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Childhood idiopathic generalized epilepsy with phantom absences: A case report

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Abstract

Idiopathic generalized epilepsy with phantom absences manifests in adults with phantom absences, generalized tonic-clonic seizures and absence status epilepticus. Phantom absences are inconspicuous seizures that are usually discovered on electroencephalogram recommended after the onset of convulsive seizures. Phantom absences have been very rarely reported in children, in which they have been recognized only after absence status epilepticus. We report an 8 year-old girl presenting with phantom absences as the first overt symptom of idiopathic generalized epilepsy. She fulfilled the diagnostic criteria for phantom absences, including mild impairment of consciousness associated with brief (< 5 seconds) generalized spike and slow wave discharges >2.5 Hz on electroencephalogram. Diagnostic requirements for IGE were present, including normal examination, electroencephalography background activity and brain magnetic resonance imaging. Treatment with valproate was effective. Identification of phantom absences is important, since patients may suffer from phantom absences for years before a more dramatic epileptic event occurs.

Keywords

Absences; epilepsy; EEG; children.

Introduction

Absence seizures and the related epileptic syndromes are an area of controversy owing to possible overlap in clinical presentation that may pose difficulties in classification, prognosis and treatment [1]. Syndromes of Idiopathic Generalized Epilepsy (IGE) with typical absences recognized by the International League Against Epilepsy (ILAE) include childhood absence epilepsy, juvenile absence epilepsy, juvenile myoclonic epilepsy and epilepsy with myoclonic absences [1,2]. However, there are other probable syndromes of IGE with typical absences to consider, including IGE with phantom absences [3]. Recognition of

this condition may be difficult, as phantom absences are so mild that they may be undetected even by the patient himself [4]. IGE with phantom absences has been described in adults, and it is characterized by the triad of phantom absences, Generalized Tonic-Clonic Seizures (GTCS) and absence status epilepticus (SE) [5]. GTCS are usually the first overt clinical manifestation, although absences may have started much earlier [6]. Phantom absences have been very rarely reported in childhood with IGE, in which they have been recognized only after absence SE [7,8].

Herein we report a young girl presenting with phantom absences, whose clinical, EEG and brain MRI features were consistent with IGE. Informed consent was obtained from the patient's parents for recording and publishing the data.

Case Presentation

An 8 year-old girl was admitted to our hospital for assessment of a suspected seizure occurring in the early morning, after a nocturnal sleep disturbed by cough. Her parents noted that she was suddenly motionless and unresponsive, with blank stare and upward eyes rolling. The spell could not be interrupted and lasted 2-3 minutes, after which the patient recovered quickly, although she was not able to recall any details about the episode. Past medical history was remarkable for a febrile seizure occurring at the age of 3 years, while development during early childhood, sleep and school results were normal. Family history was negative for seizures, epilepsy or other neurological disorders. General and neurological examinations were normal and age-appropriate. Electroencephalogram (EEG) showed 3-4 Hz Generalized spikes/multiple Spikes and Slow Wave Discharges (GSWD) of brief duration (less than 5 seconds), occurring soon after hyperventilation, with normal background activity (Figure 1). GSWD were associated with staring and impairment of consciousness. Interictal short transients of localized spikes or sharp waves were also present. Sleep EEG revealed brief runs of generalized polyspikes and slow waves (Figure 2). Brain magnetic resonance imaging (MRI) was unremarkable. Valproate was started and titrated to 15 mg/Kg/d, following which the patient's staring spells did not recur. Two weeks after starting therapy, sleep EEG showed a clear improvement, with only a few polyspikes during sleep (Figure 3). At 1 year follow-up, no further seizure was reported.

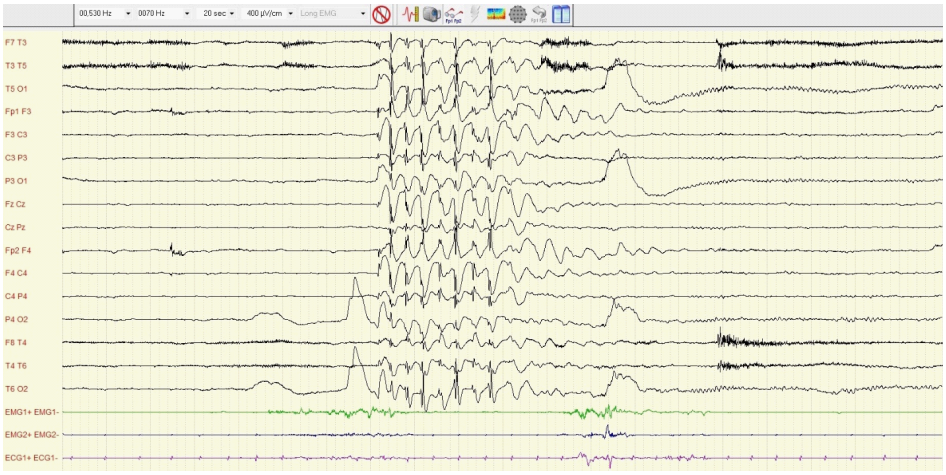

Figure 1: Ictal EEG: brief generalized discharge of high-amplitude 3-4 Hz spikes/multiple spikes and slow waves. Head movement and muscle artifacts are evident before and after the discharge.

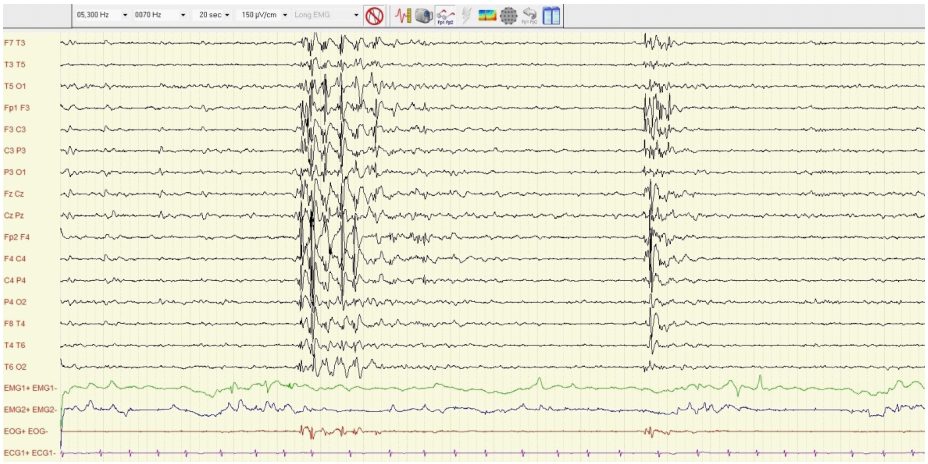

Figure 2: Interictal EEG: generalized discharges of polyspikes and slow waves during NREM sleep, stage 2.

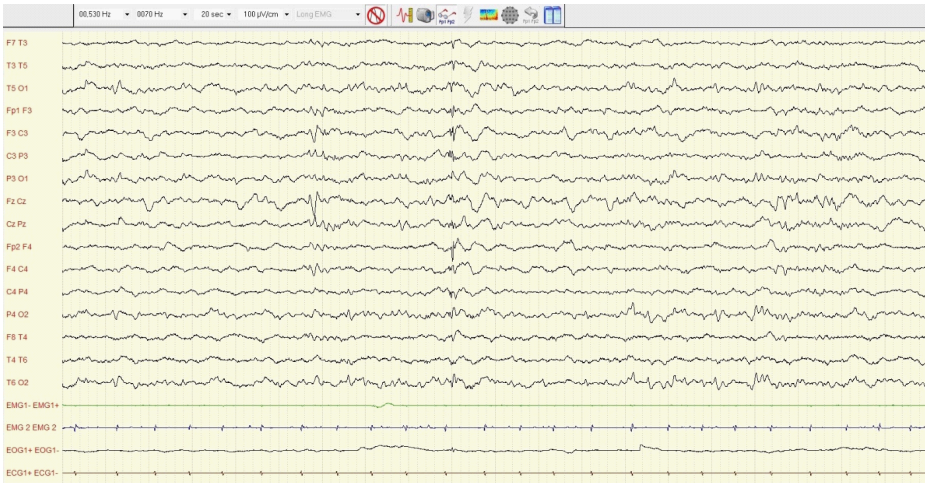

Figure 3: After starting antiepileptic therapy, only a few polyspikes during sleep were evident.

Discussion

Our patient fulfilled the diagnostic criteria for phantom absences, including mild impairment of consciousness associated with brief (usually no more than 5 seconds) > 2.5-Hz GSWD on EEG [3,9]. Diagnostic requirements for IGE were also present, including normal examination and intellect, normal background EEG activity and brain MRI [9].

Phantom absences are typical absence seizures with the mildest impairment of consciousness, which are inconspicuous to the patient and imperceptible to the observer [5]. Panayiotopoulos et al. described an IGE in adults manifested by phantom absences, GTCS and absence SE [5]. Because of their mildness and brevity, phantom absences had passed unnoticed by all patients and their relatives, and they were only discovered on EEG recommended after the onset of GTCS, which are usually the first overt clinical manifestation of this epilepsy syndrome [5,6].

Phantom absences as overt symptom of IGE have been very rarely reported in childhood [7,8]. Panayotopolous et al. presented a normal 11 year-old girl who had six episodes of absence SE starting at age 10 years, in which the video-EEG documented numerous typical absence seizures, whose occurrence was totally unsuspected by either the patient or their parents [7]. Adams et al. reported an 8 year-old boy who

experienced two episodes of absence SE, followed by staring episodes lasting a few seconds over the next months [8]. In our patient, phantom absences were discovered by an EEG obtained a few hours after the seizure occurring at home, which followed poor sleep owing to cough. This finding seems to be consistent with the view that extending sleep EEG by a prolonged recording on awakening with one or multiple sessions of hyperventilation may reveal possible absences [9]. The seizure occurring at home could indeed represent a brief cluster of phantom absences that did not evolve into absence SE [5].

In IGE with phantom absences, all tests apart from the EEG are normal [6]. The background activity is normal, although 50% of patients have EEG focal paroxysmal abnormalities, occurring either independently or in association with the generalized discharges. Ictal EEG consists of 3-4 Hz spikes/multiple spikes and slow wave with occasional fragmentations, which are typically brief (2-4 seconds), lasting usually no more than 5 seconds [6]. Distinctive EEG features have been described, such as abundance of polyspkipes and fast paroxysmal activities during sleep in the interictal EEG, and runs of polyspikes preceding the absence-related spike-wave discharge in the ictal EEG [4]. A polyspike component appears to be an EEG characteristic of IGE with phantom absences, reinforcing the view that polyspikes are not necessarily associated with myoclonic seizures [9]. Focal paroxysmal abnormalities and polyspikes were also present in our patient.

Clinicians should be aware of childhood onset cases of IGE with phantom absences. This rare epilepsy syndrome is not yet recognized by the ILAE, and further case reports and series are warranted to better understand it [8]. Early identification of phantom absences is important, since patients may suffer from phantom absences for years before a more dramatic epileptic event occurs, such as GTCS or absence SE [10].

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