

Commentary

Commentary for 'Rare Seizure Presentation in 3-year-Old Male: Case of Focal Epilepsy Associated with Squatting and Running'

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The case report by Musa et al. (2025) highlights the challenges faced by pediatricians in diagnosing pediatric epilepsy, especially when focal epileptic episodes present similarly to benign childhood behavior. This case of a 3-year-old male initially presumed to have breath-holding spells (BHSs) but later diagnosed with focal seizures illustrates how atypical clinical features in children can lead to misinterpretation and delayed diagnosis. This case further demonstrates the importance of electroencephalography and neuroimaging as critical diagnostic tools. A notable strength of the report is its emphasis on atypical seizure presentations and the need for timely neurological workups. Unlike adult seizures, pediatric episodes do not present with the expected motor features, stiffness, twitching, and urinary incontinence, which typically necessitates early electroencephalogram and neuroimaging. Focal seizures in children may manifest with subtle behaviors that resemble benign childhood phenomena such as tantrums or breath-holding spells. This overlap complicates timely diagnosis and may delay treatment initiation.

Breath-holding spells, while distressing to parents, are benign and often self-limiting. They are usually triggered by frustration or pain and characterized by transient apnea, cyanosis, and possible transient loss of consciousness. Importantly, BHSs lack postictal phenomena and sustained neurological sequelae. By contrast, the patient described in this case began to display additional features, including gaze deviation, urinary incontinence, and spontaneous crying. These symptoms went beyond the scope of BHSs, illustrating the need for providers to maintain a high suspicion when evaluating prolonged or recurrent paroxysmal childhood events. The MRI findings of periventricular leukomalacia (PVL) further reveal the neurological complexity underlying this child's seizures. While PVL is often associated with prematurity and hypoxic-ischemic injury, consequences, such as heightened seizure risk, may not appear until later during development. Early identification of atypical seizures not only helps provide appropriate treatment but also prevents potential long-term neurological effects. Uncontrolled seizures can result in structural and functional brain changes leading to cognitive impairment, memory deficit, and behavioral challenges. Therefore, prompt imaging may help provide an explanation for subtle seizure phenotypes.

This case report contributes meaningfully to the literature by

illuminating how atypical epilepsy can present as benign conditions, thereby delaying care. General pediatricians should be vigilant when BHSs present with additional neurologic features. Referral for EEG and neuroimaging should occur without hesitation in order to provide adequate treatment for these pediatric patients. However, it is important to acknowledge that in underserved regions, the turnaround time between a pediatrician's initial recognition of atypical events and formal evaluation by a neurologist may be significantly prolonged. Such a delay arises due to limited access to subspecialists and extended scheduling intervals for neurology evaluations. This highlights the importance of expanding subspecialty resources and capacity. For these reasons, providers must stay attentive for early and accurate diagnosis in order to ensure equitable and timely care for underserved pediatric populations.

Citation:

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