■ Multimedia teaching material

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Epileptic Disorders

Early detection and evolution of hypsarrhythmia in a patient with subcortical band heterotopia

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A two-week-old full-term baby with XY genotype, but female phenotype, and bilateral ventriculomegaly on prenatal ultrasound presented with failure to thrive and episodes of right arm shaking. Interictal EEG was initially normal (*figure 1A*). Episodes of right arm shaking were not captured on EEG. Two months later, repeat EEG after a witnessed bilateral clonic seizure revealed abundant, independent bitemporal spikes (*figure 1B*). MRI of the brain showed band heterotopia along the lateral ventricles and mild ventriculomegaly (*figure 2*).

At five months, EEG obtained after breakthrough bilateral clonic seizures showed hypsarrhythmia (*figure 1C*). After two weeks of starting prednisolone, the hypsarrhythmia background improved noticeably with reduced amplitude and relative organization, although multifocal epileptiform discharges persisted (*figure 1D*). No additional seizures were noted. The patient had global developmental delay since birth, however, no regression. Genetic testing with epilepsy and brain malformation gene panels was unrevealing for an etiology of subcortical band heterotopia (SBH). SBH can present with infantile spasms with or without hypsarrhythmia and is often related to mutations in the *PAFAH1B1* or *XLIS* genes [1]. Notably, our patient did not develop infantile spasms. *PPP1R12A* gene testing, to rule out *PPP1R12A*-related urogenital and/or brain malformation syndrome, was negative.

These findings support the importance of keeping a low threshold for follow-up EEGs for hypsarrhythmia in patients with malformations of cortical development, such as band heterotopia, despite unrevealing initial EEGs and an inconsistent semiology. Early detection and treatment of hypsarrhythmia have been associated with an improved prognosis depending on the underlying etiology [2, 3]. ■

Supplementary material.
Summary slides accompanying the manuscript are available at www.epilepticdisorders.com.

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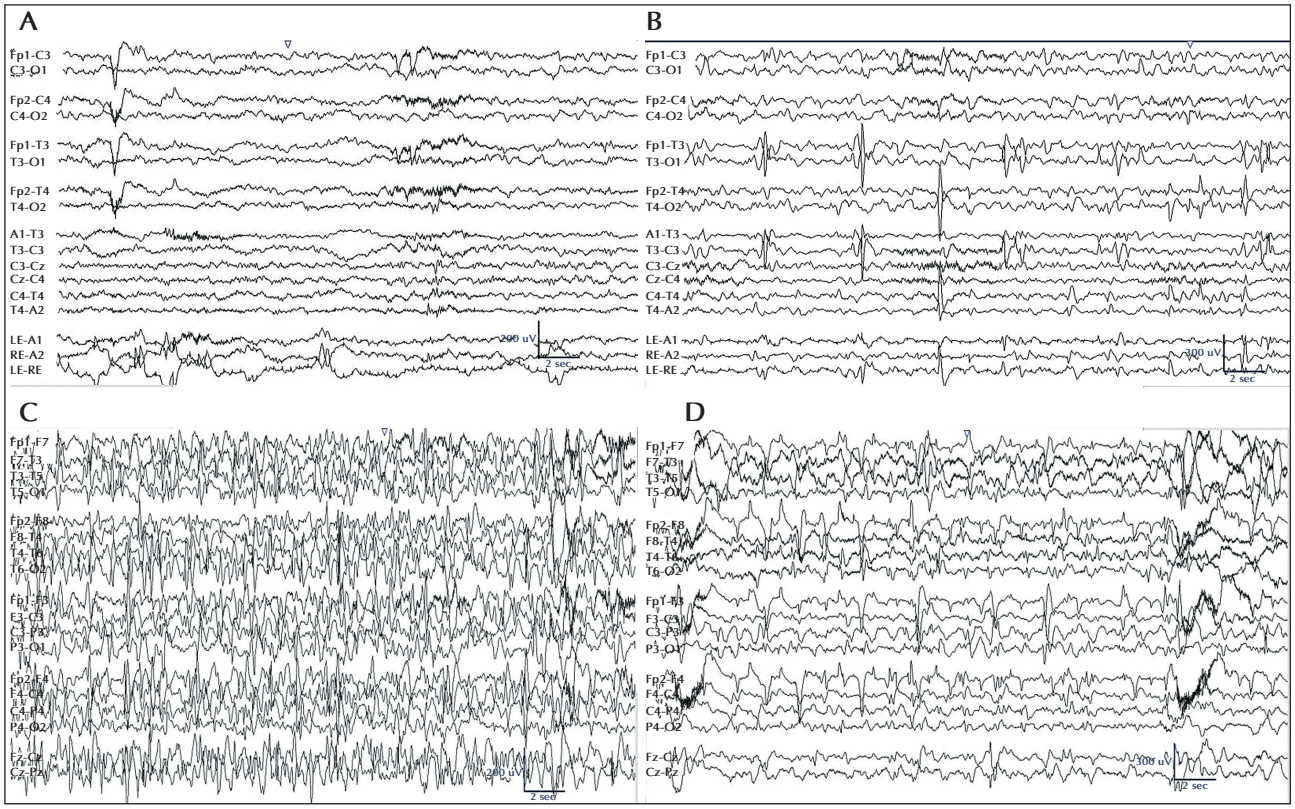
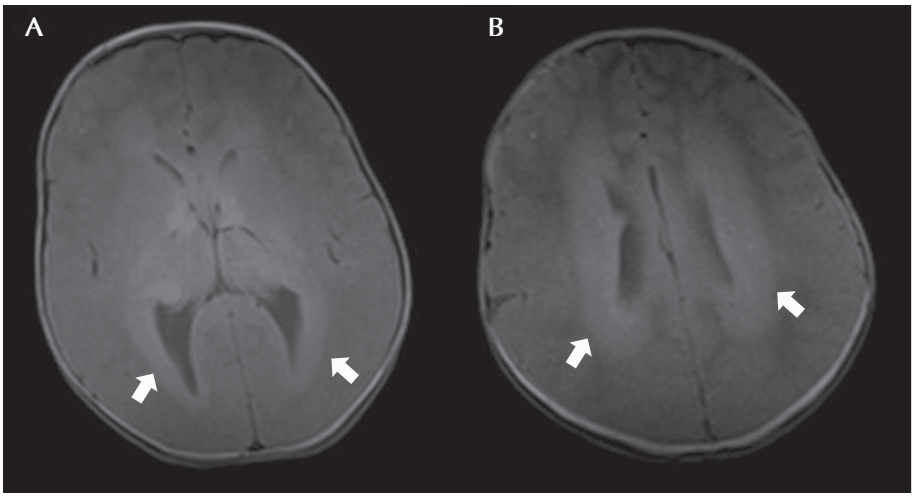


■ **Figure 1.** Evolution of hypsarrhythmia on serial EEG. (A) Normal EEG at two weeks of age on neonatal montage. (B) Abundant bitemporal spikes at two months of age on neonatal montage. (C) Hypsarrhythmia at five months of age. (D) Improvement in background, two weeks after initiation of prednisolone, although with persistence of interictal abnormalities. EEG settings: (A, C) sensitivity 10 uV/mm, LFF 1 Hz, HFF 70 Hz; (B, D) sensitivity 15 uV/mm, LFF 1 Hz, HFF 70 Hz.



■ **Figure 2.** (A, B) Brain MRI showing a band of abnormal signal surrounding the lateral ventricles symmetrically, which follows gray matter on T1 axial images, and bilateral ventriculomegaly.

TEST YOURSELF

(1) A nine-week-old girl with subcortical band heterotopia presented with right arm and leg clonic seizures. Interictal EEG showed frequent left temporal spikes. Topiramate was initiated resulting in effective seizure control. Molecular genetic testing confirmed mutations in the *PAFAH1B1* gene. Six weeks later, the patient's mother brought her to the emergency room (ER) following new activity concerning for multiple bilateral clonic seizures followed by a gradual return to baseline. What is the next best step in management?

A. Send for repeat genetic testing
B. Perform repeat brain MRI to look for new lesions
C. Admit for EEG monitoring for possible hypsarrhythmia
D. Add a new seizure medication without additional testing and discharge from the ER

(2) Which of the following can be seen in patients with subcortical band heterotopia?

A. Infantile spasms
B. Mutations in *PAFAH1B1* or *XLIS* genes
C. Hypsarrhythmia
D. All of the above

Note: Reading the manuscript provides an answer to all questions. Correct answers may be accessed on the website, www.epilepticdisorders.com.