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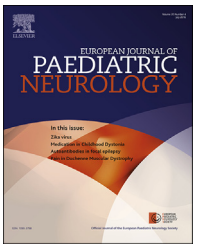

Case study

Myoclonic absence seizures with complex gestural automatisms

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ABSTRACT

Epilepsy with myoclonic absences is a rare generalized epilepsy syndrome with distinctive seizures. Two unrelated children had mild developmental impairment and onset of myoclonic-absences at 3 and 8 years. Seizures were characterized by bilateral 3 Hz myoclonic jerks superimposed on tonic abduction of the upper limbs. Events lasted 10–60 s, and complex gestural automatisms were often observed; in one case, a boy undid his seatbelt and attempted to exit a moving vehicle. Post-ictally, both children immediately regained awareness without recollection of their actions. Ictal EEGs showed 3 Hz generalized spike-wave. Complex automatisms have not been described in myoclonic absence seizures. This generalized seizure type can be confused with focal seizures when these ictal behaviours occur.

1. Introduction

Myoclonic absence seizures are a rare type of generalized seizure involving tonic upper limb abduction and ~3 Hz clonic jerking.[1] Events typically last 10–60 s and autonomic features, such as urinary incontinence and apnoea, are commonly associated.[1] Partial or complete loss of awareness occurs during the seizure, with an instantaneous return to full awareness post-ictally.[1] Ictal EEG shows generalized spike-wave discharges at 3 Hz, coinciding with the clonic jerks.[1] Myoclonic absence seizures are classically associated with the syndrome of epilepsy with myoclonic absence seizures (EMA),[1] but have also been reported in Angelman syndrome,[2] GLUT1 deficiency syndrome[3] and Dravet syndrome.[4]

Here, we present two patients with EMA in which myoclonic absence seizures involved complex automatisms, an ictal feature not previously reported.

2. Case studies

Patient 1: A nine-year-old boy had absence seizures from four years. The events initially involved blank staring and eyelid

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flickering, lasting 10–60 s, and tended to cluster with up to 40 events over 20 min. By eight years, the events involved tonic arm abduction with superimposed rhythmic upper body jerking, in addition to purposeful actions of which the patient was unaware (Supplementary Video). For example, while riding in the car with his mother, the patient had a seizure during which he undid his seatbelt and tried to exit the moving vehicle. Post-ictally, he immediately returned to baseline awareness with no recollection of his actions. The seizures were provoked by hyperventilation and occurred many times per day despite trials of valproate, levetiracetam, topiramate, lamotrigine, carbamazepine and ethosuximide. Interictal EEG showed generalized spike-wave and polyspike-wave, and generalized photoparoxysmal spikes at 10 and 12 Hz photic stimulation; ictal EEG showed 3 Hz spike-wave occurring in tandem with upper body clonic movements (Fig. 1A and C).

Supplementary video related to this article can be found at https://doi.org/10.1016/j.ejpn.2017.12.003

Early developmental milestones were normal but he had borderline intellectual disability. There was no family history of seizures. *SLC2A1* sequencing was negative. Brain MRI was normal.

Patient 2: A 13-year-old girl had absence seizures from three years, sometimes associated with rhythmic upper limb jerking. During events, parents noted she would sometimes walk in the wrong direction or continue with activities such as drawing, clapping, dancing or singing; and would not recall her actions afterwards (Supplementary Video). Seizures typically lasted 10–15 s, and were provoked by sunlight and stress. Post-ictally, she returned immediately to baseline awareness. Interictal EEG showed fragments of frontally-predominant generalized spike-wave and polyspike-wave; ictal EEG demonstrated 3 Hz generalized spike-wave co-occurring with rhythmic synchronous shoulder abduction (Fig. 1B and D).

She initially required dual therapy with valproate and lamotrigine to control seizures, but on most recent review had been seizure-free on lamotrigine monotherapy for four years. Topiramate was trialed early in her course and did not improve seizure frequency.

Early milestones were normal; however, she had borderline intellectual disability. She exhibited anxiety, depression and aggressive behaviours, all of which improved with sertraline, but worsened when valproate was weaned. There was no history of regression.

She had dysmorphic features including frontal bossing, receding hairline, hirsutism and hypertelorism, as well as symmetrically large growth parameters (height 172 cm and weight 62.4 kg at 12 years, both >95th percentile); these features led to a suspicion of Sotos or other genetic overgrowth syndrome. Comparative genomic hybridization microarray and sequencing for *SLC2A1*, *SCN1A* and *NSD1* were all negative. Family history was significant for a paternal cousin with a single febrile seizure, a paternal great-uncle with seizures associated with poliomyelitis, and a distant maternal relative with two tonic-clonic seizures and mild intellectual disability. Brain MRI was normal.

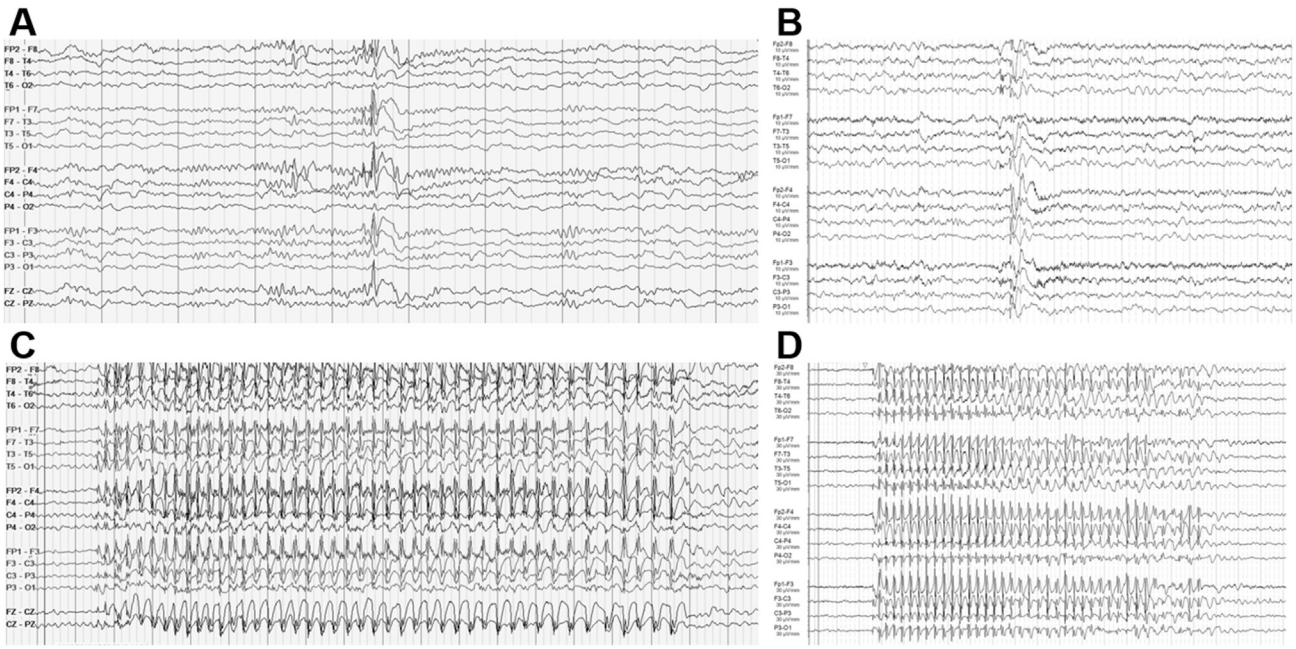

Fig. 1 – Interictal and Ictal EEG – Interictal EEG shows fragments of generalized polyspike-wave discharges in light sleep for patient 1 (A) and in wakefulness for patient 2 (B). Ictal EEG during 15–16 s myoclonic absence seizures for patients 1 (C) and 2 (D) demonstrates evolving generalized spike-wave, initially at 3 Hz and slowing as the seizure progresses. For all studies, bipolar montage used and thickened vertical lines represent 1 s intervals. Sensitivity was set to 10 μV/mm for interictal recordings (A and B) and 30 μV/mm for ictal EEG (C and D).

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