

## *Differentiation between Complex Tic and Eyelid Myoclonia with Absences: Pediatric Case Report and Brief Review of the Literature*

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### Abstract

Eyelid Myoclonia with Absences (EMA) can often be clinically mistaken for a tic disorder. This case report describes a 4-year-old female who presented with eye fluttering and blinking accompanied by a hand gesture described as a "brow wipe" that was exacerbated by sunlight, warm temperatures, anxiety, and excitement. She was diagnosed with a transient tic disorder, and aripiprazole and clonazepam treatment partially decreased symptom intensity. She was stable for months, after which symptoms worsened with constant tics while in the sun and seeking light stimuli to induce an episode. Video electroencephalogram confirmed self-induced photosensitive seizures with generalized epileptiform discharges consistent with generalized seizures. The EMA diagnosis was confirmed by these characteristic eye closure-related discharges, and the patient was successfully treated with valproate. We also present a brief review of the literature that underscores the clinical similarities between EMA and tics, as well as the importance of timely implementation of appropriate treatment in EMA.

**Keywords:** Absence Seizures; Eyelid Myoclonia with Absences; Jeavons Syndrome; Pediatric Absence Seizures; Pediatric Seizure Disorders

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### Introduction

Eyelid Myoclonia with Absences (EMA), also known as Jeavons syndrome, is an epileptic syndrome characterized by eyelid myoclonia associated with brief absences and photosensitivity [1, 2]. EMA was originally classified as reflex syndrome of idiopathic generalized epilepsy, but is currently identified as a myoclonic form of epilepsy [3, 4]. Typical age of onset is between 6 and 8 years, and more females are affected than males [5]. The etiology of EMA is thought to be genetic [5-10].

During an episode, a patient with EMA will assume a posture of head slightly bent backwards when he/she is staring at the sun or a source of bright light while shaking a hand in front of their face [11-13]. The patient's eyelids exhibit rhythmic jerks and consciousness is impaired only mildly and briefly following eyelid closure. These absences, which can occur more than three times daily, last approximately three to six seconds in the

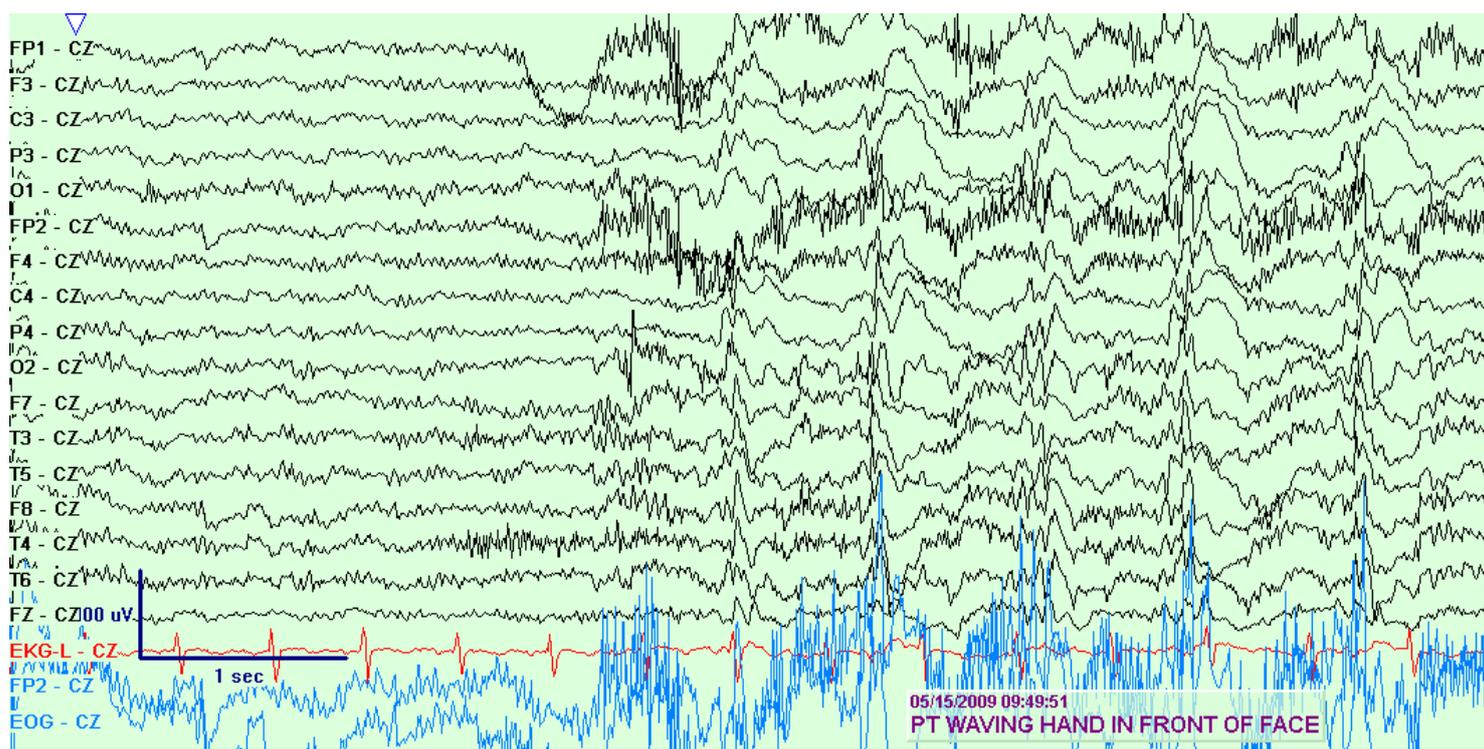
presence of uninterrupted light; this is in contrast with other forms of photosensitive epilepsies that are sensitive to flickering lights. Approximately 50% of patients with EMA will display myoclonic limb jerks and tonic-clonic seizures triggered by sleep deprivation, fatigue, and/or alcohol intake [14]; however, this is more common in adults than children [15, 16]. Photosensitivity often decreases with age and antiepileptic drug (AED) treatment [14].

EMA diagnosis is confirmed through video EEG, which will typically display frequent high-amplitude 3-6 Hz generalized polyspike-slow-wave discharges and therefore rule out non-epileptic paroxysmal eyelid movements [4, 5]. Notably, neuroimaging and clinical examination will be normal in patients with EMA [15]. Reports suggest that EMA is often misdiagnosed as a tic disorder, [17-19] and it is essential that patients, especially school-age females who present with photosensitivity, be evaluated comprehensively to rule out photosensitive epilepsy through video EEG. The goal of this paper is to aid in differential diagnosis by calling attention to the clinical features of EMA that differentiate it from other conditions by presenting a case report of a 4-year-old girl who was initially diagnosed with a tic disorder. Further investigation suggested a diagnosis of EMA, which was subsequently confirmed through video EEG and appropriate treatment was implemented.

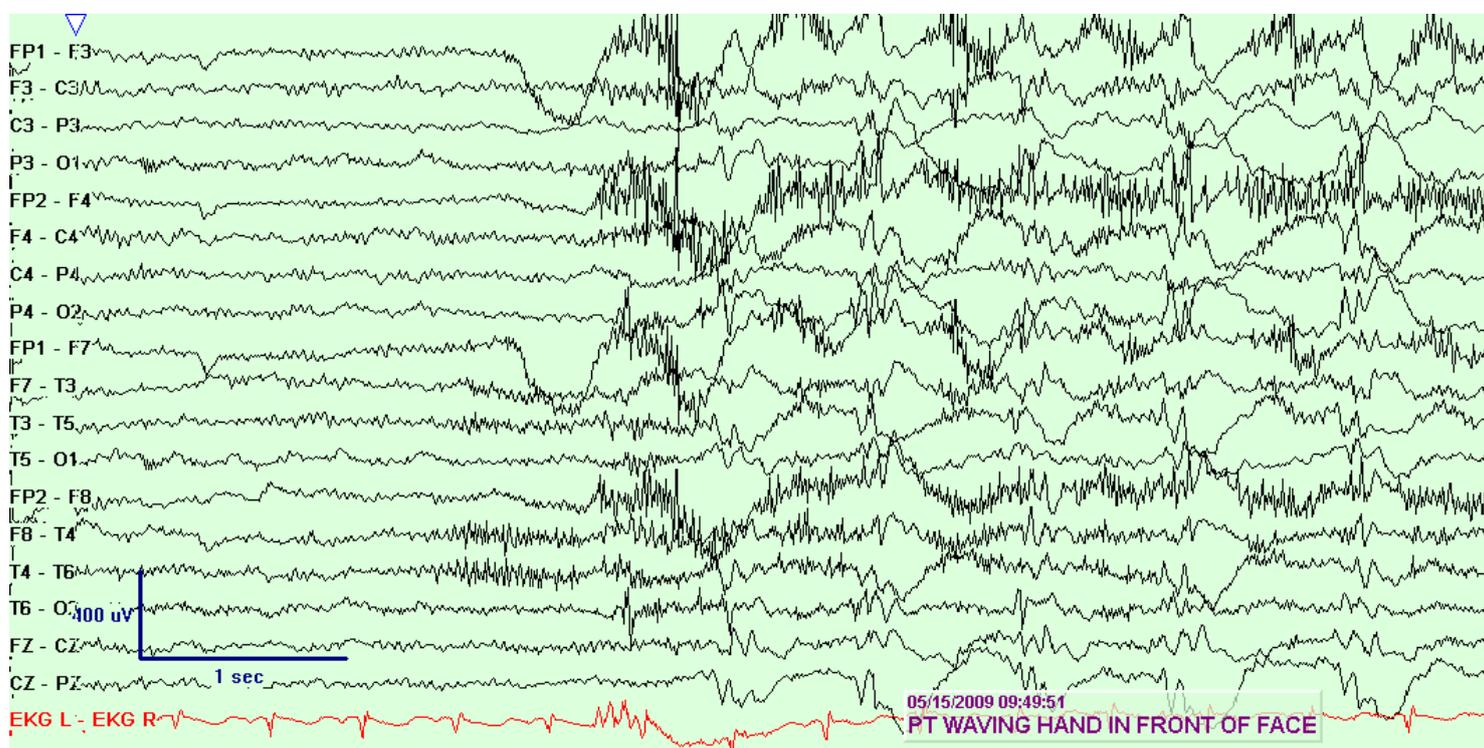
### Case Report

The patient is a previously healthy 4-year-old female with a family history significant for motor tics and autism. She presented with eye fluttering and repetitive blinking that occurred throughout the day and worsened when anxious or exposed to warmer temperatures. The eye fluttering occurred concurrently with a complex movement of a hand gesture described as a brow wipe that became more frequent and intense in the sunlight. Her neurologist considered stereotypic movement disorder and transient tic disorder, but when her episodes continued to worsen, the patient was referred to child

psychiatry. After evaluation, the patient was started on clonazepam 0.25mg one-quarter tablet once a day, and eventually increased to 0.25mg one tablet twice a day. Two months later, aripiprazole 1.5mg daily was added to her medication regimen, as well as behavior intervention. She was stable for six months, after which her behaviors worsened, as she began to seek sunlight to induce the repetitive movements that eventually became nonstop when exposed to the sun. In addition, the patient's social functioning began to suffer, as she displayed constant brow swiping during recreational outdoor time and lost interest in social activities. The possibility of a seizure disorder characterized by the self-induction of repetitive behaviors and brow swiping through seeking sunlight, a condition known as EMA, was then considered, and she was referred to a pediatric neurologist specializing in seizure disorders. Per neurology, findings from the evaluation revealed a normal neurological examination. Video EEG displayed generalized polyspikes and maximal bioccipitally in conjunction with her episodes. The patient's habitual episodes of brow wiping were recorded. Clinically, the patient would stare at the light and move her left hand from the thumb to the pinky across her face. The spike was usually at the time her pinky cleared the left eye. On the EEG, generalized spikes at 1.5 and 2 Hz were present. The EEG showed a normal background. Photoc stimulation was performed without abnormality. The patient was then allowed to do her "tic movements;" these distinctly showed reactivity in the background with bilateral parasagittal spike and wave discharges at approximately 1 Hz. Spikes were not present in the absence of this "brow wiping" movement, and were more pronounced when the patient was next to a window and performed the movement looking into the sunlight, suggesting features of self-induced photosensitive epilepsy with generalized epileptiform discharges. The patient was subsequently treated with lamotrigine and levetiracetam without symptom relief. However, successful treatment with valproate followed and patient was doing well on follow-up.



EEG before and during typical Eye Brow wiping (Referential montage)



Same EEG epoch with bipolar Montage

## Discussion

This case demonstrates a challenge in appropriate and timely diagnosis of EMA in a child. The reasons for misdiagnosing EMA for tic disorder stems from various similarities between EMA and tic disorders. For example, patients with EMA often report inducing seizures by waving their fingers within their line of vision or repetitively blinking their eyes, [20, 21] which are often signs that are mistaken for tics. Notably, the movements observed in tic disorders are often less rhythmic and do not involve tonic muscle contractions. An urge towards sunlight to induce tics has been reported in cases of Tourette's syndrome [22]. Also similar to tic patients, pediatric EMA patients report premonitory sensations and sense of relief after the absence [23]. The compulsive need to induce a seizure may be analogous to the compulsory tic behavior in patients with tics [23]. This type of presentation shares overlap with stereotypic movement disorder, in that these complex repetitive movements can be triggered by an environmental event. Despite similarities in clinical presentation, a patient with EMA will have EEG characteristic abnormalities, unlike a patient with tics or stereotypies.

EMA treatment can also present a clinical challenge. The effect of uncontrolled seizures on neurocognitive development and the potential adverse effects of appropriate pharmacotherapy on a child's physical, emotional, and intellectual well-being will need to be considered. A case series has described patients with EMA who did not respond to AEDs developing EMA-associated intellectual disability as a consequence of uncontrollable seizures [9]. In contrast, the patients who did respond to AEDs did not display impairment neurological or executive function, as their seizures were controlled. Specifically, absences have been successfully controlled with valproic acid, lamotrigine, levetiracetam, ethosuximide, and clonazepam [14, 24]. However, ethosuximide and lamotrigine have demonstrated some worsening of myoclonic jerks, and ethosuximide, as well as clonazepam to a lesser extent, has been known to exacerbate generalized tonic-clonic seizures [14]. As well as exhibiting benefit in absences, valproic acid has demonstrated efficacy in

controlling myoclonic jerks and generalized tonic-clonic seizures, [14] as was the case in our patient.

EMA diagnosis is confirmed when the characteristic seizures, photosensitivity, and a video EEG demonstrating eye closure-related EEG discharges are present [25]. Due to similar clinical presentations, EMA is often misdiagnosed as tics, [17, 18] and reassessment of a patient with tic-like movements who also presents with photosensitivity is advisable. Child psychiatrists and neurologists should be aware of EMA and appropriately differentiate it from non-epileptic conditions, such as tics, stereotypies, and exaggerated normal eye movements. Making the timely diagnosis could spare the patient from inappropriate therapies and potential deterioration in emotional and intellectual function if seizures go untreated.

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