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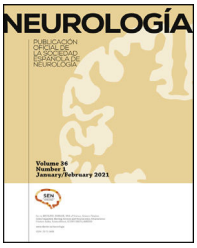

ORIGINAL ARTICLE

Case series of epilepsy with eyelid myoclonia

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KEYWORDS
Eyelid myoclonus with and without absences;
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Photosensitivity;
Sociodemographic factors;
Drug resistance

Abstract
Introduction: epilepsy with palpebral myoclonus is an idiopathic generalised childhood epilepsy, which is underdiagnosed because it is confused with tics or behavioral actions. Patients and Methods: quantitative, observational, descriptive of 16 patients with epilepsy and myoclonic eyelids treated at an institution specialised in neurology, between the years 2017 and 2022. Clinical histories and videoelectroencephalograms were evaluated.
Results: of 16 patients, 11 were women (68.8%), the median age was 17.5 years (IQR 12.5). The first diagnosis most frequently received by patients was idiopathic generalised epilepsy. Stressful situations were the most reported precipitant. 93.75% of the patients initially presented palpebral myoclonic seizures and after several years presented generalised tonic-clonic seizures. The time elapsed between the first seizure and the diagnosis varied between 1 and more than 40 years, being greater among patients with a subsidised health system. Men had a high frequency of family history of epilepsy in the first and second degrees of consanguinity, had a longer delay in diagnosis and reported greater drug resistance. In both women and men, the posterior regions (occipital, temporoccipital and parietoccipital) presented a greater focal epileptiform activity. The photoparoxysmal response occurred with greater predominance in women, with the Waltz type III and IV responses being the most frequent.
Conclusion: the importance of suspecting this pathology before palpebral myoclonus in children should be highlighted, guaranteeing quality and timely management.

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PALABRAS CLAVES
Mioclonía palpebral con y sin ausencias;
Síndrome de jeavons;
Epilepsia generalizada idiopática;
Fotosensibilidad;
Factores sociodemográficos;
Farmacorresistencia

Serie de casos de epilepsia con mioclonías palpebrales

Resumen
Introducción: la epilepsia con mioclonías palpebrales es una epilepsia generalizada idiopática infantil, la cual es infradiagnosticada ya que se confunde con tics o una acción conductual. Pacientes y métodos: cuantitativo, observacional, descriptivo de 16 pacientes con epilepsia y mioclónicas palpebrales atendidos en una institución especializada en neurología, entre los años 2017 al 2022. Se evaluaron las historias clínicas y videoelectroncefalogramas.
Resultados: de 16 pacientes, 11 eran mujeres (68,8%), la mediana de edad fue de 17,5 años (RIC 12,5). El primer diagnóstico que los pacientes recibieron con mayor frecuencia fue epilepsia idiopática generalizada. Las situaciones estresantes fueron el precipitante más reportado. El 93,75% de los pacientes presentaron inicialmente mioclonías palpebrales y al cabo de varios años presentaron convulsiones tónico-clónicas generalizadas. El tiempo transcurrido entre la primera convulsión y el diagnostico varió entre 1 y más de 40 años, siendo mayor entre los pacientes con sistema de salud subsidiado. Los hombres tenían una alta frecuencia de antecedentes familiares de epilepsia en primer y segundo grado de consanguinidad, presentaron mayor demora en el diagnóstico y reportaron mayor farmacorresistencia. Tanto en mujeres como en hombres las regiones posteriores (occipital, temporoccipital y parietoccipital) presentaron mayor actividad epileptiforme focal. La respuesta fotoparoxistica se presentó con mayor predominio en las mujeres, siendo la repuesta tipo III y IV de Waltz las más frecuentes.
Conclusión: se debe resaltar la importancia de sospechar esta patología ante mioclonías palpebrales en población infantil, garantizando calidad y manejo oportuno.

Introduction

Epilepsy with eyelid myoclonia (EEM), previously known as Jeavons syndrome (JS), consists of an idiopathic generalised childhood epilepsy that shares electroclinical features with other idiopathic or genetic generalised epilepsies. No precise epidemiological data on this syndrome are available due to the diagnostic challenges it presents and a lack of knowledge among medical professionals. EEM is calculated to represent 2.7% to 12.9% of generalised epilepsies,[1] with a prevalence of 3% in adults and 13% in the paediatric population with idiopathic generalised epilepsy with absence seizures,[2] and accounts for 0.56% to 2.7% of all cases of epilepsy.[3] EEM manifests during childhood, between the ages of 2 and 14 years (mean age 6.5 [2.5] years),[4] and predominates in women, with a female-to-male ratio of 2:1.[1,5] EEM is assumed to be as frequent as juvenile myoclonic epilepsy; however, it is underdiagnosed because it is mistaken for tics or a behavioural action.[3,4]

EEM is characterised by the following triad of symptoms: eyelid myoclonia with or without absence seizures,[6] seizures with an EEG pattern showing short bilateral activity with generalised polyspike wave discharges at 3—6 Hz induced by eyelid closure in bright environments,[3] and photosensitivity or photoparoxysmal response,[6] which tends to decrease over time.

The aetiology of this syndrome is still being studied. A genetic cause has been proposed, based on the results of studies of cases of concordant monozygotic twins.[3] Mutations that have been associated with the disease include variants of *KCNB1*, *KIAA2022*, *NAA10*, and *CHD2* (chromodomain helicase DNA binding protein 2).[6] Four genes have been described in association with EEM with absence seizures (*SYNGAP1*, *KIA02022/NEXMIF*, *RORB*, and *CHD2*); different alterations have been reported in 3 genes (*LC2A1*, *NAA10*, and *KCNB1*) of some patients, although more evidence is needed to establish a clearer association.[7] Approximately 20% of patients with EEM present a positive family history.[5] It is believed to manifest with an alpha rhythm generator malfunction in the occipital cortex[6]; it has been proposed that this brain area may initiate a generalised epilepsy network including the brainstem and thalamocortical and transcortical pathways.[8,9]

The most widely used treatment is valproic acid; however, due to its teratogenic effect, the drug is avoided in women of childbearing age and wishing to become pregnant[4,6]: levetiracetam is preferred in this population. Other useful medications include lamotrigine, ethosuximide, and zonisamide, which may be administered in monotherapy or in combination with valproic acid.[2,10]

Patients with EEM tend to present drug-resistant epilepsy and therefore require a combination of multiple drugs together with other therapeutic alternatives.[2,10,11]

To date, there are no published epidemiological data on EEM in Colombia nor clear statistical registries of data on clinical and sociodemographic characteristics, electroencephalographic findings, treatment response, comorbidities, type of syndrome manifestation, or time

Table 1 Sociodemographic and clinical characteristics by sex.

	Female ($n = 11$) *n* (%)	Male ($n = 5$) *n* (%)
Age in year 2022[a]	16 (10.5)	19 (29)
Social security system regime		
Subsidised	4 (36.4)	3 (60)
Contributory	3 (27.2)	1 (20)
Private/prepaid healthcare	4 (36.4)	1 (20)
Level of schooling		
Primary education	6 (54.6)	3 (60)
Complete secondary education	3 (27.3)	1 (20)
Technician/technologist	1 (9)	—
Qualified professional	1 (9)	—
Special education	—	1 (20)
History of growth and developmental alterations	5 (45.5)	3 (60)
Family history of epilepsy	3 (27.3)	3 (60)
First diagnosis		
Idiopathic generalised epilepsy	4 (36.4)	2 (40)
Other type of epilepsy	4 (36.4)	1 (20)
Motor tics	1 (9)	—
Other diagnoses	2 (18.2)	2 (40)

[a] Median (interquartile range).

from diagnosis to treatment onset. Therefore, the aim of our study was to describe the sociodemographic and clinical characteristics of a series of Colombian patients with EEM.

Material and methods

This is a quantitative, observational, descriptive study of 16 patients with EEM who were attended at the Fundación Instituto Neurológico de Colombia (FINDEC) between 2017 and 2022. Data were gathered from the clinical record and video-EEG study of each patient. We included all patients with a confirmed diagnosis of EEM or JS and excluded all those for whom video-EEG data were not available.

For each sex, quantitative variables are expressed as medians and interquartile range (IQR), and qualitative variables as absolute and relative frequencies. We used the free software Jamovi 2.2.5.

This study was approved by the ethics committee of our hospital (meeting minutes number 128). During all the stages of the research, patient data were anonymised and remained confidential.

Results

Between 2017 and 2022, we identified 16 patients with a confirmed diagnosis of EEM. A total of 68.8% (11/16) of the sample were female; in 2022, patients were aged between 9 and 54 years (median: 17.5; IQR: 12.5).

The majority of patients were born and resided in the towns of the Valle de Aburrá region (9); one patient was from Venezuela. Most patients belonged to large nuclear families.

Regarding perinatal history, 2 patients were born at home, without complications; one patient was born pre-term, showing intrauterine growth restriction and respiratory difficulty; one was born after a long dystocic delivery; and another was born with microcephaly and developed neonatal jaundice.

The majority of female patients benefited from the contributory and complementary regime of the Colombian health and social security system (63.3%), whereas males more frequently benefited from the subsidised regime. More than 80% of patients had completed primary or secondary education (Table 1).

Of the 16 patients, 8 presented growth and developmental alterations, with the cognitive domain being the most frequently affected (75%; 6/8), and language impairment being the most frequently reported alteration (62.5%). Before onset of seizure symptoms, no history of head trauma was reported. Relevant personal history included: renal tubular acidosis in one female patient, one male with solitary kidney, and 2 patients with cerebral palsy. Two patients had undergone karyotyping, with one showing alterations: a female patient presenting a duplication chromosomopathy of unknown origin, with karyotype 47XX + mar, involving genes from the region 18p11.32p11.21.

Men presented a high frequency of family history of epilepsy among first- and second-degree relatives. The most frequent diagnosis was idiopathic generalised epilepsy (2 male and 4 female patients); other diagnoses included febrile seizures, paroxysmal episodes, and cortical blindness (Table 1).

Semiological description of seizures corresponds to the characteristics reported in the clinical records before the diagnosis of EEM was established. The most frequently reported trigger was stressful situations. In terms of seizure frequency, the most common manifestation was daily seizures, at any time of the day. Only one patient

Table 2 Clinical characteristics of seizures by sex.

	Female ($n = 11$) *n* (%)	Male ($n = 5$) *n* (%)
Triggers		
Stressful events	7 (63.6)	2 (40)
Sleep deprivation	4 (36.4)	1 (20)
Photosensitivity	3 (27.3)	—
Alcohol consumption	1 (9)	—
Seizure frequency		
Daily	7 (63.6)	3 (60)
Weekly	3 (27.3)	1 (20)
Rarely during the year	1 (9)	1 (20)
Time of day		
Sleeping hours	6 (54.5)	3 (60)
Waking hours	11 (100)	5 (100)

	Median (IQR)	Range	Median (IQR)	Range
Age at the first eyelid myoclonus (years)[a]	6 (3)	0.3—11	5 (2)	2—9
Age at first GTC seizure (years)[a]	9 (2.5)	8—13	9 (14)	3—41
Age at diagnosis	10 (7)	5—46	8 (24)	7—46
Time to diagnosis (years)[b]	5 (7)	1—40	6 (11)	2—42

Healthcare system regime	*n* (%)	Median (IQR)	Range	*n* (%)	Median (IQR)	Range
Subsidised	4 (36.4)	7 (13)	3—40	3 (60)	6 (18)	5—42
Contributory	3 (27.2)	2 (2)	1—5	1 (20)	2 (0)	—
Private/prepaid	4 (36.4)	9 (6)	5—14	1 (20)	32 (0)	—
Treatment delay (months)[c]	—	2 (2)	0—6	—	7 (2)	5—9

GTC: generalised tonic-clonic; IQR: interquartile range.
[a] Median (IQR).
[b] Years between the first seizure and the diagnosis of epilepsy with eyelid myoclonia.
[c] Time in months between video-EEG diagnosis and onset of specific pharmacological treatment.

reported daily seizures during morning hours. Medical records reported seizures during waking periods in 100% of patients, and during sleep periods in 50% (Table 2).

A total of 93.75% of patients initially reported eyelid myoclonic seizures, and presented generalised tonic-clonic seizures after several years.

The first eyelid myoclonic seizure was most frequently reported during childhood; it only occurred before one year of life in 2 patients. The number of years between the first seizure and the diagnosis of EEM ranged from 1 year to over 40 (median: 5.5; IQR: 8), in both sexes. Regarding social security regime, patients in the subsidised regime presented the longest diagnostic delay. The time between video-EEG diagnosis and onset of specific treatment varied, although it was shorter than 10 months in all cases (Table 2).

Video-EEG revealed that more than 50% of female patients presented epilepsy aura, which was not the case in males. Furthermore, eyelid myoclonia in female patients was more frequent when closing the eyes (72%) than when opening (18%). More than half of patients presented an increase in seizures upon hyperventilation. During sleeping and waking hours, the most frequent pattern of generalised epileptiform discharges in female patients was polyspike-waves, whereas males most frequently presented polyspikes and spike-waves during waking hours and polyspikes during sleeping hours. In both sexes, epileptiform activity was most frequently observed in posterior regions (occipital, temporo-occipital, and parieto-occipital) (Table 3).

Photoparoxysmal response to intermittent photic stimulation manifested more predominantly in female patients: Waltz type III (parieto-occipital spikes and waves extending to the frontal region), and type IV (generalised spikes and waves) were the most frequent.[12] The frequencies most frequently triggering photoparoxysmal response were between 1 and 30 Hz (Table 3).

Some patients underwent further testing: neuroimaging (CT scan in 2, MRI in 11, both in 1), polysomnography (1), and visual (2) and auditory (1) evoked potentials. Three patients presented alterations in these diagnostic tests.

Neuroimaging findings were normal in the majority of cases. In one patient with history of cerebral palsy, MRI revealed hypoxic-ischaemic encephalopathy, Wallerian degeneration, and right hippocampal sclerosis. In the auditory evoked potential test, another patient displayed mild hearing loss in the right ear and abnormal visual evoked potentials, with latency on the right side and absence on the left side. The polysomnography study revealed absence of REM sleep, apnoea, hypopnoea, and EEG abnormalities with slow spike-and-wave and generalised polyspike patterns. Another patient presented complete absence of cortical response to the flash stimulus and flash pattern in both eyes in the visual evoked potential test.

Table 3 Video-electroencephalography characteristics.

	Female ($n = 11$) *n* (%)	Male ($n = 5$) *n* (%)
Aura	6 (54.5)	0
Sensitivity to eye closure	8 (72.7)	3 (60)
Sensitivity to eye opening	2 (18.2)	3 (60)
Increased with hyperventilation	8 (72.7)	3 (60)
Generalised epileptiform discharges when awake		
Polyspike-wave	8 (72.7)	2 (40)
Polyspike	5 (45.5)	3 (60)
Spike-wave	7 (63.6)	3 (60)
Generalised epileptiform discharges when asleep		
Polyspike-wave	6 (54.5)	3 (60)
Polyspike	2 (18.2)	4 (80)
Spike-wave	5 (45.5)	1 (20)
Focal epileptiform activity		
Anterior regions (frontal and frontocentral)	3 (27.3)	2 (40)
Posterior regions (occipital, temporo-occipital, and parieto-occipital)	8 (72.7)	3 (60)
Photoparoxysmal response during intermittent photic stimulation		
Photoparoxysmal response	10 (91)	2 (40)
Waltz I	0	0
Waltz II	1 (9)	0
Waltz III	3 (27.3)	0
Waltz IV	3 (27.3)	1 (20)
Frequency of the light stimuli provoking the photoparoxysmal response		
1—30 Hz	4 (36.4)	2 (40)
Higher than 31 Hz	1 (9)	0
Both at 1—30 Hz and higher than 31 Hz	4 (36.4)	0

Valproic acid was the most frequently prescribed drug both before and after diagnosis. The second most frequently prescribed drug before diagnosis was confirmed was levetiracetam in female patients and lamotrigine in male patients. After diagnosis is confirmed, valproic acid was the most frequently prescribed drug. Monotherapy was more frequently used in female patients, achieving seizure control in most of the cases. Bitherapy was more frequently prescribed in male patients; this treatment achieved partial control, with patients showing increased drug resistance (Table 4).

Non-pharmacological treatments (blue Z1 lenses, vagus nerve stimulation, responsive neurostimulation of the thalamus) are not recorded in the patient's clinical records. One case was reported of a patient treated with ketogenic diet, which adequately controlled seizures, and another of a female patient treated with cannabidiol, which achieved good control of epileptiform discharges.

Discussion

The results of our study enable us to make 2 main analyses. The first focuses on the similarities with the findings reported in the literature on EEM, and the second is associated with the effect of the Colombian healthcare system on patients with EEM. Both analyses offer a general understanding, both at the national level and specifically in the city of Medellín, of the characteristics of the syndrome and the implications of patients' socio-economic context at the time of diagnosis and treatment.

In our study, we describe the sociodemographic and clinical findings obtained from the medical records and video-EEG studies of 16 patients with EEM and attended at a specialised neurology institution in the city of Medellín. Due to the specific characteristics of seizures, electroclinical findings, and treatment resistance, EEM is underrecognised, and is often misdiagnosed or diagnosed late, which makes it difficult to accurately estimate its prevalence.[13—15] In the literature, it has been reported to account for 2.7%—12.9% of cases of generalised epilepsy, and 0.56%—2.7% of all epilepsy.[1,16,17] Our findings regarding age and sex are similar to those reported by other authors: onset typically occurs during childhood,[4] and with predominance in the female sex.[3,13] However, other studies report that the sex distribution is almost equal.[14,15] This unequal sex distribution may be due to differences in the genetic background of different populations.[15] Regarding age, we should underscore that 2 patients presented EEM in the first year of life. Articles published in the literature describe clinical subtypes of this epilepsy; the cases of these 2 patients would correspond to an early-onset EEM.[4,6]

Half of these patients had family history of epilepsy, affecting first- and second-degree relatives in 37.5% of cases. This is consistent with the findings of several stud-

Table 4 Characteristics of treatment response.

	Female ($n = 11$)		Male ($n = 5$)	
	n (%)		*n* (%)	
Drug (active ingredient)	Before diagnosis	After diagnosis[a]	Before diagnosis	After diagnosis[a]
Valproic acid	8 (72.7)	7 (63.6)	5 (100)	4 (80)
Carbamazepine	2 (18.2)	0	3 (60)	1 (20)
Levetiracetam	3 (27.3)	3 (27.3)	3 (60)	2 (40)
Lamotrigine	2 (18.2)	1 (9)	4 (80)	2 (40)
Phenobarbital	1 (9)	0	1 (20)	0
Phenytoin	1 (9)	0	0	0
Topiramate	1 (9)	0	1 (20)	0
Other antiseizure drugs	2 (18.2)	1 (9)	0	1 (20)

	Female ($n = 11$) *n* (%)	Male ($n = 5$) *n* (%)
Monotherapy	9 (81.8)	1 (20)
2 drugs	1 (9)	4 (80)
No antiseizure drugs	1 (9)	0
Seizures controlled with current drugs		
Yes	4 (36.4)	1 (20)
No	0	1 (20)
Partial	2 (18.2)	2 (40)
Not reported	5 (45.4)	1 (20)
Continuation of treatment reported in the CH		
Yes	3 (27.3)	3 (60)
No, due to lack of supply by the HIP	0	1 (20)
No, by decision of the patient or guardian	1 (9)	1 (20)
Not reported	7 (63.6)	0
Drug resistance[b]	2 (18.2)	3 (60)

CH: clinical history; HIP: health insurance provider.
[a] Drugs taken at the time of the study.
[b] Failure of treatment with 2 antiseizure drugs, in monotherapy or in combination, which are well tolerated and appropriately selected and used, to achieve sustained seizures freedom. Variable reported in the clinical history by a neurologist.

ies, in which family history of epilepsy in EEM is reported in 33%—83% of cases.[15,18,19]

Other types of seizures have been described in EEM, in addition to eyelid myoclonia. In our case series, 43.75% had presented generalised tonic-clonic seizures at least once in their lifetime; these seizures are reported in more than half of patients in other studies, and are usually unavoidable in the long term.[13,20]

Photoparoxysmal response was observed in all young untreated patients; this phenomenon may decrease with age or with antiseizure drugs.[5,21,22] In our study, it was reported in 75% of the patients, particularly in the female sex.

The most frequently reported seizure trigger was stressful situations (10 patients), followed by sleep deprivation (5 patients), drug discontinuation (3 patients), exposure to intermittent lights or use of electronic devices (3 patients), and alcohol consumption, among others. These data are similar to those reported by Pérez-Errazquin et al.[23] Two patients reported triggers other than those found in the literature. One female patient identified an increase in episodes during the menstrual period, whereas other associated it with prolonged fasting. These data demonstrate that triggers are varied and differ between patients. The identification of these triggers during consultations is essential for intervening or preventing an increase in seizures and thus improving the patient's quality of life.

Regarding MRI, 93.7% of patients presented normal findings; this is consistent with previous reports in the literature, in which MRI findings were also normal or non-specific in this population.[3,6]

Academic performance in our series was reported as poor in 80% of patients, with 33.3% repeating a year; patients mainly presented difficulties acquiring knowledge, lack of attention, and difficulties following instructions. They also presented neurocognitive function with a tendency towards the lower limit, without being classified as intellectual disability.[14,24,25] Academic performance was generally poor; however, no study has determined whether this learning difficulty is inherent to the syndrome, as it may be associated with the multiple epileptiform discharges or be the consequence of the use or the inappropriate prescription of antiepileptic drugs.[24]

The mean time of diagnosis of EEM after the first episode amounted to 11.64 years. This time is higher than the 9 years reported by Reyhani and Özkara,[15] or the 10 years or less reported by other researchers.[3,26] In the majority of patients, the first diagnosis was associated with some type of epilepsy, especially idiopathic generalised epilepsy and focal

epilepsies. Other diagnoses included tics, cortical blindness, febrile seizures, and attention-deficit/hyperactivity disorder, among others. EEM is still a poorly understood and unrecognised condition among healthcare professionals, and tends to be mistaken for other types of disorders, which delays diagnosis.[15,27]

Misdiagnosis of these patients leads to contraindicated treatments. Of the 16 patients, 5 were treated with carbamazepine and oxcarbazepine. The use of these drugs is not recommended in this type of epilepsy, as they exacerbate seizures,[6] in addition to the adverse effects they may cause, which may lead to treatment discontinuation.

Analysis of social security system regime and diagnostic delay revealed that patients with a delay of over 5 years were managed under the subsidised system. It is well known that the most basic healthcare needs remain unmet in developing countries, and that the situation worsens when high-quality healthcare is required, including specialised medical staff and equipment,[28] as is the case in EEM. In addition to the challenge of correctly diagnosing the disease, patients face barriers in accessing medical care. Although the Colombian Social Security and Healthcare System has increased its population coverage, there was also an increase in healthcare demand, including human resources and specialised equipment.[29] Furthermore, inequality between regions results in a lack of access to healthcare services and to a shortage of specialised medical staff.[30]

The majority of patients in this study presented increased focal epileptiform activity in posterior regions (occipital, temporo-occipital, and parieto-occipital), similarly to the results reported by Nilo et al.[27] in 60% of the patients in their cohort. These results are explained by the functional neuroimaging findings, which have revealed a state of hyperexcitability in the occipital cortex of patients with EEM.[31] This activity may be limited to this region or may also involve the brainstem, activating thalamocortical and transcortical networks that trigger generalised seizures.[8]

It was not possible to assess drug resistance in all patients based on data from their clinical records, due to the lack of recording of this variable in some registries. However, we observed greater rates of combination therapy and poorer seizure control in male patients. Articles in the literature report that EEM is a lifelong disease that is usually highly resistant to treatment, and that eyelid myoclonia usually persists until adulthood.[32] Smith et al.[3] consider that generalised tonic-clonic seizures and seizures other than absence seizures may be predictors of drug-resistant epilepsy. Zawar and Pestana[5] report better prognosis in men than in women, contrary to the results of our study; however, we should not forget that the number of patients in this series was small and this finding may have been incidental.

Regarding the use of other non-pharmacological therapies, the ketogenic diet was prescribed in one patient. This type of diet therapy represents an alternative in patients with treatment-resistant epilepsies. Ruiz Herrero et al.[33] report 18 years of follow-up data from 160 paediatric patients with epilepsy and treated with ketogenic diet; they report that 12%—15% remained seizure-free, with 41.9% presenting a good response at 3 months and 16.2% at 24 months of follow-up. However, adverse effects are frequent and may affect patients' nutritional status and growth. In previous studies, 15% of patients were seizure-free and more than 30% presented decreases of over 50% in seizure frequency.[34—36] The literature does not include specific studies of EEM. Another patient in our series was treated with cannabidiol. Se Hee et al.[37] reported that cannabidiol is an efficacious antiseizure drug in Dravet syndrome and Lennox-Gastaut syndrome; however, it did not improve the quality of life of the 41 patients assessed. Furthermore, Zawar et al.[38] describe the use of cannabidiol in 2 patients with EEM, reporting exacerbation of eyelid myoclonia, which remitted after discontinuation of the drug. Further studies are needed in this population to establish the effectiveness of this compound.

The limitations of our study include its retrospective design and the use of clinical notes and documentation as the data source. Some variables were not recorded, and the monitoring of drug efficacy was not systematically documented in all cases.

Conclusions

EEM or JS is a convulsive disorder that manifests in childhood and causes lifelong symptoms. We should underscore the importance of suspecting this disease in paediatric patients with eyelid myoclonia, in order to establish a correct diagnosis and avoid the use of not-indicated drugs, thus improving the quality of life of these patients.

Understanding the pathophysiology of the disease and recognising its semiological and electroclinical characteristics are relevant not only for the neurologist or epileptologist, but for the general physician to perform an appropriate initial approach and achieve an adequate remission. %

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Declaration of competing interest

The authors have no conflicts of interest to declare regarding the research, authorship, or publication of this study.

References

1. Caraballo RH, Fontana E, Darra F, Chacon S, Ross N, Fiorini E, et al. A study of 63 cases with eyelid myoclonia with or without absences: type of seizure or an epileptic syndrome? Seizure. 2009;18(6):440—5, http://dx.doi.org/10.1016/j.seizure.2009.04.004.
2. Sobolova G, Drobny M, Tulejova L, Ucnova S, Saniova BD. Jeavons syndrome, Eyelid Myoclonus with Absences (EMA) with epileptic status and hypoactive delirium. Acta Medica Martiniana. 2018;18(3):30—41, http://dx.doi.org/10.2478/acm-2018-0015.

3. Smith KM, Youssef PE, Wirrell EC, Nickels KC, Payne ET, Britton JW, et al. Jeavons syndrome: clinical features and response to treatment. J Pediatr Neurol. 2018;86:46—51, http://dx.doi.org/10.1016/j.pediatrneurol.2018.06.001.
4. Covanis A. Jeavons syndrome—updated review. J Epileptol. 2015;23(2):113—23, http://dx.doi.org/10.21307/joepi-2015-0033.
5. Zawar I, Pestana Knight EM. An overview of the electroencephalographic (EEG) features of epilepsy with eyelid myoclonia (jeavons syndrome). Neurodiagn J. 2020;60(2):113—27, http://dx.doi.org/10.1080/21646821.2020.1750879.
6. Zawar I, Knight EP. Epilepsy with eyelid myoclonia (Jeavons Syndrome). J Pediatr Neurol. 2021;121:75—80, http://dx.doi.org/10.1016/j.pediatrneurol.2020.11.018.
7. Mayo S, Gómez-Manjón I, Fernández-Martínez FJ, Camacho A, Martínez F, Benito-León J. Candidate genes for eyelid myoclonia with absences, review of the literature. Int J Mol Med Sci. 2021;22(11):5609, http://dx.doi.org/10.3390/ijms22115609.
8. Viravan S, Go C, Ochi A, Akiyama T, Carter Snead O III, Otsubo H. Jeavons syndrome existing as occipital cortex initiating generalized epilepsy. Epilepsia. 2011;52(7):1273—9, http://dx.doi.org/10.1111/j.1528-1167.2011.03068.x.
9. Striano S, Capovilla G, Sofia V, Romeo A, Rubboli G, Striano P, et al. Eyelid myoclonia with absences (Jeavons syndrome): a well-defined idiopathic generalized epilepsy syndrome or a spectrum of photosensitive conditions? Epilepsia. 2009;50:15—9, http://dx.doi.org/10.1111/j.1528-1167.2009.02114.x.
10. Pérez-Errazquin F, Chamorro-Muñoz MI, García-Martín G, Romero-Acebal M. Does Jeavons syndrome exist? A report of a series of 10 cases. Rev Neurol. 2010;50(10):584—90, http://dx.doi.org/10.33588/rn.5010.2009481.
11. Kokkinos V, Urban A, Sisterson ND, Li N, Corson D, Richardson RM. Responsive neurostimulation of the thalamus improves seizure control in idiopathic generalized epilepsy: a case report. Neurosurgery. 2020;87(5):E578—83, http://dx.doi.org/10.1093/neuros/nyaa001.
12. Waltz S, Christen H-J, Doose H. The different patterns of the photoparoxysmal response—a genetic study. Electroencephalogr Clin Neurophysiol. 1992;83(2):138—45.
13. Striano S, Striano P, Nocerino C, Boccella P, Bilo L, Meo R, et al. Eyelid myoclonia with absences: an overlooked epileptic syndrome? Neurophysiol Clin. 2002;32(5):287—96, http://dx.doi.org/10.1016/s0987-7053(02)00343-x.
14. Wang XL, Bao JX, Liang S, Tie M, Deng YC, Zhao G, et al. Jeavons syndrome in China. Epilepsy Behav. 2014;32:64—71, http://dx.doi.org/10.1016/j.yebeh.2013.12.016.
15. Reyhani A, Özkara Ç. Pitfalls in the diagnosis of Jeavons syndrome: a study of 32 cases and review of the literature. Epileptic Disord. 2020;22(3):281—90, http://dx.doi.org/10.1684/epd.2020.1162.
16. Giannakodimos S, Panayiotopoulos CP. Eyelid myoclonia with absences in adults: a clinical and video-EEG study. Epilepsia. 1996;37(1):36—44, http://dx.doi.org/10.1111/j.1528-1157.1996.tb00509.x.
17. Covanis A. Eyelid myoclonia and absence. Adv Neurol. 2005;95:185—96. PMID: 15508923.
18. Giuliano L, Fatuzzo D, Mainieri G, Maira G, Elia M, Ferlazzo E, et al. Eyelid myoclonia with absences: electroclinical features and prognostic factors. Epilepsia. 2019;60(6):1104—13, http://dx.doi.org/10.1111/epi.15157.
19. Sadleir LG, Vears D, Regan B, Redshaw N, Bleasel A, Scheffer IE. Family studies of individuals with eyelid myoclonia with absences. Epilepsia. 2012;53(12):2141—8, http://dx.doi.org/10.1111/j.1528-1167.2012.03692.x.
20. Panayiotopoulos CP. Syndromes of idiopathic generalized epilepsies not recognized by the International League Against Epilepsy. Epilepsia. 2005;46:57—66, http://dx.doi.org/10.1111/j.1528-1167.2005.00314.x.
21. Abanoz YG, Abanoz Y, Özkara Ç. Ongoing photosensitivity in an elderly patient with Jeavons sydrome. Neurologist. 2018;23(3):94—7, http://dx.doi.org/10.1097/NRL.0000000000000175.
22. Capovilla G, Striano P, Gambardella A, Beccaria F, Hirsch E, Casellato S, et al. Eyelid fluttering, typical EEG pattern, and impaired intellectual function: a homogeneous epileptic condition among the patients presenting with eyelid myoclonia. Epilepsia. 2009;50(6):1536—41, http://dx.doi.org/10.1111/j.1528-1167.2008.02002.x.
23. Pérez-Errazquin F, Chamorro-Muñoz M, García-Martín G, Romero-Acebal M. ¿ Existe el síndrome de Jeavons? Aportación de una serie de 10 casos. Rev Neurol. 2010;50(10):584—90, http://dx.doi.org/10.33588/rn.5010.2009481.
24. Fournier-Goodnight AS, Gabriel M, Perry MS. Preliminary neurocognitive outcomes in Jeavons syndrome. Epilepsy Behav. 2015;52:260—3, http://dx.doi.org/10.1016/j.yebeh.2015.09.022.
25. Arvio M, Sauna-Aho O, Nyrke T, Bjelogrlic-Laakso N. Intellectual disability in patients with epilepsy with eyelid myoclonias. SAGE Open Med Case Rep. 2018;6, http://dx.doi.org/10.1177/2050313X18777951, 2050313X18777951.
26. Senol PN, Tezer FI, Saygi S. Eyelid myoclonia seizures in adults: an alternate look at the syndrome paradox. Epilepsy Behav. 2015;45:265—70, http://dx.doi.org/10.1016/j.yebeh.2014.12.042.
27. Nilo A, Crespel A, Genton P, Macorig G, Gigli GL, Gelisse P. Epilepsy with eyelid myoclonias (Jeavons syndrome): an electro-clinical study of 40 patients from childhood to adulthood. Seizure. 2021;87:30—8, http://dx.doi.org/10.1016/j.seizure.2021.02.028.
28. Kruk ME, Gage AD, Arsenault C, Jordan K, Leslie HH, Roder-DeWan S, et al. High-quality health systems in the Sustainable Development Goals era: time for a revolution. Lancet Glob Health. 2018;6(11):e1196—252, http://dx.doi.org/10.1016/S2214-109X(18)30386-3.
29. Ochoa Zuluaga LF, URL: https://repository.urosario.edu.co/server/api/core/bitstreams/91923aa8-fd82-4558-9cfc-26721e95e0c1/content, 2018.
30. Bonet-Morón J, Acosta-Ordoñez K, Pérez-Valbuena GJ, Ayala-García J, Guzmán-Finol K, URL: https://repositorio.banrep.gov.co/bitstream/handle/20.500.12134/9402/LBR_2017-12?sequence=1&isAllowed=y, 2017.
31. Vaudano AE, Ruggieri A, Tondelli M, Avanzini P, Benuzzi F, Gessaroli G, et al. The visual system in eyelid myoclonia with absences. Ann Neurol. 2014;76(3):412—27, http://dx.doi.org/10.1002/ana.24236.
32. Abdulla H, Mirza M, Osman S, Dunne K. Jeavons syndrome. Bahrain Med Bull. 2016;38(2):105—7.
33. Herrero JR, Villarroya EC, Peñas JJG, Alcolea BG, Fernández BG, Macfarland LAP, et al. Terapias dietéticas cetogénicas en epilepsia: experiencia en 160 pacientes durante 18 años. An Pediatr. 2022;96(6):511—22.
34. Caraballo R, Vaccarezza M, Cersósimo R, Rios V, Soraru A, Arroyo H, et al. Long-term follow-up of the ketogenic diet for refractory epilepsy: Multicenter Argentinean experience in 216 pediatric patients. Seizure. 2011;20(8):640—5, http://dx.doi.org/10.1016/j.seizure.2011.06.009.
35. Henderson CB, Filloux FM, Alder SC, Lyon JL, Caplin DA. Efficacy of the ketogenic diet as a treatment option for epilepsy: meta-analysis. J Child Neurol. 2006;21(3):193—8, http://dx.doi.org/10.2310/7010.2006.00044.
36. Keene DL. A systematic review of the use of the ketogenic diet in childhood epilepsy. J Pediatr Neurol. 2006;35(1):1—5, http://dx.doi.org/10.1016/j.pediatrneurol.2006.01.005.

37. Kim SH, Choi HS, Koo CM, Joo B-R, Park B-J, Lee HK, et al. Effects of cannabidiol on adaptive behavior and quality of life in pediatric patients with treatment-resistant epilepsy. J Clin Neurol. 2022;18(5):547—52, http://dx.doi.org/10.3988/jcn.2022.18.5.547.

38. Zawar I, Franic L, Kotagal P, Knight EP. Exacerbation of eyelid myoclonia in patients with epilepsy and eyelid myoclonia receiving cannabidiol. Epileptic Disord. 2021;23(6):906—10, http://dx.doi.org/10.1684/epd.2021.1338.