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EARLY ONSET JEAVONS SYNDROME – A CASE REPORT

Paediatrics

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ABSTRACT

Eyelid myoclonia with absences, also known as Jeavon's syndrome [1](JS) is one of the underreported childhood onset epileptic syndromes and is characterized by eyelid myoclonia, eye closure-induced seizures or electroencephalography (EEG) paroxysms, and photosensitivity[ps].Though a genetic contribution is likely present, the genes responsible are not identified.[1] **Clinical forms of JS**: we have identified four forms of JS; early onset (< 4 years), mild form, classical form and an ELMA-JME form. If diagnosed can be treated effectively with sodium valproate and benzodiazepines.

KEYWORDS

Eyelidmyoclonus, videoelectroencephalography, photossensitive, sodiumvalproate.

I. INTRODUCTION

Eyelid myoclonus with absence (EMA) or Jeavons syndrome is a rare idiopathic generalized epilepsy (IGE) that occurs in childhood. The symptomatic features of eyelid myoclonus were first reported by Radovici et al [5] more than 80 years ago. The study of epileptic discharges and seizures caused by closing the eyes was reported in 1955 . This epileptic syndrome was first fully described by Jeavons in 1977. It is characterized by eyelid myoclonus, with or without absence of seizures, eyelid closure induces epileptic discharge and/or seizures, and photosensitivity. Subsequently, Eyelid myoclonus is a hallmark of Jeavons syndrome[1]. Usually, it is associated with repeated myoclonic jerks of the eyelids, eyeballs roll upward, and the head may move slightly backward. When accompanied by impairment of consciousness, it is named eyelid myoclonus with absence. Eyelid myoclonic status may be spontaneous or light stimulated. It consists of recurrent and discontinuous seizures of eyelid myoclonus, accompanied by mild absence [3]. Many patients with IGE have visual precursors associated with photosensitive and focal epilepsy, EMA is common in IGE patients with photosensitivity. The occipital cortex can be activated by eye closure and IPS, and this excitability may spread to the brain stem, resulting in eyelid myoclonus. As the intensity of the photic stimulus increases, the excitability can be projected into the central frontal cortex via transcortical and thalamocortical pathways, and then generalized seizures and absence seizures may occur [4]. However, focal epileptic discharge is also present in patients with IGE. Gungor-Tuncer et al had demonstrated that epileptic discharges of focal origin can still occur in some IGE patients. The interictal focal discharge independent of localization in some patients suggests that there may be an overlap between focal epilepsy and generalized epilepsy [5]. Eyelid myoclonus has some overlap with other seizures, especially photosensitive epilepsy, and reflex epilepsy; therefore, the differential diagnosis of EMA is very important. Patients with eyelid myoclonic status are even less common in clinical practice. Giuliano et al [6] found that 17.6% of patients with eyelid myoclonus had epileptic status. Striano[1] also reported that one in five patients might experience eyelid myoclonus status. Valproate is widely recognized as the most effective treatment for photosensitive epilepsies [5]. Therefore, we present a very rare case of eyelid myoclonus status followed by an occipital origin of tonic–clonic seizure with the clinical manifestations, electroencephalogram (EEG) characteristics, differential diagnosis, and therapeutic effect in detail.

II. Case Report

A three-year-old male child admitted in the Guntur government hospital (Department of Pediatrics, with the main complaint of seizure like activity, clonic movements of all four limbs, up rolling of eyeballs lasting for 5 min, post ictal confusion for 5 minutes, involuntary micuration present. The similar episodes occur 6 to 7 times over a period of 3 months. on examination child had continuous blinking, upward rolling of the eyeball, backward movement of the head, and accompanied by decreased consciousness. Physical examination showed poor condition, continuous eyelid blinking with unclear consciousness; neck strength (–), Babinski sign (–), limb muscle tone was normal, and muscle strength grade V and all reflexes were normal. Physical examination of other nervous systems was normal. After hospitalization, video electroencephalography (VEEG) was performed immediately. It showed background activity consists of 7-8 HZ ,40 to 50 microvolts of alpha activity arising from occipital areas and getting attenuated by eye opening. (Figure 1), [figure -2]. Photo paroxysmal response noted. After a seizure, his eyelid myoclonus symptoms disappeared, and simultaneously, EEG discharge decreased significantly. However, the child was heavily drowsy, and slow-wave increased. Each convulsion is manifested as loss of consciousness, backward movement of the head, teeth closed, limbs stiff and shaking, second incontinence, lasting for about 1 min, and can be relieved by itself. . He was tired and went to sleep after convulsion. During the one year, prior to admission, the child developed frequent eyelid blinking and decreased consciousness following frequent eye closure. Combined with the patient's clinical manifestations, EEG, closed eye sensitivity, photosensitivity, and history, we diagnosed the patient as with eyelid myoclonic status. He had no positive family history. After admission, the biochemical examination was completed. There was no significant abnormality on his biochemical examination, and no abnormality on MRI brain. The IQ for the age of child is normal. After being diagnosed, the child was given sodium valproate 20 mg per kg per day. and the patient responded well to valproic acid, without obvious side effects. On the 4 th day of admission, the child had no convulsions and was discharged. In addition, we recommended to the patient to check the liver and kidney function, blood routine, and blood drug concentration detection regularly, drug increase or decrease according to clinical manifestations, and EEG examination. Moreover, mental tests should be performed to determine whether the cognitive aspects of the patient are affected. His parents were advised to watch for if other types of epilepsies occur and avoid light stimulation. The child was followed for two months, and no recurrence is seen.

Fig: 1

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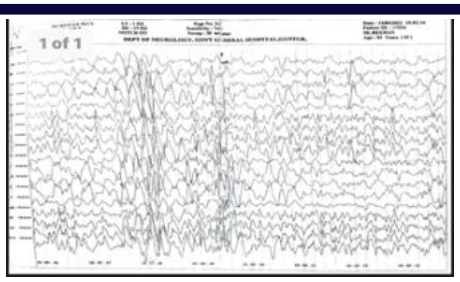

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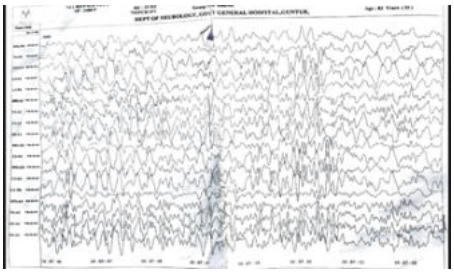

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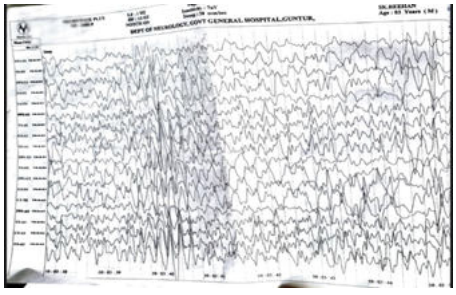

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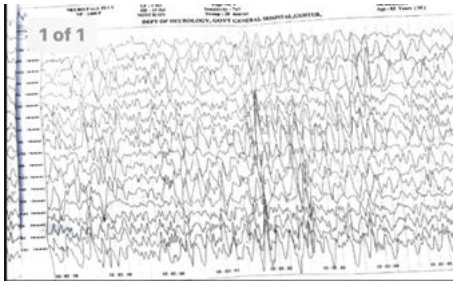

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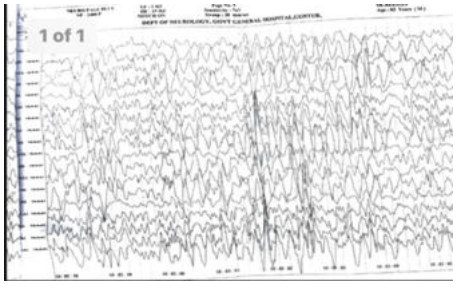

Fig: 6

I. DISCUSSION

Eyelid myoclonia, by definition, is always present and is expressed by eyelid trembling, flickering, fluttering or myoclonia with a concomitant upward deviation of the eyes and head, associated with a brief conspicuous or inconspicuous absence seizures and simultaneous with generalized paroxysmal EEG activity. The eyes never deviate to the side as in some cases with complex partial seizures[7]

ELMA of early onset: under the age of 4 years old A. Typical form. The characteristic and diagnostic electroclinical phenomena easily differentiate Jeavons.

1. syndrome from any other epilepsy appearing early in life. The repeated eye closure phenomena in bright light are unavoidable and characteristic. The child often puts his arm in front of his eyes or rubs the eyelids to show the uncomfortable and very disturbing feeling. In contrast to the other forms of ELMA, blinking by confrontation and passive eye closure may occasionally induce clinical and EEG paroxysmal discharges reflecting a stronger genetic expression. The response to treatment and prognosis of early onset Jeavons syndrome is worse compared to the mild and classical forms, but comparable to absence epilepsies of early onset, with almost 75% of the cases having moderate to severe educational problems. The MRI is normal. In some of these children the electroclinical phenomena, including the response to IPS, gradually disappear before puberty and the child starts showing signs of educational improvement.
2. Atypical form. The presentation is with frequent GTCS in the first year of life. Giving sodium channel blocker drugs will provoke absence and myoclonic seizures. Eyelid jerking and other myoclonic jerks usually become apparent during infancy [8].
3. Mild form ELMA In these patients' eyelid fluttering, after eye closure, is observed for months or years before seeking medical advice for an occasional GTCS or be discovered by an expert in the field. The EEG shows abnormalities on eye closure and some positive response to IPS.
4. Classical ELMA In the classical ELMA there is marked jerking of the eyelids, often with upward deviation of the eyes and retropulsive movements of the head, immediately after eye closure, associated with general wave discharges in the EEG. In all cases, eyelid myoclonia is associated with absences and the positive response to IPS is usually marked. In some cases, during IPS, the head, instead of jerking, is drawn towards the light, as if by a magnet. The command "close your eyes" fails due to eyelid jerking and synchronous eye opening.

Self-induction in Jeavons syndrome:

Self-induced seizures in Jeavons syndrome are rare. Eyelid myoclonia is a seizure and not an attempt for self-induced seizures. These patients do not need to produce conditions of intermittent photic stimulation for self-induction. Simply closing the eyes would be more potent.

IV. Diagnosis

All tests apart from the EEG are normal. Video-EEG is the single most important procedure for the diagnosis of eyelid myoclonia with or without absences. It shows frequent high- amplitude 3–6 Hz generalized discharges of mainly polyspikes and waves. These are brief (1–6 s, commonly 2 or 3 s) and they are typically related to eye closure, i.e., they occur immediately (within 0.5–2 s) after closing the eyes in an illuminated recording room. Eyelid myoclonia of varying severity often occurs during these EEG discharges. Photoparoxysmal discharges induced by photic stimulation occur in all untreated young patients but may be absent in older patients or those on medication. Sleep EEG patterns are normal and generalized discharges are more likely to increase during sleep but may also decrease. The EEG and clinical manifestations deteriorate consistently after awakening. A normal EEG is rare, even in well-controlled patients.

A. Differential Diagnosis

The diagnosis of Jeavons syndrome is simple because the characteristic eyelid myoclonia, if seen once, will never be forgotten, or confused with other conditions. Furthermore, the EEG with the characteristic eye-closure-related discharges and photosensitivity leaves no room for diagnostic error. Nevertheless, eyelid myoclonia is often misdiagnosed as facial tics, sometimes for many years. The symptom/seizure of eyelid myoclonia alone is not sufficient to characterize Jeavons syndrome, as it may also occur in symptomatic and cryptogenic epilepsies, which are betrayed by developmental delay, learning difficulties, neurological deficits, and abnormal MRI and background EEG.[8]

B. Management

Based on anecdotal evidence, the drugs of choice are those used for other idiopathic generalized epilepsies.[5][6] Valproate alone, or most probably in combination with clonazepam, levetiracetam, lamotrigine or ethosuximide, appears to be the most effective regimen. The choice of the second drug depends on the main seizure type. Clonazepam is highly efficacious in eyelid myoclonia and myoclonic jerks. Of the newer antiepileptic drugs, levetiracetam may be the most effective, because of its anti-myoclonic and anti-photosensitive properties. Lamotrigine is very effective in absence seizures but may exaggerate

myoclonic jerks. Contra-indicated drugs are Carbamazepine, gabapentin, oxcarbazepine, phenytoin, pregabalin, tiagabine and vigabatrin. Lifestyle and avoidance of seizure precipitants are important. Non-pharmacological treatments used for photosensitive patients (such as wearing special glasses or the newly commercially available blue Z1 lenses) should be employed in Jeavons syndrome when photosensitivity persists.

C. Prognosis

Jeavons syndrome is a lifelong disorder, even if seizures are well controlled with antiepileptic drugs. Men have a better prognosis than women. There is a tendency for photosensitivity to disappear in middle age, but eyelid myoclonia persists. It is highly resistant to treatment and occurs many times a day, often without apparent absences and even without demonstrable photosensitivity.

V. CONCLUSION

Jeavons syndrome is an idiopathic (genetic) epilepsy syndrome that shows electroclinical heterogeneity relevant to the genetic heterogeneity. It is characterized by eyelid myoclonia induced immediately after eye closure, associated with generalized EEG paroxysms, brief conspicuous or inconspicuous absence seizures in almost all cases, myoclonic and GTCS in some and photosensitivity. Recently the ILAE Task Force on Classification of seizures has recognized Jeavons syndrome as a separate seizure entity (Fisher et al., 2015).

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