

EYELID MYOCLONIA (JEAVONS SYNDROME) IN 11- YEAR OLD GIRL: A CASE STUDY AND REVIEW OF THE LITERATURE

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Abstract

We report a case of an 11-year old girl with episodes of squinting and upward deviation of the eyes. Awakening EEG showed generalized spike and wave discharges and photosensitivity. After valproic acid therapy she became free of these episodes, but a follow-up EEG remained abnormal. A video EEG demonstrated considerable eyelid myoclonia and absence seizures.

Jeavons syndrome is a rare and frequently overlooked epileptic syndrome. The video EEG proved to be the only diagnostic tool for recognition of the full range of symptoms. In the discussion we present a short review of the recent articles on this syndrome.

Keywords: eyelid myoclonia, absences, EEG, Video EEG

Rezumat

Raportam cazul unei paciente de 11 ani cu episoade de devierea inaintu si in sus a globilor oculari. EEG de veghe a aratat descarcari generalizate de complexe varf-unda si fotosensibilitate. După începerea tratamentului cu valproat de sodiu aceste episoade au dispărut, dar EEG a rămas anormal. Video EEG a demonstrat numeroase episoade de absențe cu mioclonii palpebrale.

Sindromul Jeavons este rar si este adesea nedignificat. Video EEG s-a dovedit a fi singurul instrument valoros pentru recunoasterea tuturor simptomelor. In partea de discutii prezentam o scurta trecere in revista a datelor din literatura in legatura cu acest sindrom.

Cuvinte cheie: sindrom Jeavons, absențe, EEG, Video EEG

Introduction

Eyelid myoclonia with absences (Jeavons syndrome, EMA) is a syndrome not yet recognized by the International League Against Epilepsy (ILAE) ¹, and not listed in the proposed diagnostic scheme published by Engel in 2001 ². It is present on the List of Syndromes and Idiopathic Generalized Epilepsies not Recognized by ILAE ³. The syndrome was described originally by Jeavons in 1977⁴.

The prevalence of Jeavons syndrome is around 3% among adult patients with epilepsy⁵. The characteristic seizure is a brief episode of marked jerking of the eyelids with upward deviation of the eyes, associated with a

generalized spike – wave discharge (GSWD), occurring on closure of the eyes ^{6,7}. All patients are photosensitive ⁶⁻¹¹. The period of absence follows the eyelid myoclonia, while the eyelid jerking becomes less violent than at the onset. Impairment of consciousness is usually mild, manifested by cessation, repetition, errors, and delays in counting. Automatism are not observed ^{6,7}.

Eyelid myoclonia, not absence seizures, are the hallmark of this syndrome^{6,7}.

General tonic-clonic seizures (GTCS) are also reported, as well as sporadic absence status epilepticus and myoclonic jerks other than eyelid myoclonias^{6,7,9-13}.

The mean age of onset is 6-8 years (range 2-14), being earlier than in other photosensitive

epilepsies^{6,7,9,11,13}. Most authors support the view that it is an idiopathic epileptic syndrome, genetically determined, affecting otherwise normal children, with preponderance of females^{6,7,9}. Consistently with the idiopathic epilepsy definition, patients have normal neurological status and the neuroimaging studies reveal no pathology. Slight mental deficiency or poor academic performance are not considered to be exclusion criteria^{10,14,15}.

The symptom of eyelid myoclonia alone is not sufficient to characterize EMA because it may occur in cryptogenic or symptomatic epilepsies, which are betrayed by developmental delay, learning difficulties, neurological deficits, abnormal MRI, and abnormal EEG background¹⁶.

All tests apart from the EEG are normal. Video EEG is the single most important procedure for a diagnosis.

Case report

An 11-year old girl was admitted to our hospital because of episodes of squinting and upward deviation of the eyes, which occurred many times a day.

She was born as the third child in a family. Pregnancy and delivery were uneventful. The family history and developmental milestones were normal and she attended elementary school, with average results.

A routine awake EEG showed a brief, 3-6HZ generalized spike, polyspike and wave discharges, with a preponderance to the left side, and photosensitivity.

Epilepsy treatment with valproic acid at a dosage of 500mg per day was initiated, with a rapid cessation of seizures within several next days. A follow-up EEG after 10 months of therapy revealed multiple generalized discharges as described above, active on eye closure and hyperventilation. The patient and her mother continued to deny any epileptic episodes.

A full video EEG (awake, asleep, awakening - sleep deprivation) was performed to confirm or exclude any epileptic presentations. During the video EEG considerable eyelid

myoclonia, with or without speech and counting interruption was observed, mostly after awakening, and correlating well with the 2-6HZ generalized spike and wave discharges (GSWD), lasting 0,5-7,0 sec., active on eye closure (Fig.1) and hyperventilation.

Brain MRI scans were normal except for a slight asymmetry of the lateral ventricles. Neurological examination did not reveal any abnormalities.

The dose of valproic acid was increased to 800mg per day. A follow-up EEG after 1 month of modified therapy showed a decreased number of GSWD. The patient continued to deny epileptic episodes.

Discussion

There are only few cases of EMA in the recent literature^{5,8,9,11,14,17-21}.

Most of them are reported as a single case reports. The largest groups of patients are found retrospectively among the patients with abnormal EEG and eyelid flutter, eyelid blinking, ticks, idiopathic generalized epilepsies, clinical absences, atypical absences and photoparoxysmal response^{10,15}. EMA patients were found in 75 patients from 288 with diagnosed IGE in Joshi's study¹⁵ and in 35 patients from 469 with IGE in Striano's study¹⁰. Patients with EMA could also be found from those with typical absence status epilepticus^{12,19,20}.

Presented patient showed all the diagnostic criteria of Jeavons syndrome:

- eyelid myoclonia,
- eyelid myoclonia with absences,
- eye closure –induced seizures, EEG paroxysms, or both,
- photosensitivity^{6,7}.

The age of onset was also typical.

Presence of absence seizures is reported by many authors^{5,9,10,12,15,17,21}. In the group of 50 cases studied by Covanis by sleep-wake video EEG after sleep deprivation all patients demonstrated absence seizures¹³.

GTCS are not rare. According to different authors their incidence ranges from 50%¹¹ up

to 100%^{5,21}. We didn't observe them in our patient.

Our patient didn't present myoclonic jerks except for eyelid myoclonia, which was also reported by other authors^{5-7,12,13}. Destina Yalcin described 4 female patients with eyelid myoclonia associated with absences, myoclonic jerks causing falling down and rare GTCS. These patients show the characteristics of both EMA and juvenile myoclonic epilepsy (JME) syndromes. The study supports the opinion that EMA and JME might be dynamic syndromes that tend to evolve one into another²¹.

Photosensitivity is a constant sign in EMA^{8,11,14,17,18}. In Covanis group (50 patients) photosensitivity was present in 92% and it was significant in 76%¹³.

Photosensitivity was present only in the first EEG in our case, before valproic acid (VPA) therapy. Clinical and EEG signs of photosensitivity decrease with age, and can be modified by AEDs^{5-7,10}.

The presence of characteristic eyelid myoclonia is the hallmark of EMA. However, eyelid jerks are often misdiagnosed as facial tics, or as a mannerism. Absences may be easily overlooked, as in our case. Eyelid myoclonia should not be confused with: the rhythmic closing of the eyes seen in other forms of IGE with absences, or in the typical absence seizures of childhood absence epilepsy^{6,7,10,15}.

The electroclinical findings in our study are consistent with a diagnosis of EMA.

The EEG background was age adequate. Some authors support the view that slowing background is not an exclusion criteria^{9,14} but most of them believe that normal EEG background is typical^{6,7,10,11,15}. Hyperventilation could aggravate GSWD and provoke absence seizures^{6,7} as in our case.

The most potent precipitating factor is eye closure, whether this is voluntary, involuntary or reflex. Almost all of the seizures are induced immediately after eye closure in the presence of uninterrupted light. Eye closure in total darkness is ineffective. The paroxysmal discharges could be induced

without eye closure when fixation is eliminated by e.g. Frenzel glasses¹⁷.

Contrary to other forms of photosensitive epilepsies, which are sensitive only to flickering lights, patients with EMA are also sensitive to bright non-flickering lights^{6,7,10}. Some authors consider the eyelid myoclonia as a maneuver used by patients to self induce intermittent photic stimulation and elicit seizures^{8,9}. In Panayiotopoulos study based on numerous video EEG recordings and interviews from 17 patients, self induced seizure were probable in 2 cases²². Also Striano reported only one case with occasionally self-induced seizures, mostly by closely watching TV¹⁰.

Other precipitating factors are sleep deprivation, AEDs discontinuation, awakening, alcohol, menstruation^{6,7,12,19}.

Awakening and sleep deprivation together with eye closure were also triggering factors in our patient.

Jeavons syndrome is a lifelong disorder, even when seizures are well controlled with AEDs. Men have a better prognosis than women.

Eyelid myoclonia is highly resistant to treatment and occurs many times a day, often without apparent absences, and even without demonstrable photosensitivity^{5-7,9,11}. Of 35 patients studied by Striano 5 (14,2%) were drug resistant despite polytherapy¹⁰.

The drug of choice is valproic acid, alone or in combination with clonazepam, ethosuximide or lamotrigine. Our patient responded well to VPA monotherapy.

Carbamazepine, gabapentine, oxcarbazepine, phenytoin, tiagabine and vigabatrin are contraindicated^{6,7,23}. The patient's lifestyle and an avoidance of seizure precipitants are important. Non-pharmacological treatments used for photosensitive patients (i.e. blue glasses) can be beneficial^{6,7,24,25}.

The family background of epilepsy was found in 8/21 patients reported by Isnard¹¹, 17/35 by Striano¹⁰ 14/18 by Parker²⁶. Of the 18 patients with EMA 4 patients had other family members affected by the same syndrome²⁶. There was not family history in our patient.

This case concerns the first published EMA patient in Poland. We support the view that it is a frequently overlooked epileptic syndrome.

The video EEG proved to be the only diagnostic tool for recognition of the full range of symptoms of Jeavons syndrome.

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