Jeavons Syndrome

Hafsa Abdulla, MD, BSc* Mahwish Mirza, MBBCh, BAO* Shatha Osman, MRCPCH, MD**
Kevin Dunne, MBBCh, FRCPCS, DCH***

Jeavons syndrome is a rare generalized childhood epileptic condition characterized by eyelid myoclonia with or without absences. Other features of the syndrome include photosensitivity and electroencephalography (EEG) findings.

An eight-year-old male was referred to the pediatric clinic from the ophthalmology clinic with a chief complaint of repetitive abnormal eye movements. The patient did not exhibit any photosensitivity but his EEG was diagnostic for primary generalized epilepsy. Treatment with levetiracetam revealed significant improvement.

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Jeavons syndrome (JS) is a seizure disorder characterized by eyelid myoclonia (with or without absences), photosensitivity and characteristic EEG findings. Eyelid myoclonia consists of jerking movements of the eyelids associated with upward movements of the eyeball and extension of the neck; these seizures are brief (lasting a few seconds) and occur several times during the day.

Video EEGs show "frequent and brief high-amplitude (3 to 6 Hz) generalized spike and wave discharges of polyspikes, which occur after eye closure" and are frequently associated with eyelid myoclonia[1]. Typically, generalized tonic-clonic seizures do not occur, and all young untreated patients are photosensitive. Jeavons described it in 1977, JS has an onset in childhood (ranging from the age of 2 to 14 years with a peak at the age of 8), lasts a lifetime and is two-times more common in females[2,3]. Jeavons syndrome is not usually associated with abnormal development or intellectual disability[4,5]. Some identical twin studies support a genetic component to the syndrome[2]. Antimyoclonic drugs, such as levetiracetam have shown good clinical result.

The aim of this report is to raise awareness about Jeavons syndrome. Early recognition leads to timely treatment, which enhances the quality of life for the affected children.

THE CASE

An eight-year-old Egyptian male presented in October 2014 to the Ophthalmology Outpatient Clinic with a one-month history of recurrent attacks of abnormal repetitive eye movement.

Ophthalmological conditions were ruled out, and the patient was referred to the General Pediatric Clinic. The attacks consisted of abnormal movements mainly repetitive upward deviation of both eyes with movement of the head in the same direction. Such attacks occurred at least 5 to 6 times a day and would happen approximately every day lasting for only a few seconds each time. These attacks were not associated with any disturbance in consciousness level. There were no attacks of tonic-clonic seizures or absence seizures; no history of aura or postictal phenomena was documented. The patient was completely conscious during such attacks and was performing his normal daily activities. His past medical history was significant for three attacks of simple febrile seizures, a measles infection and a tonsillectomy. His neonatal and developmental history was not significant. Developmentally and socially, he was within normal including developmental milestones and his school performance. The patient had no family history of epilepsy, seizure disorders or any other relevant medical disorders.

His systemic examination was within normal, and a neurological assessment did not reveal any abnormality. MRI was reported normal; however the EEG report did reveal significant findings. The following are the main conclusions of the EEG report:

"There was an alpha rhythm of 10 Hz over the occipital regions which was symmetrical and blocked to eye opening. The record was symmetrical. Occasional generalized bursts of spike waves occurred from time to time without any reported clinical manifestation. The child did not appear to be photosensitive, but high voltage generalized discharges were again evident post-hyperventilation. It is abnormal EEG with generalized epileptic discharges without any clinical correlate. The findings were consistent with a Primary Generalized Epilepsy."

Based on the patient's clinical history and the video provided by the father, the significant EEG report and the normal MRI, a provisional diagnosis of Jeavons syndrome was made and the patient was started on levetiracetam. He was started on a dose of 30 mg/kg/day which was titrated up every few weeks while monitoring his condition. Currently, his family is reporting more than 75% reduction in the frequency of seizures.

DISCUSSION

Jeavons syndrome is known as eyelid myoclonia with absences; it is a distinct idiopathic generalized epilepsy characterized by a well-defined triad of clinical manifestations and EEG findings that consist of eyelid myoclonia (with or without

* Intern
** Senior Registrar
*** Consultant Pediatrics
Department of Pediatrics
King Hamad University Hospital
Kingdom of Bahrain
E-mail: hafsa.bucheery@gmail.com; mahwish.m.mirza@gmail.com

absence seizures). The seizures are precipitated by eyelid closure and photosensitivity and EEG bursts[1]. The prevalence is around 3% amongst people with epilepsy and 13% amongst patients suffering from idiopathic generalized epilepsy with absence seizures[1]. Its etiology is unknown, but a hypothesis supported by several familial and twin studies alludes to a genetic predisposition[6].

The typical seizures observed in Jeavons syndrome consist of eyelid myoclonia which could be described as repetitive upward jerky movements of the eyeball associated with head movements. In some instances, the patient may suffer from impairment of consciousness. Such attacks are repetitive, occurring several times a day and lasting for no more than a few seconds each time.

Other seizures associated with Jeavons syndrome include absence seizures, generalized tonic-clonic seizures, myoclonic jerks and eyelid myoclonic status epilepticus.

The first clinical description of a patient suffering from Jeavons syndrome was 70 years ago; a young male presented with a history of being bullied by his schoolmates as a child, they used to force him to gaze directly at the sun, which in turn provoked his undiagnosed seizures, almost instantly causing him to go into rhythmic eye and head movements and then falling to the ground and losing consciousness[2].

Several factors have been identified as precipitants for the episodes in Jeavons syndrome: light, flickering or uninterrupted. Other factors include eyelid closure, sleep deprivation, alcohol consumption, hyperventilation and inappropriate antiepileptic use, particularly carbamazepine, gabapentin, oxcarbazepine, phenytoin, pregabalin, tiagabine and vigabatrin[1]. All the drugs above are contraindicated in any of the myoclonic epilepsy syndromes as they are known to aggravate such conditions.

One important differential of Jeavons syndrome is facial tics. This disorder is described as partially controllable repetitive spasmodic movements of the facial muscles producing distinct movements, such as blinking of eyes or scrunching of the nose. However, this condition could be easily differentiated from Jeavons syndrome as it is usually involuntary but could be temporarily suppressed; the patient also described as having an irresistible desire to produce such movements and could only suppress it for a short period. It may be part of a spectrum of diseases and behavioral abnormalities, such as transient tics disorder and/or Tourettes syndrome.

Physical and neurological examination of the patients usually yields no clinically significant findings. MRI brain is not of great clinical relevance. A study found that 3 out of 6 MRI had abnormal findings[7]. One patient had nonspecific increased signal intensity at bilateral frontal white matter. Another had a Chiari 1 malformation, and the last had a dysembryoplastic neuroepithelial tumor in the right frontal region.

Video telemetry is the gold standard for diagnosis, demonstrating the classical EEG findings occurring while the patient is suffering from a seizure. A study has postulated that patients affected by Jeavons syndrome fall into two categories: "a predominantly male group with frontal predominant epileptiform discharges, eyelid myoclonia and eyes rolling up and a predominantly female group with occipital predominant epileptiform discharges with eyelid myoclonia alone"[8].

Jeavons syndrome is treated with antiepileptic medications including levetiracetam, valproate, clonazepam and ethosuximide. Levetiracetam seems to be the most popular since it has an effect on both the myoclonia and the photosensitivity. The exact antiepileptic mechanism of action of levetiracetam is unknown but several theories have been proposed: (1) it inhibits voltage-dependent calcium channels, (2) it binds to synaptic proteins that control neurotransmitter release and (3) it facilitates GABA-like inhibitory transmission. In a prospective clinical trial, it was found that levetiracetam was most effective if given at a dose of 50-60 mg/kg/day[3].

Jeavons syndrome is a lifelong disorder that is usually highly resistant to treatment; even though most patients outgrow the absence seizures and photosensitivity, the eyelid myoclonia usually persists into adulthood.

CONCLUSION

Jeavons syndrome is a seizure disorder seen in childhood and has lifelong symptoms. The most important tool for diagnosis, after clinical evaluation of the patient, is an EEG. The main mode of treatment is anti-epileptic medications; the most effective is levetiracetam. Although symptoms do not usually disappear totally; but, most patients report an improved quality of life with treatment.

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