MedCrave
Step into the World of Research

Journal of Neurology & Stroke

Case Report

Open Access

CrossMark

Jeavon's syndrome: a case report

Volume 14 Issue 1 - 2024

Cristina Pozzi, Rafael Bertoldi Torres, Pamela Emilyn dos Santos, Luiz Alfredo Lonzetti
Universidade do Vale do Itajaí, Brazil

Correspondence: Rafael Bertoldi Torres, Universidade do Vale do Itajaí, Brazil, Email torresrafaelbertold@gmail.com

Received: February 15, 2024 | **Published:** February 27, 2024

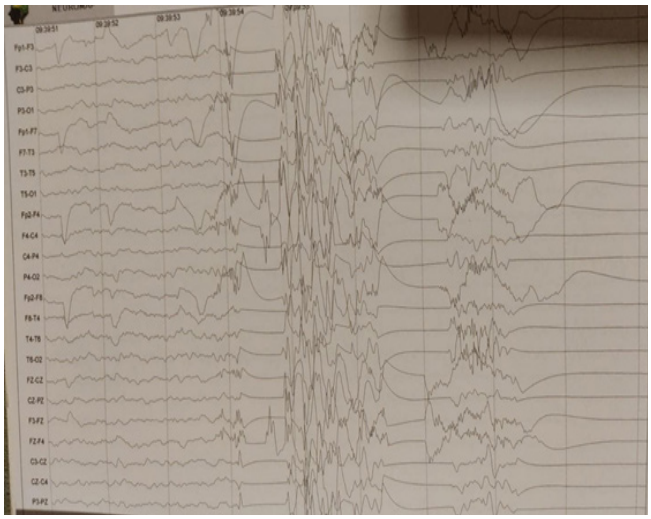
Introduction and literature

The Jeavon's Syndrome is a generalized and idiopathic epileptic disorder of childhood, characterized by the triad of peaceful myoclonus, photosensitivity and characteristic findings on the electroencephalogram, whether or not it may be associated with absences. Eyelid myoclonus consists of rhythmic and continuous movements of the eyelids, associated with eye movements and head extension, which can occur several times a day and can be confused with behavior problems, psychiatric disorders or another type of rare epilepsy such as Sunflower Syndrome. The pathology typically occurs between 2 and 14 years of age, with a predominance of females. It is responsible for 2.5-2.7% of all epilepsy cases and 7.3-12.9% of generalized epilepsies of genetic origin.[1]

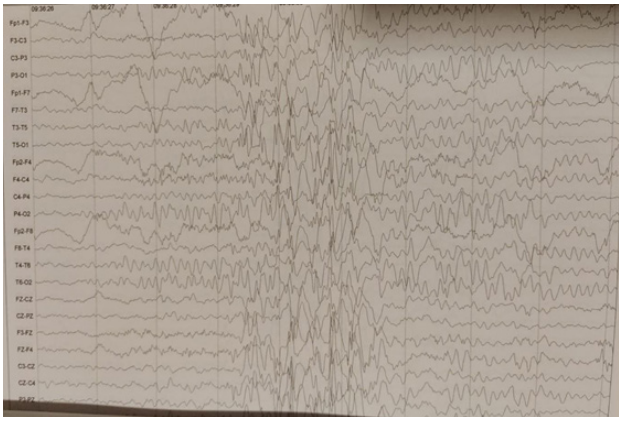
Case report

J.M.C., female, 8 years old, brazilian. Scholar with no perinatal history of pathologies and regular neuropsychomotor development. At the age of two years old, started with frequent blinking episodes alone, with no limbs shaking or behavioral arrest syndrome. During this occasion, the electroencephalogram (EEG) demonstrated frequent wave tip paroxysms and generalized wave polytip at 3Hz. Due to the diagnosis of epilepsy, used valproic acid with cession of the crisis. Thirty months after the first approach, the EEG was repeated and proved to be normal, being the patient instructed to discontinue the antiepileptic. After three months with no medication, the crisis started over with the same characteristics and photosensitivity. With reintroduction of valproic acid there was no response.

A new EEG was performed and showed epileptogenic activity with spike and wave complex and polyspike slow wave with diffuse projection in sleep and wakefulness, not accentuated by photostimulation, with recording episodes of seizures with upward ocular deviation and cervical extension of 2 to 3 seconds as follows:



a)



b)

Figure 1 Recording episodes of seizures

Treatment with valproic acid 1000 mg/day was then started again with partial control, and continued for two and a half years, with improvement in the condition, although no seizure cessation. After this period, a new electroencephalography was performed, which showed rare and diffuse epileptogenic activity with spike and wave complexes, moderate to elevate amplitude.

Throughout this follow-up period, the family sought out several neuropediatricians who adjusted the dose of valproic acid, without improvement, and the use of risperidone was even proposed under the hypothesis of motor tics. At the age of 8, as there was no control over the crises, the parents brought him for diagnostic evaluation and therapeutic guidance, with this history. Awaiting neuroimaging examination.

At school, she has a good performance, but with a mathematics handicap. Regarding comorbidities, hypothyroidism being treated with Levothyroxine and enuresis, urinary urgency, with a history of urinary incontinence until the age of 7, already under follow-up with an urologist. There is a not completely known father's history of epilepsy, treated with valproic acid in adolescence, for 5 years.

The current neurological examination showed several episodes of eyelid myoclonus when looking at light and hyperpnea, without other motor particularities. She presents with an unmotivated smile, inattention and anxious behavior.

Given the clinical picture and the encephalographic record, she was then diagnosed with Jeavons Syndrome. Divalproic acid was proposed with gradual increase, until 1000 mg/day, but she presents with side effects and a tonic clonic seizure occurred. Therefore, it is needed to keep the following regularly with a neuropediatrician, in order to adjust medication and evaluate more carefully the development of the child.

Discussion

Jeavon's syndrome is a rare epileptic genetically linked syndrome that occurs in patients generally less than fourteen years old and requires clinical and electroencephalographic criterias to be diagnosed. Thus, the child reported in our case is a classical form of this disease. The first description of a patient presenting eyelid myoclonia with absences was reported more than 70 years ago. The patient described in the report was marked by involuntary movements of the head and the eyes in times in which he was exposed to photic stimulation. It was reported that the patient was suffering from rhythmic movements of the head when directing it towards sunlight, associated with eyelid movements and, in situations that the sunlight was more intense, the boy progressed with loss of consciousness and fall.[2]

Only in 1977, however, Jeavons described clearly this condition as an eyelid myoclonia and absences, with a typical EEG pattern of bilateral spike and wave activity, with the characteristic of that spike-waves do not occur in the dark, showing the association with a photic stimulation.[3] Therefore, the EMA (eyelid myoclonia with absences) or JS (Jeavons syndrome) is marked by a triad; frequent occurrence of eyelid myoclonia with or without absences associated with an electroencephalography pattern of generalized epileptiform activity and triggered by eye closure, generalized photoparoxysmal EEG response, with visually induced seizures and onset in childhood. Thence, EM (eyelid myoclonia) is the mainly semiologic pattern of the syndrome and it is associated or not with absences.[4]

In such way, the history of eye-closure-related discharges in addition of photosensitivy leaves no room for misdiagnosis.[4] It is considered rare the occurrence of myoclonic jerks in others body parts than the eyelids, such occurrence being considered and exclusion criterion.[4]

In the electroencephalogram tracing, the characteristic pattern is the high-amplitude generalized polyspikes or polyspike-wave complex, usually followed by brief discharges (less than 6 seconds) of rhythmic spike or polyspike-wave complexes at 3 or more per second. This pattern, often, is triggered by active eye-closured, immediately on closing eyes, with darkness ending, totally or partially, this response. Hyperventilation could trigger the EM and absences.[4]

The photosensitivity has a tendency to decrease with age and antiepileptic drugs could be efficient in the suppression of this, even if EM persists. Just as expected for an IGE (idiopathic generalized epilepsies), mental status is, usually, normal and only in rare cases and slightmental retardation is reported.[5]

The JS is considered a myoclonic rather than an absence epilepsy, supporting the idea of the efficacy of anti myoclonic drugs.[6] However, some patients show resistance to antiepileptic drugs.[4] A considerable number of patients have drug resistant epilepsy, requiring multiple antiepileptic agents. Valproic acid has been commonly used and levetiracetam has been shown to be efficacious.[7]

Another relevant consideration is about the genetic influence on the syndrome. Taking into consideration some studies, JS is thought to have a genetic etiology.[8–10] In several reported cases of photosensitivity epilepsy, CHD2 was the only shared gene with deletion in the chromosome 15q26.1 region, with the frequency of unique variations in CHD2 been the highest in JS than others epilepsy syndromes.[11]

The correct diagnosis of Jeavon's Syndrome is sometimes challenging, because not only other epileptic syndromes can have a similar clinical presentation, but some psychiatric disorders such as autism. Nevertheless, it is important to follow logical reasoning concerning the possibility of a differential diagnosis, as an example of our case.

Acknowledgments

None.

Conflicts of interest

The authors declare no conflicts of interest.

References

1. Abdulla H, Mirza M, Osman S, et al. Jeavons Syndrome. *Baharin Medical Bulletin.* 2016;38(2):105–107.
2. Radovici MMA, Misrliou VL, Gluckman M. Reflex Epilepsie reflexe provoquee par excitations optiques des rayons solaires. *Rev Neurol.* 1932;1:1305–1308.
3. Jeavons PM. Nosological problems of myoclonic epilepsies in childhood and adolescence. *Dev Med Child Neurol.* 1977;19(1):3–8.
4. Striano S, Capovilla G, Sofia V, et al. Eyelid myoclonia with absences (Jeavons syndrome): a well-defined idiopathic generalized epilepsy syndrome or a spectrum of photosensitive conditions? *Epilepsia.* 2009;50 Suppl 5:15–19.
5. Altıokka-Uzun G, Özdemir O, Uğur-İşeri S, et al. Investigation of SLC2A1 gene variants in genetic generalized epilepsy patients with eyelid myoclonia. *Epileptic disorders.* 2018;20(5):396–400.
6. Covanis A. Jeavons Syndrome. In: Panayiotopoulos CP, editor. *Atlas of Epilepsies*. Springer, London. pp.1081–1091.
7. Parissis D, Ioannidis P, Karacostas D. Levetiracetam as alternative treatment in Jeavons syndrome. *J Neurol Sci*. 2014;341(1-2):147–149.
8. Sadleir LG, Vears D, Regan B, et al. Family studies of individuals with eyelid myoclonia with absences. *Epilepsia.* 2012;53(12):2141–2148.
9. Lu Y, Yang L, Worrell GA, et al. Dynamic imaging of seizure activity in pediatric epilepsy patients. *Clin Neurophysiol*. 2012;123(11):2122–2129.
10. Adachi M, Inoue T, Tsuneishi S, et al. Eyelid myoclonia with absences in monozygotic twins. *Pediatr Int.* 2005;47(3):343–347.
11. Galizia EC, Myers CT, Leu C, et al. CHD2 variants are a risk factor for photosensitivity in epilepsy. *Brain.* 2015;138(Pt 5):1198–1207.