Mystery Case: Bilateral posterior periventricular heterotopias

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 tonsilar 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. Epilepsy surgery in bilateral pPNH has a less favorable outcome.

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, study concept or design, accepts responsibility for conduct of research and final approval. Beate Diehl: drafting/revising the manuscript, 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.

REFERENCES
Mystery Case Responses

The Mystery Case series was initiated by the Neurol-
yogy® Resident & Fellow Section to develop the clin-
ical 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 Consor-
tium 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 asso-
ciated with diffuse cortical trophy. The most com-
plete answer came from Dr. Felippe 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 illus-
trates a classic neuronal migration disorder that can
present with epilepsy both in childhood and adult age.

Dragos A. Nita, MD, PhD, FRCPC
Division of Neurology, The Hospital for Sick Children, Uni-
versity of Toronto, Canada
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Stjepana Kovac, Caroline Micallef, Beate Diehl, et al.

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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