Epilepsy with Myoclonic Absence: Proximal Myoclonus in Absence Seizures Is the Clue

Chandana Bhagwat[1] Pradeep Kumar Gunasekaran[1] Anju Gupta[1] Arushi Gahlot Saini[1]

[1] Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh, India

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Address for correspondence Arushi Gahlot Saini, MD, DM, Department of Pediatrics, Postgraduate Institute of Medical Education and Research, Chandigarh 160012, India (e-mail: doc.arushi@gmail.com).

A 7-year-old boy, second born to nonconsanguineously married parents, with normal perinatal period, presented with proximal shoulder jerking associated with uprolling of eyes and loss of awareness. Each episode lasted 10 to 15 seconds, and he had 12 to 15 episodes a day, both in awake state and during sleep for the past 7 to 8 months. He had no significant family history. He had a good scholastic performance. His anthropometry, systemic, and neurological examinations were within normal limits, without any neurocutaneous markers. Awake ictal electroencephalogram (EEG) revealed generalized 3- to 3.5-Hz spike-and-wave discharges coinciding with myoclonic jerks on hyperventilation (►**Video 1**). An electroclinical diagnosis of epilepsy with myoclonic absences was made and the patient was started on sodium valproate, followed by ethosuximide and lamotrigine. A ketogenic diet was not tried. At 2 years of follow-up, he showed improvement with 60 to 70% reduction of seizure episodes. He was controlled on ethosuximide alone, and the other two were tapered and stopped.

Video 1

Video electroencephalographic (VEEG) recording in the index case during hyperventilation with outstretched hands (common average montage, sensitivity: 50 µV/mm, sweep speed: 30 mm/s) induces the typical ictal event as cessation of ongoing activity, unrolling of eyes, proximal jerks in the upper limbs and head lasting for a few seconds, and rapid regain of consciousness. The child had no memory of the event. The corresponding EEG shows abrupt onset of high-amplitude, monomorphic, bifrontal-dominant, generalized 3- to 3.5-Hz spike-and-wave discharges. The seizure starts a few seconds after the onset of discharge. Online content including video sequences viewable at: https://www.thieme-connect.com/products/ejournals/html/10.1055/s-0044-1790247.

Tassinari's syndrome, also known as epilepsy with myoclonic absence (EMA), is a rare childhood generalized epilepsy syndrome, first described by Tassinari et al in 1969.[1,2] EMA is characterized by the rhythmic, myoclonic jerking of the head, extremities, or both, with impairment of awareness.[3] The mean age of onset of seizures is 3.5 to 5.2 years.[2,4] Affected children may have normal intellect before the onset of seizures, but most have global developmental delay, language impairment, or cognitive slowing after the onset of symptoms.[2] The ictal EEG pattern in EMA includes bilateral, synchronous, and rhythmic spike-and-wave complexes at 3- to 3.5-Hz frequencies, time-locked with myoclonus.[2,4] The preferred antiseizure medications (ASMs) reported in the literature are sodium valproate, lamotrigine, and ethosuximide, similar to the index case.[2,4] The other epileptic syndromes that overlap with EMA are childhood absence epilepsy, myoclonic-astatic epilepsy, and juvenile myoclonic epilepsy. Transient response to ketogenic diet has been reported.[2] Long-term prognosis of EMA is guarded, and drug-refractory epilepsy is common.[2,4] In children with absence seizures, proximal jerks can be the diagnostic clue for Tassinari's syndrome.

Authors' Contribution

C.B., P.K.G., A.G. and A.G.S. contributed to the study design, writing, editing, and drafting. A.G.S. was responsible for

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the intellectual content of the study. All the authors were involved with critical revision and final approval of the manuscript.

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Conflict of Interest
None declared.

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