Case Report

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The Blink Beyond Control: A Case of Jeavons Syndrome

Paediatrics Section

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ABSTRACT

Photosensitive epilepsy syndromes are often underdiagnosed or misdiagnosed, despite their increased prevalence in children. These syndromes may present as frequent eye blinking, with or without altered awareness, which is commonly dismissed as normal behavior. Such blinking can be the first clinical manifestation of a neurological disorder. The differential diagnosis includes epilepsy syndromes, movement disorders, and behavioral issues. This present report describes a case of a five-year-old male child with frequent eye blinking and poor academic performance. An Electroencephalogram (EEG) showed high voltage generalised epileptiform discharges consisting of sharp waves intermixed with theta activity on eye closure, suggestive of photosensitive epilepsy. The child was diagnosed with Jeavons Syndrome (JS) following a detailed clinical and neurological workup. Based on the neurologist's recommendation, oral valproate therapy was administered, and the child has remained seizure-free since the initiation of treatment, with no recurrences reported during follow-up. Early recognition of subtle seizure presentations, such as eyelid myoclonus, is crucial for timely diagnosis and management. Increased awareness among clinicians can prevent misdiagnosis and improve long-term outcomes in children with JS.

Keywords: Children, Eyelid myoclonus, Photosensitivity, Reflex epilepsy, Seizure disorder

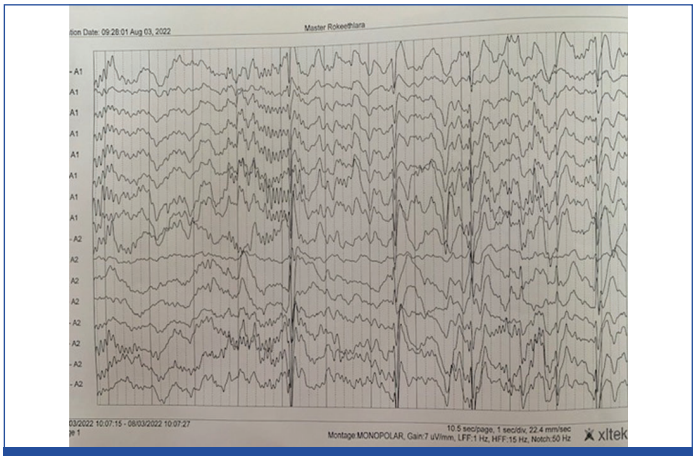
CASE REPORT

A five-year-old male child, the second-born of a non-consanguineous marriage, presented to the Department of Paediatrics at Sree Balaji Medical College and Hospital with episodes of frequent eye blinking lasting 10-15 seconds, occasionally associated with brief unresponsiveness (<1 minute), for the past 6 months. These episodes occurred 2-3 times daily and were more frequent during mobile phone usage. There was no history of limb jerking, eye-rolling, frothing, or incontinence. There was no significant antenatal, perinatal, or family history. Developmental milestones were appropriate, but the mother reported a decline in scholastic performance over the last year. The child's screen time was approximately 3-4 hours per day.

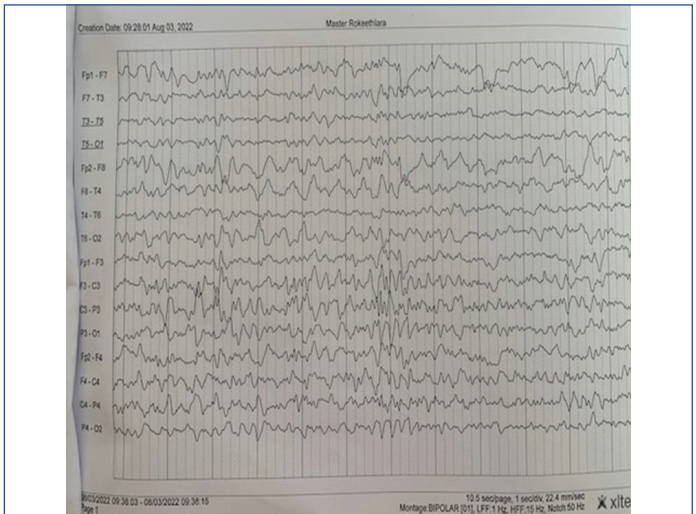
Clinical and systemic examinations were unremarkable. Routine investigations, including complete blood count, liver and renal function tests, and electrolytes, were within normal limits. Magnetic Resonance Imaging (MRI) of the brain revealed no abnormalities. While under observation, the child had an episode of rapid eyelid blinking lasting <10 seconds, followed by brief unresponsiveness for 5-6 seconds.

The EEG showed high voltage generalised epileptiform discharges consisting of sharp waves intermixed with theta activity on eye closure, suggestive of photosensitive epilepsy. The EEG recording showed generalised high-amplitude epileptiform discharges with polyspike and slow-wave complexes (3-6 Hz) interspersed with theta activity. This activity was prominent during eye closure, consistent with features of photosensitive epilepsy characteristic of JS. (Montage: Monopolar, Gain 7 µV/mm, Low-Frequency Filter (LFF): 1 Hz, High-Frequency Filter (HFF): 15 Hz) [Table/Fig-1]. Bipolar montage EEG revealed generalised spike-and-wave discharges with eye closure sensitivity. The epileptiform discharges were consistent with Eyelid Myoclonia with or without Absences (EMA), as seen in JS. (Montage: Bipolar, LFF: 1 Hz, HFF: 15 Hz, Notch: 50 Hz) [Table/Fig-2].

A provisional diagnosis of JS was made, ruling out other differentials such as facial tics, Juvenile Myoclonic Epilepsy (JME), sunflower syndrome, and occipital lobe epilepsy based on clinical history, the absence of motor manifestations, and EEG findings. The child received an initial intravenous loading dose of sodium valproate at 20 mg/kg, followed by oral maintenance therapy at 10 mg/kg twice daily. He has completed one year of maintenance therapy and continues to be reviewed every three months. Based on the neurologist's recommendation, oral valproate therapy will be



[Table/Fig-1]: EEG recording showing generalised high-amplitude epileptiform discharges with polyspike and slow-wave complexes (3-6 Hz) interspersed with theta activity.



[Table/Fig-2]: Bipolar montage EEG revealing generalised spike-and-wave discharges with eye closure sensitivity.

continued for an additional year. The child has remained seizure-free since the initiation of treatment, with no recurrence reported during follow-up.

DISCUSSION

JS is a rare idiopathic generalsed epilepsy characterised by a triad of eyelid myoclonus (with or without absences), photosensitivity, and eye-closure-induced seizures. Typically, onset occurs between 6-8 years of age [1]. The prevalence is reported to be between 2.5% and 2.7% among all epilepsy patients; however, it is likely underdiagnosed due to its subtle clinical presentation and overlap with behavioral or movement disorders.

In JS, light-induced activation of the occipital cortex can trigger epileptic discharges that spread through thalamocortical pathways, leading to generalised or frontocentral spikes. These interactions contribute to the development of myoclonic jerks, spike-and-wave patterns, and absence seizures. The brainstem also plays a role in generating photomyoclonic responses [2].

Smith KM et al., emphasised the diagnostic challenges in JS, where eyelid myoclonia is often mistaken for tics or behavioral issues. Their international consensus highlighted that careful video-EEG monitoring and photic stimulation are essential for diagnosis. In the present case, the EEG confirmed generalised epileptiform discharges with eye closure sensitivity, consistent with features described in the literature [1].

Kuriakose ME et al., and Pozzi C et al., described similar cases of children with frequent blinking episodes who were ultimately diagnosed with JS after detailed EEG evaluations [2,3]. Differentiating JS from other disorders such as facial tics, JME, sunflower syndrome, and occipital lobe epilepsy is vital. Unlike tics, JS is associated with EEG abnormalities and photosensitivity. JME often has a later onset and involves limb myoclonus upon awakening. Sunflower syndrome, although also light-induced, presents with hand-waving and lacks consistent EEG findings characteristic of JS [2]. Sheetal S et al., also reported a child misdiagnosed with a head-nodding tic, which further supports the need for clinician awareness [4].

Pharmacological management includes anti-myoclonic drugs. Valproic acid remains the first-line treatment and has shown efficacy in controlling eyelid myoclonus and absences. However, as Yuan Y et al., highlighted in their report, some cases may exhibit drug resistance and require combination therapy [5]. Levetiracetam is another effective agent, especially in photosensitive epilepsy [2]. Spurgeon AL et al., reported involvement of the PLCB-1 gene in early-onset epilepsies, from birth to encephalopathic epilepsies [6]. Previous literature, such as that by Nilo A et al., stated that intellectual disability and psychological disorders like anxiety, irritability, and psychosis are also associated [7]. The present case contributes to existing literature by reinforcing the need for early identification of JS in pediatric patients with frequent blinking and brief unresponsiveness. Prompt diagnosis facilitates appropriate management and avoids unnecessary labeling as behavioral or psychological disorders.

CONCLUSION(S)

Jeavons Syndrome though uncommon, should be suspected in children with frequent blinking and brief episodes of altered awareness, especially when exacerbated by visual stimuli. Timely diagnosis prevents unnecessary mislabeling as behavioral or tic disorders and ensures better outcomes through early treatment and counseling.

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