Transmantle focal cortical dysplasia in a patient with drug-resistant epilepsy

Varun Padmanaban ,¹ Jennifer Baccon,^{2,3} Jayant Acharya,⁴ Michael Sather¹

¹Department of Neurosurgery, Penn State Health Milton S. Hershey Medical Center, Hershey, Pennsylvania, USA

²Department of Pathology and Laboratory Medicine, Akron Children's Hospital, Akron, Ohio, USA

³Department of Pathology, Northeast Ohio Medical University, Rootstown, Ohio, USA

⁴Department of Neurology, Penn State Health Milton S. Hershey Medical Center, Hershey, Pennsylvania, USA

Correspondence to

Dr Michael Sather;
msather@pennstatehealth.psu.edu

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DESCRIPTION

Focal cortical dysplasia (FCD) is a malformation of cortical development and is a common cause of drug-resistant epilepsy in children and adults.¹ We review images in a patient who was found to have FCD after suffering for years with drug-resistant epilepsy. Her imaging and pathological findings were classic for FCD type IIB (transmantle dysplasia). Her case highlights the importance of a thorough workup by a multidisciplinary epilepsy surgery team with an epilepsy-specific MRI scan.

The patient is a middle-aged woman with medically resistant epilepsy. She began having seizures around the age of 3. The semiology of her seizures includes rare generalised tonic clonic seizures with frequent night-time auras, which occasionally progress to complex partial seizures. She describes her auras as 'gasping for air' and a feeling that she cannot catch her breath. She had failed five antiepileptic agents, and prior to definitive surgical treatment, she was on two agents with moderate control. She also had a vagal nerve stimulator (VNS) placed with minimal improvement in her symptoms many years prior at an outside facility.

On referral to our epilepsy surgeon for replacement of her VNS generator, an epilepsy protocol MRI brain was performed. This revealed a classic 'transmantle sign' with fluid-attenuated inversion recovery (FLAIR) signal abnormality along the right frontal lobe extending to the ventricle, consistent with FCD (figure 1, arrow). She underwent

Figure 2 (A) H&E section showing dysmorphic neurons (balloon cells, star) in background of cortical disorganisation. (B) Neurofilament immunohistochemical staining highlighting balloon cells.

a craniotomy with placement of subdural and depth electrodes for invasive seizure monitoring. Multiple stereotypical bilateral asymmetric tonic seizures were observed during sleep. Electroencephalography (EEG) findings were localised to the lesion seen on MRI. She underwent extended lesionectomy with motor mapping and maximal safe resection. Pathology noted cortical dysplasia type IIB with dysmorphic neurons on H&E staining (figure 2A, star) and balloon cells seen with neurofilament protein (NFP) staining (figure 2B). Following surgery, she had a mild left-sided weakness which resolved by her first postoperative clinic visit. She has been seizure free for over 7 years with significant improvement in her quality of life and memory, and she has regained her driver's license.

The International League Against Epilepsy has published a clinicopathological classification system describing three subtypes of FCD.² Typical FCD imaging findings include cortical thickening, blurring of the grey-white matter junction as well as T2 and FLAIR hyperintensity within the grey and/or white matter. Type II FCD is the most common and homogenous type of FCD and is typically found within the frontal lobe often with a characteristic 'transmantle sign'.^{3,4} Advanced imaging, including epilepsy protocol MRI with a 3 T magnet, has significantly improved our ability to find FCD lesions.⁵ Treatment for FCD should include surgical evaluation with rates of seizure-freedom ranging from 62% to 90%.^{6,7}

It is the responsibility of neurologists and neurosurgeons to critically evaluate and re-evaluate decisions in patients with epilepsy, including with placement or replacement of VNS. Epilepsy-specific MRI protocols such as the Harmonized Neuroimaging of Epilepsy Structural Sequences MRI protocol (HARNESS-MRI) are required for any patient with epilepsy and should be repeated if initial images are suboptimal or not available as in the case of this patient.^{8,9}

Figure 1 Coronal fluid-attenuated inversion recovery (FLAIR) sequence MRI showing transmantle dysplasia involving the right superior frontal gyrus.

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Patient's perspective

I feel the epilepsy team helped me wonderfully. I was able to go back to school (and) got my graduate equivalency degree (GED). I was not able to do anything—I had very poor memory due to my seizures. I have five kids and it was tough on my husband as well. My kids were basically taking care of me. We came to the agreement of going through with the surgery and now I am totally seizure free. I never thought I would be able to go back to school but was able to get my GED.

Learning points

- ▶ Focal cortical dysplasia is a common cause of drug-resistant epilepsy in adults and should prompt surgical epilepsy evaluation.
- ▶ Epilepsy protocol MRI is integral to workup of drug-resistant epilepsy and should be repeated if initial images are suboptimal or not available.
- ▶ Focal cortical dysplasia type IIb classically involves the frontal lobe and transmantle sign is nearly pathognomonic. This classic radiographic sign is correlated with dysmorphic nuclei and balloon cells on pathology.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

ORCID iD

Varun Padmanaban <http://orcid.org/0000-0002-9538-7708>

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