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.

REFERENCES
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 trophy. The most complete 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 illustrates a classic neuronal migration disorder that can present with epilepsy both in childhood and adult age.

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