Mystery Case: Bilateral posterior periventricular heterotopias

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Figure MRI of bilateral posterior periventricular heterotopias

Coronal T1-weighted MRI shows the extent of posterior periventricular nodular heterotopias (pPNH) (A, B, E) and a magnification of the right periventricular area (D). Coronal T2-weighted images show the bilateral pPNH (C) and a magnification of the right periventricular area (F). Note that the nodules of pPNH have the same intensity as cortical and hippocampal gray matter and are more pronounced on the right side. The arrows point to the pPNH. The hippocampus is marked with an asterisk.

A 33-year-old man presented with a 2-year history of focal seizures. A previous brain MRI scan was reported to show dilation of third and fourth ventricles with possible aqueduct stenosis and tonsillar ectopia. Repeated brain MRI revealed bilateral posterior periventricular nodular heterotopias (pPNH), a malformation of cortical development, lining the occipital and temporal horns of both ventricles (figure, A–F), which was retrospectively visible in the first MRI scan. pPNH may present with epilepsy but can easily be missed. White matter volume decreases and other associated brain abnormalities are often seen in pPNH and should prompt careful review of the periventricular region.¹ Epilepsy surgery in bilateral pPNH has a less favorable outcome.²

AUTHOR CONTRIBUTIONS

Stjepana Kovac: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval. Caroline Micallef: drafting/revising

the manuscript, accepts responsibility for conduct of research and final approval, interpretation of data—namely of the MRI brain scan. Beate Diehl: drafting/revising the manuscript, study concept or design, analysis or interpretation of data, accepts responsibility for conduct of research and final approval. John S. Duncan: drafting/revising the manuscript, accepts responsibility for conduct of research and final approval.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

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

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MYSTERY CASE RESPONSES

The Mystery Case series was initiated by the *Neurology*® Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses were solicited through a group e-mail sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media. All the answers that we received came through social media, from individuals rather than groups. Most of the respondents (75%) correctly indicated the presence of bilateral periventricular nodular heterotopias (PNH) on the brain MRI associated with diffuse cortical atrophy. The most complete answer came from Dr. Felipe Borlot (Clinical Fellow, Toronto Western Hospital and University of Toronto, Canada). In his response, he pointed out that PNH is a brain malformation due to abnormal

neuronal migration in which a subset of neurons fails to migrate into the developing cerebral cortex and remains as nodules that line the ventricular surface. Classical PNH is a rare X-linked dominant disorder, generally associated with prenatal lethality in males. X-linked PNH is associated with mutations in the *filamin A* gene, mapping to chromosome Xq28. Recently an autosomal recessive form of PNH due to mutations of the *ARFGEF2* gene was also reported. *ARFGEF2* gene protein product is involved in vesicular trafficking within the cell and plays an important role in controlling the migration of neurons during the development of the brain. This Mystery Case illustrates a classic neuronal migration disorder that can present with epilepsy both in childhood and adult age.

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Neurology 2013;81:e163-e164

DOI 10.1212/01.wnl.0000436613.73952.4e

This information is current as of November 25, 2013

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