2 hyperintensity (figure 1). The patient was treated with biotin and riboflavin and diagnosis of biotin-responsive basal ganglia disease was confirmed by *SLC19A3* gene analysis. Three years

later, MRI examination showed resolution of these changes (figure 2).

Biotin-responsive basal ganglia disease is an autosomal recessive disease associated with *SLC19A3* gene mutation that encodes thiamine transporter 2. Caudate heads and putamen swelling and T2 hyperintensity are highly suggestive of this diagnosis in a child with unexplained encephalopathy.^{1,2}

Figure 2 Axial brain fluid-attenuated inversion recovery MRI

Images 3 years after initiation of the treatment show almost complete resolution of the caudate head, basal ganglia, thalami (A), and cortical frontal lobe changes (B).

Download teaching slides:
Neurology.org

From the Radiology Department, King Faisal Specialist Hospital and Research Centre, Riyadh, Saudi Arabia.

Go to Neurology.org for full disclosures. Funding information and disclosures deemed relevant by the authors, if any, are provided at the end of the article.

AUTHOR CONTRIBUTIONS

M. Bin Saeedan: data acquisition, drafting of manuscript. M. Dogar: data acquisition and manuscript revision.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

REFERENCES

1. Kassem H, Wafaie A, Alsuhbani S, Farid T. Biotin-responsive basal ganglia disease: neuroimaging features before and after treatment. *AJNR Am J Neuroradiol* 2014;35:1990–1995.
2. Tabarki B, Al-Shafi S, Al-Shahwan S, et al. Biotin-responsive basal ganglia disease revisited: clinical, radiologic, and genetic findings. *Neurology* 2013;80:261–267.

Neurology®

Teaching *NeuroImages*: MRI findings of biotin-responsive basal ganglia disease before and after treatment

Mnahi Bin Saeedan and Mohammed Asif Dogar

Neurology 2016;86:e71-e72

DOI 10.1212/WNL.0000000000002372

This information is current as of February 15, 2016

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/86/7/e71.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2016/02/14/WNL.0000000000002372.DC1
References	This article cites 2 articles, 2 of which you can access for free at: http://n.neurology.org/content/86/7/e71.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Medical/Systemic disease http://n.neurology.org/cgi/collection/all_medical_systemic_disease MRI http://n.neurology.org/cgi/collection/mri
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

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2016 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.