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Jeavons syndrome - clinical spectrum, diagnostic challenges and innovative treatment strategies

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Abstract

Introduction:
Jeavons syndrome (JS), also known as epilepsy with eyelid myoclonus (EEM), is a rare form of genetically generalized epilepsy (GGE) characterized by light-triggered seizures, eyelid myoclonus, and often concurrent absence seizures and GTCS. The condition is sometimes misdiagnosed as neural tics, leading to significant diagnostic delays. In recent years, there has been growing interest in new treatments for JS, including alternative approaches in cases of treatment resistance.
Objective:
The purpose of this study is to provide an update on Jeavons syndrome, including clinical presentation, genetic factors, diagnostic methods and the latest therapeutic strategies.
Methods:
A non-systematic review of the scientific literature from 2020-2025 was performed using PubMed, Google Scholar, Web of Science and Scopus databases. Publications describing JS, treatment of drug-resistant epilepsy, eyelid myoclonus, dietary therapy and modern brain stimulation methods were included.
Results:
The most commonly used first-line drugs are valproic acid, levetiracetam and lamotrigine. In refractory cases, lacosamide, zonisamide, and VNS and RNS have also shown efficacy. The potential of diet therapy (ketogenic, Atkins, low-GI diets) and lentiviral therapy in patients with photosensitivity was identified. The study emphasized the role of genetic diagnosis in personalizing treatment.
Conclusions:
Jeavons syndrome requires a comprehensive therapeutic approach. Standard pharmacotherapy is sometimes insufficient, so adjunctive therapies are playing an increasingly important role. Early diagnosis, consideration of genetic factors and implementation of individualized treatment strategies can significantly improve seizure control and quality of life for patients.

Keywords: Jeavons syndrome, Jeavons syndrome treatment, eyelid myoclonus, genetic generalized epilepsy, photosensitive epilepsy,absence seizures, drug-resistant epilepsy.

Introduction

Jeavons syndrome (JS) or epilepsy with eyelid myoclonus (EEM) is a rare neurological disorder associated with genetically determined forms of epilepsy (GGE). In addition to epilepsy with eyelid myoclonus and seizures induced by light (PPR), patients may experience absence seizures and general tonic-clonic seizures (GTCS). Myoclonus is characterized by flickering, twitching/fluttering eyelids and usually occurs after the eyelids are slowly closed. Sometimes there is head retraction and the eyes are open and may twitch with an upward turn of the eyeballs.[1] The syndrome was described by Jeavons in 1977, who noted that “brief episodes of absentmindedness can occur spontaneously and are accompanied by 3 Hz needle-wave discharges. Their presence on a routine EEG is a strong signal that light stimulation may be causing the abnormalities."[2] Importantly, the seizures are very difficult to control and require polypharmacotherapy or show significant resistance to treatment. [2].

Purpose

The purpose of this article is to bring together the latest information and clinical, genetic and therapeutic perspectives on JS.

Methods

The review was based on publicly available PubMed and Google Scholar, Web of Science and Scopus databases from 2020-2025 using the following phrases: Jeavons syndrome, Jeavons syndrome treatment, eyelid myoclonus, genetic generalized epilepsy, photosensitive epilepsy,absence seizures, drug-resistant epilepsy. Publications were analyzed using the non-systematic review method to create a brief synthesis of available information.

Epidemiology

Epilepsy with eyelid myoclonic seizures (EEM) most commonly occurs between the ages of 2 and 14, with the highest incidence at ages 6-8.[3] . The syndrome is demographically twice as common in females and accounts for 7.3-12.9% of generalized epilepsy cases and 2.5-2.7% of all epilepsy types.[2]In most children, intellectual development and cognitive abilities remain normal. The disorder is believed to have a genetic basis, as about one-third of patients have a family history of epilepsy.

Recent studies suggest the involvement of pathogenic mutations in genes such as CHD2, KCNB1, KIAA2022, NAA10, and a recently reported case of EEM in a patient with the ATP1A3 variant. [3]

Diagnosis

One of the main problems is the significant delay in making a proper diagnosis. Available data show that the average time from the appearance of the first symptoms to the diagnosis of the disease is between 9.6 and 10.3 years, and the average age of diagnosis is about 17 years. Such a long delay is often due to misinterpretation of eyelid twitches, which are sometimes mistaken for nervous tics or mental problems, instead of being treated as a potential symptom of epilepsy. [4]

Eyelid myoclonus is mandatory for diagnosis, regardless of whether it occurs in the presence of absence seizures. EEG is an obligatory test for recording myoclonus.[5] EEG studies during interictal periods often show generalized or partially generalized needle-wave discharges, with predominance in the frontal or occipital regions. As is common in genetically generalized epilepsies, these discharges can be induced by NREM sleep and waking, as well as by hyperventilation. During EEG seizures, general polyglot waves of 3-6 Hz are most often recorded when the eyes are closed in a bright room. Focal abnormalities are also common in JS, occurring in 16-56% of cases. PPR in JS differs from other photogenic epilepsies, such as juvenile myoclonic epilepsy (JME), because EEG discharges are often concentrated in the frontal lobe. Atypical PPRs, associated with spike waves, can also occur in JS, with synchronization of polyspike waves to light stimuli, indicating hyperactivity of the visual-occipital cortex.[6]

According to the researchers, there is a strong consesnus that genetic testing in the form of a whole panel of epilepsy genes or whole exome sequencing should be wconducted in patients who have familial epilepsy, drug-resistant epilepsy or intellectual disability.[5]. The tests allow for future individualization of specific etiologies and selection of appropriate therapeutic regimens. The most significant factors influencing the presence of epilepsy are its early onset and the presence of intellectual disability. [7]

Treatment

Successful treatment of EEM requires a thorough knowledge of its clinical picture and how the patient responds to the therapy used.[7] Namely, if JS is not properly treated there is a risk of developing drug-resistant epilepsy (DRE), the course of which is more severe and persists throughout life. [8]

During treatment, difficulties are often encountered due to the limited effectiveness of pharmacotherapy. Nevertheless, the mainstay of therapy remains the use of antiepileptic drugs. In clinical practice, substances such as valproate, lamotrigine, ethosuximide, levetiracetam and, in some cases, drugs from the benzodiazepine group are most commonly used. [9] Other methods are also prominent in treatment: vagus nerve stimulation (VNS), ketogenic diet, lentiviral therapy.

Pharmacological treatment

According to expert opinion according to the work of Kelesey M Smith et al, valproic acid, levetiracetam and lamotrigine showed the highest efficacy in the treatment of Jeavons syndrome. Valproic acid was indicated as the preferred first-line therapeutic agent, while levetiracetam and lamotrigine were considered suitable alternatives especially in the context of treating women of childbearing age. For ethosuximide and clobazam, there was a moderate level of agreement on their clinical suitability. For the other treatments, there was no consensus on their efficacy. At the same time, the need to avoid most drugs that interact with sodium channels - with the exception of lamotrigine - was emphasized because of the possibility of exacerbating disease symptoms and worsening seizure control. [10] In addition, zonisamide preparations are confirmed in retrospective scientific studies. [11]

Zawar et al. described a case that suggests lacosamide may be an effective therapeutic option for treating drug-resistant Jeavons syndrome, especially when standard approaches - such as ethosuximide, valproic acid, lamotrigine, topiramate or a ketogenic diet - prove ineffective or poorly tolerated. The patient had complete seizure control and significant improvement in EEG recording during lacosamide monotherapy, indicating the potential value of this drug in the treatment of selected cases of this rare form of generalized epilepsy. [12]

Future research on Jeavons syndrome should pay particular attention to cannabidiol (CBD) as a potential adjunctive therapy, especially in patients with drug-resistant disease. CBD, unlike THC, has no psychoactive effects, and its anti-epileptic mechanism is still not fully understood. A number of studies suggest that CBD affects various neurotransmitter systems, including modulating GPR55 and TRPV1 receptors, reducing neuronal excitability, as well as having anti-inflammatory and neuroprotective effects [13][14]. Additionally, cannabidiol has the ability to regulate endocannabinoid levels and calcium channel activity, which may be important in the context of the neuronal hyperexcitability characteristic of Jeavons syndrome [15][16]. Nevertheless, further studies are required to confirm the efficacy of CBD in this particular form of epilepsy and to identify biomarkers of treatment response [17][18].

Vagus nerve stimulation (VNS)

Caitlin Wessel et al. in their paper on the efficacy of vagus nerve stimulation in the treatment of drug-resistant epilepsy syndromes studied, among other things, the effect of this procedure on patients with Jeavons syndrome. Although the mechanism of action of VNS therapy has not yet been fully elucidated, it is speculated that stimulation of the afferent fibers of the vagus nerve leads to activation of the

parasympathetic nervous system, which, as a consequence, may reduce excessive synchronization of cortical activity . A retrospective study showed that among patients with JS, seizure frequency was reduced, by as much as 65.4% after the VNS procedure. In the group of patients analyzed, there were no statistically significant differences in either the age at which the vagus nerve stimulator was implanted , or the time elapsed from the onset of the first epileptic seizure to the time of implantation. Also, a comparison between patients who responded to VNS therapy and those who showed no improvement showed no significant difference in the time interval between the first unconscious seizure and VNS insertion. The study concluded that the procedure, which is the surgical implantation of VNS, is a well-tolerated procedure. During it, there were single postoperative complications such as : apnea, voice change, wound infections. VNS therapy in the future may be a new therapeutic method in the treatment of drug-resistant epilepsies. [8]

A study on reactive neurostimulation of the thalamus in a patient with EEM additionally demonstrated the effect of reactive neurostimulation (RNS), which targeted the centromedial/ventral/lateral area. The procedure resulted in a marked decrease in the frequency of daily seizures from 60 to ≤ 10. [19].

Dietary therapy

Analyzing a study on the treatment of epilepsy with eyelid myoclonus, patients participating in the study showed relatively little experience with dietary interventions in EEM. In an attempt to obtain a consensus on the effectiveness of dietary therapy in different age groups, the results were inconclusive. The limited number of responses indicated a moderate consensus that less than a quarter of patients who respond positively to dietary therapy achieve sustained improvement without the need for concomitant antiepileptic drugs (ASMs). The panelists' statements indicated that diets considered potentially effective include the modified Atkins diet, the ketogenic diet and the low glycemic index diet. [10]

A randomized clinical trial evaluating the efficacy of a modified Atkins diet as adjunctive therapy in adults with drug-resistant focal epilepsy showed a significant reduction in seizure frequency in the diet group compared to the control group. Nevertheless, only moderate clinical improvement, defined as a seizure reduction of more than 25%, was achieved among participants who completed the dietary intervention. [20]

Analyzing the Jo Sourbron et al. study, we can conclude that the ketogenic diet shows significant efficacy in reducing seizure frequency in children and adolescents with drug-resistant epilepsy, with relatively good tolerance and no serious side effects. Although the results are promising, further research is needed on biomarkers of response to therapy and its impact on patients' quality of life.[21]

Lens therapy

There are few studies demonstrating the effectiveness of Z1 blue lens therapy in patients who are hypersensitive to light. [22] The blue lens reduces and filters red light (about 700nm), which has been found to be the most triggering of the primary colors. [23]. In a retrospective study by Zawari et al. in 2022, eight patients out of eleven treated with lens therapy responded to treatment [22]. The use of this type of lens has sometimes been limited due to difficulties in tolerating them and low patient acceptance, which is associated with a significant reduction in luminance levels. However, newer blue-tinted lens models, which allow more light to pass through, show potential in reducing photoparoxysmal reactivity (PPR) and may be better tolerated. [23]

Conclusion

Jeavons syndrome (JS) is a rare but clinically significant variant of genetic generalized epilepsy, the diagnosis of which is sometimes significantly delayed due to nonspecific symptoms and diagnostic difficulties. Effective treatment requires an individualized therapeutic approach based on accurate diagnosis and understanding of the specifics of the disease. Although standard antiepileptic drugs such as valproic acid, levetiracetam and lamotrigine remain the mainstay of therapy, their effectiveness is sometimes limited, and some patients require alternative therapies. Promising results have been reported with the use of lacosamide, vagus nerve stimulation (VNS), reactive neurostimulation (RNS), dietary therapies and photosensitive lenses - particularly in patients with drug-resistant forms of the disease. Despite the availability of a variety of treatments, future research should focus on developing more accurate diagnostic biomarkers to detect Jeavons syndrome more quickly. Genetic testing should also be deepened to individualize therapy, and the efficacy of new drugs such as lacosamide and cannabidiol should be evaluated. It is also important to further explore alternative therapies, including the ketogenic diet, lentiviral therapy and brain stimulation, and their impact on patients' quality of life.

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