Teaching NeuroImages: Atrophy in epileptic encephalopathy

Ingo Helbig, MD, and Laura Adang, MD, PhD

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Correspondence

Dr. Helbig
helbigi@email.chop.edu

Figure MRI of the brain

Brain MRI demonstrates diffuse atrophy (A) and bilateral and symmetrical T2/fluid-attenuated inversion recovery hyperintense signal intensity in the thalami (B). Prior MRI at the age of 1 and 5 months were unremarkable. Magnetic resonance spectroscopy at the age of 5 months and 3 years was unremarkable.

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Neuroimaging in a 6-year-old girl with an unknown neurodegenerative disorder showed atrophy and bilateral thalamic T2/fluid-attenuated inversion recovery hyperintense signal intensity (figure). A mitochondrial or metabolic condition was suspected. Through a targeted gene panel, the patient was found to have a de novo *KCNT1* mutation, a gene known to cause of a broad range of epileptic encephalopathies.¹

Channelopathies may present with features suggesting neurodegenerative or neurometabolic disorders, or leukodystrophies.² Epilepsies due to de novo mutations are more common by probably more than an order of magnitude and atypical presentations may be on the differential for unsolved neurodegenerative disorders.

From the Division of Neurology, The Children's Hospital of Philadelphia, PA.

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

Author contributions

Ingo Helbig: study concept of design and acquisition of data.
Laura Adang: study concept of design and critical revision of manuscript for intellectual content.

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The authors report no disclosures relevant to the manuscript.
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