Epileptic Disorders

Crossing the lines between epilepsy syndromes: a myoclonic epilepsy variant with prominent eyelid myoclonia and atonic components

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Epilepsy syndrome diagnosis

- Effective classification of epileptic seizures and syndromes is essential for appropriate management and prognosis.

- **Diagnostic challenge:**

  Patients with unusual manifestations whose electroclinical features cannot fit neatly into a recognized epilepsy syndrome or cases where epilepsy syndromes appear to overlap.

ILAE

Eyelid myoclonia with or without absences

- Phenotypic heterogeneity has been documented in the literature in several cases marked by the presence of Eyelid Myoclonia with or without Absences (EMA- EM)

- **Jeavons syndrome** hallmark:
  - (i) eyelid myoclonia with or without absences
  - (ii) eye closure-induced seizures, EEG paroxysms, or both
  - (iii) photosensitivity

Case presentations

- We describe two female paediatric patients who presented with EMA but demonstrated clear atypical features that cross the lines between epilepsy syndromes, such as prominent myoclonic seizures, atonic components on video-EEG polygraphy, and cognitive impairment.

- Photosensitivity can be suppressed by antiepileptic medication and may diminish with age, as demonstrated in the case of the second patient.

- Of note, the patients were not on any medications which could have induced these symptoms.

Case 1

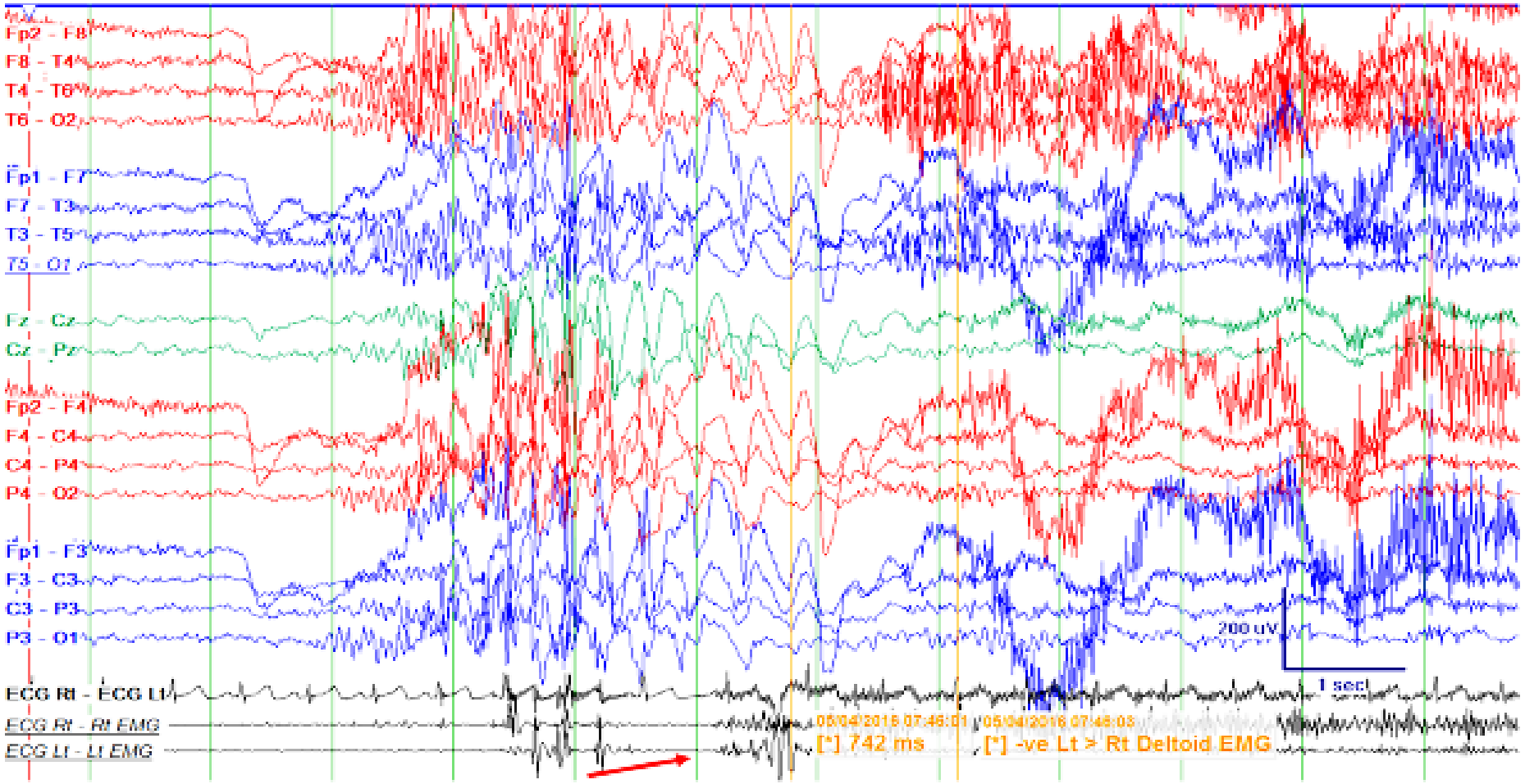

Figure 1: Ictal EEG with polygraphy showing atonia on the deltoid EMG; left > right (duration: 742 msec).

Case 2 please include arrow showing atonia

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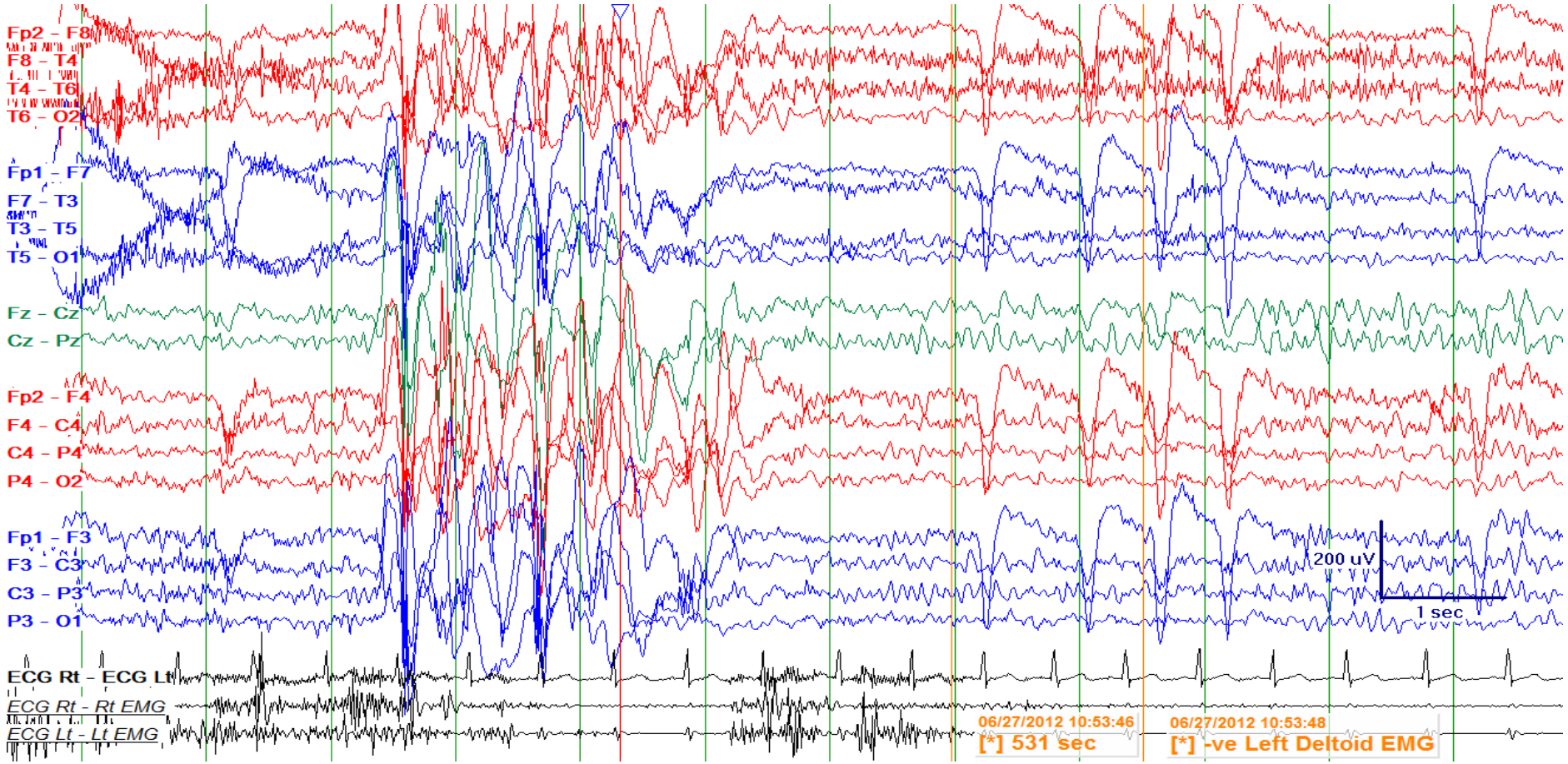

Figure 2: Ictal EEG with polygraphy showing atonia on the left deltoid EMG (duration: 531 msec).

Discussion

- A few reports in the literature have described similar cases with unusual presentation of Eyelid Myoclonias with Absences (EMA) in the clinical course of childhood epilepsy, suggesting that their occurrence alone is not sufficient to characterize a definite epilepsy syndrome.
- Either a diagnosis of a Jeavons syndrome variant was proposed or EMA was considered an electroclinical feature of a distinct genetic generalised epilepsy syndrome.
- Our description of atonic components enlarges the spectrum of seizure types that can be associated with eyelid myoclonia with and without absences, adding to the variability of “overlap” phenotypes.